Exploring the application of Psychiatric Genetic Counselling within the UK

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Abstract

Substantial progress is being made in molecular psychiatry. Psychiatric genetic counselling (PGC), which may address *how* our knowledge from genetic studies is delivered to patients, is likely to become more routinely available as our aetiological understanding of psychiatric illness increases. The present study explores, using mixed-methods, potential consumers' understanding and beliefs about the aetiology and pathology of psychiatric illness; their awareness of genetic counselling and perceptions of the purpose of the service, and their attitudes towards receiving PGC. Results indicate that there is an interest and keenness in receiving PGC, however also highlights potential issues that may arise through the provision of genetic counselling for psychiatric conditions, including that awareness of the service is low and misconceptions exist regarding its purpose; as well as concerns amongst respondents that it may cause psychological distress and also that it may be associated with eugenic-type values and practices which raises considerations of an ethical nature. The study overall highlights a need for further exploration of the findings presented and their wider implications in regards to future efforts to implement PGC within the UK.

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List of abbreviations

ASHG – American Society of Human Genetics

BPD – Bipolar disorder

C – Concern (variable)

CACNA1C - Voltage dependent L type calcium channel, alpha 1C subunit

EA – Attribution of mental illness to environmental factors

EAC – Certainty regarding attribution of mental illness to environmental factors

GA – Attribution of mental illness to genetic factors

GAC – Certainty regarding attribution of mental illness to genetic factors

GC – Genetic counselling

GCOS - The Genetic Counselling Outcomes Scale

GWAS – Genome-Wide Association Studies

MDD – Major depressive disorder

MHC – Major histocompatibility complex

NSGC – National Society of Genetic Counsellors

OCD – Obsessive compulsive disorder

PGC – Psychiatric genetic counselling

PROM – Patient reported outcome measure

PTSD – Post traumatic Stress disorder

SCZ – Schizophrenia

SCZAD – Schizoaffective disorder

1. Psychiatric Genetic Counselling within the UK – An Introduction

Following advances in psychiatric genetics we are now starting to elucidate the genetic architecture of psychiatric disorders including schizophrenia (SCZ), bipolar disorder (BPD) and major depressive disorder (MDD). Researchers and clinicians now face the challenge of how to translate genetic findings to improve the lives of patients and families. The potential genetic counselling (GC) holds in addressing this has been discussed for many years; however there is a lack of consensus on the best way forward in delivering this service. Exploring future UK service user's beliefs and perceptions about aetiology and familial risk, and their attitudes regarding the service, may be helpful in providing insight into how this may be best achieved to guide future delivery of psychiatric genetic counselling (PGC) within the UK.

This research study uses a mixed-methods approach to examine beliefs about aetiology and familial risk and awareness and perceptions of the UK public with regard to PGC, to explore its implementation within the UK specifically. It will focus on UK individuals only as proper consideration of individuals from other countries and thus healthcare systems is beyond the scope of this research project.

This chapter begins by exploring the background literature supporting the rationale behind PGC, followed by a review of the current literature specifically regarding PGC. It concludes with the aims and objectives that this study intends to address.

1.1 Defining mental illness

Mental disorders are extremely common across the world (Ormel et al. 1994, Steel et al. 2014). There are many different types of mental disorders with different presentations (Insel et al. 2010, WHO 2014). The World Health Organization's International Classification of Diseases (ICD) is the official world classification system for psychiatric disorders (WHO 1993, WHO 2010, Tyrer 2014), and is the system used in the UK for diagnosis. ICD-10 groups mental disorders into blocks on the basis of a common aetiology or similar presentation of symptoms (American Psychiatric Association 2000, WHO 2010, see figure 1).

In comparison, the Diagnostic and Statistical Manual of Mental Disorders (DSM) is the standard classification of mental disorders used by mental health professionals in the United States, however the main groups of psychiatric disorder are diagnosed similarly by the two classification systems (Tyrer 2014).

This literature review will focus on mood disorders and psychoses as these account for a substantial proportion of psychiatric disorders globally (Wittchen and Jacobi 2005, Wittchen et al. 2011).

(Fig. 1)

Chapter V
Mental and behavioural disorders
(F00-F99)

Incl.: disorders of psychological development

Excl.: symptoms, signs and abnormal clinical and laboratory findings, not elsewhere classified (R00-R99)

This chapter contains the following blocks:

F00-F09	Organic, including symptomatic, mental disorders
F10-F19	Mental and behavioural disorders due to psychoactive substance use
F20-F29	Schizophrenia, schizotypal and delusional disorders
F30-F39	Mood [affective] disorders
F40-F48	Neurotic, stress-related and somatoform disorders
F50-F59	Behavioural syndromes associated with physiological disturbances and physical factors
F60-F69	Disorders of adult personality and behaviour
F70-F79	Mental retardation
F80-F89	Disorders of psychological development
F90-F98	Behavioural and emotional disorders with onset usually occurring in childhood and adolescence
F99-F99	Unspecified mental disorder

Asterisk categories for this chapter are provided as follows:

F00*	Dementia in Alzheimer disease
F02*	Dementia in other diseases classified elsewhere

Figure 1: ICD-10 classification of mental and behavioural disorders

(Figure from World Health Organization 2015)

Disorders that are symptomatically similar are classified into blocks. The ICD-10 is the standard classification of mental disorders used by clinicians in the UK. This review focuses on psychoses (F20-F29) and mood disorders (F30-F39).

1.1.2 Mood disorders

According to ICD-10 ‘mood disorders’ encompasses characteristics affecting either depression or elation (Arnow et al. 2015) and is usually associated with a secondary change in the patient’s overall physical activity (Emerson and Williams 2015).This includes bipolar affective disorder, recurrent depressive disorder, mania, and persistent mood disorders (WHO 2015). Mood disorders are often recurrent, with individual episodes typically related to stressful events or periods (WHO 2015).

Depressive disorder, recurrent (Major depressive disorder)

Major depressive disorder (MDD) is the most common mood disorder and is characterised by persistent sad or low mood, and/or anhedonia (NICE 2011). Other symptoms include fatigue, insomnia or hypersomnia, reduction in energy and inability to concentrate (Kendler and Gardener 1998, Arnow et al. 2015).

MDD can include recurrent episodes of depressive reaction, recurrent episodes of major depression, and seasonal depressive disorder (WHO 2015). There is no history of mania, defined as independent episodes of mood elation and increase in energy, although the individual may experience brief episodes of mild mood elation and overactivity following a depressive episode; if the individual experiences an episode of mania, the ICD-10 states the diagnosis should be changed to bipolar disorder (BPD) (WHO 2015, see figure 2).

(Fig. 2)

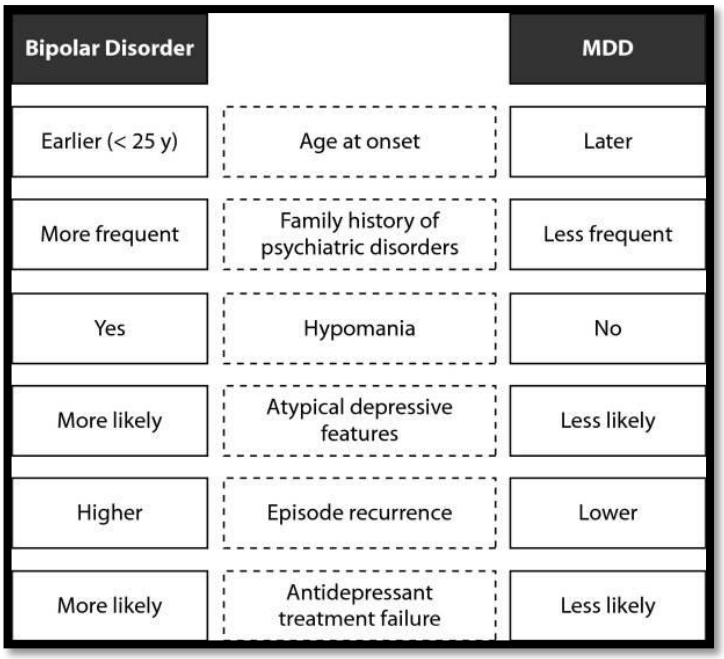


Figure 2: Key Differentiating symptoms between MDD and depressive symptoms in BPD.

(Figure from Culpepper 2014)

The overlap between clinical symptoms can be problematic for diagnosis and can result in deterioration of symptoms and increase treatment costs.

Bipolar disorder (BPD)

BPD is episodic disorder characterised by recurrent, periodic episodes of depression and mania (Cosgrove and Suppes 2013, Culpepper 2014). Subtypes of BPD include BPD type I (depressive and manic episodes); BPD type II (depressive and hypomanic episodes); and cyclothymic disorder (hypomanic and depressive symptoms that do not meet criteria for depressive episodes) (Phillips and Kupfer 2013, Culpepper 2014). Depressive symptoms are usually more common and longer-lasting than symptoms of elation and contribute to most overall morbidity, predominantly due to suicidality (Anderson 2012). Additionally, around 50% of manic episodes also contain psychotic elements (Cosgrove and Suppes 2014, see figure 3).

The diverse presentation of symptoms in BPD can make diagnosis a challenge and misdiagnosis - often of MDD - is a clinical problem (see figure 2) that often results in inappropriate treatment, typically involving overuse of antidepressants and underuse of more effective treatment options, ultimately resulting in deterioration of symptoms for patients and significantly increasing direct treatment costs (Matza et al. 2005, Culpepper 2014).

1.1.3 Psychosis

The term ‘psychosis’ describes a group of disorders in which a person’s mood, thoughts, perceptions and behaviour are significantly altered (Yung and McGorry 1996, Austin and Honer 2008, NICE 2014, see figure 3). Psychosis can include schizophrenia (SCZ), schizoaffective disorder (SCZAD) and severe depression, and is also often experienced during the manic phase of BPD (Mental Health Foundation 2007, Cosgrove and Suppes 2013).

The overlap in symptomology between diagnostic boundaries can be clinically problematic (Malhi et al. 2008, Cosgrove and Suppes 2013, Wilson et al. 2014, see figure 3) as evidence-based outcomes have shown that early and appropriate treatment interventions can be critical in the effective management of psychotic disorders (Birchwood et al. 1998, McGorry 2005), including reducing suicide rates, shorter duration of hospitalisations, and longer employment periods (Chan et al. 2015).

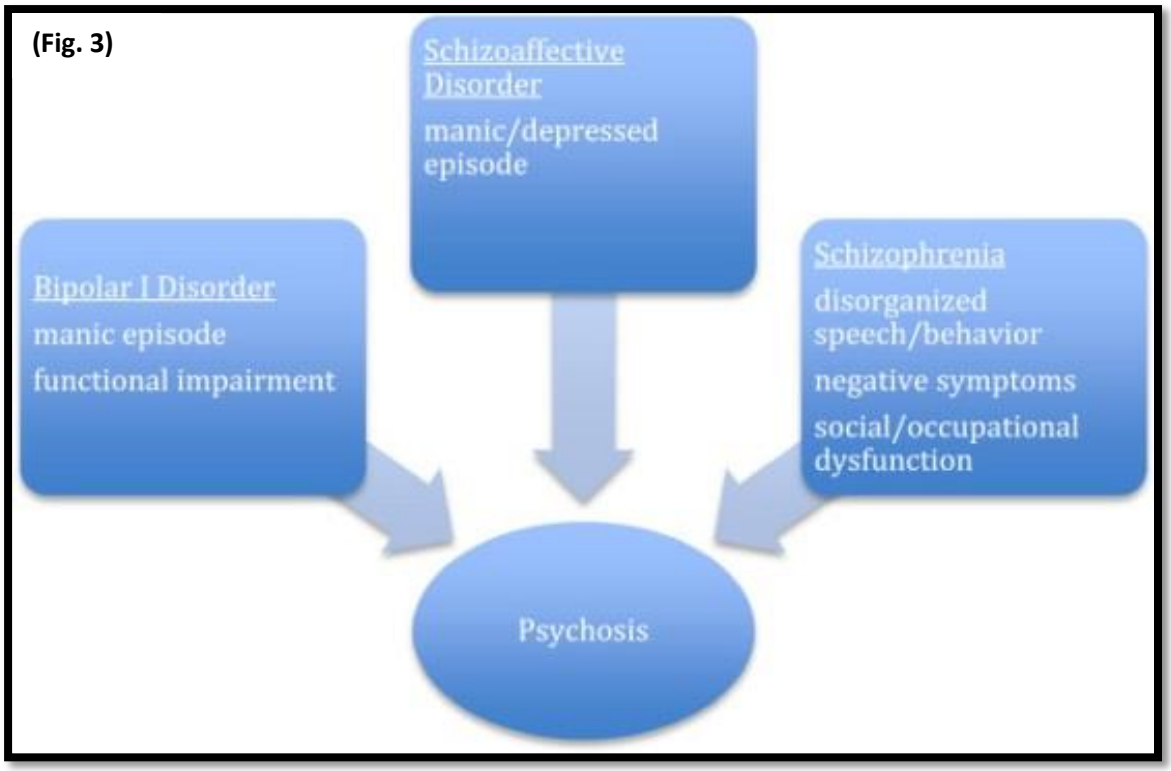


Figure 3: Typical features of BPD, SCZ and SCZAD.

(Figure from Cosgrove and Suppes 2013, p.2)

Whilst hallucinations and delusions are typically considered the hallmark symptom of SCZ and mood fluctuations of BPD; psychosis may be present in both. SCZAD, has been proposed to represent a mid-point on the phenotypic spectrum between BPD and SCZ. In clinical practice, symptomatic overlap between diagnostic boundaries presents clinical problems.

Psychosis – symptoms

The two major symptoms of psychosis are hallucinations and delusions (see figures 4 and 5, following pages); other common pathophysiological phenomena in psychosis include thought disorder, negative symptoms and cognitive impairment. Symptoms of psychosis vary in nature and severity across patients and the course of the illness; and each affected individual will develop their own unique combination of symptoms and experiences that often change over course of the illness and are dependent on their personal circumstances and life experiences (Tandon et al. 2009, Schmitt et al. 2014, NICE 2014).

Definitions of hallucinations and delusions in psychosis, along with examples, are provided in the figures in the following pages.

Figure 4: Hallucinations in psychosis

Definitions in the literature

- "A sensory perception that has the compelling sense of reality of a true perception but that occurs without external stimulation of the relevant sensory organ" (American Psychiatric Association 1994, p.767)
- "Perceptual experiences not shared by others" (Mueser and McGurk 2004, p.2064)
- "The perception of an object or event (in any of the 5 senses) in the absence of an external stimuli" (Teeple et al. 2009, p.26)
- "Percepts, experienced by a waking individual, in the absence of an appropriate stimulus from the extracorporeal world" (Blom 2015, p.433)

Hallucinations in psychosis

- Hallucinations may affect any of the 5 senses (auditory, visual, olfactory, gustatory or tactile) (Mueser and McGurk 2004)
- Auditory hallucinations are the most common in psychotic illness (Mueser and McGurk 2004, Tandon et al. 2009)
- Auditory hallucinations typically involves voices conversing amongst themselves (Tandon et al. 2009, p.4), i.e. ("a running commentary") (World Health Organization 1993 chapter F25; or accusatory or threatening voices speaking directly to the individual (Tandon et al. 2009, p.4)

Figure 5: Delusions in psychosis

Definitions in the literature

- "Persistent delusions… that are culturally inappropriate and completely impossible" (World Health Organization 1993, chapter F20)
- "Fixed, false beliefs that are not shared by an individual's cultural/religious group" (Austin 2005, p.329)
- "Markedly unusual or bizarre ideas" (NICE 2014, p.102)

Common types of delusions in psychosis

- **Grandiose:**
  e.g. "Thinking that one has superhuman powers… …(believing that one is) able to control the weather, or… in communication with aliens from another world (World Health Organization 1993 chapter F20);
  e.g. Thinking one is Jesus Christ" (Mueser and McGurk 2004, p.2064);
- **Persecutory (paranoid):** "Abnormal attention to threat related-stimuli" (Bentall et al. 1994, p.331)
  e.g. "An evil spirit is out to kill me" (Freeman and Garety 2014, p. 1179).
- **Erotic:**
  e.g. The false belief that another person (often of higher status, a stranger, or somebody famous) has fallen in love with the affected individual and is making "amorous advances towards him/her" (Kennedy et al. 2002, p.1)
- **Control:** *The belief that others can interfere with the affected individuals thoughts and/or actions (Mueser and McGurk 2004, p.2064)*
- **Somatic:** involves an "irrational preoccupation" with one's health or body (Kamara et al. 2009, p.1-2), however this belief is "false" or untrue, and that there is sufficient contradictory evidence to prove as such (Maher and Ross 1984, p.383)

Schizophrenia

Schizophrenia (SCZ) affects around 1% of the population (Mueser and McGurk 2004, National Institute for Mental Health 2015). In addition to positive symptoms (psychosis) affected individuals experience negative symptoms including blunted affect (lack of emotional reactivity), alogia (lack of speech) and anhedonia (lacked capacity to experience pleasure) (Rector et al. 2005, WHO 2015, Tsapakis et al. 2015); and cognitive impairments including deficits in working memory, attention and processing (Mikell et al. 2009, Tsapakis et al. 2015, see figure 6).

SCZ is the 'flagship' disorder of the Psychiatric GWAS *(Genome Wide Association Studies)* Consortium and resultantly its genetic dissection is more advanced than other psychiatric disorders (Gratten et al. 2014), which have much smaller sample sizes. This review therefore gives particular attention to the aetiology of SCZ, as described in section 1.3.1

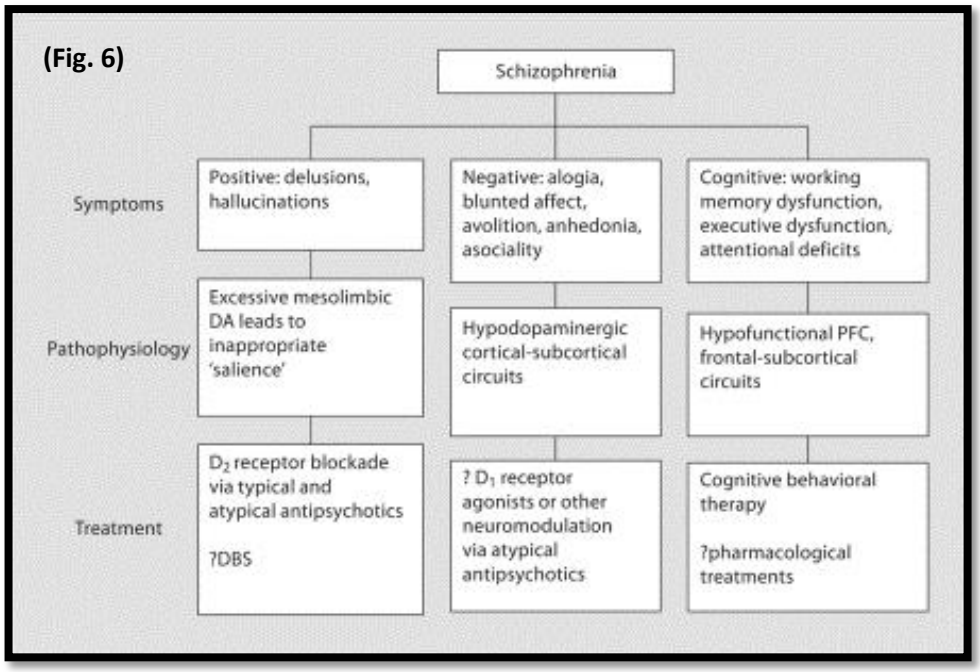


Figure. 6: The three major symptom domains of SCZ

(Adapted from Mikell et al. 2009, p.257)

The three major symptom domains of SCZ are positive symptoms, negative symptoms, and cognitive impairments. Hypotheses regarding proposed pathophysiology underlying impairment have guided suggested pharmacological treatment pathways, as shown on the diagram. The efficacy of pharmacological treatments in psychiatry are poor and very few drugs of proven efficacy have been developed; the therapeutic stagnation is largely due to limited aetiological understanding.

1.2 Significance of mental disorders

1.2.1 Global and national Significance

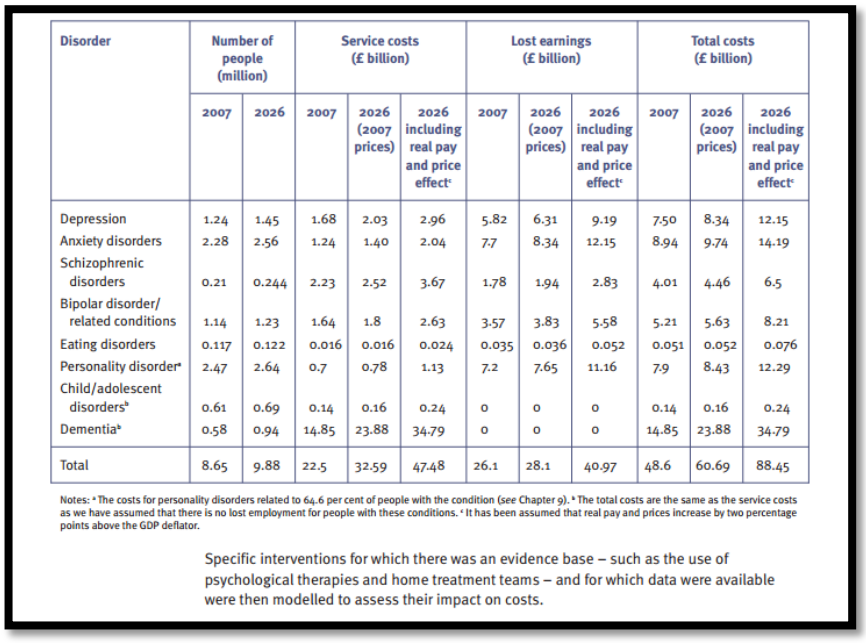
A WHO report in 2001 estimated that 450 million people worldwide suffer from mental health problems (WHO 2001, Mental Health Foundation 2007) placing mental disorders amongst the leading causes of illness and disability globally. This is now believed to have reached almost 500 million (Tawar et al. 2014) and is projected to rise exponentially as the population continues to grow; life expectancy increases; as well as additional contributory socioeconomic factors including increased immigration and warfare (Mawani 2014) and growing global poverty (Tilleczek et al. 2014). These figures are likely to be underestimations due to many affected individuals with mental health problems not seeking professional help and/or not receiving a proper psychiatric diagnosis.

The growing global burden of mental health disorders has major social, economic and human rights consequences across the world (WHO 2014). All mental disorders can be chronic, lifelong conditions that can cause significant long-term functional impairment and disability (NICE 2011) meaning that, in addition to medical treatments (e.g. medication, therapy), affected individuals with mental illness often require social care and support within the community such as finding housing and employment and accessing education (WHO 2014). It has thus been urged that mental disorders should be made a public health priority (Whiteford et al. 2013); this could have both quality of life and economic benefits for hundreds of thousands, potentially millions, of people world-wide (Collins et al. 2011).

In Britain specifically, one in four adults will experience mental health problems in any year (Singleton et al. 2001, Mental Health Foundation 2014). Direct costs of mental health in England are over £22.5 billion a year and are projected to continue growing as prevalence increases (McCrone et al. 2008, see table 1). Despite this, mental health services in the UK are facing increasing funding cuts (see figure 7). This period of austerity makes efficiency in mental health services imperative; evidence-based prevention and early intervention and treatment for mental disorders are becoming increasingly important in ensuring maximum efficacy of mental health services in the UK, and overall optimal patient outcomes (McDaid and Knapp 2010).

Table 1 Number of UK affected individuals with specific disorders and current and projected costs

(From McCrone et al. 2008, p.18)



Disorder	Number of people (million)		Service costs (£ billion)			Lost earnings (£ billion)			Total costs (£ billion)		
	2007	2026	2007	2026 (2007 prices)	2026 including real pay and price effect[c]	2007	2026 (2007 prices)	2026 including real pay and price effect[c]	2007	2026 (2007 prices)	2026 including real pay and price effect[c]
Depression	1.24	1.45	1.68	2.03	2.96	5.82	6.31	9.19	7.50	8.34	12.15
Anxiety disorders	2.28	2.56	1.24	1.40	2.04	7.7	8.34	12.15	8.94	9.74	14.19
Schizophrenic disorders	0.21	0.244	2.23	2.52	3.67	1.78	1.94	2.83	4.01	4.46	6.5
Bipolar disorder/ related conditions	1.14	1.23	1.64	1.8	2.63	3.57	3.83	5.58	5.21	5.63	8.21
Eating disorders	0.117	0.122	0.016	0.016	0.024	0.035	0.036	0.052	0.051	0.052	0.076
Personality disorder[a]	2.47	2.64	0.7	0.78	1.13	7.2	7.65	11.16	7.9	8.43	12.29
Child/adolescent disorders[b]	0.61	0.69	0.14	0.16	0.24	0	0	0	0.14	0.16	0.24
Dementia[b]	0.58	0.94	14.85	23.88	34.79	0	0	0	14.85	23.88	34.79
Total	8.65	9.88	22.5	32.59	47.48	26.1	28.1	40.97	48.6	60.69	88.45

Notes: [a] The costs for personality disorders related to 64.6 per cent of people with the condition (see Chapter 9). [b] The total costs are the same as the service costs as we have assumed that there is no lost employment for people with these conditions. [c] It has been assumed that real pay and prices increase by two percentage points above the GDP deflator.

Specific interventions for which there was an evidence base – such as the use of psychological therapies and home treatment teams – and for which data were available were then modelled to assess their impact on costs.

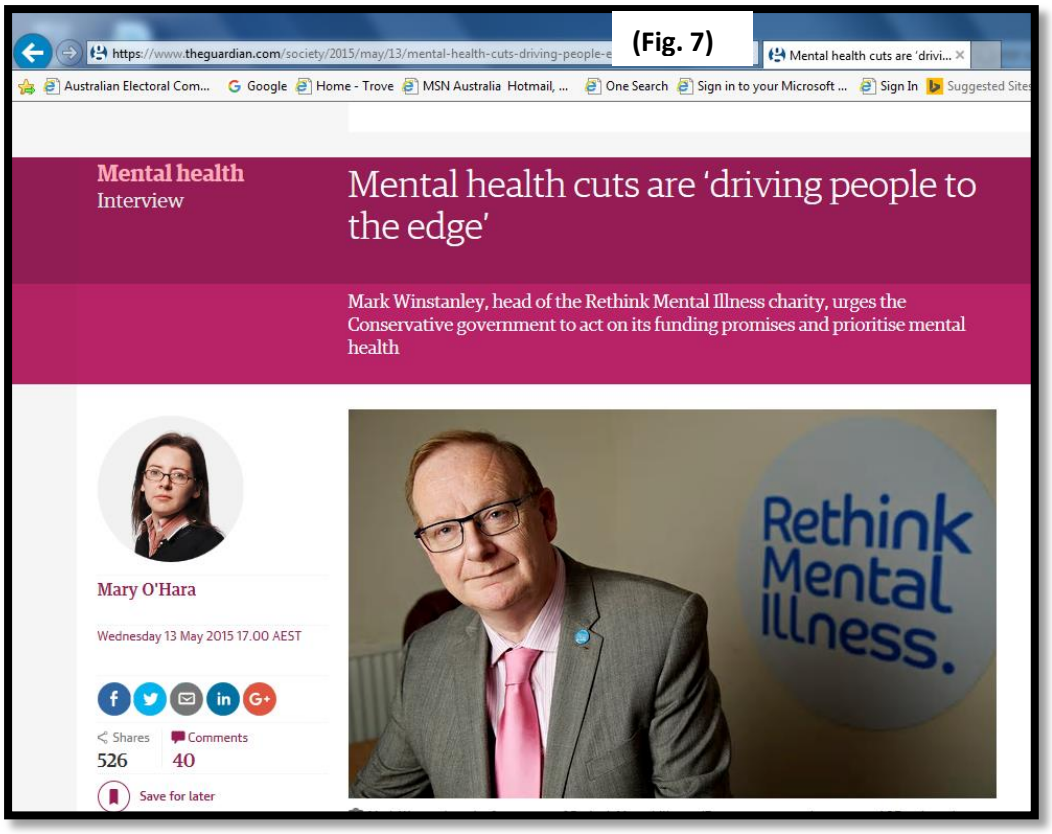


Figure 7: Guardian article featuring Mark Winstanley

(Source: O'Hara 2015).

Chief executive of Rethink mental illness charity, warning of the impact of spending cuts in the UK. Despite the growing prevalence of psychiatric conditions, services are facing huge cuts with major implications on care and support.

1.2.2 Morbidity and mortality in mental disorders

Individuals with mental health problems have shorter life expectancy, especially those with more serious psychiatric disorders (Dickey et al. 2004). Individuals with SCZ, for example, have a lifetime expectancy that is 20% shorter than that of the general population (Newman and Bland 1991, Hennekens et al. 2005, Munitz 2010).

The mortality gap is largely explained by natural causes owing to morbidity with other, non-psychiatric medical diseases (Gardner-Sood et al. 2015), including higher prevalence of metabolic diseases such as hypertension, diabetes and hyperglycaemia (Koran et al. 1989, Briskman et al. 2012, Gardner-Sood et al. 2015); HIV and infectious hepatitis (Rosenburg et al. 2001, Goff 2005) and poorer dental health (Mirza et al. 2001, Kisely et al. 2015). Under-diagnosis and under-treatment of hospitalised psychiatric patients in comparison to non-psychiatric patients (Briskman et al. 2012); less effective self-care; adverse health behaviours such as smoking (Naylor et al. 2012) or alcohol abuse, potentially to self-medicate symptoms (Duffy et al. 2007); and poorer quality of life (Naylor et al. 2012) have been proposed as confounding factors. Additionally, cardiac distress in psychosis is also believed to act as a specific potential cardiovascular risk factor (Rasul et al. 2005).

Furthermore, psychiatric comorbidity, whereby an individual meets the diagnostic criteria for two or more psychiatric conditions, is also common with studies reporting that over 50% of affected individuals diagnosed with a common mental disorder also meet diagnostic criteria for at least one more psychiatric condition (Andrews et al. 2002, Maj 2005, NHS information Centre for Health and Social Care 2009). Psychiatric comorbidity is associated with increased severity of symptoms, longer duration of illness, greater functional disability, poor adherence to self-care regimens and increased use of public health services (NHS information Centre for Health and Social Care 2009, Katon et al. 2011).

Higher rates of suicide and suicidal tendencies also account for the excess mortality in affected individuals with psychiatric disorders. Suicide is strongly associated with psychiatric conditions (Cheng 1995, Duffy 2014). For example, for affected individuals with SCZ there is a lifetime suicide risk exceeding 10% (Becker 1988, Inskip et al. 1998, Enger et al. 2004, Giusti-Rodríguez and Sullivan 2013); and suicide is the leading cause of premature death (Kwon et al. 2013). Additionally of all psychiatric disorders, mood disorders account for the greatest proportion of suicides (Pritchard et al. 2005).

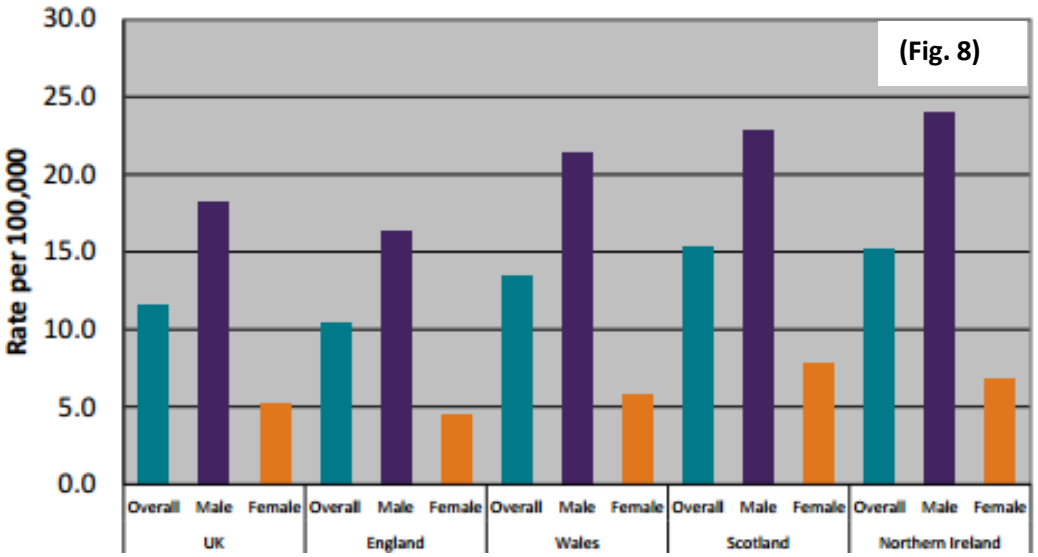


Figure 8 : Suicide rates per 100,000 in the UK in 2012.

(Figure from Scowcroft 2014, p.8).

Suicide rates per 100,000 in the UK in 2012. British men are three times higher to die by suicide than British women. The so-called gender paradox in suicide is largely attributed to social factors, especially that pressures to fit the traditional image of male masculinity may promote maladaptive coping strategies including substance abuse, emotional inexpressiveness and increased reluctance to seek medical help.

In the UK specifically, in 2012, more than 5,900 people died by suicide, and British men are over three times more likely to die by suicide than British women (Scowcroft 2014, see figure 8). The UK also has one of the highest rates of self-harm in Europe, at 400 per 100,000 (Singleton et al. 2001, Mental Health Foundation 2007). Furthermore, prevalence of suicidal phenomena is actually much higher than indicated from such data, as the number of affected individuals with psychiatric disorders that *experience* suicidal thoughts, and even *attempt* suicide, will be far greater than those who *successfully commit* suicide.

The high morbidity and mortality rates seen in psychiatric patients thus emphasise the urgent need to improve both psychological and physical care provision to affected individuals with psychiatric conditions.

1.2.3 Psychosocial aspects of mental disorders – stigma

Stigma is defined as a mark, label or attribute that usually links a person to undesirable characteristic (Goffman 1963, Jones et al. 1984, Link and Phelan 2001, Thornicroft et al. 2007) and sets a person apart from the rest of society (Austin and Honer 2005, Steel et al. 2014). Affected individuals with mental illness are amongst the most stigmatised in society; indeed the association between stigma and psychiatric illness has been explored many groups (see e.g. Phelan et al. 1998, Thara and Srinivasan 2000, Corrigan et al. 2001, Byrne 2001, Link et al. 2004, Steel et al. 2014).

Typical negative connotations associated with mental illness that induce stigmatising attitudes and prejudice include beliefs and assumptions that affected individuals can be violent, weak, lacking intelligence and dirty (Olmsted and Durham 1976, Phelan 2002, Schulze and Angermeyer 2003, Steel et al. 2014).

Stigma can manifest as open-discrimination in areas such as employment and housing (Corrigan et al. 2003, Thornicroft et al. 2007, Brohan et al. 2010) to more subtle expressions such as the negative portrayal of characters with mental illness in television series (Phelan 2002). Stigma can also be a major barrier to help-seeking behaviours in mental health, such as medication adherence, help-seeking at time of onset, and following medical advice, which results in deterioration of symptoms and poorer management of the condition (Schulze and Angermeyer 2003, Steel et al. 2014, Mental Health Commission of Canada 2015).

Stigma thus has a direct impact on an individual's self-esteem and psychological well-being (Link et al. 1991, Link 2001, Link and Phelan 2001, Austin and Honer 2005, Steel et al. 2014, Government of Western Australia Mental Health Commission 2014); their economic and physical well-being through reducing life and employment opportunities and health-related quality of life (Schulze and Angermeyer 2003, Corrigan et al. 2003). Indeed, for many affected individuals the effects of stigma can be more debilitating than the mental illness itself (Schulze and Angermeyer 2003, Thornicroft et al. 2007).

Equally family members of affected individuals with mental illness can also be affected by stigma (Goffman 1963, Phelan 2002, Corrigan and Miller 2004). Common examples include family members being blamed for causing their relative's illness or relapse (Phelan 2002, Corrigan and Miller 2004, Larson and Corrigan 2008, Girma et al. 2014); being rejected or avoided out of fear of contamination of the mental illness (Corrigan and Miller 2004); or having their own mental health questioned (Phelan 2002, Austin and Honer 2005). These prejudices can result in feelings of shame, guilt, fear and isolation from society amongst family members (Thara and Srinivasan 2000, Austin and Honer 2005), may reduce the support network, make them less able to be proactive in their loved one's care (Major and O'Brien 2005); and may cause psychological distress (Ostman and Kjellin 2002).

1.2.4 Psychosocial aspects of mental illness - Shame and guilt

Although used synonymously, shame is defined as a negative judgement of oneself in response to failing to meet personal or social standards; whilst feelings of guilt result from negative evaluations of a specific behaviour (Averill et al. 2002).

Affected individuals affected by mental illness and their relatives often experience profound feelings of guilt and shame (Miller and Mason 2005, Austin and Honer 2005); this has been reported across different cultures and nations including the UK (Gilbert et al. 1994), Americans (Morrison 1985), Indians (Thara and Srinivasan 2000), American Asians (Chow et al. 2003) and Hispanic Americans (Strug and Mason 2002). These feelings can be intense and remain even after successful treatment or management of the symptoms of the mental illness (Miller and Mason 2005).

Although multiple factors can contribute to feelings of guilt and shame, it is well established that oversimplified ideas and misconceptions about the causes of mental illness, often when there is a lack comprehensive understanding and in place affected individuals develop their own explanations (Meiser et al. 2005, Austin and Honer 2005, Austin and Honer 2007, Jaremo et al. 2011, Inglis et al. 2014, Costain et al. 2014a, Costain et al. 2014b), commonly induce feelings these feelings amongst families. This may include extensive attribution of the mental illness to environmental factors and life experiences such as events that occurred during childhood that the mental illness is later attributed to (Meiser et al. 2005, Austin and Honer 2005, Hill and Sahaar 2006, Finn and Smoller 2006, Austin and Honer 2007, Peay et al. 2008, Inglis et al. 2014); and also oversimplified ideas about genetic contributions to mental illness; for example, parents may feel responsible for ‘passing on’ the condition to their children (Austin 2005, Austin and Honer 2007, Austin and Honer 2008).

Experiencing guilt and shame can have a direct effect on self-esteem (Lewis 1987), such as feelings of humiliation, disgrace, or failure in fulfilling one's responsibilities (Miller and Mason 2005). Shame and guilt can also have important treatment implications as they may act as a barrier to help-seeking behaviours such as accessing professional mental health services (Thara and Srinivasan 2000, Miller and Mason 2005), for example out of fear of a label or diagnosis that carries such a negative stereotype (Miller and Mason 2005), or, amongst relatives, fear of being blamed for causing their or their loved one's illness (Austin and Honer 2005).

1.3 Aetiology of Psychiatric disorders

1.3.1 Psychiatric Genetics

Epidemiological studies

Family, adoption and twin studies have indicated a significant genetic contribution to psychiatric disorders (Gottesman and Shields 1966, Cardno et al. 1999, Shih et al. 2004). Familial aggregation has been consistently demonstrated for psychiatric disorders including SCZ, SAD and BPD (Kendler 1988, Tsuang 2000, Shih et al. 2004, Laursen et al. 2005). The percentage risk for these psychiatric disorders is correlated to the degree of relatedness of the affected individuals in the family (Craddock and Jones 1999, Tsuang 2000, see figures 9 and10), and despite the fact that a substantial majority of affected individuals diagnosed with SCZ, SAD or BPD have no family history of the disorder, having a positive family history remains the single greatest risk factor for developing mental illness (Merikangas and Risch 2003, Laursen et al. 2005, Austin 2005, Finn and Smoller 2006, Hunter et al. 2010, Craddock and Sklar 2013, Meiser et al. 2013, see figures 9 and 10).

(Fig. 9)

Relationship to person with schizophrenia	Lifetime risk
General population	1%
First-degree relative	
Identical twin	40%–48%
Fraternal twin	10%–17%
Sibling	9%
Parent	6%–13%
Offspring	13%
Second-degree relative	
Aunt/uncle	2%
Niece/nephew	4%
Grandchild	5%
Third-degree relative	
First cousin	2%

Figure 9: Estimated lifetime risks for SCZ.

(Figure from Hill and Sahaar 2006, p.508).

Empirical risk estimates are derived from over 2 decades of standardised, family-based studies that have resulted in relatively comprehensive risk data.

(Fig. 10)

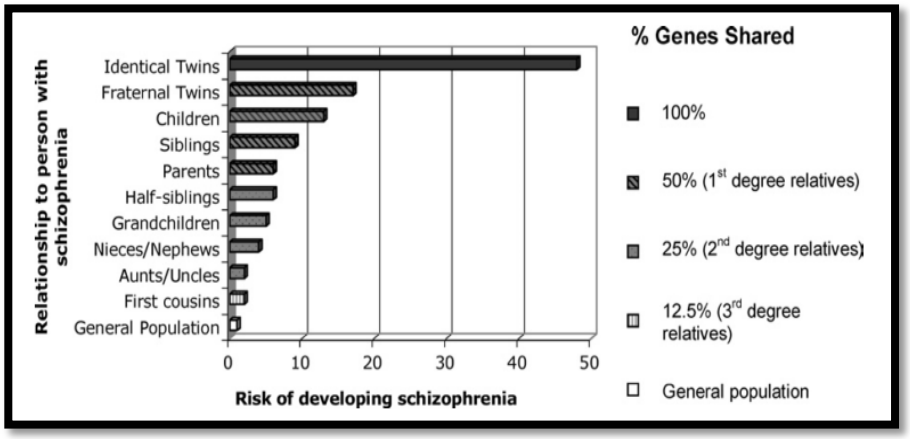


Figure 10: Empirical risks of developing SCZ for relatives of an individual with SCZ.

(Figure from Austin 2005, p. 331, adapted from data from Gottesman 1991).

The percentage risk for these psychiatric disorders is correlated to the degree of relatedness of the affected individuals in the family.

Heritability of psychiatric disorders

Twin studies have consistently reported substantial and similar heritability estimates for psychiatric disorders, with estimates typically ranging from 60-85% (Cardno et al. 1999, Wray and Gottesman 2012, see table 2, following page), indicating that around 80% of vulnerability is determined by genetics (Nature 2010).

Notably the heritability for psychiatric illnesses including BPD and SCZ is substantially higher than for other complex medical including breast cancer, diabetes and heart disease - diseases widely recognised by the public as having a genetic component (Plomin et al. 1994, Austin 2005).

Despite this whilst affected individuals with or at risk of breast cancer, diabetes and heart disease are routinely referred for GC this is not currently the case in psychiatry (see table 2), indicating current provision of genetic information for psychiatric disorders lags behind that for physical disorders.

Table 2: Heritability estimates of multifactorial diseases including SCZ, BPD and MDD

Disease	Heritability estimate	Reference
Schizophrenia	60-85%	Cardno et al. 1999, Wray and Gottesman 2012
BPD	60-85%	McGuffin et al. 2003, Wray and Gottesman 2012
MDD	30-50%	Hamet and Tremblay 2005,Lohoff 2010,Wray and Gottesman 2012
Asthma	70-80%	Thomsen et al. 2010
Diabetes*	26-70%	Poulsen et al. 1999, Almgrem et al. 2011
Spina Bifida*	60-70%	Copp et al. 2015
Age-related macular degeneration*	50-70%	Klaver et al. 1998
Breast Cancer*	25-55%	Schildkraut et al. 1989, Czene et al. 2002, Cancer Research 2015.
Coronary heart disease*	35-50%	Katzmarzyck et al. 2000

Note: * denotes GC is available in the UK for affected individuals with/at risk of having specified condition.

Genetic studies in psychiatric genetics

Despite evidence from family studies, progress in psychiatric genetics had until recently been slow with early attempts to identify risk loci proving disappointing.

Linkage studies, for instance, which attempt to identify large segments of chromosomes that are inherited with disease, were a major focus of early psychiatric genetic studies. Although some psychiatric genetic linkage study groups reported positive linkages in regions, some of which achieved replication by other study groups, including 22q11-12 for SCZ (Coon et al. 1994, DeLisi et al. 2002), and 13q32 for BPD (Stine et al. 1997, Detera-Wadleigh et al. 1999) and SCZ (Brzustowicz et al. 1999, Mulle et al. 2005, Gadelha et al. 2012), most studies neither achieved 'genome-wide' levels of significance nor replicated pre-existing findings (Owen et al. 2004, Owen et al. 2005, Craddock and Sklar 2013).

Similarly, findings of candidate gene studies, which have also traditionally dominated psychiatric genetic approaches (Varga et al. 2011) with over 1000 studies of SCZ (Allen et al. 2008) and hundreds of studies of BPD (Craddock and Sklar 2013), have been inconsistent, with initial positive findings also lacking replication in subsequent, independent studies (Tabor et al. 2002, Collins et al. 2012).

However these approaches were not an overt failure as they provided insight into the inheritance pattern of common psychiatric which finally enabled settlement of the longstanding the "rare" Vs. "common" variant debate in psychiatric genetics (Collins and Sullivan 2013). Providing strong evidence that Mendelian-like mutations (i.e. highly penetrant) could be ruled out in psychiatric pathogenesis, the insight gained through these early genetic studies helped guide researchers in designing studies that are adequately powered to detect the common, low-penetrance loci involved in conferring susceptibility to psychiatric disorders. This resulted in the transition to genome-wide association studies (GWAS), which compare frequencies of genetic variants between cases and controls for a large set of genetic markers distributed across the genome (Collins and Sullivan 2013).

At the time of writing this thesis, genome-wide association studies (GWAS) have identified 108 independent loci for SCZ (Psychiatric Genomics Consortium SCZ Working Group 2013, see figure 11 next page) and eight loci for BPD (Charney et al. 2013 cited Gratten et al. 2014) that reach genome-wide significance. Crucial to the success of psychiatric GWAS has been the adoption of rigorous statistical standards (Pe'er et al. 2008, Sullivan 2010, Panagiotou and Ioannidis 2011), meaning that the biology of a gene plays no role in establishing its association and the studies are thus unbiased in their approach (Sullivan 2010, Collins and Sullivan 2013), and also international collaborations between study groups which has resulted in the accrual of historically massive sample sizes and resultantly increased the power of detection of modest-effect size alleles (Wellcome Trust Case Control Consortium 2007, Ferreira et al. 2008).

Conversely no association achieving genome-wide significance for MDD has been made to date (Cohen-Woods et al. 2013, Major Depressive Disorder Working Group of the Psychiatric GWAS Consortium 2013, Flint and Kendler 2014). As a higher-prevalence, low heritability disorder, the most likely explanation for the lack of success is that GWAS studies have been underpowered to detect loci (Wray et al. 2012, Major Depressive Disorder Working Group of the Psychiatric GWAS Consortium 2013, Flint and Kendler 2014). Similarly, for obsessive compulsive disorder (OCD), anorexia nervosa and Tourette's, published data are sparse and GWAS sample sizes are small by current standards (Collins and Sullivan 2013). It is anticipated that as sample size increases power of detection will increase and loci will be identified for such common, lower heritability psychiatric conditions.

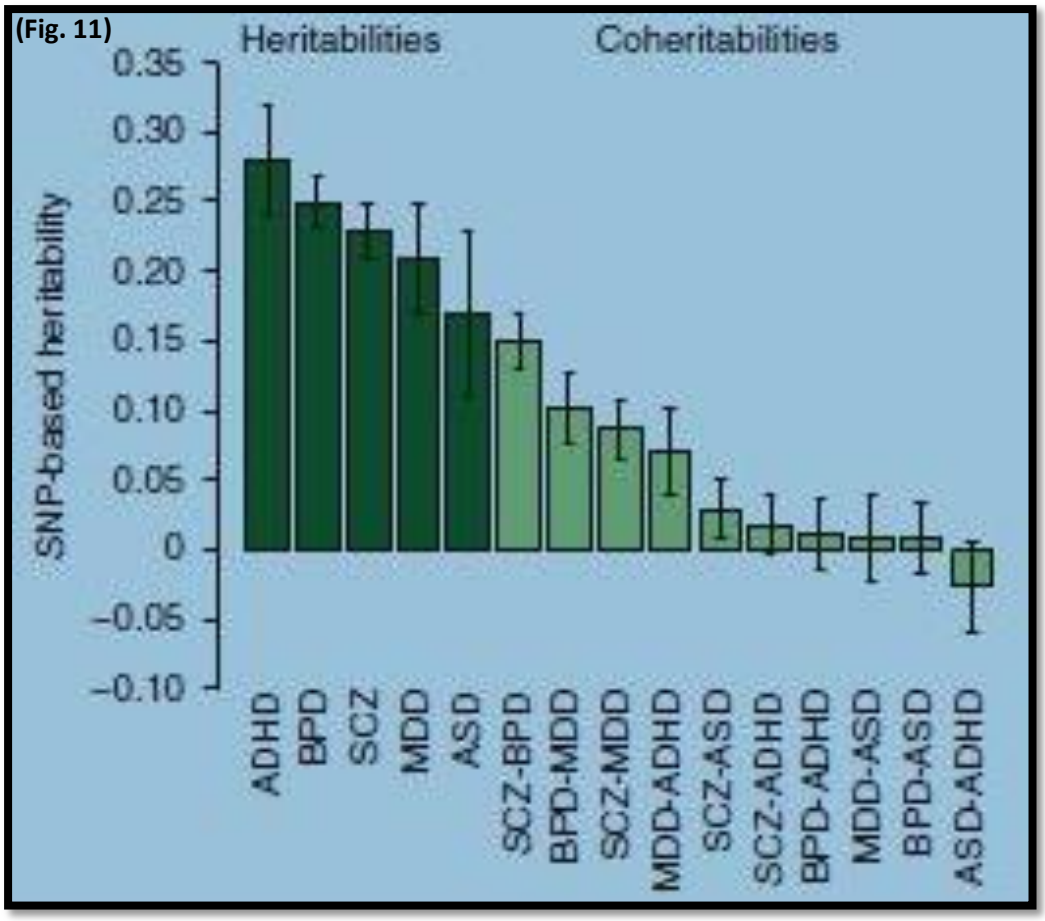


Figure 11: Figure showing i) Proportion of variance in liability (SNP-based heritability) and ii) proportion of covariance in liability between disorder (SNP-based coheritability) for five major psychiatric conditions.

(Figure from Cross-disorder group of the Psychiatric Genomics Consortium 2013, p.23).

Common genetic variation accounted for up to 30% of the variance in liability. Among pairs of disorders (light green), SCZ and BPD shared ~16% of the same common genetic variation ('coheritabilities').

GWAS has thus finally provided insight into the genetic architecture of psychiatric disorders. Psychiatric disorders are now understood to be highly polygenic, with many different genetic loci conferring risk - estimates suggest that variation at around 8,300 independent loci will ultimately be found to account for up to 50% of the genetic risk to SCZ (Ripke et al. 2013) and it is predicted that this is likely true for most psychiatric disorders (Gratten et al. 2014). Additionally it is now understood that risk is dominated by smaller-effect, common variants (Ripke et al. 2013, Gratten et al. 2014) with statistical analyses on GWAS data (described in Ripke et al. 2013) indicating that common variants of small effect (i.e. 0.05% or less) account for a substantial proportion of heritability (between one third to half) for SCZ (Gratten et al. 2014, see figure 10), and that this is likely to be the case for other psychiatric disorders including for BPD and MDD (Lee et al. 2013) and OCD and TS (Davis et al. 2013).

Thus, heritability for psychiatric disorders is not "missing" – a concept that has attracted much coverage in the media (Hirsch 1999, Wade 2010), and online blogs and articles (Latham and Wilson 2010, Joseph 2013) - but rather "hidden" by inadequately powered studies (Gershon et al. 2011, Giusti-Rodríguez and Sullivan 2013, Gratten et al. 2014).

Furthermore, GWAS has revealed that some genes associated with psychiatric disorders are non-specific, having associations across the diagnostic boundaries including SCZ, BPD, intellectual disability, and autism (Rudan 2010, Gratten et al. 2014, see figure 10). Cross-disorder GWAS meta-analyses has identified three loci for a shared SCZ-BPD phenotype (SCZ Psychiatric Genome-Wide Association Study (GWAS) Consortium et al. 2011) and four loci for a broader psychiatric genotype spanning ASD, ADHD, BPD, MDD and SCZ (Cross-Disorder Group of the Psychiatric Genomics Consortium 2013).

This finding of shared genetic risk factors across traditional psychiatric diagnoses is consistent with overlapping clinical presentations and it is hoped this insight may help develop a more effective classification system based on underlying biological causes rather than symptomology-based diagnoses in psychiatry today (Cross-disorder group of the Psychiatric Genomics Consortium 2013).

Biological insights from GWAS

As well as providing glimpses into the genetic architecture of psychiatric conditions, GWAS findings also enable insights into the biological underpinnings of psychiatric disorders. Such understanding is valuable as it is hoped it will enable development of better management of psychiatric conditions (e.g. medication and treatment options), better prevention measures for those deemed 'at-risk,', and more accurate diagnosis.

A key theme to emerge for example is the implication of calcium signalling in SCZ and BPD pathogenesis. A recent GWAS associated the gene encoding the α_{1c} subunit of L-type, voltage-dependent calcium channels, CACNA1C at the intronic SNP rs 1006737 with SCZ (Ripke et al. 2013), replicating previous associations that did not reach the threshold of genome-wide significance, for both schizophrenia (Green et al. 2010) and BPD (Ferreira et al. 2008). Notably, this provides genetic molecular evidence of shared aetiology between BPD and SCZ (Cross-disorder group of the Psychiatric Genetics Consortium 2013).

Additionally one of the most statistically robust findings for SCZ GWAS involves genetic variation within the major histocompatibility complex (MHC) region (Shi et al. 2009, Stefansson et al. 2009, Collins et al. 2012). Although the MHC encodes over 400 genes involved in immunity (McAllister 2014), it has traditionally been a major focus of early genetic studies, largely driven by epidemiological findings that have reported increased prevalence of autoimmune and inflammatory diseases, including caeliac disease and rheumatoid arthritis, in SCZ affected individuals and their relatives (Eaton et al. 2006) as well as reports of immune dysregulation in affected individuals including CNS inflammation in SCZ post-mortem brain tissues (Smyth and Lawrie 2013), and abnormal cytokine levels in tissues in SCZ affected individuals (Watanabe et al. 2010, Di Nicola et al. 2013. The implication of MHC by GWAS thus raises the possibility of an aetiological role for an immune, autoimmune, or infection process in psychiatric pathogenesis (Sullivan 2010). Mechanistically, it has been hypothesised that epigenetic interactions may be involved in mediating vulnerability to SCZ conferred by MHC risk variants (see 1.3.2). Interestingly, this thus highlights how GWAS findings may be helping explain epidemiological puzzles, such as increased rates of immune disorders amongst psychiatric patients (Gratten et al. 2014).

1.3.2 Environmental (non-genetic) factors in psychiatric pathogenesis

Epidemiological data

Family studies have also demonstrated an environmental (non-genetic) contribution to pathogenesis of mental illness.

Despite high heritability, concordance rates for psychiatric illness amongst monozygotic twins is substantially less than 100% (see table 3). In SCZ for example, although heritability is estimated to be 60-85% (Cardno et al. 1999, Wray and Gottesmann 2012), concordance in monozygotic twins is only 40-48% (Gottesman and Wolfgram 1991). This indicates environmental factors play a critical role in mediating susceptibility to illness onset (Tsuang 2000, Hill and Sahhar 2006).

From a clinical perspective this knowledge is especially valuable as it may help identify factors which i) protect mental health or ii) increase risk, to facilitate strategies to better manage and protect mental health for those with or at risk of developing psychiatric conditions.

Table 3: Concordance rates (%) for schizophrenia in monozygotic (MZ) and dizygotic (DZ) twin studies.

(from Tsuang 2000, p.211, adapted from data from Gottesman 1991).

	MZ		DZ	
Study	Pairs	Rate	Pairs	Rate
Finland 1963, 1971	17	35	20	13
Norway 1967	55	45	90	15
Denmark 1973	21	56	41	27
UK 1968, 1987	22	58	33	15
Norway 1991	31	48	28	4
U.S. 1969, 1983	164	31	268	6
Pooled (excluding U.S.)				
Median	146	48	212	15
Weighted mean		48		16
Pooled (all studies)				
Median	310	46	480	14
Weighted mean		39		10

Identifying environmental factors involved in psychiatric pathogenesis

Identification of environmental factors is notoriously complex. Sophisticated studies addressing genetic confounding by controlling for genetic risk have been conducted along with extensive meta-analyses in order to comprehensively aid identification. To this degree, some notable progress has been made over the past two decades.

What is now understood that factors or events that occur both pre- and peri-natally mediate environmental risk (Mueser and McGurk 2004, Maric and Svrakic 2012), and that these risk factors may be biological, physical and/or psychosocial (Vilain et al. 2013).

Indeed some environmental factors have now been consistently associated with increasing risk of psychosis (Mueser and McGurk 2004, Uher 2009, van Os et al. 2010, Maric and Svrakic 2012) including being born in spring (Barry and Barry 1961, McDonald and Murray 2000), increased paternal age (Dalman and Allebeck 2002, Crow 2003, Rees et al. 2011, Goreily et al. 2013), growing up in an urban environment (Frissen et al. 2015) cannabis use (Semple et al. 2005, Parakh and Buso 2013), minority group position (van Os et al. 2010, Suvisaari et al. 2014), obstetric complications (Lewis and Murray 1987, Cannon et al. 2002, Ballon et al. 2008), and childhood trauma (Rutter 1965, Janssen et al. 2004, Schäfer and Fisher 2011, Dvir et al. 2013). Additionally, early trauma or stress- including separation, bereavement, family problems, neglect and child abuse (Ford and Kidd 1998, Kendler et al. 2004, Mandelli et al. 2015); occupational stress (Cohen and Wills 1985, Stansfeld et al. 2012, Theorell et al. 2015) and social isolation or reduced social support (Cohen and Wills 1985, Nordentoft 2007, Voisin et al. 2015) have been consistently associated with MDD and suicidal behaviour (Paykel 1976, Mandelli and Serretti 2013).

Focus has now switched to attempts to elucidate the biological underpinnings of putative gene/environmental interactions in psychiatric pathogenesis (see figure 7). These investigations typically involve studies that attempt to identify the influence environmental factors have on gene expression and activity (i.e. epigenetic interactions) in order to elucidate the underlying molecular mechanism(s) that may confer susceptibility (Marik and Svrakic 2012, Mandelli and Serretti 2013 for reviews).

The consistent association of psychiatric illness with increased paternal age, for example, has been attributed to accumulation of de novo mutation with age in the male germline which are subsequently passed on to offspring (Dalman and Allebeck 2002, Crow 2003,Uher 2009, Rees et al. 2012, Fromer et al. 2014, Milekic et al. 2015). It has been hypothesised that the mutations alter biological mechanisms, such as methylation (Jenkins et al. 2014, Milekic et al. 2015), to influence “risk pathways” and result in neurodevelopmental alterations that ultimately increase risk to neuropsychiatric illness (Malaspina et al. 2015). Interestingly these de novo mutations are believed to increase risk across a spectrum of neurodevelopmental disorders, providing further evidence of shared aetiology between traditional diagnostic boundaries (van Os et al. 2010).

Additionally, inflammatory imbalance - potentially mediated by both genetic contributions, such as MHC risk variants (as discussed in 1.1.3.1.2) and environmental contributions such as maternal infection - have been proposed as a biological mechanism conferring vulnerability to onset of psychosis (McAllister 2014). A leading hypothesis is that the resulting chronic changes in immune molecules in the brain may affect neurodevelopmental processes later in life, such as nerve cell maturation, signalling, differentiation, proliferation, and survival, ultimately resulting in the neurological defects associated with SCZ and/or psychosis (Smyth and Lawrie 2013).

Gene-environment interactions have also been proposed to explain the association between cannabis use and psychosis (Henquet et al. 2008). Available evidence indicates that genetic variation may influence sensitivity to the psychosis-inducing effects of cannabis (Decoster et al. 2012). For example, the polymorphism Val158Met in the gene encoding catechol-O-methyl transferase (COMT) - one of the most studied hypothesis-driven candidate genes (Collins et al. 2012)- results in respectively in high and low-activity forms of the enzyme, which degrades dopamine (Al-Asmary et al. 2014). Studies have reported that carriers of the COMT Val homozygous alleles have a rapid dopamine metabolism and subsequently low cortical and high midbrain dopamine levels (Sagud et al. 2010) and have been found to be at increased risk of psychosis if they use cannabis during adolescence (Caspi et al. 2002, Caspi et al. 2005, Maric and Svrakic 2012). Cannabis is known to cause a significant decrease in cortical dopamine and increase in midbrain dopamine, and it has therefore been proposed that pre-existing heritable dopamine dysfunction may be amplified by cannabis to ultimately induce psychosis (Maric and

Svrakic 2012). Other candidate variants hypothesised to influence cannabis-induced risk are NRG1 (Pelayo-Téran et al. 2012, Long et al. 2013, Suárez-Pinilla et al. 2015) and AKT1 (van Winkel et al. 2011, Decoster et al. 2012, Radhakrishnan et al. 2014). Notably, it has been proposed that other environmental factors such as childhood trauma or urbanicity may also influence differential dopamine sensitisation effects, to have a synergistic effect with cannabis and further increase risk of psychosis (Pelayo-Téran et al. 2012, Radhakrishnan et al. 2014).

Thus, current understanding is that genetic vulnerability to psychiatric illness is partially mediated by differential sensitivity to numerous risk factors which affect brain functioning and/or influence brain development (van Os et al. 2010). It is hoped that longitudinal studies alongside the adoption of multidisciplinary translational approaches will enable deeper exploration of gene-environment interactions and help identify the underlying mechanisms (van Os and Kapur 2009, Nature 2010, van Os et al. 2010) which may in turn help increase biological understanding of psychiatric illness and thus advance clinical management and treatment strategies (van Os et al. 2010, Schmitt et al. 2014).

1.3.3 Current aetiological understanding of psychiatric conditions: Implications for clinical practice

What is currently understood about the aetiology of psychiatric disorders raises the clinical question of how this information can effectively delivered to patients and their families.

The aetiological information regarding the relative contributions of genetic and environmental factors, and protective factors in mental health, is especially complex and surrounded by uncertainty with much yet to be understood, and this needs to be effectively managed and counselled around by the clinician (Peay et al. 2008, Hippman 2013).

Genetic counsellors, whom are specially trained to convey complex information and in the provision of supportive counselling, have been proposed to be ideally placed to deliver such information (Austin and Honer 2005, Austin and Honer 2008, Peay et al. 2008) This is discussed further in sections 1.5.1 and 1.5.2

1.4 Genetic counselling

1.4.1 Genetic counselling – A growing profession

Genetic counsellors today are qualified healthcare professionals that have specific expertise in identifying and educating patients at risk for inherited conditions (AGNC 2015). They are specially trained to personalise, interpret, and communicate complex science into information that is helpful for the patient (Mester et al. 2012).

As our understanding of the human genome continues to unfold and we gain more insight is gained into the genetic underpinnings of disease and health, genetic counsellors are becoming an increasingly important part of the healthcare team. Over recent decades GC has evolved to embrace an increasing number of fields in clinical medicine (Resta et al. 2006, Guttmacher et al. 2007, Skirton et al. 2015, see table 2) including oncology, cardiology, and endocrinology; with the skillset of genetic counsellors continuing to expand in response to the increasing complexity and diversity of the issues of their presenting patients (Kasparian et al. 2007, Mester et al. 2012).

1.4.2 Genetic counselling - Definition of practice

The NSGC's most recent definition of practice of genetic counselling describes it as:

> "The process of helping affected individuals adapt to the medical, psychological and familial implications of genetic contributions to disease" (Resta et al. 2006, p. 77)

Under this definition of practice, the process should integrate the following:

- *"Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.*
- *Education about inheritance, testing, management, prevention, resources and research*
- *Counseling to promote informed choices and adaptation to the risk or condition"*

(Resta et al. 2006, p.77)

However the goals of and approaches to genetic counselling have undergone significant changes since early practice, and to this day, whilst there has been overall agreement in the general tasks encapsulated in the process of genetic counselling, as described in the NSGC's definition of practice, there remains a lack of consensus regarding the *process* itself. Indeed, the development of a consistent model of practice is a topic has engaged much recent discussions (see for example, Biesecker 2001, McCarthy Veach et al. 2006, McCarthy Veach et al. 2007, Hartmann et al. 2013).

1.4.3 Genetic counselling – A brief history

Origins, Sheldon Reed, and associations with eugenics

The term ‘genetic counselling’ was originally coined by Sheldon Reed in 1947, who described it as the provision of

> “supportive counselling and genetic information about inheritance patterns and recurrence risks” (Reed 1947, cited Reynolds and Benkendorf 1999 p.375).

Early genetic counselling was of a social rather than medical nature, and Reed himself described it primarily as a form of “social work without eugenic connotations” (Reed 1975, p.335). For example, the single most common purpose of early genetic counselling enquiries was to evaluate a newborn for adoptive placement (Stern 2012), which typically related to matters of race or ancestry, e.g. to determine a child’s skin colour or to obtain an evaluation for other racial characteristics (Resta 2006, see figure 12). Other purposes for early enquiries included obtaining recurrence risk for conditions including epilepsy, intellectual disability, and SCZ (Resta 2006, Stern 2012) as well as questions relating to consanguinity and mate-choice (Resta 2006, Stern 2012).

(Fig. 12)

"Letter from Miss (…), Bureau of Child Welfare, regarding adoption of (a boy), a 'near white' by a white mother. Usual question as to whether his children could show prominent Negroid characteristics."

Figure 12: Extract detailing an early genetic counselling enquiry from the Dight Records, Dight Institute Inquiries, August 30, 1948.

(Cited Resta 2006, p.270).

Enquiries regarding skin colour, such as the above, were common lines of enquiry in early GC practice.

Thus, the early era of genetic counselling had a predominantly "public health-centered approach"(Resta et al. 2006, p.270), often with the wide aim of bettering the well-being of society rather than that of benefitting the individual patient. It has been reflected that, in reality, it was sometimes difficult to disentangle early practice with the eugenics movement of the early 20th Century (Resta 2006). This will be reviewed in greater detail in section 1.5.5

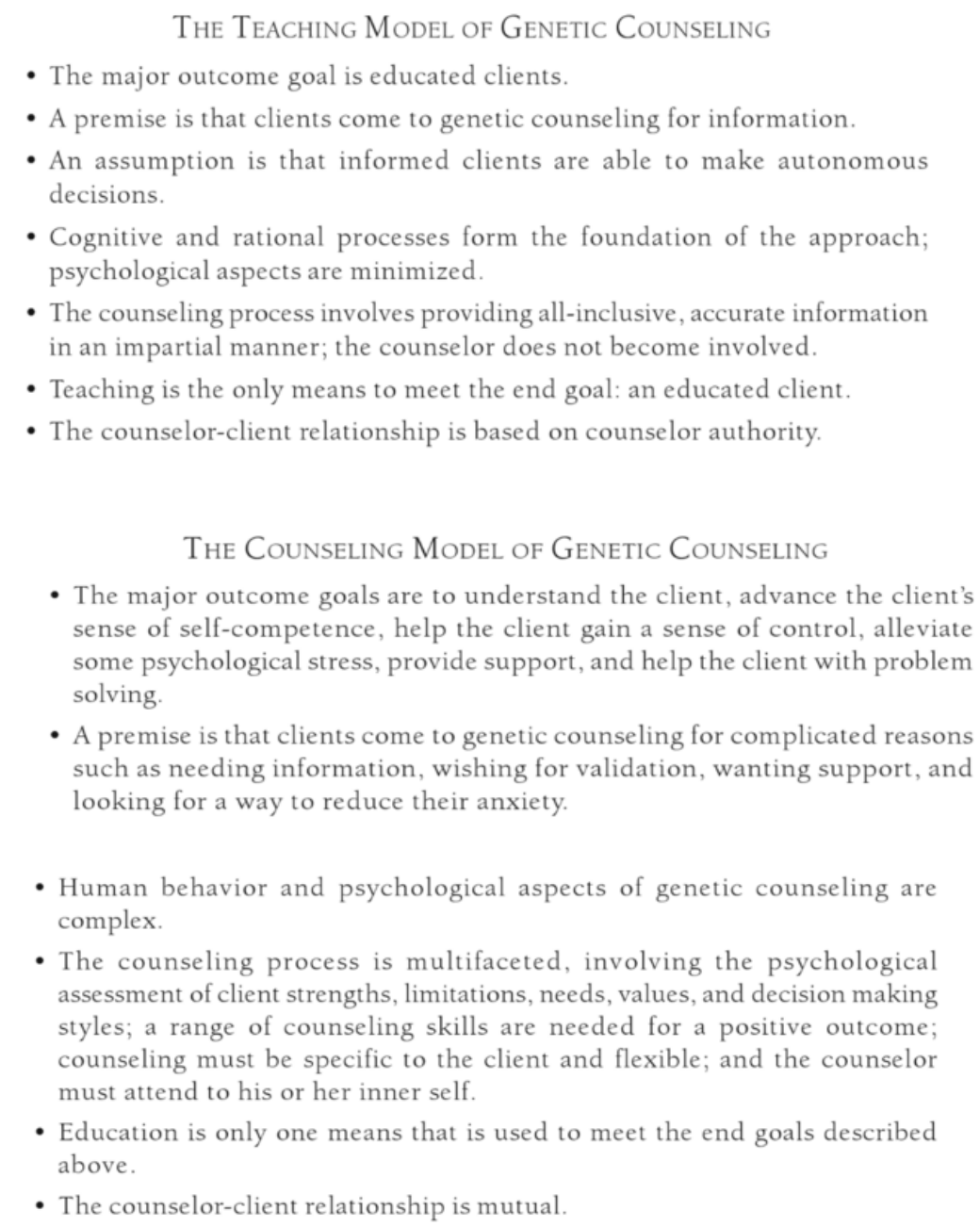
In the 1980s there was a general shift from a 'public-health' approach towards preventative medicine in regards to provision and practice of GC. Whilst the overall goal of genetic counselling still focussed on preventing genetic defects, there was increasing support for an information model that facilitated client-informed decision-making, and had a more person-centered approach (Biesecker 2001).

In contemporary practice, non-directiveness is a guiding principle, with the counsellor promoting autonomy of the individual patient and helping them make decisions which are in line with their own personal, social and cultural beliefs and practices (Biesecker 2001, Resta 2006, McCarthy Veach et al. 2007). This has been proposed, at its most basic level, as the counterpoint to a eugenic approach (Maio et al. 2013).

1.4.4 Genetic counselling - Models of practice

Traditionally there have been, two major schools of thought in regards to approaches to genetic counselling - education, and counselling (Kessler 1997). These are captivated by the two prominent models used to conceptualise genetic counselling - the ‘teaching’ model, common to healthcare, and the ‘counselling’ model, common to psychology (Hartmann et al. 2013). These models are distinctive in both their approach and goals (Kessler 1997, Roter et al. 2005, McCarthy and McCarthy Veach et al. 2006, see fig. 13, following page).

(Fig. 13)



THE TEACHING MODEL OF GENETIC COUNSELING

- The major outcome goal is educated clients.
- A premise is that clients come to genetic counseling for information.
- An assumption is that informed clients are able to make autonomous decisions.
- Cognitive and rational processes form the foundation of the approach; psychological aspects are minimized.
- The counseling process involves providing all-inclusive, accurate information in an impartial manner; the counselor does not become involved.
- Teaching is the only means to meet the end goal: an educated client.
- The counselor-client relationship is based on counselor authority.

THE COUNSELING MODEL OF GENETIC COUNSELING

- The major outcome goals are to understand the client, advance the client's sense of self-competence, help the client gain a sense of control, alleviate some psychological stress, provide support, and help the client with problem solving.
- A premise is that clients come to genetic counseling for complicated reasons such as needing information, wishing for validation, wanting support, and looking for a way to reduce their anxiety.

- Human behavior and psychological aspects of genetic counseling are complex.
- The counseling process is multifaceted, involving the psychological assessment of client strengths, limitations, needs, values, and decision making styles; a range of counseling skills are needed for a positive outcome; counseling must be specific to the client and flexible; and the counselor must attend to his or her inner self.
- Education is only one means that is used to meet the end goals described above.
- The counselor-client relationship is mutual.

Figure 13: A comparison of the teaching model and counselling model of genetic counselling.

(Figure from McCarthy Veach et al. 2006, p.30-31).

Historically, the two traditional approaches to GC have been the teaching model, and the counselling model. These models are distinctive in their approach and goals.

The teaching model of genetic counselling

The teaching model centres on the transmission of information between counsellor and client. It aims to facilitate understanding of the occurrence, probable course, and available management of the condition (Berkenstadt et al. 1999, Davey et al. 2005), with the overall goal of educating the patient. Consequently genetic counselling under the teaching model has been described as health education rather than counselling (Kessler 1997). The central tenet underlying this model is that patients seek genetic counselling in order to obtain genetic information, and so it has an educational purpose (McCarthy Veach et al. 2007).

In support, to an extent, of the teaching model of practice, studies have found that delivery of factual information is the most frequent interaction between client and counsellor (Michie et al. 1997, Roter et al. 2005, Austin et al. 2014). Indeed, traditionally many research studies evaluating effectiveness of genetic counselling sessions have focused on client knowledge gain (Kasparian et al. 1997, Davey et al. 2005, Roter et al. 2005, Austin et al. 2014).

The counselling model of genetic counselling

The counselling model of genetic counselling, on the other hand, rests upon the assertion that the relationship between the client and the counsellor is psychotherapeutic in its nature, with the psychological well-being of the patient being the integral aspect of the exchange (Biesecker 2001). The central tenet to this model is that there are diverse reasons for which people seek genetic counselling, and these will be individual to each patient (McCarthy Veach et al. 2007); and that the most effective genetic counselling will involve recognition, and subsequent addressing, of these needs.

Historically, although counselling based-approaches are largely accredited to Joan Marks, director of the first Genetic Counseling Program at the Sarah Lawrence College in 1969, and the counselling theories of Carol Rogers, a leading psychologist who devised ‘client-centered’ counselling (Resta 2006 p.271, NICE 2014), the psychotherapeutic aspects of genetic counselling had been discussed by early practitioners of genetic counselling some decades previous (Resta 2006).

Under the counselling model of GC, key goals would be: understand the patient's needs and concerns, help the client to personalise the genetic information and use it in a way that *is personally meaningful* to them (Biesecker 2001), increase their perceived sense of control and self-efficacy, reduce or minimise psychological distress, provision of support, help the patient with autonomous decision-making (McCarthy Veach et al. book, Biesecker 2001, Biesecker and Peters 2001), and, ultimately, to enhance the patients' ability to adapt to the condition, or risk of the condition, over time (Resta 2006). Thus, the key underlying philosophy of this model is that education is not considered an end in itself, unlike under the teaching model, but rather a means to facilitating other, psychological, goals and outcomes for patients (Kessler 1997).

Specifically, the counselling model recognises that communicating about genetic information, and especially about risk, can have a major emotional impact for patients. For example, obtaining this information can often be a stressful or 'threatening' event for patients (Davey et al. 2005, p.198) and can induce diverse negative psychological feelings and emotions including shame, guilt, anxiety, feelings of loss of personal control, bereavement, reduced self-esteem, social isolation, and stigmatisation (Biesecker 2001). The counselling model advocates that a counsellor should deliver information in such a way that not only minimises the potential negative impact of such information, but also reduces psychological distress (Biesecker 2001, Resta et al. 2006). This may be achieved, for example, through being aware of the client's emotions – their hopes, fears and rationalisations; and building a supportive relationship through which these feelings may be discussed and addressed (Bernhardt et al. 2000, McCarthy Veach et al. 2007).

Furthermore, the counselling model recognises that scientific explanations are only one way to explain risk. In practice, individuals think in varied, complex and abstract ways which can be influenced by numerous factors (Sivell et al. 2008). This can allowing for personal interpretation and meaning of genetic information, and the overriding of what may be considered 'logical' by the client's emotions (Biesecker 2001). Under the counselling model, the clinician should have an awareness of these psychological and psychosocial complexities, and be able to effectively counsel around them, to ultimately facilitate better comprehension amongst the patient (Biesecker and Peters 2001).

In order for genetic counselling training courses to be accredited by the Accreditation Council for Genetic Counseling (ACGC) there is the requirement for the curricula to incorporate psychotherapeutic skills and competencies in regards to genetic counselling (Resta 2006, ACGC 2013 cited Semaka et al. 2014). Thus, theoretically, counselling approaches are integral to current day practice.

In spite of this, there is some evidence indicating that a teaching model is more prevalent than a counselling model in regards to practice (Kessler 1997, Biesecker and Peters 2001, McCarthy Veach et al. 2007, Roter et al. 2005). Additionally it has been proposed that in the current so-called 'genomic era,' with increasingly sophistic genomic technologies, increasing genetic testing options and surging interest of the public in genetics in regards to health, genetic counselling is shifting more towards being a form of health education and thus teaching-based approaches (Biesecker 2001, Austin et al. 2014).

1.4.5 Genetic counselling – Contemporary practice

There has been growing awareness of the need to develop a consensus, international model of practice of GC. This is due to a number of factors including, and not limited to, growing understanding of the role of genetic contributions in common diseases and consequently a bigger scope of genetic counselling; increased public interest and awareness in genetics research in relation to health; increasing sophistication of genomic technologies; and growing support for the importance of evidence-based practice in healthcare interventions (Bisecker 2001). Resultantly program directors and special task forces have met over the past decade to discuss models of practice to try and achieve more, international, agreement in regards to the practice of GC (see for example Biesecker 2001, Roter et al. 2005, McCarthy Veach 2007, Hartmann et al. 2013).

Lack of consensus on a model of practice has been partially put down to inconsistencies in regards to measuring outcomes of interventions and lack of validated outcome measures, largely due to lack of clarity in regards to the best outcomes to measure (McAllister et al. 2011).

Model of empowerment and GCOS-24

In an attempt to address the existing gap in consensus of GC practice, McAllister et al. (2011) developed and validated the Model of Empowerment.

Under the model of empowerment, the key goal of GC would be to increase empowerment, defined as:

> "A set of beliefs that enable a person from a family affected by a genetic condition to feel that they have some control over and hope for the future"

(McAllister et al. 2011, p. 125).

Key outcomes would be that, following the intervention, the patient:

- *Can make informed life decisions ('decisional control').*
- *Has adequete information and understanding about the genetic condition, e.g. familial risk to self and relatives; treatment, management and prevention options available; support that is available to them ('cognitive control').*
- *Can make effective use of the health and social care systems for both self and relatives ('behavioural control').*
- *Can manage one's feelings about having a genetic condition in the family ('emotional regulation').*
- *Has hope for the future incuding in terms of fulfilling family life, for oneself and living and future descendants ('hope').*

(adapted from McAllister et al. 2011, McAllister 2015, pers comms, 15 February 2015).

Thus, at its most basic level, the model of empowerment embraces both 'teaching' aspects and 'counselling' aspects of traditional genetic counselling approaches. The model of empowerment was the key construct for the development of the Genetic Counselling Outcomes Scale, a ***validated*** 24-question self-reported patient reported Outcomes Measure (PROM). GCOS-24 has been translated or is currently undergoing translation into Dutch, Danish, Urdu, Arabic and Japanese, and is being used to evaluate service evaluations for routine clinical practice and patient benefits from new interventions in research, including novel PGC interventions (Inglis et al. 2014). It is the outcomes measurement supported by the National Society of Genetic Counselors (NSGC 2016).

1.5 Genetic counselling for psychiatric disorders - A review of the literature

A review of the literature regarding PGC was undertaken in order to formulate a research question and guide study design. This review process is described in more detail in chapter 2, materials and method: Literature review.

Genetic counselling for psychiatric conditions – An introduction

PGC is a novel but currently minor sub-specialism of genetic counselling that is as yet largely untested, predominantly because PGC is not routinely provided for psychiatric conditions.

Despite this, there has in fact been long-standing interest in the application of GC for affected individuals with psychiatric disorders, with supporting literature spanning over 5 decades (Heston 1966, Kessler 1980, Schulz et al. 1982, Moldin and Gottesman 1997, Rutter et al. 1997, Hodgkinson et al. 2001, Collier et al. 2009, Inglis et al. 2014).

These groups have generally advocated GC may be valuable clinical tool in delivering genetic information about psychiatric conditions to patients and providing supportive counselling around related concepts. Genomic advances in psychiatry, which have provided insights into the aetiology of psychiatric disorders, has seen renewed interest in the potential application of GC in psychiatry over the past decade (Hodgkinson et al. 2001, (Austin and Honer 2007, Austin and Honer 2008, Meiser et al. 2013, Austin et al. 2014).

Additionally, whilst focus has typically centered on providing GC for SCZ and BPD, as more is understood about the genetic architectures and aetiology of other mental illnesses there is growing consensus that GC may have a wider application and be provided for other, more common, mental disorders including OCD and MDD (Meiser et al. 2013, Austin et al. 2014).

1.5.1 Goals of PGC

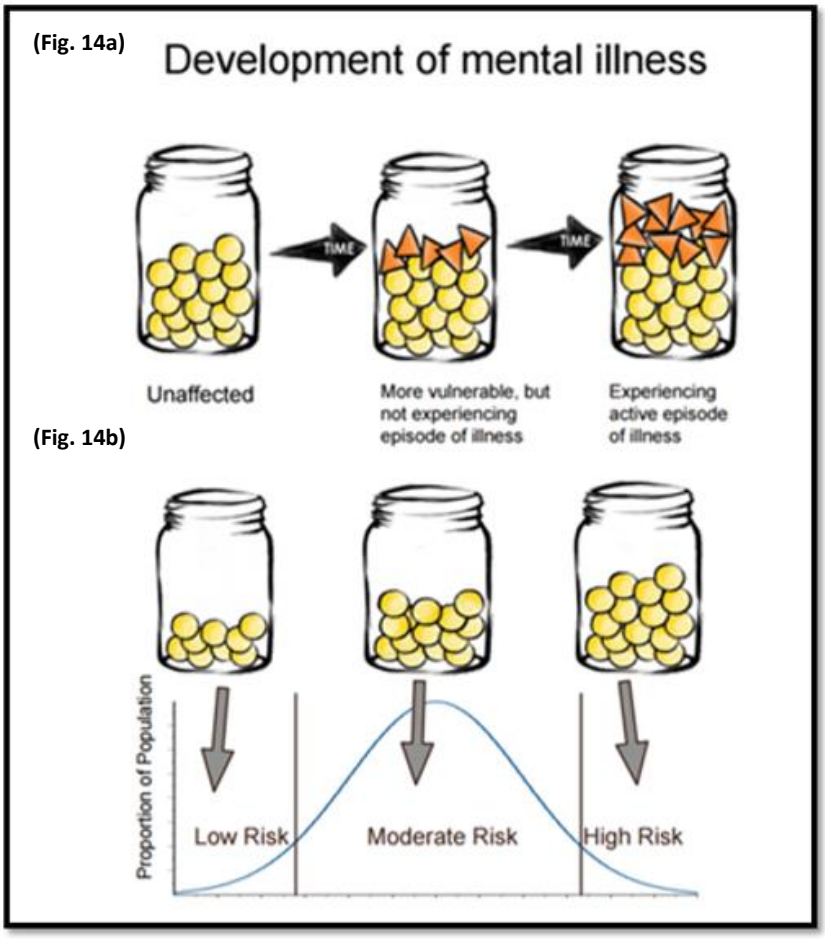
There has been general consensus in currently available literature in regards to goals of PGC interventions. Identified potential outcomes include increasing aetiological understanding, identifying protective factors and influencing health-related behaviours, increasing understanding of familial recurrence risks, providing information, support, and facilitating decision-making around genetic testing, reducing stigma, and psychotherapeutic aspects including reducing shame, guilt and blame.

However these discussions remain largely hypothetical, as there is to date little available outcomes data assessing actual practice of PGC interventions, meaning it remains a largely untested sub-specialism of GC. Clinical research groups are putting greater emphasis on assessing interventions to enable development of an evidence-base (Costain et al. 2014a, Costain et al. 2014b, Austin 2015, pers comms, 15 February 2015).

Goals of PGC: Increasing aetiological understanding

A consistently identified major goal of PGC is to increase understanding about the causes (genetic and environmental) of mental illness amongst affected individuals and their relatives (Austin and Honer 2005, Finn and Smoller 2006, Hill and Sahaar 2006, Austin and Honer 2007, Lyus 2007, Austin and Honer 2008, Costain et al. 2014a, Costain et al. 2014b, Inglis et al. 2014).

Specifically, PGC should increase understanding of multifactorial models of inheritance and comprehension of gene X environment interactions. These concepts are often explained using pictorial aids (Austin and Honer 2007, Austin and Honer 2008, see figures 14 and 15) to enhance understanding. Incorporating the individual's family history of mental illness may also facilitate understanding of complex concepts, .e.g. explaining why some affected individuals develop psychiatric disorders and others do not, or why some affected individuals develop one psychiatric condition and their relatives develop a different condition (i.e. genetic overlap between psychiatric diagnostic boundaries) (Austin and Honer 2007, see figure 14); and may also help affected individuals contextualise the information, e.g. understanding why periods of stress may have influenced their illness onset or worsened their symptoms (Peay et al. 2008).



Figures 14a and 14b: Quantitative trait models used in PGC sessions

(Morris 2015, pers comm., 13 January. Figures © J. Austin, 2015).

Yellow balls represent genetic factors and orange triangles represent environmental factors. These diagrams can help explain concepts such as i) relative contributions of factors involved in pathogenesis (figure 14a) and ii) why some affected individuals may never develop mental illness (figure 14b).

Studies that have researched attribution perceptions for mental illness amongst the general public have reported that, although consistent with medical models of practice, affected individuals and relatives commonly attribute a multifactorial models of causation for mental illness (Gamm et al. 2004, Meiser et al. 2005, Meiser et al. 2007, Peay et al. 2008, Baines and Wittkowski 2013) there have been indications that comprehensive understanding of pathophysiology is lacking and uncertainty still exists regarding causal explanations (Hodgkinson et al. 2001, Holzinger et al. 2003, Austin and Honer 2005, Costain and Bassett 2012, Costain et al. 2014a).

In support of these findings, studies exploring public understanding of genetics more widely have shown that knowledge of genetic concepts related to health and disease is limited and that common misconceptions exist (Lanie et al. 2004, Molster et al. 2009, Potokar et al. 2012). For example, the terms ‘genetic’ and ‘hereditary’ are commonly thought of as synonymous, indicating potentially lack of full understanding of the implications of genetic contributions to disease (Costain et al. 2014b).

Thus, it has been proposed that there is a potential need regarding the provision of aetiological and genetic information amongst this population (Austin and Honer 2005, Finn and Smoller 2006, Hill and Sahaar 2006, Austin and Honer 2007, Lyus 2007, Austin and Honer 2008, Costain et al. 2014a, Costain et al. 2014b, Inglis et al. 2014).

Providing further supporting for the case of providing aetiological information for affected individuals and their families, and therefore it has been proposed, for the provision of PGC, groups exploring potential impact of PGC (e.g. Hodgkinson et al. 2001, Austin and Honer 2005, Finn and Smoller 2006, Lyus 2007, Austin and Honer 2007, Peay et al. 2008, Austin and Honer 2008, Inglis et al. 2014) have advocated that research from health psychology has indicated increased aetiological understanding can be a critical factor in facilitating psychological adaptation to the disease (e.g. Skirton and Eiser 2003, Walter et al. 2004, Husson et al. 2011, Johannson et al. 2014).

Furthermore it has been asserted that this may be particularly useful in psychiatry due to the incomplete understanding regarding aetiology of psychiatric disorders; uncertainty regarding familial risk; and psychotherapeutic aspects of mental illness associated to causation including shame, guilt, and stigma (Hodgkinson et al. 2001, Austin and Honer 2005, Finn and Smoller 2006, Lyus 2007, Austin and Honer 2007, Peay et al. 2008, Austin and Honer 2008, Inglis et al. 2014).

PGC thus potentially provides the opportunity to explore the patient's existing perceptions of cause, address misconceptions, explore emotional implications of misconceptions, and redevelop more positive attitudes towards the mental illness on a basis of an improved aetiological understanding (Austin and Honer 2007, Peay et al. 2008, Inglis et al. 2014), which may have important outcomes such as decreasing perceived burden of the illness, increasing coping ability and health-related quality of life, reducing family-based conflict; to ultimately facilitate better adaptation to the mental illness (Austin and Honer 2007).

The hypothesis that PGC is helpful in facilitating better understanding of aetiology is now some supported with some data from GC outcomes studies. For example, in a pilot study, 92% of participants (n=12) reported they had learned new information about the causes of mental illness, and 78% (n=7) of participants reported that GC had decreased confusion regarding causes of mental illness (Austin and Honer 2008). Costain and colleagues (2012, 2014) reported significant and lasting improvements in knowledge and perceived knowledge of aetiology for patients with schizophrenia (Costain et al. 2014a) and relatives of affected individuals with schizophrenia (Costain et al. 2014b) following PGC. Further, Costain et al. (2014a) reported PGC reduced self-blame alongside increasing perceived and objective aetiological knowledge amongst affected individuals with SCZ as well as general reduction in anxiety. No other relevant outcome data is currently available, however.

Goals of PGC: Identifying protective factors and influencing health-related behaviours

Studies have identified that PGC may facilitate better management of (/risk of) mental illness by identifying protective factors and influencing health-related behaviours of affected individuals and their relatives.

Practically, it has been identified that PGC provides a useful forum to presenting and discussing what is currently knows with affected individuals and relatives known from research about risk contributors to psychiatric illness such as smoking cannabis or methamphetamine use (Austin and Honer 2005, Austin and Peay 2006, Austin et al. 2007, Austin and Honer 2008, Inglis et al. 2014).

It has also been postulated that PGC provides an environment for patients to identify effective research-informed 'protective factors', such as coping strategies for dealing with stress (e.g. adequate sleep, regular exercise)((Austin and Honer 2005, Austin et al. 2008), again often explained using pictorial aids (see figures 15a and 15b). This may enable patients to be better informed in making decisions that may help prevent relapse or development of psychiatric illness (Austin and Honer 2005, Austin et al. 2008).

In GC these discussions often incorporate the patients' *personal* experiences of mental illness or stress (Austin and Honer 2005, Inglis et al. 2014) for example, life events that occurred around the time of onset of illness; environmental factors that may be personal 'triggers' to onset or relapse, such as smoking marijuana; or life events occurring around periods of excess stress or anxiety. It has been asserted that this may help affected individuals identify, on an individual level, strategies that may be particularly effective in facilitating better protection and management of their *own* mental health (Austin and Honer 2005, Inglis et al. 2014). This may also provide a useful opportunity for counselling regarding emotional implications of these specific events which may in turn alleviate guilt, shame, blame and stigma (Austin and Honer 2005. see previous section).

Fig. 15a

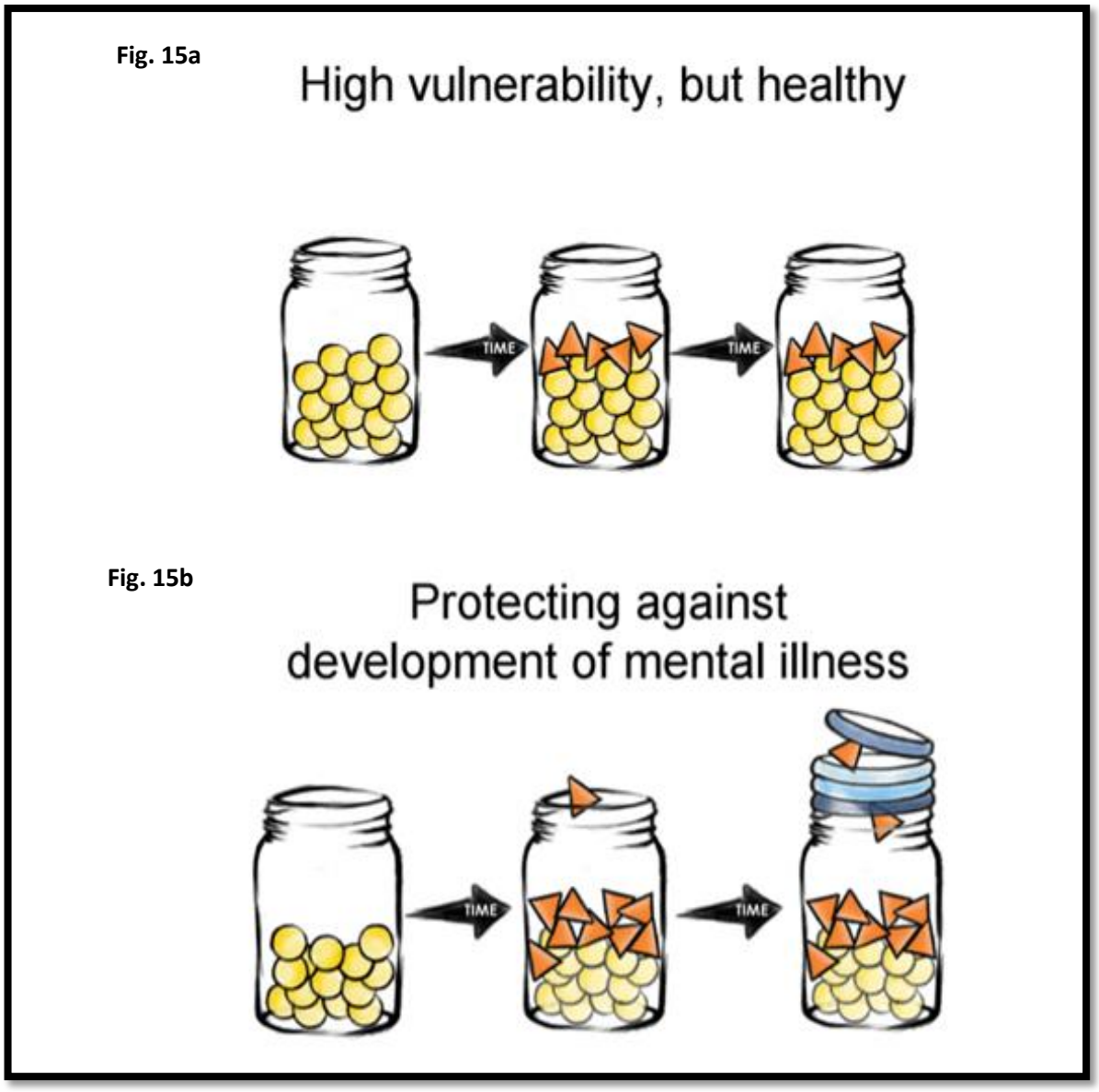


Fig. 15b

Figures 15a and 15b: Pictorial aids representing mental illness pathogenesis/recovery

(Figures from Morris et al. 2015, pers comm, 13 January. Figures ©J. Austin 2015).

Visual aids can help explain i) how putative environmental factors (orange triangles) can be avoided and so blocked from the jar, to reduce risk of illness (e.g. avoiding drugs) and ii) how putative protective factors (blue rings) can protect against risk by making it harder for the jar to fill and the threshold for onset of symptoms to be met (e.g. therapy, medication adherence, exercise). This may also help reduce feelings of genetic fatalism.

Furthermore, studies have proposed that the information provided through PGC may have a *psychological impact* on management of mental illness (Papdimitriou and Dikeos 2003, Austin 2005, Austin and Honer 2007, Meiser et al. 2007, Peay et al. 2009, Hippman et al. 2013). In support of this, it is well established from health psychology that aetiological understanding can influence health-related behaviours (Taylor 1983, Williams and Healy 2001, Brown et al. 2001, Walter et al. 2004, Husson et al. 2011, Oflaz et al. 2015); for example discussions about aetiology and the relative contributions of both genetic and behavioural risk emphasise that behaviour changes can reduce overall risk and may therefore increase perceptions of *value* and/or *cost* of certain health-related behaviours, and thus likelihood of their uptake or avoidance (Zubin et al. 1983, Austin 2005, Austin and Honer 2007, Sivell et al. 2008, see figures 15a and 15b). This may influence decisions regarding health-related behaviours, risk avoidance, and accessing medical services, to facilitate better self-management (Sivell et al. 2008, Austin and Honer 2007).

Similarly, research has shown increased aetiological understanding can also empower an affected individuals' sense of management of their illness by increasing their perceived personal control (Thompson et al. 1993, Davey et al. 2005) which may help further promote help-enhancing behaviours in mental illness (Zubin 1983, Landsverk and Kane 1998, Merinder 2000, Gamm et al. 2004, Austin and Honer 2007, Meiser et al. 2007, Hippman et al. 2013). Increasing sense of empowerment is recognised as particularly important for complex diseases, such as mental illness, as genetic attributions to disease may result in fatalistic attitudes and feelings of hopelessness regarding the illness (Nuffield Council on Bioethics 2002, Alper and Beckwith 1993, Rose 1995, Chakravarti and Little 2003, see figure 15b) which may deter from uptake of health-related behaviours (Nuffield Council on Bioethics 2002, Chakravarti and Little 2003, Walter et al. 2004). As fatalistic viewpoints have been reported amongst members of families affected by psychiatric illness (Biesecker and Peay 2003, Peay et al. 2009), it has been discussed that PGC may be especially helpful in this regard (Austin and Honer 2005, Finn and Smoller 2006, Austin and Honer 2007, Peay et al. 2008), through helping affected individuals accept that "genes are not necessarily destiny," (Papdimitriou and Dikeos 2003 p. 240) and that environmental factors can have a substantive protective effect.

That PGC may be helpful in facilitating better management and protection of mental health is now supported by some outcomes data. In a pilot study, parents of affected individuals reported that PGC gave them hope, including in regards to their child's recovery and managing their mental illness (Austin and Honer 2008). Additionally, a study evaluating impact of PGC found significant increases in Empowerment, as measured by the GCOS scale, and also Self-Efficacy as measured by the Illness Management Self-Efficacy Scale, which measures an individuals' confidence to self-manage the illness (Inglis et al. 2014). There is, however, no currently available outcomes data in regards to the impact of PGC on health-related behaviours specifically, although this is currently being explored by groups (Austin and Inglis 2015, pers comms, 12th February).

Goals of PGC: increasing understanding of familial recurrence risk

Providing information about familial recurrence risks has been identified as another important goal of PGC (Tsuang et al. 1994, Hodgkinson et al. 2001, Austin and Honer 2005, DeLisi and Bertisch 2005, Austin and Peay 2006, Austin et al. 2006, Finn and Smoller 2006, Hill and Sahaar 2006, Austin and Honer 2007, Austin et al. 2008, Austin and Honer 2008, Peay et al. 2009, Hunter et al. 2010, Costain and Bassett 2012, Gershon and Alliey-Rodriguez 2013, Costain et al. 2014a, Costain et al. 2014b).

Using family history, genetic counsellors provide a tailored risk estimate for psychiatric illness (Tsuang 1994, Hogkinson et al. 2001, Papdimitriou and Dikeos 2003, Austin and Peay 2006, Austin et al. 2008). Given the lack of clinically available genetic testing, this remains the most accurate method of assessing risk for psychiatric illness (Merikangas and Risch 2003, Finn and Smoller 2006, Hunter et al. 2010, Meiser et al. 2013).

Theoretically, obtaining a more accurate perception of risk can be helpful for a number of reasons. Firstly, risk assessments may assist in decision-making around family-planning (Finn and Smoller 2006, Austin et al. 2008, Hunter et al. 2010, Costain and Bassett 2012), as well as decisions around other important lifestyle choices and behaviours which may in turn reduce risk for illness development, facilitate better management of the condition, and promote mental well-being amongst both affected and unaffected individuals (Austin and Honer 2005, Austin et al. 2008, Meiser et al. 2013).

Additionally, it has been proposed that more accurate perception of familial risk may be useful in further enhancing perceived personal control of risk and thus enhancing empowerment (Austin et al. 2008, Peay et al. 2009), as well as also helping to further reduce feelings of guilt, shame and blame within families e.g. through providing reassurance that parenting or personal life choices did not play a major role in causing the illness (Costain and Bassett 2012).

Studies that have explored perceptions of risk amongst affected individuals and their relatives have generally reported that there are misconceptions about genetic risk, although there has been some inconsistencies in findings. Some, earlier, study groups reported that affected individuals with SZ or BPD underestimated risk for

family recurrence (Targum et al. 1981, Schulz et al. 1982); conversely, other groups have reported that affected individuals overestimate risk (Trippitelli et al. 1998, Quaid et al. 2001, Costain et al. 2014a); and that overestimation of genetic risk may be associated with reproductive decisions favouring fewer or no children (Austin et al. 2006, Meiser et al. 2007, Wilde et al. 2010, Meiser et al. 2013). Relatives of affected individuals have been reported to overestimate risk (Targum et al. 1981, Schulz et al. 1982, Austin et al. 2006, Costain et al. 2014b).

Additionally in a study amongst parents of affected individuals, Austin and Honer (2008) reported familial risk was a source of anxiety and worry for respondents, with 84% of participants (n=11) identifying that they were concerned about other relatives becoming ill. However there is, to best knowledge, no other evidence that has researched implications (i.e. rather than risk *perceptions)* of familial risk amongst affected individuals and their relatives.

Thus, the findings from studies indicate that misconceptions about familial risk exist amongst affected individuals and their families, and that it may be a source of concern for individuals affected with psychiatric conditions and their relatives. It has therefore been hypothesised that PGC may be helpful in addressing this, as well as having other important outcomes (Austin 2005, Austin et al. 2006)

This supposition is now supported by some data from outcomes studies, demonstrating that PGC facilitates more accurate perception of risk amongst affected individuals and their relatives (Costain et al. 2014a, Costain et al. 2014b); and that it reduces concern about other relatives becoming ill, most likely due to increased understanding of risk (Austin and Honer 2008). There is a need for further research in this regard.

Goals of PGC: Providing information, support, and facilitating decision-making around genetic testing.

It has been explored that PGC may also be useful in providing information, support, and decision-making around genetic testing.

PGC provides a clinical framework for patients to discuss the potential impact of genetic information prior to testing, and for support in interpreting results. Which may help patients make informed decisions regarding testing, facilitate comprehension of results, and provide supportive counselling around the new information, and uncertainty that may stem from the test results (Austin et al. 2006, Wray and Visscher 2010, Ram et al. 2012). PGC may also ease increased demands on practitioners, who may not have the time, experience and/or knowledge to interpret test results (Wilde et al. 2010, Salmm et al. 2014).

Although clinical testing is not currently available, some groups have proposed it may become a possibility for future genetic risk prediction (Hodgkinson et al. 2001, Wray and Visscher 2010).Specifically, there is a particular research focus on copy number variants, which have a higher penetrance than common variants (Collier, St Clair, Vassos, Kirov, Gershon and Alliey-Rodriguez 2013), although the clinical efficacy remains to be determined (Collins 2010 Wray and Vischer 2010)

Indeed, there has been concern raised about the potentially harmful effects of genetic testing including living with uncertainty if genetic testing could only indicate probability and not certainty of developing illness, which may raise levels of anxiety (Meiser et al. 2005, Hippman et al. 2013), trigger negative behavioural changes by family members towards affected individuals, and especially children, who may be labelled 'at-risk' (Meiser et al. 2005, Finn and Smoller 2006), and potentially influence important life decisions including marriage and reproductive choices (Meiser et al. 2005).

Testing in relation to psychiatric diagnoses opens many ethical concerns, but the question of whether or not testing will, and should, become clinically routine is somewhat overshadowed by the rise of direct-to-consumer testing (Collins 2010, Wray and Vischer 2010). Given that high hypothetical demand for genetic testing for psychiatric conditions has been reported (Smith et al. 1996, Jones et al. 2002, Meiser et al. 2005), it has been proposed that this puts an emphasised need to provide GC for this population (Austin et al. 2006, Austin and Honer 2007, Wilde et al. 2010).

Goals of PGC: Addressing stigma

Studies have proposed that PGC may also be helpful in addressing mental illness-related stigma.

It is well established that uncertainty and myths regarding the origin of mental illness can contribute to stigma (Austin and Honer 2005, Meiser et al. 2013 Costain et al. 2014b). On this basis it has therefore been proposed that improving understanding about what is understood about the causes of mental illness may therefore help dispel misconceptions and reduce uncertainty and fear, to alleviate stigmatising attitudes, and therefore that GC may be helpful in this regard (Papdimitriou and Dikeos 2003, Austin and Honer 2005, Hill and Sahaar 2005, Austin and Honer 2007, Lyus 2007, Peay et al. 2008, Meiser et al. 2013, Costain et al. 2014b).

For example, attributing mental illness to sources such as stress or life experiences may help 'de-mistify' the illness which may result in less social avoidance and discrimination (Mechanic et al. 1994, Martin and Pescosolido 2000, Corrigan et al. 2003).

Genetic attributions to mental illness may also reduce stigma (Meiser et al. 2005, Austin and Honer 2005, Hill and Sahaar 2006, Austin and Honer 2007, Costain and Bassett 2012, Gershon and Alliey-Rodriguez 2013), as it may move 'the locus of control and responsibility away from the individual towards the role of hereditary' (Meiser et al. 2005).

Furthermore it has been explored by some groups that, on a population-level, PGC may help address possible negative relationships between neurobiological explanations for mental illness by providing public education which may enhance understanding and dispel misconceptions (Costain et al. 2014b). In this sense, groups have proposed that GC may empower affected individuals and families to share knowledge with relatives, friends and peers, which may reduce both perceived and experienced stigma (Austin and Honer 2005, Austin and Honer 2007, Austin and Honer 2008).

Consistent with hypotheses, there is now some available outcomes data linking PGC with decreases in stigma for affected individuals (Costain et al. 2014a) and family members of adults with SCZ (Costain et al. 2014b). Additionally, reported increases in self-efficacy and empowerment following PGC (Austin and Honer 2007, Inglis et al. 2014), which are frequently thought of as being the opposite of internalised stigma, have also been reported, this, it has been proposed, providing further support that GC may reduce stigma (Inglis et al. 2014).

1.5.2 The PGC session

Although a nascent discipline, a relatively clear picture regarding the basic concepts of PGC has been established with groups showing overall agreement in their approach (see for example: Tsuang et al. 1994, Papadimitriou and Dikeos 2003, Austin and Honer 2005, Austin and Honer 2008).

Numerous studies have surmised that GC for psychiatric conditions is very similar to that for other complex disorders (Biesecker and Peay 2003, Austin and Honer 2008) in that it is a dynamic process that involves both information gathering and provision, and provision of support and counselling (Austin and Honer 2007, Austin and Honer 2008). As in GC for physical disorders, typical stages of the process involve: information gathering, information provision and support, supportive decision-making.

However it has also been acknowledged that PGC sits within a more challenging context: aetiological understanding is limited, recurrence risk provision is difficult, and there are considerable psychosocial implications of mental illness for relatives and families (Austin and Honer 2005, Finn and Smoller 2006, Austin and Honer 2007).

It has been thus postulated that resultantly clinicians may feel uncomfortable and/or inexperienced to discuss psychiatric illness with patients, which may be, partially, why GC for psychiatric conditions is not practised (Peay et al. 2008). Out of awareness of this there is an albeit small but growing number of online resources now available to guide clinicians not necessarily trained in psychiatry in regards to the provision of PGC (for example: Genetic Alliance 2008, NSGC 2015 ; NCHPEG 2015). Additionally Peay et al. (2008) have proposed a guiding framework for GC and clinicians (see table 4), partially out of growing recognition that they may feel uncomfortable or inexperienced to engage in such discussions.

Table 4: Suggested framework for guiding discussions regarding patient's psychiatry history in a clinical setting

(From Peay et al. 2008, p.11)

Question to consider	Discussion
Is a personal or family history of a psychiatric disorder the reason for indication?	Yes: Always discuss in detail
Does the client actively bring up a personal or family history of a psychiatric disorder as an area of interest?	Yes: Always discuss, level of detail varies based on indication and competing interests in the session; can simply make referral
Is the personal or family history of psychiatric disorder immediately relevant to diagnosis, risk, or decision making? Example 1. A pregnant woman on a psychiatric medication: psychiatric illness may affect decision making and perceived risk Example 2. Evaluation of a child with behavioral issues, including ADHD: in this case, a family history of bipolar disorder might affect differential diagnoses	Yes: Always discuss in relevant detail; reproductive counseling in a woman with a personal or family history of a mood/psychotic disorder, post-partum depression (PPD), or post-partum psychosis (PPP) should always include a discussion of risk for PPD or PPP
Is the family history ancillary to the indication and of limited concern to the client?	Yes: Indicate that risk may be increased for others in the family, and offer referral; NOTE: When the client has a striking family history, increase the emphasis on risk to other family members and consider shifting the session's focus to include psychiatric risk assessment
In your practiced professional opinion, will a discussion of psychiatric history detract from something that is time-limited and important?	Yes: Mention hereditary nature of psychiatric illness, and consider making a referral for later discussion

Information Gathering

The information gathering process of PGC typically involves identifying the patient's needs and concerns and obtaining family history and individual's psychiatric history (Tsuang et al. 1994, Hodgkinson et al. 2001, Papadimitriou and Dikeos 2003, Austin and Honer 2007).

During the information gathering process it is important to establish the patient's reasons for receiving the genetic information, both in terms of the information they wish to obtain and reasons behind it (Tusang et al. 1994, Papadimitriou and Dikeos 2003, Austin and Honer 2007), to identify needs and expectations (Hodgkinson et al. 2001, Finn and Smoller 2006). Groups have also discussed the importance of identifying the patient's existing disease construct and potential misconceptions and uncertainty around this (Hodgkinson et al. 2001, Papdimitriou and Dikeos 2003, Austin and Honer 2005, Austin and Honer 2007).

Family history for psychiatric illness should be taken along with other important health information (e.g. substance abuse, age of onset of symptoms, history of psychiatric hospitalisations) (Hodgkinson et al. 2001, Finn and Smoller 2006). Family history for other physical conditions should also be taken which may identify presence of another underlying genetic condition causing psychiatric symptoms, such as 22q syndrome, which will influence recurrence risk (Finn and Smoller 2006, Austin and Honer 2007).

Assessment of the patient's emotional, psychological and intellectual capacity is important to ensure the patient is able and well enough to receive the information (Tsuang 1994, Papadimitriou and Dikeos 2003, Austin and Honer 2007).

Information provision and support

Information about aetiology should include discussions about both genetic and environmental factors in psychiatric pathogenesis (Papdimitriou and Dikeos 2003, Austin and Honer 2007).

Throughout this process it has been highlighted that it is important to address misconceptions expressed regarding aetiology (Papdimitriou and Dikeos 2003, Austin and Honer 2007). Emotional aspects of both misattributions and of newly learned genetic information, e.g. fatalistic attides/genetic determinism, should also be discussed and explored (Papdimitriou and Dikeos 2003), as should concepts of uncertainty stemming from the limitations of current aetiological understanding which can have emotional implications for patients (e.g. anxiey, feelings of disempowerment) (Austin and Honer 2007, Peay et al. 2008).

It has been discussed in the literature that provision of recurrence risks is recognised a particularly challenging element of PGC (Hodgkinson et al. 2001, Finn and Smoller 2006, Austin and Peay 2006, Austin and Honer 2007). Firstly, concepts relating to familial risks are often a source of considerable anxiety and stress for many patients (Hodgkinson et al. 2001, Austin and Honer 2005, Finn and Smoller 2006) and it is thus important to clarify what the patient wishes to know (Austin and Honer 2007).

Additionally perception of risk is a complex and subjective process and so risk information should be conveyed in several formats (Hodgkinson et al. 2001, Hill and Sahaar 2006, Austin and Honer 2007, see table 5), as for other genetic counselling settings, and the genetic counsellor should also be unbiased in their delivery of risk information to prevent influencing the patient's perceptions (Hodgkinson et al. 2001, Hill and Sahaar 2006).

It is also paramount that the limitations of risk assessment for psychiatric illnesses based on empirical data are discussed, and resultant emotions explored and counselled around (Papdimitriou and Dikeos 2003, Hill and Sahaar 2006, Austin and Honer 2007, Austin et al. 2008).

Table 5: Suggested formats for presenting risk information in PGC

(From Austin et al. 2008, p.20)

	Morbid Risk[a]	Risk Ratio[b]
Expressed as:	A percentage (X%)	A ratio (1 in X)
Range:	0–100%	Negative to positive infinity
Provides information about:	The probability of a disorder occurring in a certain type of relative (e.g. sibling)	How much more likely it is for a biological relative of an affected individual to become affected, as compared to the general population risk
Example of using this type of risk information in context for first-degree relatives:	The risk for the disorder in first-degree relatives of affected individuals is X%	The risk for the disorder is X times greater among first-degree relatives of affected individuals than in the general population
Aggregation in families is indicated if:	Risks to relatives of affected individuals are greater than risks to the control group	Value is greater than 1

[a] It is important to notice whether or not morbid risks are age corrected—age corrected risks are more accurate, because there may be a period of several years during which a person is at risk, which typically begins in the late teens (Nurnberger and Berrettini 1998).
[b] Risk ratio information can put morbid risk into context. For example, a high morbid risk will not seem so genetic if the risk ratio is low due to high population prevalence. In contrast, a small morbid risk will seem very genetic if the risk ratio is high due to low population prevalence.

Support and decision-making

Relevant decisions patients may have to make may include treatment or medication options during pregnancy (Austin and Honer 2005), family planning (Hodgkinson et al. 2001, Papdimitriou and Dikeos 2003), or whether to undergo genetic testing (Hodgkinson et al. 2012).

It is important than the genetic counsellor helps affected individuals make informed decisions that are in line with their cultural, ethnic and religious backgrounds, and their individual or family goals (Tsuang 1994, Papdimitriou and Dikeos 2003, Austin and Honer 2005); and provides support and helps the patient adjust to their decision (Hodgkinson et al. 2001, Austin and Honer 2005, Austin and Honer 2007).

It is fundamental that the genetic counsellor is non-directive and promotes the autonomy of the patient in regards to facilitating decision-making (Tsuang 1994, Hodgkinson et al. 2001, Papdimitriou and Dikeos 2003, Austin and Honer 2005, Finn and Smoller 2006, Austin and Honer 2007). It has been asserted that this may be especially important in regards to providing GC for psychiatric conditions, to prevent potentially advocating (even unintentionally) stigma and discrimination against individuals with a mental illness (Austin and Honer 2007).

Follow-up

Follow-up typically involves sending a summarising document of the session to the patient. It is an important component of all genetic counselling interventions, including PGC (Tsuang et al. 1994, Hodgkinson et al. 2001, Finn and Smoller 2006). It provides the opportunity to reinforce the information covered (Tsuang et al. 1994, Hodgkinson et al. 2001, Hill and Sahaar 2006), facilitates sharing of information with the patient's family and/or clinician, and encourages the patient to contact the genetic counsellor if any new questions or new diagnostic information arises, which may require revision of recurrence risks (Tsuang et al. 1994, Hodgkinson et al. 2001).

1.5.3 PGC – Indications of a need for its implementation

Available evidence indicates that currently many healthcare practitioners do not feel prepared to deliver psychiatric genetic information.

For example, surveys of psychiatrists have reported limitations in knowledge regarding both medical and psychiatric genetics (Finn et al. 2005, Hoop et al. 2008, Klitzmann et al. 2014), and also uncertainty regarding testing options and interpretation (Klitzmann et al. 2014). Despite this, affected individuals with psychiatric illness are rarely referred to specialist genetics services such as GC (Hodgkinson et al. 2001, Austin and Honer 2007, Hunter et al. 2009, Costain et al. 2014a, Austin 2014, pers. comm., 14 November).

Additionally, a recent study also reported that the majority of neurologists and psychiatrists that ordered genetic tests did not have access to a genetic counselling service (Salmm et al. 2014).

Furthermore, studies amongst genetic counsellors have found that they would not feel prepared (Peay and McInnerney 2002) or comfortable (Feret et al. 2011, Martin et al. 2012) to provide GC for psychiatric disorders, largely due to a lack of familiarity with the illnesses (Peay and McInnerney 2002) and stigma related to psychiatric illness (Feret et al. 2011, Martin et al. 2012).

On the basis of such evidence it has been proposed that a gap seemingly exists in the provision of genetic and aetiological information to affected individuals and their families, and some research groups have asserted that this may mean that, currently, medical informational needs of patient may not be being met (Finn and Smoller 2006, Meiser et al. 2013).

Further research into concepts relating to i) informational needs of prospective patients and ii) knowledge and perceptions of healthcare providers may thus be helpful in aiding further exploration.

1.5.4 Demand for PGC

Interest and uptake.

Evidence exploring interest in receiving genetic counselling for psychiatric conditions amongst the American and Canadian populations have generally demonstrated that PGC is favourably viewed by potential future service-users. These studies have typically explored interest by presenting respondents with the hypothetical situation of receiving PGC, and the majority of respondents (~62-75%) consequently indicating that they would wish to receive the service if it were available and/or that they believed it would be helpful (Schulz 1982, Quaid et al. 2001, DeLisi and Bertisch 2006, Lyus 2007).

Furthermore, some studies have demonstrated relatively high uptake of the service when offered, providing further evidence that PGC is positively regarded (Austin and Honer 2008, Costain et al. 2014a, Costain et al. 2014b).

There is no published data regarding interest amongst the UK population specifically in regards to PGC and so no indications of a demand for, nor even interest in receiving, the service. Such data is important because evidencing demand is particularly important in justifying new healthcare interventions, especially in the age of austerity in which mental health services are facing such spending cuts (Costain et al. 2014a, Costain et al. 2014b).

Awareness and perceptions of GC and PGC

Other concepts relating to interest in PGC are awareness of the service and perceptions of its purpose.

There are speculations and some available evidence that awareness is low and that there are misconceptions about GC and especially its role within psychiatry, amongst both prospective service-users and clinicians (Lyus 2007, Hunter et al. 2010). Further, this has recently been proposed as a fundamental reason for low rate of referrals to a British Columbia Provincial Medical Genetics Program (BCPMG), which provides the world's first specialised PGC clinic (Hunter et al. 2010).

In support of this, a study conducted in 2007 that explored perceptions of PGC amongst the American population, reported low awareness of the service, with only 28% (n=19) of affected individuals with SCZ and 48% (n=71) of relatives having previously come across GC (Lyus 2007). Additionally, the study found that misconceptions amongst respondents regarding GC and PGC, were common, including false beliefs that a GC could provide genetic testing for SCZ, and a number of respondents reporting they did not believe a GC would provide emotional support.

More widely, that there is low awareness and misconceptions regarding GC has been reported by other study groups assessing perceptions in other fields of GC. For example, retrospective studies that have surveyed affected individuals who have received GC have found that they attended the session typically unaware of the content and structure (Hallowell et al. 1997, Bernhardt et al. 2000, Metcalfe et al. 2007) and specifically that the counselling aspect of the session came as a pleasant surprise (Bernhardt et al. 2000).

Additionally, a study *prospectively* studying awareness and perceptions of GC amongst the Canadian general population found that 69% of affected individuals had not heard of GC, and that a substantial proportion had misconceptions about its purpose, particularly relating to perceptions that GC involves prevention of inheritable diseases, helping couples to have desirable characteristics, and advising couples whether to have children, indicating perceptions that GC is based on Eugenic-type values (Maio et al. 2013).

Concepts regarding awareness and perceptions of genetic counselling are important because research has demonstrated they can impact engagement and also patient outcomes (Brown et al. 1999, Pieterse et al. 2005, Joseph et al. 2010, Albada et al. 2012a, Albada et al. 2012b, Maio et al. 2013).

For example, GC can result in better outcomes when clients have a better understanding of what to expect (Brown et al. 1999, Pieterse et al. 2005, Joseph et al. 2010, Albada et al. 2012a, Albada et al. 2012b, Maio et al. 2013). This may reduce anxiety (Austoker and Ong 1994. Hallowell et al. 1997, Davey et al. 2005, Metcalfe et al. 2007) which may enable the client to be a more active participant in the session; build rapport with the genetic counsellor; have a more active role in decision-making (Metcalfe et al. 2007); and enable the genetic counsellor to ensure the client's needs are met (Pieterse et al. 2005, Babul-Hirji et al. 2010, Albada et al. 2012b, Maio et al. 2013), leading to overall better outcomes and adaptation to the illness (Hack et al. 2005, Metcalfe et al. 2007). Better awareness of the GC process may also help affected individuals better prepare in advance, for example formulating questions individual to their own circumstances (Hallowell et al. 1997, Brown et al. 1999, Metcalfe et al. 2007), enabling them to use the session more effectively.

1.5.5 PGC: Criticisms and controversies

Opposition to biogenetic models in psychiatry – stigma, and eugenics

In the modern day, the majority of academics and clinicians within the mental health field embrace a holistic approach which acknowledges both genetic and environmental models of illness and treatment, and the previous dichotomy of the 'nature or nurture' conflict in psychiatry (for example, see Kessler 1980, Chakravarti and Little 2003) is, generally, regarded accepted as over-simplistic (Rutter 2006, Jaffee and Price 2007, Peay et al. 2008). Considering both genetic and non-genetic contributions in the underpinnings of human behaviour is widely accepted as the most accurate and clinically helpful explanation for mental illness (Phelan 2002, Phelan 2005, Austin 2015 pers comm.,13th April , Mayers 2015 pers comm., 14th April) and, for GC, the best approach to take to ensure best outcomes for patients (Austin and Honer 2007, Meiser et al. 2007, Peay et al. 2008, Mesier et al. 2013).

However, some research groups have expressed strong concern, or outright objections, over adoption of genetic and/or medical approaches to mental illness (Conrad 1992, Beresford and Wilson 2002, Membis 2009, Beresford 2015 pers comm., 24th April). These objections stem predominantly from concern that such approaches may exacerbate stigma, prejudice and discrimination (Haslam 2000, Beresford and Wilson 2002, Membis 2009, Howell et al. 2011), and may induce adoption of divisive mentalities (Bennett et al. 2008), i.e. an 'us and them' way of thinking. Furthermore, there have been concerns raised that such approaches could form a fundamental basis for discrimination and even re-emergence of eugenics-based policies against individuals with mental illness (Nuffield Council on Bioethics 2002).

Indeed, there is a deep and ugly historical link between psychiatry and the eugenics movement of the early 20th Century (Nuffield Council on Bioethics 2002). Influenced by the breeding programs of domesticated plants and animals, in which humans altered characteristics of species by replacing natural reproduction with artificial selection so they adapted specific characteristics in order to subsequently meet human needs (Brüne 2007), some researchers, clinicians and politicians came to believe that 'self-domestication' of humans could be achieved (Brüne 2007, p.1), whereby the qualities of the human race could be improved by selective breeding (Nuffield council on bioethics 2002). Resultantly, in the false belief that they might

therefore be able to reduce the presence of 'bad genes' within the human population and prevent so-called degeneration of 'erbgut,' or genetic material (Brüne 2007, p.1), they came to not only toy with the idea, but actually conduct genetic experimentations and approaches in humans. The results had profound and devastating consequences.

The eugenics movement became more wide-spread across the US, Europe and elsewhere, with eugenic explanations being used to justify discriminatory doctrines, policies and practices against those deemed as having undesirable characteristics, including mental illness , as well as the poor and the physically disabled (Kevles 1985, Dikköter 1998, Phelan 2002). Through marriage restrictions, involuntary sterilisation, segregation and institutionalisation, the reproductive rights of tens of thousands of people of societies' most stigmatised individuals were not only controlled but sometimes completely diminished, purely on the basis that they were judged to be genetically inferior (Dikköter 1998, Phelan 2002, Nuffield Council on Bioethics 2002).

Ultimately, the idea of improving the qualities of the human race by selective breeding was used in partial justification of the genocide of individuals –including thousands the mentally ill - under the Nazi regime (Nuffield Council on bioethics 2002). Thus, even though carried out by politicians, the fundamental basis for these policies was provided by scientists (Harper 2010), explaining the entrenchment between science, and especially genetics, and eugenics (Nuffield Council on Bioethics 2002).

Indeed many early clinicians, especially geneticists, openly supported eugenics (Harper 2010), and five of the six first presidents of the American Society of Human Genetics served on the board of the American Eugenics Societies during their presidencies (Paul cited Resta 1997). Although they were typically critical of the *method* of eugenics programs, there was widespread consensus that eugenic goals of 'improving' the human race could be achieved via genetic approaches (Resta 1997, see figure 16 Nuffield Council on Bioethics 2002).

(Fig. 16)

'The improvement of human genetic quality by eugenic methods would take a great load of suffering and frustration off the shoulders of evolving humanity, and would much increase both enjoyment and efficiency. Let me give one example. The general level of genetic intelligence could theoretically be raised by eugenic selection; and even a slight rise in its average level would give a marked increase in the number of the outstandingly intelligent and capable people needed to run our increasingly complex societies.

How to implement eugenic policy in practice is another matter. The effects of merely encouraging well-endowed individuals to have more children, and vice versa, would be much too slow for modern psychosocial evolution. Eugenics will eventually have to have recourse to methods like multiple artificial insemination by preferred donors of high genetic quality.'[11]

Figure 16: Extract from 'Man and His Future,' 1963.

(Nuffield Council on Bioethics 2002, p.15, cited 'Man and His Future, 1963.)

In 1962 an international meeting of scientists was held to discuss anthropological and evolutionary concepts relating to the future of mankind. Eugenics were a major focus of discussions, as demonstrated by the above extract.

This association between genetics and eugenic-type values extended down to practice of GC. With typical outcomes involving reducing the occurrence of what were considered 'undesirable' characteristics by influencing high-risk families not to have children; and ensuring the upholding of societal racial boundaries through maintaining racial homogeneity within families (Stern 2012), its early, public health-centered practice, was sometimes hard to disentangle between eugenics (Resta 1997, Resta 2006, see figures 17 and 18).

(Fig. 17)

"These data are evidence that genetic counseling tends to have the desired effect; that is, to influence high risk families not to have further children… It is probably a long way off, but… the day may come when the effect of genetic counseling may well be felt in a significant way in the general population."

Murray 1968

Figure 17: Extract regarding GC by Robert F. Murray Jr, a paediatric geneticist at Howard University, 1968.

(Cited Resta 2006, p.271)

(Fig. 18)

"The counselor must not only be concerned with the specific problem in inheritance raised by a given family but must also attempt to make some assay of the total genetic endowment of the persons in question… most people would agree that it would be advantageous for reproduction to cease in a family producing successive crops of idiots and imbeciles… Generally,… advice concerning hereditary that is sound and advantageous for the individual family will also be found to be safe and advantageous for society as a whole."

Nash 1955

Figure 18: Extract regarding genetic counselling by Nash Herndon, an early president of the American Society of Human Genetics, 1955.

(Cited Resta 2006, p.270)

Psychiatrists, too, played a prominent role in the eugenics theorems and practices of this era. They were particularly interested in the theory of eugenics because it provided an explanation for what was then considered a degeneration of the human race - the seemingly increasing presence of psychiatric illness within the population (Brüne 2007), which did not seem fitting with evolutionary theory. The eugenic movement thus became closely linked to the study of hereditary in mental illness (Schulze et al. 2009), as psychiatric genetic research aimed to find a Mendelian form of inheritance to justify, some have argued, policies and practices that restricted their reproductive rights (Kösters et al. 2015).

Further associations between psychiatric genetics and eugenics were also formed by fact that leading profiles in the field of psychiatric genetics, such as Ernst Rüdin, Carl Schneider and Alfred Ploetz, became key protagonists of the movement and policies of eugenics and racial hygiene within Germany after the Nazi takeover (Ritter and Roelcke 2005, Roelcke 2007). These figures served as the 'Expert Committee on Questions of Population and Racial Policy' under Reich Interior Minister Wilhelm Frick. The Committee's policies for so-called 'racial hygieneity' towards the development of any Aryan race involved mass sterilisation and extermination of men, women and children with physical disabilities, learning disabilities, and serious psychiatric illness, and especially SCZ (Ritter and Roelcke 2005, Roelcke 2007). Indeed, Rudin had a particular interest in SCZ, which he believed to be caused by a Mendelian-recessive gene, and his theories of inheritance subsequently inspired many researchers, clinicians and politicians that were involved in the development of mass sterilisation and extermination policies of individuals with SCZ (Fuller Torrey and Yulken 2010). It is estimated that between 220, 000 and 270,000 individuals with SCZ were sterilized or killed, respresenting between 73% and 100% of all individuals with SCZ living in Germany between 1939 and 1945 (Fuller Torrey and Yulken 2010).

Whilst behavioural genetics formed a fundamental basis of eugenic policies, it has been asserted that this does not imply that contemporary approaches to behavioural genetics is in any driven by eugenic theories nor associated with eugenic values, and indeed genetics research today is heavily regulated by ethical research bodies (Nuffield Council in Bioethics 2002). However, there still remains views amongst some, especially in regards to hereditary of intelligence, that such research remains *fundamentally* eugenic, and concerns that it could lead to the re-establishment of eugenic policies (see figure 19).

(Fig. 19)

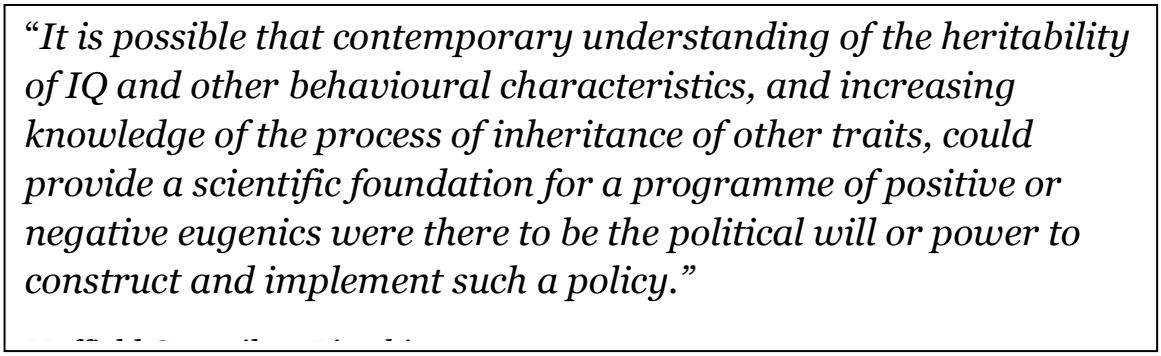


Figure 19: Nuffield Council on Bioethics' statement regarding link between eugenics and behavioural genetics research in context of contemporary practice.

(Nuffield council on Bioethics 2002, p.22)

The utility of PGC

The limitations of current aetiological understanding of psychiatric disorders, due to the complex archaeology, has also been explored as a potential barrier to the provision of PGC (Austin 2005, DeLisi and Bertisch 2006, Hippman et al. 2013), as it has led to some questioning of the clinical utility of the service.

For example, common variants explain a limited proportion of risk, their functional consequences are not yet understood, none are causative of illness, and there is overlap between diagnostic boundaries (Hippman et al. 2013). Additionally understanding of gene-environment interactions is in its infancy (Cornelis et al. 2010, van Os et al. 2010). These caveats limit the specificity of aetiological information that can be delivered to patients and also means comprehensive understanding and identification of protective factors in mental health – an important component of information provision during the PGC session - is somewhat limited (Papdimitriou and Dikeos 2003, Austin 2005).

Moreover, genetic testing does not meaningfully aid with risk estimation which reduces accuracy of risk information, figures for which are instead derived from empirical data and family history (Hippmann et al. 2013).

However, in turn, this process of estimation is clinically challenging due diagnostic uncertainty in regards to psychiatric diagnoses; incomplete psychiatric family histories: and genetic phenomena including reduced penetrance, variable expressivity and genetic heterogeneity (Hodgkinson et al. 2001, Costain and Bassett 2012, Costain et al. 2014b). Additionally, the presence of multiple distinctive psychiatric diagnoses within a family, and the presence of affected individuals on both sides of the family - not an uncommon phenomena - can make recurrence risk estimates challenging as in these cases, there is little empirical data to guide the clinician (Austin and Peay 2006).

In practice this means categorical answers are not available for many questions pertaining to aetiology, familial risk and risk-reduction strategies that patients and their families may have (DeLisi and Bertisch 2006) and therefore there is the possibility that the information provided during PGC may not only be unhelpful, but may even be confusing (Hippman et al. 2013), and may also induce anxiety due to lack of certainty (Peay et al. 2008).

Indeed a survey of genetic counsellors (Monaco et al. 2010) reported these concerns existed amongst clinicians in regards to the hypothetical provision of GC for psychiatric conditions, with some perceiving that the aetiology of psychiatric disorders as confusing for patients, and that incomplete explanations regarding causation would be frustrating for them.

However, in contrast, outcomes studies have indicated that patients accept the incomplete nature of aetiological understanding, and that PGC is still perceived to be useful in spite of the uncertainty (Austin and Honer 2008, Hippman et al. 2013). This has led to some promoting that these issues may be a projection of clinicians' concerns rather than reflecting patients' true perspectives (Hippman et al. 2013).

Furthermore, more widely in other aspects of medical genetics information on genetic risk and information very rarely provides categorical, 'yes' or 'no' answers (Smith et al. 2000, Harper 2010, Aasen and Skolbekken 2014), and indeed, GC is routinely available for other complex disorders for which clinical testing is not available and/or categorical answers cannot be provided (e.g. for patients carrying a variant of unknown significance), yet positive patient outcomes are still achieved, including for heart disease (Cirino et al. 2013) and breast cancer (Petrucelli et al. 2002, Culver et al. 2013). This has prompted, for some researchers and clinicians, the question as to why the absence of genetic testing, or indeed the inability to provide answers of greater certainty, should be a barrier for provision of GC for psychiatric illnesses when it is not for other physical illnesses (Hippman et al. 2013, Costain et al. 2014b).

1.6 Research Question: Exploring the application of PGC within the UK population

HYPOTHESES

Following the results of the literature search, the following hypotheses were deducted:

Amongst affected individuals and relatives within the UK…

1. **There will be uncertainty regarding aetiology of mental illness**

    Consistent with previous studies reporting that comprehensive understanding of pathophysiology is lacking and uncertainty exists amongst the American and Canadian populations (Hodgkinson et al. 2001, Holzinger et al. 2003, Austin and Honer 2005, Costain and Bassett 2012, Costain et al. 2014a)

2. **There will be uncertainties and negative implications of familial risk, including that:**

i). Genetic risk will be quantitatively overestimated

Consistent with findings of previous studies exploring perceptions of familial risk that have reported a tendency to overestimate risk to relatives (Targum et al. 1981, Schulz et al. 1982, Trippitelli et al. 1998, Quaid et al. 2001, Austin et al. 2006, Meiser et al. 2007, Wilde et al. 2010, Costain et al. 2014a)

ii). There will be high degrees of concern regarding risk of recurrence to relatives

Consistent with the study reporting high degree of concern amongst the siblings of affected individuals in the Canadian population (Austin and Honer 2008)

iii). There will be negative implications on family-planning decisions due to the mental illness

Consistent with findings that presence of mental illness is associated with reproductive decisions favouring fewer or no children (Austin et al. 2006, Meiser et al. 2007, Wilde et al. 2010, Meiser et al. 2013).

3. Awareness of GC will be low and there will be misconceptions about its process and purpose, including its role specifically within psychiatry

Consistent with findings of previous studies exploring awareness and perceptions of both traditional genetic counselling (Maio et al. 2013) and specifically PGC (Lyus 2007) amongst the American and Canadian populations respectively, which have found that typically less than half of respondents have previously heard of GC and that there are misconceptions about the service.
Also consistent with GC literature involving retrospective studies which have found that patients reported lacking comprehension of the content and structure of the session prior to attending the appointment (Hallowell et al. 1997, Bernhardt et al. 2000, Metcalfe et al. 2007

4. There will be hypothetical interest in receiving PGC.

Consistent with previous studies have indicated high interest rates, typically presenting respondents with the hypothetical situation of receiving GC following being provided information about the service (Schulz et al. 1982, Quaid et al. 2001, DeLisi and Bertisch 2006, Lyus 2007); and additionally studies reporting high rates of uptake of GC when offered (Austin and Honer 2008, Costain et al. 2014a, Costain et al. 2014b).

OBJECTIVES

In order to test the hypothesis, the following research objectives were consequently formulated:

i) To ascertain participants' attributions of the mental illness to i) genetic factors and ii) non-genetic factors, and measure respondents' confidence in their attributional explanations, to thus measure certainty

ii) To obtain respondents' perceived risk to first-degree relatives quantitatively, to determine accuracy of estimations of risk

iii) To explore implications of familial risk specifically in relation to respondents' concern for other relatives becoming ill, and reported impact on family-planning decisions.

iv) To explore participants' awareness of GC and beliefs about its purpose, including specifically within psychiatry

v) To determine if there is hypothetical interest in receiving PGC, by querying whether respondents would wish to receive PGC and whether they believed it would be useful to them

2. Materials and Method

Materials

Participant information sheet and participant consent forms were constructed in line with Bournemouth University's Research Ethics Code of Practice. An information document ('Cover letter') was also produced which briefly explained the rationale behind the study (See Appendices).

An online 'landing page' for the research project was developed containing a short video presented by the researcher explaining the study rationale, and a link that directed participants to the survey. It also featured downloadable PDF versions of the Participant Information Sheet, Consent Form, and Cover Letter. These are contained in the appendices.

(Fig. 20)

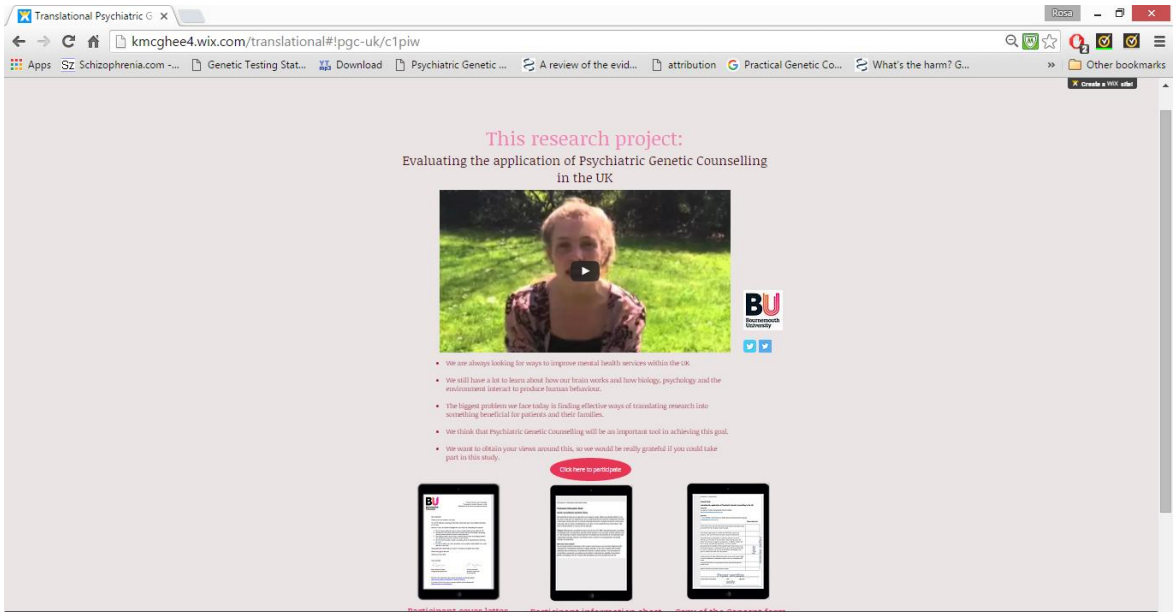


Figure 20: Screenshot of Online landing page developed for survey

(Source: Spencer-Tansley and McGhee 2015)

An additional video was produced for the survey. The script was devised and presented by Dr. Jehannine Austin PhD, MSc (Genetic Counselling), CCGC/CGC and explained the purpose and typical process of a PGC session.

The script is in included in the appendices (Appendix G). The video may also be viewed at: https://www.youtube.com/watch?v=PqnxqMnPk_g

The video was produced at Bournemouth University by Dr. Julio Montenagro.

Method

Ethics

In line with Bournemouth University (BU) Research Ethics Code of Practice (RECP), the research protocol was submitted to Bournemouth University Research Ethics Committee on 28th January 2015. A favourable opinion was reached on 11/03/2015.

The research did not require external review through the NHS National Research Ethics Service (NRES).

As the study did not involve the collection of personally identifiable data the study did not fall under the auspices of the Mental Capacity Act.

The panel was in agreement that given that participants had a copy of the participant information sheet and signed the participant agreement form (formerly consent form), then due consideration of ethical issues was offered and consent obtained, thus demonstrating informed consent.

This was clarified with the ethics panel, retrospective of the study, on 25/02/2016.

Participants

Participants were identified as any individual who has been diagnosed with a mental illness; or any relative of an individual who has been diagnosed with a mental illness.
Inclusion criterion regarding psychiatric diagnosis were: psychosis (SCZ, BPD with psychosis), mood disorder (BPD, MDD) anxiety disorders (OCD, depression with anxiety), eating disorders (anorexia, bulimia).
Whilst the majority of PGC literature has focussed on providing GC for psychotic disorders, there is growing consensus that it has wider applications and indeed is now being provided for other, common mental disorders including OCD and depression. The decision to thus include respondents with anxiety disorders and eating disorders in this research study was made, following discussions with **Dr J. Austin,** PhD, CCGC, Associate Professor, UBC Department of Psychiatry and Medical Genetics; **E. Morris**, CCGC, MSc, Clinical Instructor, University of British Columbia, Departments of Psychiatry and medical genetics; and **H. Andrighetti**, CCGC, CCGC, MSc, Clinical Instructor, University of British Columbia, Departments of Psychiatry and Medicial Genetics, University of British Columbia (UBC), Canada.

Neurodevelopmental disorders, classed as 'disabilities in the functioning of the brain that affect a child's behaviour, memory or ability to learn' which includes mental retardation, dyslexia, attention deficit hyperactivity disorder (ADHD), learning deficits and autism' (WHO 2010) were not included in the inclusion criterion as it was felt GC for these disorders may have very different constructs and also patients may have different needs and perspectives in comparison to those with 'mental disorders.'. Thus, research regarding provision of PGC for these disorders, whilst important, should be conducted separately.

Illnesses defined as 'mental disorders' were based on the ICD-10 classification system (WHO 2010).

All participants were also required to be over the age of 18.
All research participants were required to be from the UK or Ireland.

Literature search

PubMed database was searched to enable simultaneous searching of PubMed, PubMed Central, and MEDLINE. PubMed was used as it comprises more than 25 million citations for biomedical literature from MEDLINE, life science journals, and online books, and so it was believed that relevant journal articles would be identified.

Google scholar was also used as a search database to detect any publications that may have been missed by PubMed.

Search terms used across both databases were:

PSYCHIATRIC GENETIC COUNSELLING

GENETIC COUNSELLING and MENTAL ILLNESS

GENETIC COUNSELLING and PSYCHOSIS

PERCEPTIONS OF GENETIC COUNSELLING

RISK PERCEPTIONS GENETIC COUNSELLING

Certain inclusion criterion for the searches was applied. This included that only articles written in English were selected, and that, of the journal articles, only those that were peer-reviewed were considered. No limitations were applied in terms of publication date due to the limited papers regarding PGC. Furthermore it was felt that it would be interesting and important to consider beliefs, attitudes and findings about PGC over time, as there may be changes.

The literature search was conducted between October 2014 – October 2015 and so publications after this date could not have been included.

Publications were screened to assess relevance; those that were deemed to be irrelevant to the search were excluded.

Full texts of relevant articles were subsequently obtained and stored to a reference and citations manager (Mendeley desktop). The reference lists of these studies

were screened to identify any relevant publications that may have been missed from the database search process, or that were relevant to the findings of the search.

Relevant publications and articles were also suggested to the researcher from clinicians and researchers working within the relevant fields, including Dr K McGhee, PhD, Bournemouth University; Dr. J. Austin, PhD, CCGC, UBC; Dr. F Dagenhardt, M.D, University of Bonn,; Dr M. Nothen,.PhD, University of Bonn. These articles underwent the same screening process as those retrieved via database search.

Clinical tools used in PGC sessions by Dr J. Austin's research team were also sent to the researcher (see appendix H).

The remaining publications were used to conduct a literature review of the findings.

The method undertaken for the literature review is depicted in figure 21.

(Fig. 21)

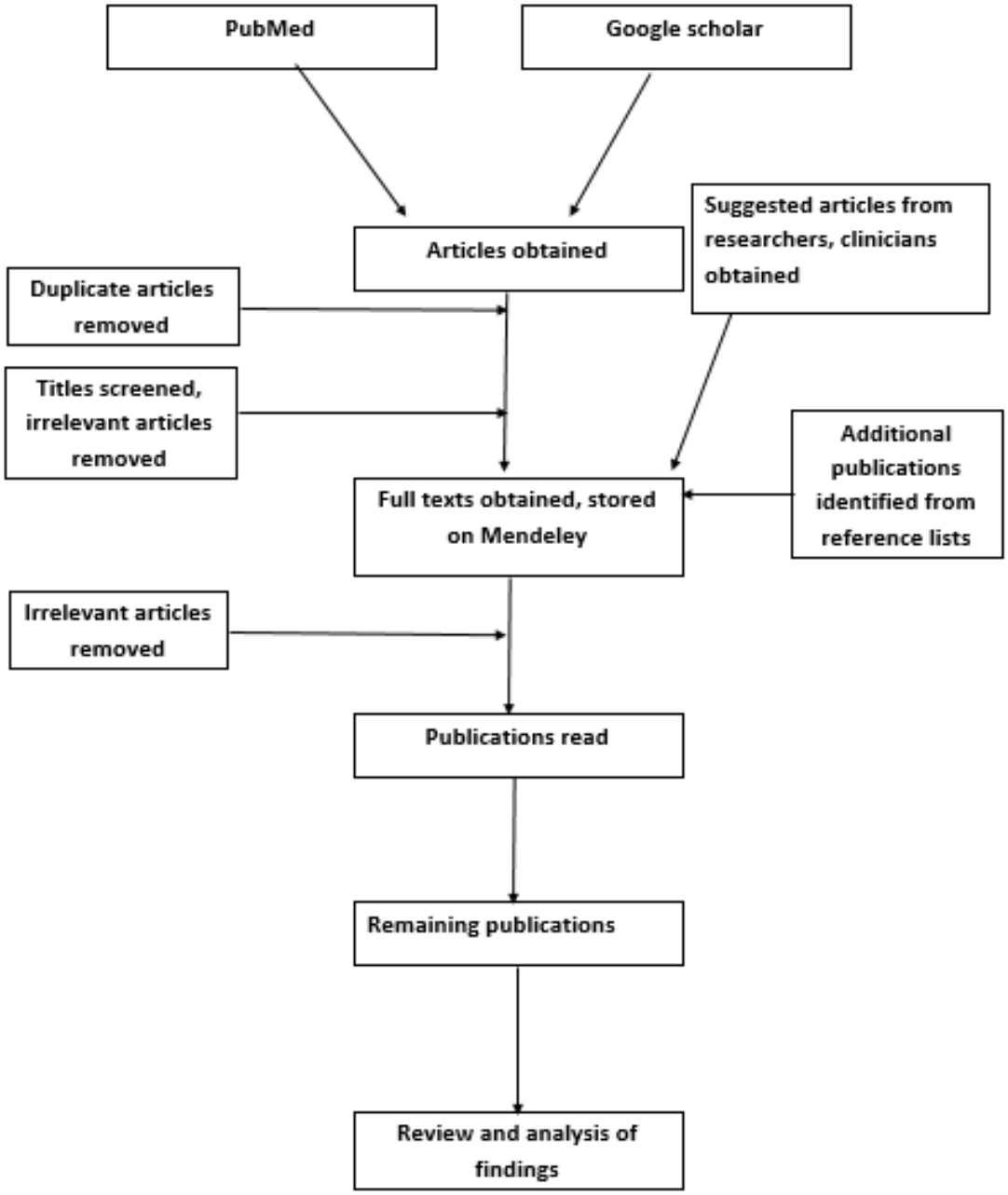


Figure 21: Diagram depicting method conducted for literature search

Choice of methodology

It was decided a mixed-methods approach would be the most effective in addressing the research aims and objectives.

There are several benefits to using mixed methods approach in research. Mixed methods enable breadth and depth of understanding and corroboration (Johnson et al 2007). Often one type of approach will not tell the whole story, whereas a mixed methods approach enables exploration of the same research problem from different perspectives, therefore enabling a more accurate and satisfactory answer to the research question, ultimately allowing for greater scope in understanding of the research area being investigated (Creswell and Clark 2011).

Indeed, sometimes the results from one method may even be contradictory to those obtained using the other method, which would have otherwise not been detected had only one method been used (Cresswell and Clark 2011), thus providing a more accurate and deeper exploration of the research question.

Furthermore, the limitations of one approach can be offset by the strength of the other (as discussed below).

Additionally, mixed-methods approaches can also be particularly useful in investigating under-researched areas (Creswell and Clark 2011), and are thus fitting for the study given that PGC is a nascent discipline with limited supporting literature.

Quantitative research aims to verify phenomena by collecting and analysing numerical data (Aliaga and Gunderson 2000). It involves observations and measurements that can be made objectively made and repeated by subsequent research groups (Hancock 1998). Quantitative research enables detection of relationships between variables, and can also be useful in gauging opinions, allowing for measurement of the extent to which particular attributes or views are held (Costain et al. 2014b).

A particular strength of quantitative research is that it is not affected by bias of the researcher (Cresswell and Clark 2011). However, although quantitative data provides a more general understanding of the research question, views of individual participants can be lost (Cresswell and Clark 2011).

Specifically, quantitative methods were used to measure attribution of illness to genetic and environmental factors, degree of certainty regarding the causes of mental illness; accuracy of estimation of risk in mental illness; awareness of GC; perceptions about PGC, and perceived usefulness and interest in receiving PGC.

Qualitative data, in contrast, is exploratory and descriptive in its nature (Hesse-Biber and Leavy 2010, Hippman et al. 2013) and useful for describing and attempting to understand and explain phenomena (Guest et al. 2012). It is insightful for studying, measuring and understanding human behaviour, opinion, emotions and responses, and gaining insight into the underlying processes influencing these factors (Creswell and Clark 2011). Such elements are often personal, subjective, complex (i.e. influenced by a variety of factors), and not easily captured by quantitative methods (Guest et al. 2012). Qualitative research honours individual participants' views, which may be lost in quantitative analysis. Additionally, qualitative data can be helpful in explaining quantitative results and identifying processes underlying them when using a mixed-methods approach (Cresswell and Clark 2011), thus allowing for greater comprehension.

Specifically, qualitative data was used to explore perceptions of GC and perceived value of PGC. This insight can be helpful in facilitating better understanding and prediction of future behavioural responses to the offer of PGC as well as deeper insight into other concepts relating to PGC that may not be detected by quantitative analysis.

Survey development

The survey questions were developed following an in-depth analysis of relevant literature ('literature search,' as described above and shown in fig.21).

The survey questions were informed by findings of previous studies, or gaps in knowledge that were identified as missing in the available literature.

Specific surveys listed in previous studies that were relevant to survey development are listed in table 6.

Throughout the process of survey development the researcher had ongoing discussions with the research supervisor.

Dr. J. Austin, E. Morris, H. Andrighettti (University of British Columbia) and a trainee genetic counsellor enrolled on the Genetic Counseling Course at UBC were also involved in the survey development process.

The researcher held a supervision session in January 2015 at UBC, Canada, in which the research project was discussed and the original questions explored, developed and additional lines of enquiry were proposed. The research team suggested alternative methods of analyses for some questions, including, specifically, exploring perceptions of GC using qualitative analysis, and using Likert-scale items to explore respondents' attributional explanations, especially as it would make responses more amenable to quantitative analysis (Austin 2014, pers comms, 26 November).

The research team are conducting a study also exploring perceptions of PGC in 2016, which will incorporate several of the questions in this study. This will also allow for comparison between the UK and Canadian populations.

Dr Austin and H. Andrighetti proof-read the survey before it was launched.

A board-certified genetic counsellor (Dr. M. Bradford, PhD, MSc Genetic Counselling, based at Plymouth University), working within the NHS, England, also proof read the survey and offered suggestions (e.g. alterations in terminology) to make the questionnaires more user-friendly.

The online survey was developed using the survey software solution ‘qualtrics.’ This allows researchers to develop, distribute surveys and collect, download and analyse the relevant data.

After a trial run of the survey was completed by both the researcher and supervisor, the online survey was made live and collection of respondents’ responses commenced.

Diagrams depicting the process of survey development are provided (Figs. 22 and 23).

Table 6: Key PGC/GC surveys from previous studies relevant to research questions and survey development

Research question and relevant studies				
Aetiological attributions	**Perceptions of risk**	**Perceptions of GC (prospective)**	**Perceptions of GC (retrospective)**	**Interest in PGC**
Gamm et al. 2004	Targum et al. 1981	Lyus et al. 2007	Hallowell et al. 1997	Schulz et al. 1982
Meiser et al. 2005	Schulz et al. 1982	Maio et al. 2013	Brown et al. 1999	Quaid et al. 2001
Meiser et al. 2007	Trippitelli et al. 1998		Bernhardt et al. 2000,	De Lisi and Bertisch 2006
Costain et al. 2014a	Quaid et al. 2001,		Pieterse et al. 2005	Lyus et al. 2007
Costain et al. 2014b	Austin et al. 2006		Metcalfe et al. 2007	Austin and Honer 2008
	Meiser et al. 2007		Joseph et al. 2010,	Costain et al. 2014a
	Wilde et al. 2010			Costain et al. 2014b
	Costain et al. 2014a			
	Meiser et al. 2013			
	Costain et al. 2014b			

(Fig. 22)

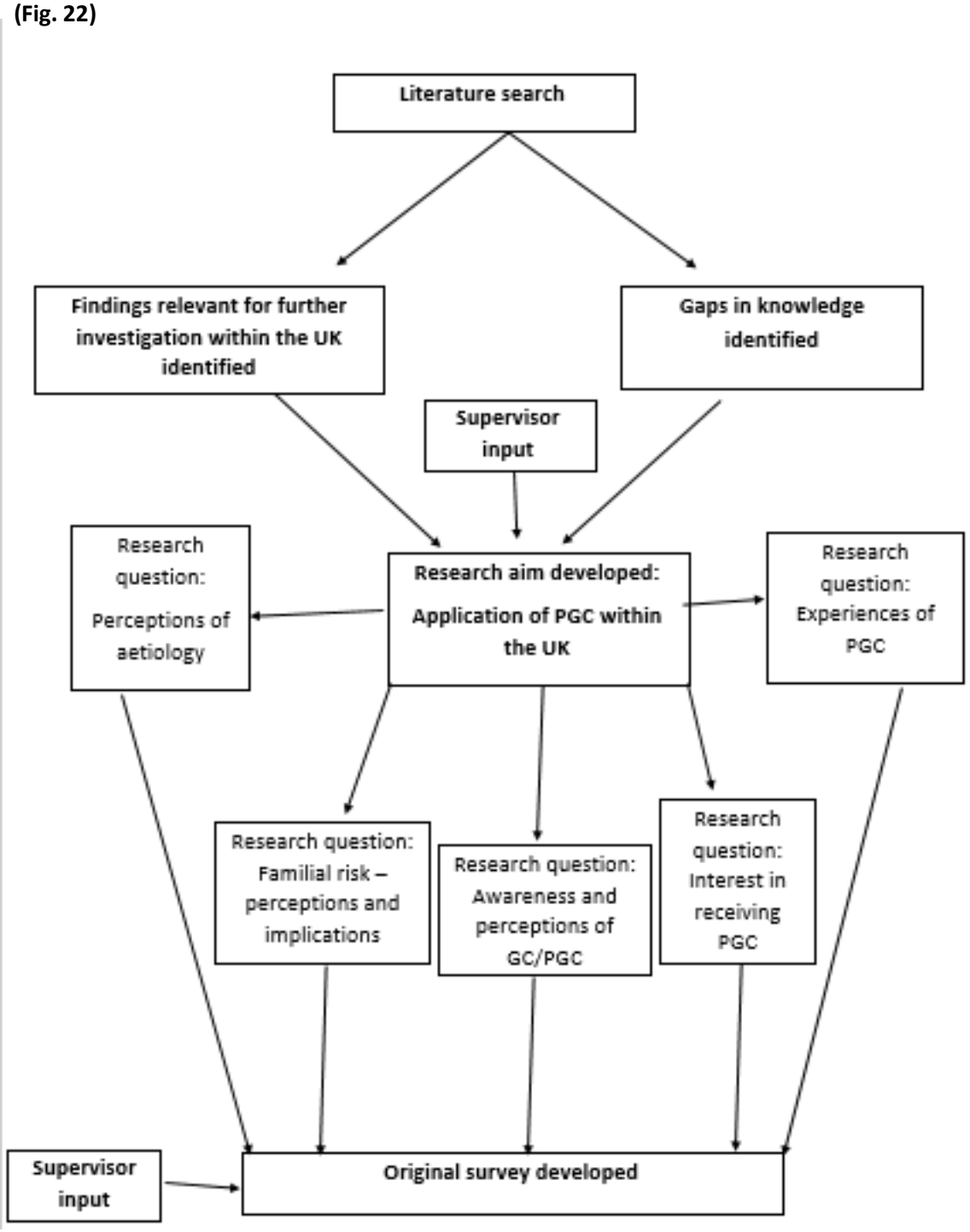


Figure 22: Diagram depicting method undertaken for survey development (1)

(Fig. 23)

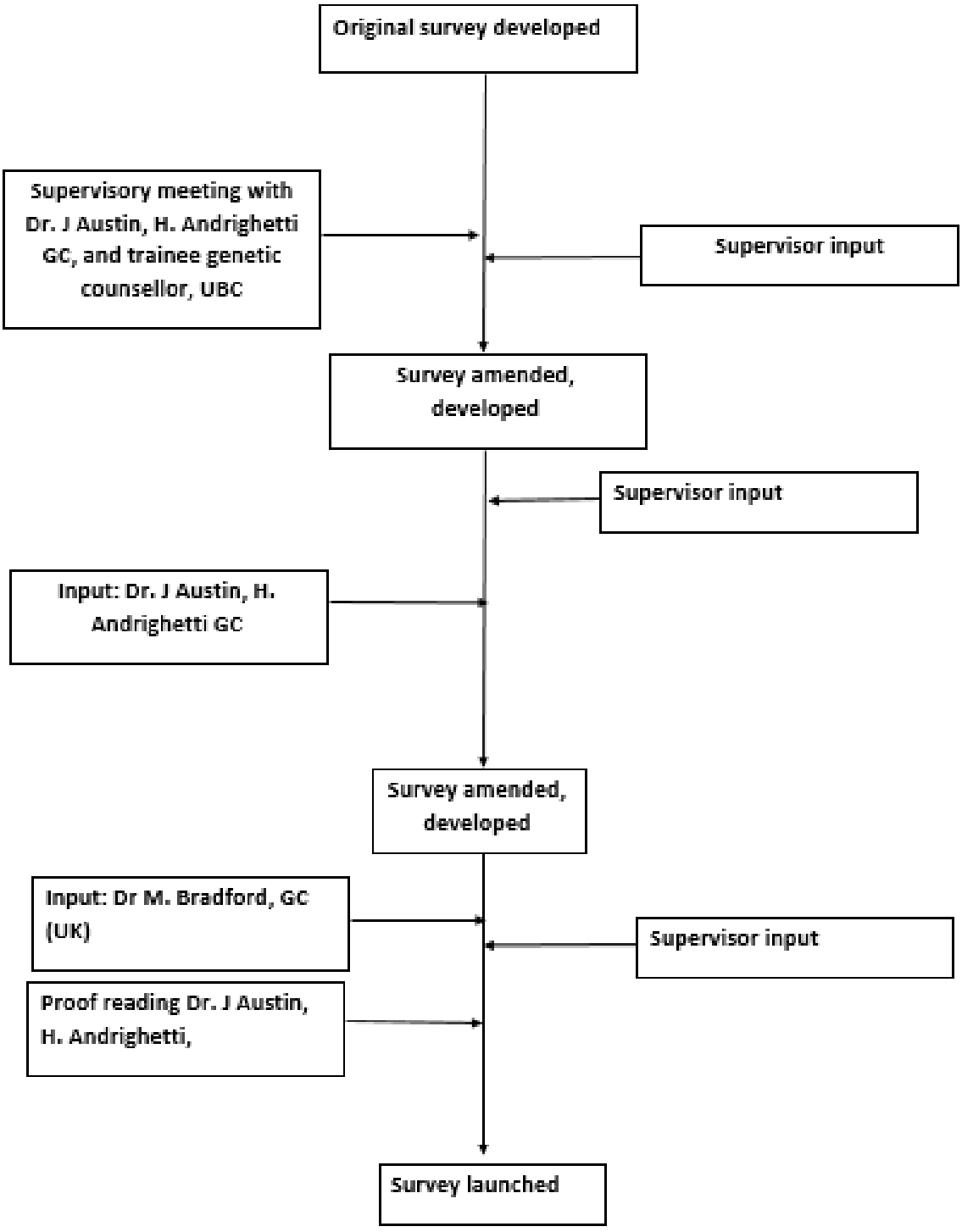


Figure 23: Diagram depicting method undertaken for survey development (2)

Rationale for using Likert-type items to categorise responses

This study used Likert-type items to measure psychological and behavioural properties of respondents (specifically, illness attributions, certainty, concern, interest in PGC, perceived usefulness of PGC).

Measuring psychological and behavioural properties such as attitudes, awareness, character and personality traits is challenging, because there are issues as to how one may transform these qualities into a quantitative measure in order to conduct data analysis (Thambirajah 2005). Likert scales have become one of the most popular tools to objectively measure such psychological properties (Clason and Dormody 1994, Hartley 2013, Maeda 2015).

A traditional Likert scale consists of multiple Likert 'items,' each of which contains a stem, e.g. a statement or question, and a scale consisting of fixed choice response alternatives (e.g. strongly disagree, disagree, agree, strongly agree) that are numerically ordered. Analysis is performed by analysing (e.g. summing or averaging) the numerical values of each response item, to develop a summated Likert-scale (Clason and Dormody 1994, Maeda 2015). Combined, these items provide a quantitative measure of opinion, attitude, or personality trait (Boone 2012). Although the disagree-agree format (as described above) is the most common form of Likert scale, other types of response options (e.g. 'below average, slightly below average, average, slightly above average, above average') are also widely used in research.

Additionally, individual Likert-scale *items* may also be analysed. In this approach, the researcher does not attempt to combine the responses from the items to produce a summated ('Likert') scale. Thus, this type of approach uses *Likert-type* items, rather than *Likert scales* (Clason and Dormody 1994). Although Likert (1932), never originally considered analysis of individual items scales (Clason and Dormody 1994) Likert-type items have become increasingly popular tools of analysis in clinical and health psychology research (Hartley 2013), and were used in this study specifically.

There are several advantages to using Likert-scale and Likert-type items. Firstly, as they are so widely used they are easily understood (Neuman 2000). They are also simple and easy to design, administer and analyse (Neuman 2000). Additionally the responses are easily quantifiable and so easily amenable to statistical analysis (Likert 1932, Hersen and Bellack 2013, Austin 2014, pers comms, November 26).

They allow for a range of responses and not concrete answers from respondents, and also allow for neutral answers, which may be more accurate in terms of reflecting respondents' true answers, and may also be preferable to the respondent (Maeda 2015).

In this study, Likert-type items were used to measure i) attributions of the mental illness to genetic factors ii) attribution of the mental illness to non-genetic factors iii) certainty regarding attributions, using a tool that had been previously developed by J. Austin's research group (see Appendix H), and is currently used within their clinics. This allows for comparison between American and UK populations in future research.

Meiser et al. (2007) similarly explored illness attributions to different factors using a 5-point Likert-type scale, using responses from 1 (not at all important) to 5 (extremely important).

Likert-type items were also used to measure iv) degree of concern for other relatives becoming ill vi) interest in receiving PGC after receiving information about the service and vii) perceived usefulness of PGC amongst respondents.

This enabled statistical analysis between variables obtained from each item, using non-parametric tests (as described later in 'analysis') to enable identification of relationships between variables and also compare responses from different groups of respondents.

Each item consisted of a 7-point scale, to offer respondents' more flexibility in their answers (Nunnally and Bernstein 1978) and also the option of a neutral response.

Recruitment

-Gatekeepers

Gatekeepers (i.e. individuals that control and/or facilitate research access) (Jupp 2006) at local and national mental health charities and organisations; carers charities; and local mental health support groups were approached by the researcher via email or telephone and requested to invite their members to participate in the research project.

Gatekeepers were sent the information document and the link to the Research Landing Page which contained the Participant Information Sheet and Consent Form. A reminder email or phone call was sent to organisations and gatekeepers that did not reply.

Methods used to promote the research project by gatekeepers included: passing on information to members, advertising the study online website and promoting it via social media, dependent upon what method was perceived most suitable for the organisation's members and framework. The researcher also attended support group meetings to provide a briefing of the research to members and invite them to participate.

-Online-presence and Social Media

Social media sites (Twitter and Facebook, see figs. 24-27) were used to promote the study. Tweets were regularly sent out during the data collection period inviting public participation in the study, and hashtags including 'Mental Health';'Genetics'; and 'Mental Health Matters' were used to make the tweet viewable to twitter users with an interest in these trends. In addition, data collection occurred during both Mental Health Awareness Week and Carers Week and 'MentalHealthAwarenessWeek' and 'CarersWeek' were used as hashtags in invitational tweets to make the tweet viewable to twitter users with an interest in these events.

Other twitter users, including mental health charities, research organisations, and leading mental health advocates retweeted these invitatory tweets out of their own initiative or after being approached by the researcher.

(Fig. 24)

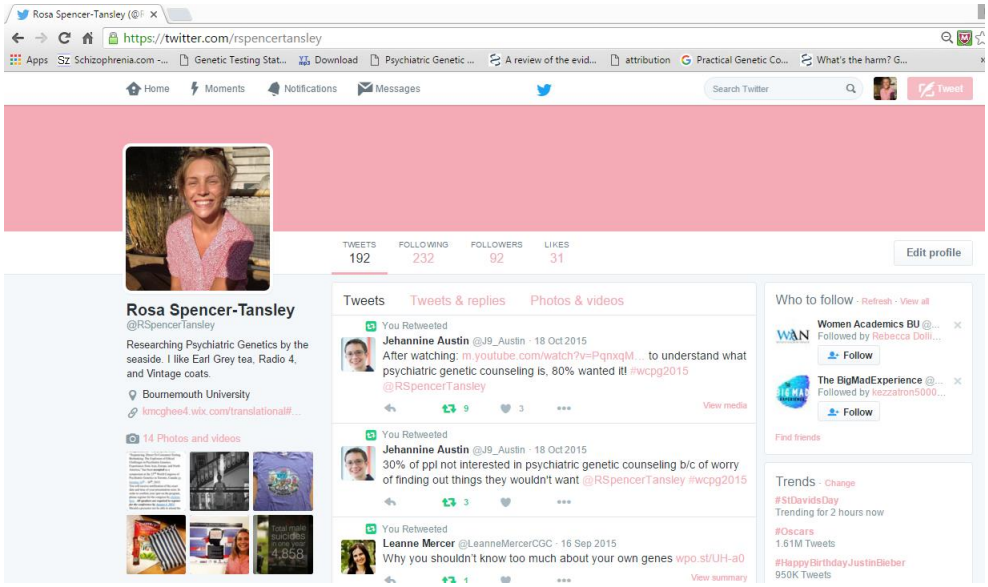


Figure 24: Screenshot of researcher's twitter page

(Source: Spencer-Tansley 2015)

Twitter was used to promote the study by tweeting invitational links. Mental health charities and organisations also promoted the study via their own twitter accounts.

(Fig. 25)

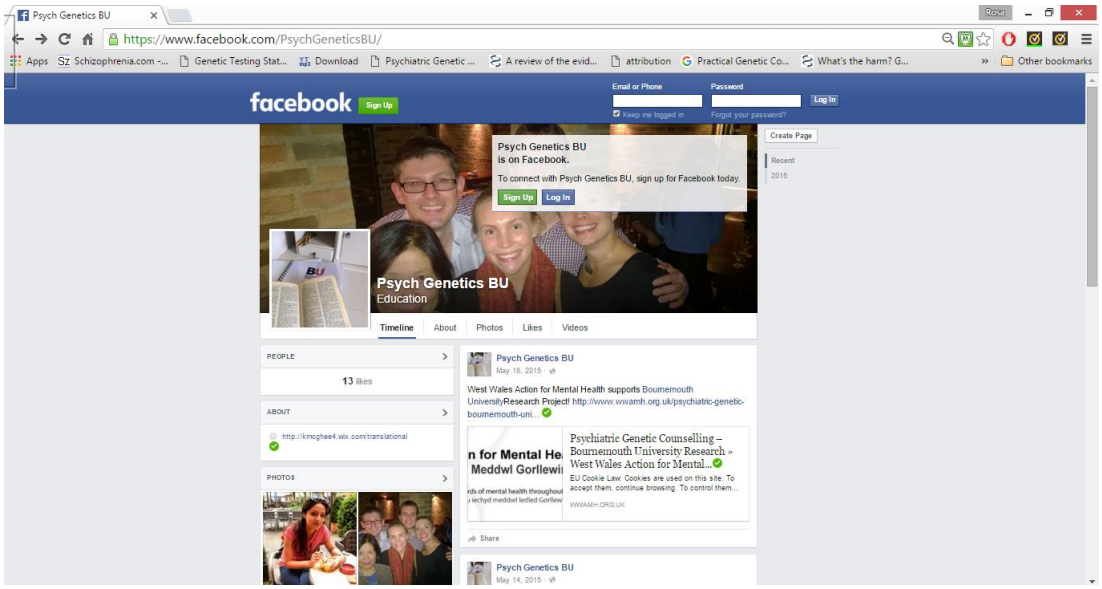


Figure 25: Screenshot showing Facebook Account ('Pysch Genetics BU)' used to promote study

(Source: Psych Genetics BU 2015).

(Fig. 26)

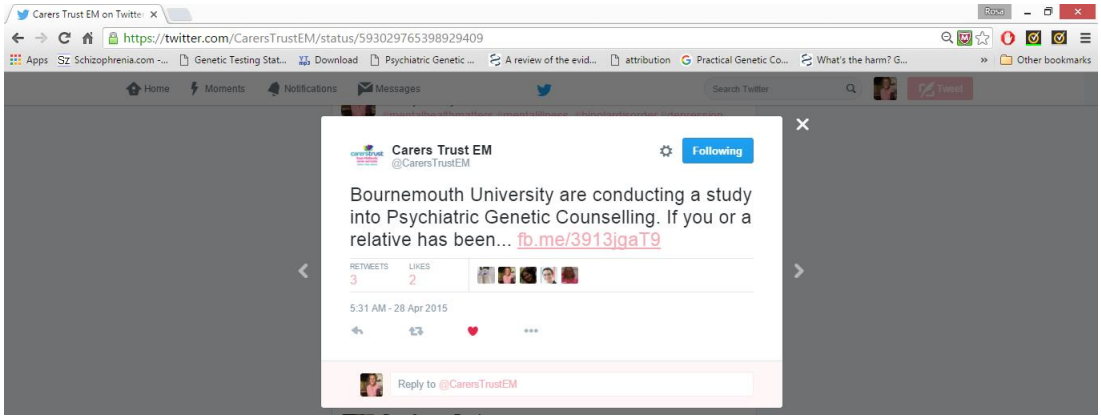


Figure 26: Screenshot - Example of charity (Carers Trust) promoting study via twitter

(Source: Carers Trust East Midlands 2015)

(Fig. 27)

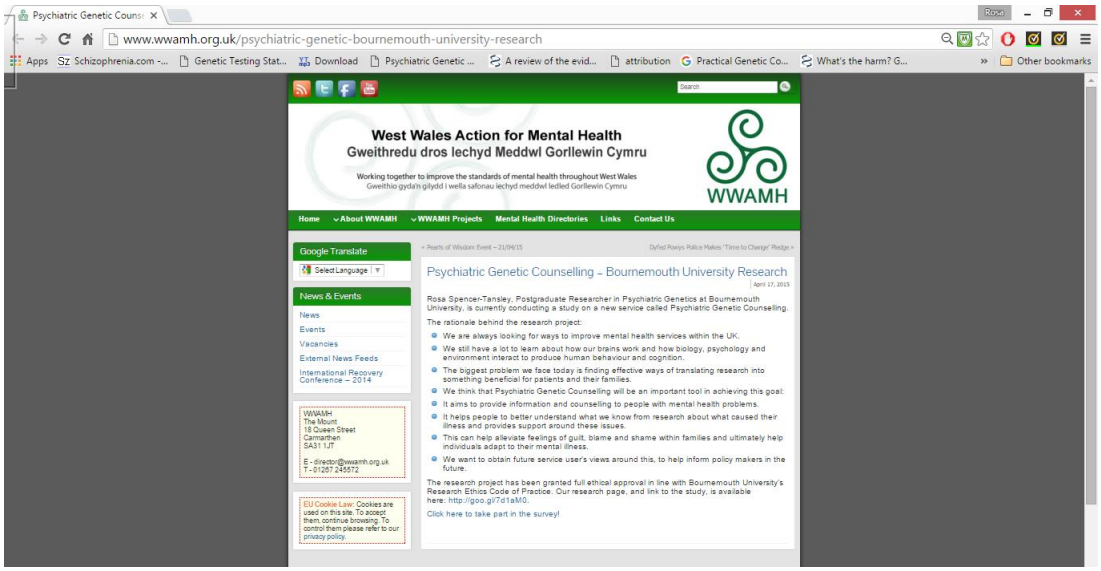


Figure 27: Screenshot - Example of charity (WWAMH) promoting study online

(Source: West Wales Action for Mental Health 2015).

Bournemouth university website and Research page also featured articles around the study containing links directing to the study page (see figure 28).

(Fig. 28)

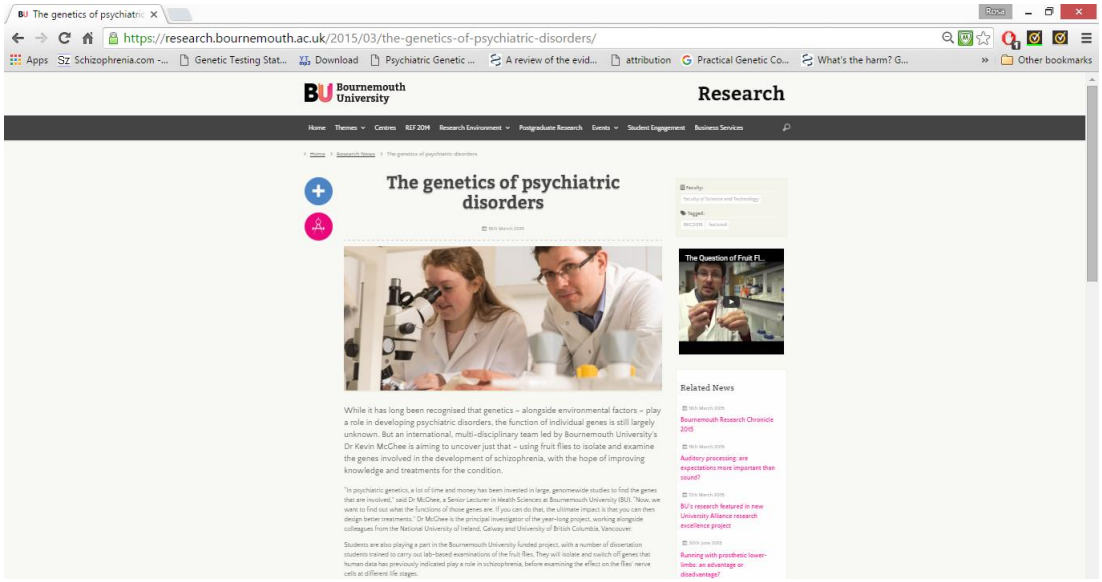


Figure 28: Screenshot of research article on Bournemouth University website regarding research group's work, and promoting study.

(Source: Bournemouth University 2015).

-Additional recruitment approaches

Participants were directly approached at various mental health awareness events across the south of the country and invited to participate in the study. The researcher also delivered presentations about the study to relevant audiences across the university including Adult Nursing students and Faculty staff members. Additionally the researcher and supervisor also spoke on a radio programme, 'Mental Health Matters,' to discuss and promote participation (see fig. 29)

(Fig. 29)

Figure 29: Screenshot showing webpage for 'Mental Health Matters' podcast link, featuring PGR and PGR supervisor discussing study

(Source: Phoenix FM 2015).

Participation

Data was collected via two online surveys – one for affected individuals with mental illness and one for relatives of affected individuals - between 25 March 2015 to 21st July 2015.

Once directed to the online Survey page, which contained the participant information sheet and consent form, informed consent was obtained. Participants were then able to commence with completion of the questionnaire. Upon completion participants were able to again download the participant information sheet and consent form.

Analysis

Data from all partially completed surveys were used if applicable questions required for an analysis were answered. Respondents that indicated they were not UK/ Ireland residents were excluded from analysis.

-Qualitative data

All qualitative responses were coded by the researcher using thematic analysis. Thematic analysis is useful for identifying, analysing, and reporting patterns (themes) within data (Braun and Clarke 2006). Thematic analysis is a widely-used analytical method that can offer a rich, thematic description of the entire dataset (Braun and Clarke 2006). A particular benefit of this analytical approach is its accessibility, meaning it is amenable to researchers whom are not overly familiar with qualitative analysis (Braun and Clarke 2007). Additionally thematic analysis may also capture elements that that are important in relation to the overall research question buy may not be quantifiable (Braun and Clarke 2006), as it offers flexibility in terms of how themes are determined – the significance of a theme is not necessarily measured by its frequency, as it may be using alternative methods of analysis (Horning-Priest 2005).

For this study, a read, re-read and code approach was undertaken to identify codes and subthemes to comprehensively categorise all the data. This started by the researcher familiarising themselves with the data set. Codes were then generated, organising the data into various groups. Throughout this process the researcher remained open-minded, generating as many codes as possible. An inductive, bottom-up approach was employed for the coding process in which analysis was not directed towards theory development (Braun and Clarke 2006). Highlighters were originally used to code all the data and identify potential patterns within it. Extracts of the data from individual responses were then copied to enable collation of each code in separate computer files. The coding was checked with an academic (research supervisor) for consistency; discrepancies were discussed and consensus reached.

From this, potential themes were identified by analysing codes and subsequently combining or modifying codes, with some codes becoming redundant. Major themes, and sub-themes within the major themes, were subsequently deduced. All the relevant coded data extracts within the identified themes were then collated. Themes and sub-themes were then reviewed and refined to ensure a coherent pattern between all of the collated extracts within a theme. Again, developed themes were discussed and checked with an academic (research supervisor) for consistency; discrepancies were discussed and consensus reached.

How widespread each view appeared to be in the sample was recorded so that it could be reported qualitatively in the text.

Quantitative data

Quantitative data was analysed using Microsoft Excel and the statistical package SPSS.

For ordinal data, the non-parametric Mann-Whitney U-Test was used to compare differences between two independent groups or samples. Mann-Whitney U-Test tests the null hypothesis that two samples come from a population with the same distribution, and therefore the distributions of both groups are identical (Butler 1985). If the p-value is small ($p<.05$), the null hypothesis that the difference is due to random sampling can be rejected and it can be instead concluded that the populations are distinct (Field 2009). In this study Mann-Whitney U-Test was used to test whether there was a significant overall difference in the magnitude of the variable of interest between affected individuals and relatives; and males and females.

Spearman's rank-order correlation coefficient is a non-parametric measure of statistical dependence between two variables (Field 20009). It is used to measure the degree (strength) and direction (positive or negative) of association between two variables. When the correlation coefficient, r_s, is close to 0 this means that there is little relationship between the variables; whilst the further away from 0 r is, in either the positive or negative direction, the greater the relationship between the two variables. A spearman correlation of +1 or -1 occurs when each of the variables is a perfect monotone function of the other (Brase 2012). In this study Spearman's rank-

order correlation coefficient was used to test for associations between testable variables.

Pearson's Chi-square test (Crosstab) was used to test the independence between two categorical variables (Field 2009). It enables evaluation of the likelihood of any observed difference between the sets of variables arose by chance; if the Pearson chi-square is significant ($p<.05$) the two variables show a relationship that is larger than what would be expected under chance alone and therefore there must be a relationship between the two variables.

3. Results

A total of 60 affected individuals with mental illness and 29 relatives responded to the survey; 3 affected individuals not from the UK or Ireland were excluded from analysis.

3.1 Demographic and diagnostic data

Table 7: Demographic data for respondents

Variable	Affected individuals	Relatives	Total sample
Gender			
Male	24	5	29
Female	32	24	56
Age			
18-24	6	6	12
25-30	7	2	9
31-35	8	3	11
36-40	6	1	7
41-45	5	2	7
46-50	10	1	11
51-55	5	3	8
56-60	6	5	11
61-65	1	6	7
66+	3	0	3
Highest level of education			
No post-school qualifications	10	5	15
Post-school qualifications	47	23	70
Employment status			
In employment	27	18	45
Self-employed	5	1	6
Not currently working	10	3	13
In full time education	2	6	8
Retired	4	1	5
Unable to work	7	0	7
Nationality			
English	32	17	49
Welsh	2	1	3
Scottish	1	1	2
Northern Irish	2	8	10
British	14	2	16
Other	6		6
Ethnicity			
White British	50	25	75
White 'other'	2	2	4
Asian British	0	1	1
Caribbean	0	1	1
Mixed – white and Asian	1	0	1
Other ethnicity	4	0	4

Note: for some questions, not all respondents provided reportable answers.

Table 8: Diagnostic data for affected individuals

Variable	**Affected individuals – Count**
Psychiatric diagnosis	
Psychosis	5
Mood disorder	44
(Bipolar disorder)	(38)
(Recurrent depression	(6)
Anxiety disorder	6
Generalised anxiety	(1)
Anxiety with depression	(1)
OCD	(2)
PTSD	(2)
Other	2
Anorexia	(1)
Borderline personality disorder	(1)
Years since diagnosed	
0-5	21
6-10	12
11-20	16
21-30	5
31+	3

Table 9: Diagnostic data for relatives

Variable	**Relatives – count**
Relationship to affected individual(s)	
Son or daughter	5
Sibling	10
Parent	14
Partner	4
Cousin	1
Niece/nephew	1
Aunt/uncle	0
Cousin	1
Grandchild	0
Relative's psychiatric diagnosis	
Psychosis	8
Mood disorder	11
(Bipolar disorder)	(10)
(Recurrent depression	(1)
Anxiety disorder	8
Anxiety with depression	(3)
OCD	(3)
PTSD	(2)
Other	1
Anorexia and bulimia	(1)
Years since relative diagnosed	
0-5	7
6-10	4
11-20	4
21-30	2
31+	0

3.2 Perceptions of aetiology

This section quantitatively explored respondents' attributions of the mental illness to genetic and non-genetic factors. Respondents' confidence in their provided answers was also queried, to measure certainty regarding their attributional explanations. In total it contained four structured questions using seven-point Likert-type response items.

Aetiological attributions

Respondent's attribution of their/their relative's mental illness to i) genetic factors and ii) environmental factors ("life experiences") was queried using two 7-point Likert-type response items (1= "did not contribute at all"; 4 = "contributed somewhat"; 7= "entirely/causal role", see figures 30a-d.).

Figures 30a-d: Likert-type response items to assess respondents' illness attributions

Fig. 30a) Likert-type response item to assess affected individuals' attribution to genetic factors in mental illness pathogenesis

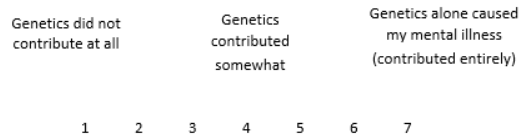


Fig. 30b) Likert-type response item to assess affected individuals' attribution to environmental factors in mental illness pathogenesis

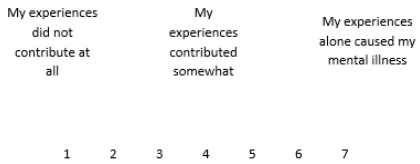


Fig. 30c) Likert-type response item to assess relatives' attribution to genetic factors in mental illness pathogenesis

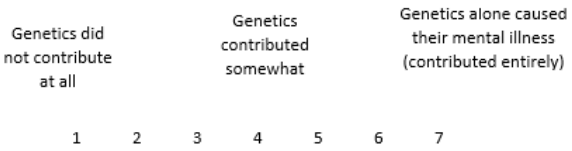
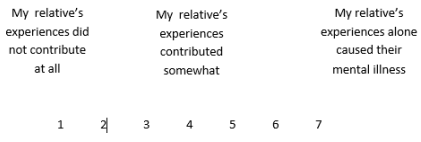


Fig. 30d) Likert-type response item to asses relative's attribution to environmental factors in mental illness pathogenesis



Aetiological attributions - results

The majority of affected individuals (61%, n=35) and relatives (62%, n=18) indicated they believed genetics played somewhat to a causal role in their or their relative's mental illness. A minority of affected individuals (7%, n=4) and relatives (7%,n=2) believed genetic factors played no role in their/their relative's mental illness.

Most affected individuals (86%, n=49) and relatives (72%, n=21) reported that they believed environmental factors played a somewhat to causal role in their/their relative's mental illness; and only 1 individual (2%) and 1 relative (3%) indicated they believed environmental factors played no role.

All data are shown in tables 10-11 and figs. 31-34.

Table 10: Frequency table for attribution variables (GA, EA) for affected individuals.

Attribution	*Attribution*							*TOTAL*
Variable	**1**	**2**	**3**	**4**	**5**	**6**	**7**	
GA	4	8	10	18	13	3	1	57
EA	1	3	4	7	20	15	7	57

Note: GA = attribution of mental illness to genetic factors; EA = attribution of mental illness to environmental factors. 1= "Not at all", 4 = "Somewhat", 7= 'Entirely."

Table 11: Frequency table showing attribution variables (GA, EA) for relatives

Attribution	*Attribution*							*TOTAL*
Variable	**1**	**2**	**3**	**4**	**5**	**6**	**7**	
GA	2	5	4	8	5	4	1	29
EA	1	3	4	4	12	5	0	29

Note: GA = attribution of mental illness to genetic factors;, EA = attribution of mental illness to environmental factors. 1= "Not at all", 4 = "Somewhat", 7='Entirely.'

Figure 31: Frequency diagram showing affected individual's attribution of the mental illness to genetic factors

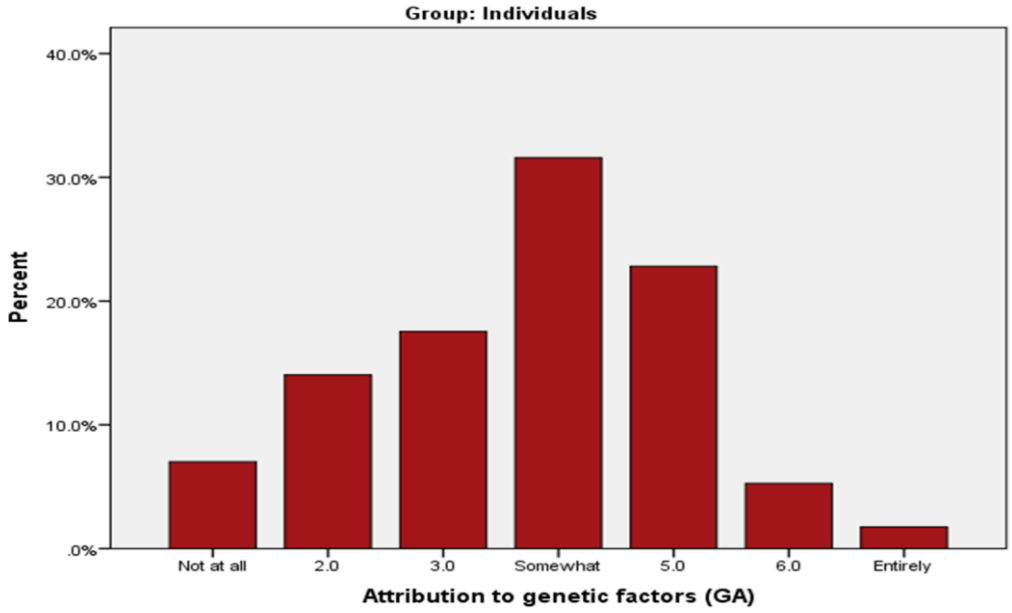


Figure 32: Frequency diagram showing affected individual's attribution of the mental illness to environmental factors

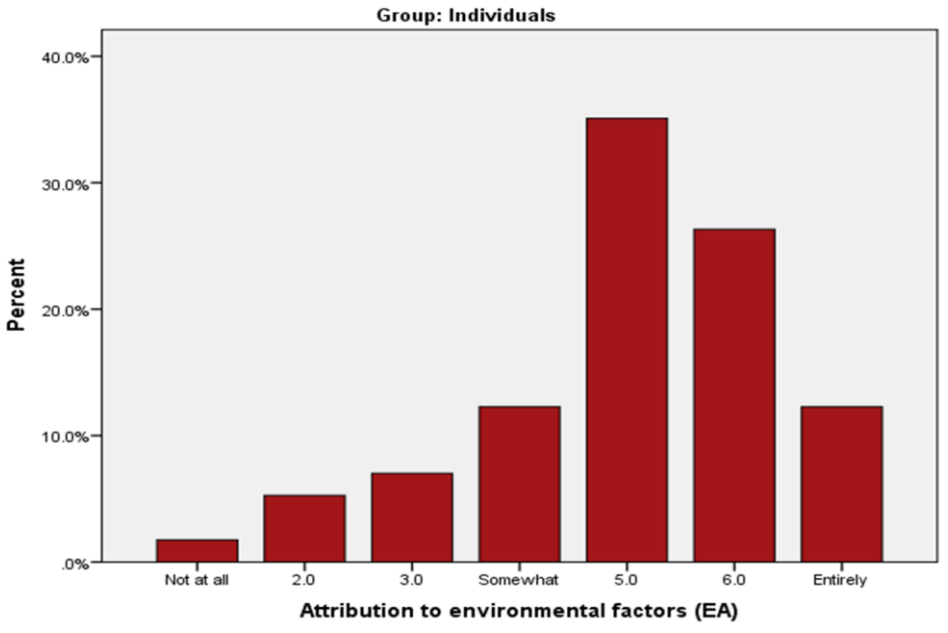


Figure 33: Frequency diagram showing relative's attribution of the mental illness to genetic factors

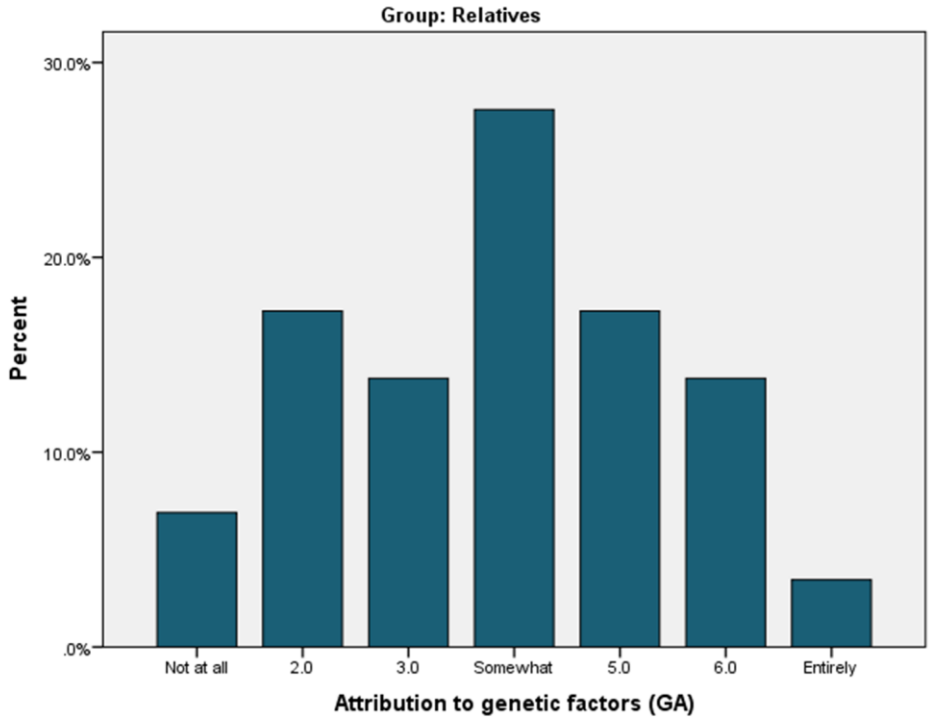
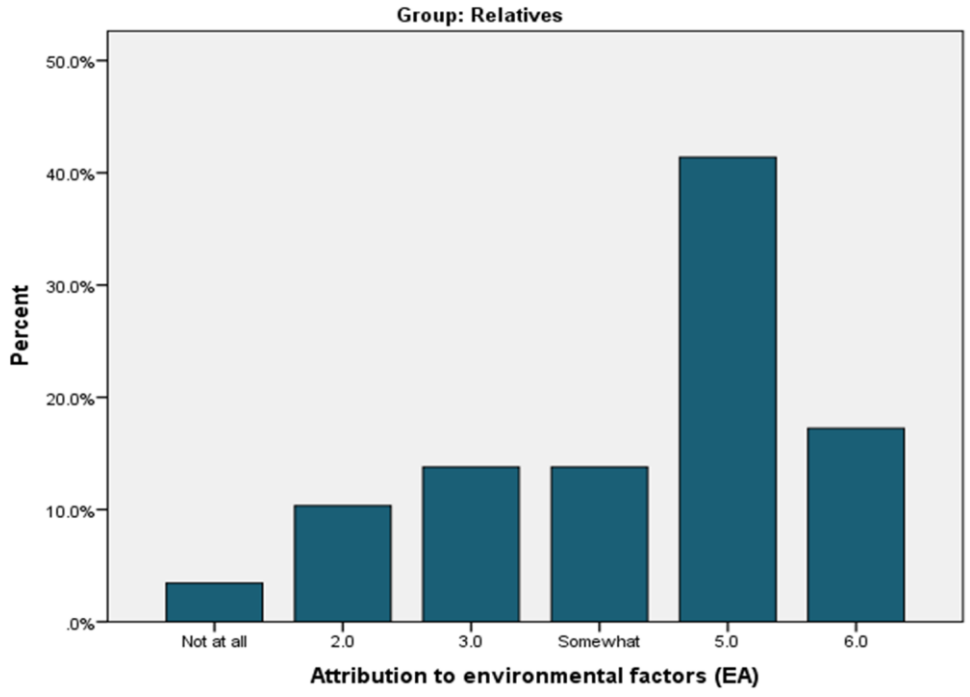


Figure 34: Frequency diagram showing relative's attribution of the mental illness to environmental factors



Certainty regarding aetiological attributions

To explore certainty regarding aetiological attributions, respondents were asked to indicate how confident they were in the answer they provided regarding their attribution of the mental illness to iii) genetics iv) environmental factors ("life experiences").

Respondents' certainty was assessed using two 7-point Likert-type response items (1= I am not sure at all; 7=I am absolutely certain, see fig. 35).

Figure 35: Likert-type response to assess respondent's certainty regarding their attribution of the mental illness to i) genetic factors ii) environmental factors.

Certainty regarding aetiological attributions – results

Overall the majority of respondents indicated relatively high levels of certainty regarding their answers provided for aetiological attribution of the mental illness.

The majority of affected individuals (77%, n=44) indicated they were relatively to extremely certain regarding their attribution of the mental illness to genetic factors (GAC); only 5% (n=3) indicated they were not at all certain.

For relatives, similarly, the majority 76% (n=22) indicated they were relatively to extremely certain about their attribution of the mental illness to genetic factors, although a proportion (17%, n=5) indicated complete uncertainty about their answers regarding the role of genetics.

For all respondents, certainty was greater regarding the role of environmental factors (EAC) with 89% (n=51) of affected individuals and (86%, n=25) indicating they were relatively to extremely certain about their attribution of the mental illness to environmental factors and only 1 individual (2%) and no relatives indicating complete uncertainty. All data are given in tables 12-13 and figs. 36-39.

Table 12: Frequency table for attribution certainty variables for affected individuals

Certainty	*Certainty*							*TOTAL*
Variable	**1**	**2**	**3**	**4**	**5**	**6**	**7**	
GAC	3	6	3	9	8	13	14	57
EAC	1	2	3	7	10	17	17	57

Note: GAC = Certainty regarding attribution of mental illness to genetic factors, EAC = Certainty regarding attribution of mental illness to environmental factors. 1= "Not at all", 7= 'Absolutely certain'

Table 13: Frequency table for attribution certainty variables for relatives

Certainty	*Certainty*							
Variable	**1**	**2**	**3**	**4**	**5**	**6**	**7**	**TOTAL**
GAC	5	0	2	5	5	7	5	29
EAC	0	0	4	1	10	9	5	29

Note: GAC = Certainty regarding attribution of mental illness to genetic factors, EAC = Certainty regarding attribution of mental illness to environmental factors. 1= "Not at all", 7= 'Absolutely certain'

Figure 36: Frequency diagram showing affected individuals' certainty regarding attribution of the mental illness to genetic factors (GAC)

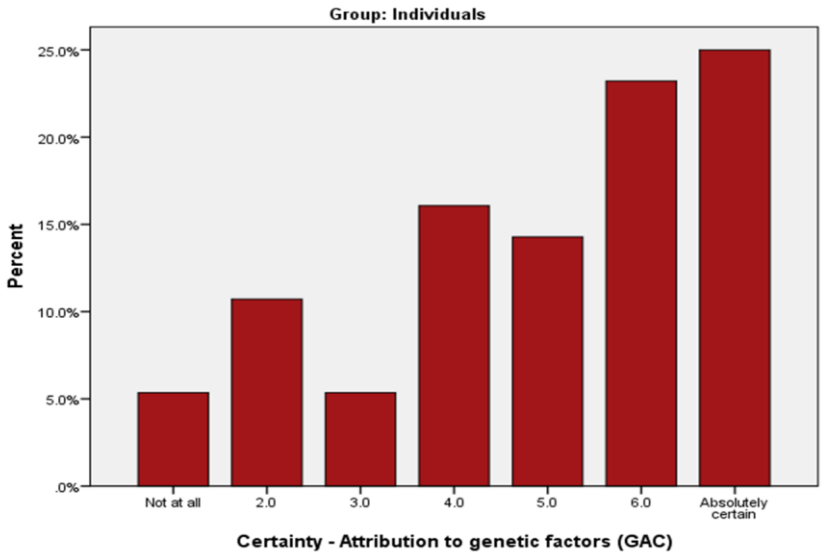


Figure 37: Frequency diagram showing affected individual's certainty regarding attribution of the mental illness to environmental factors (EAC)

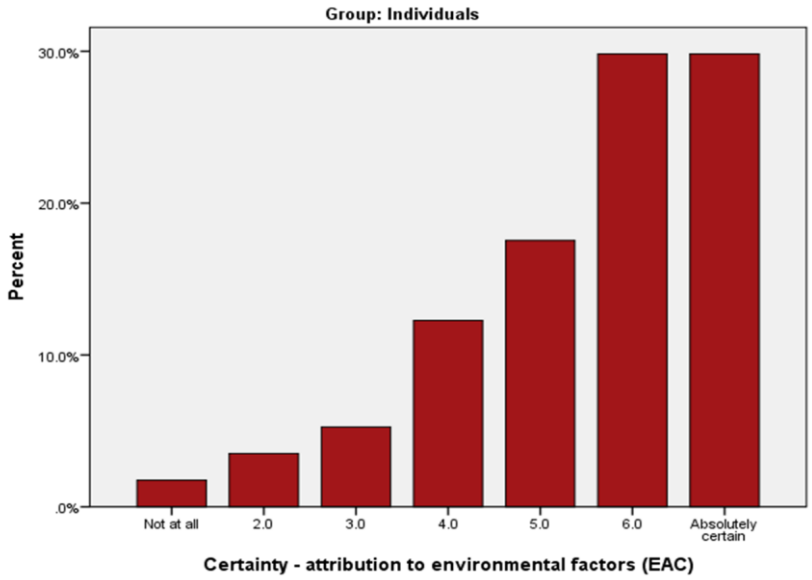


Figure 38: Frequency diagram showing relatives' certainty regarding attribution of the mental illness to genetic factors (GAC)

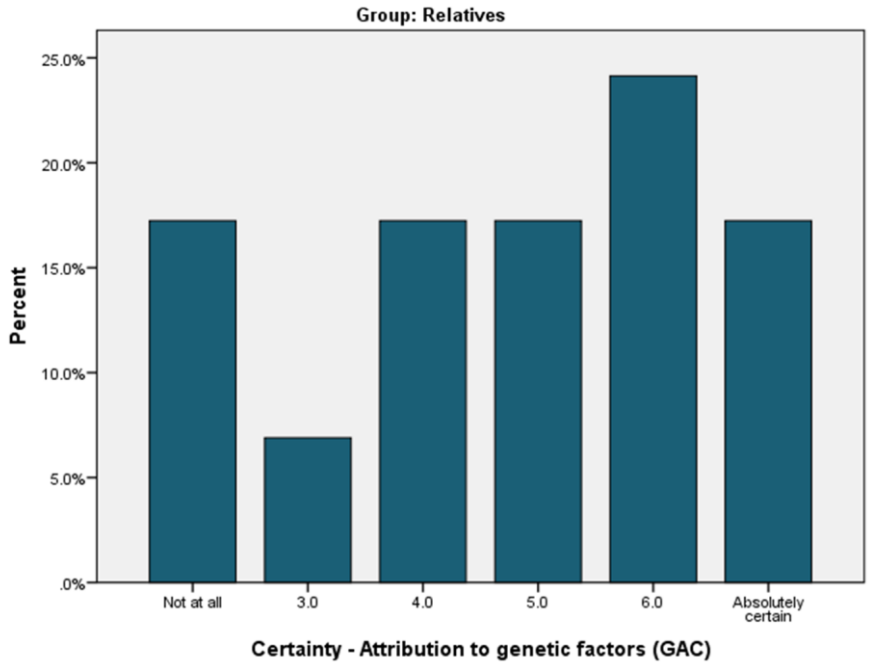
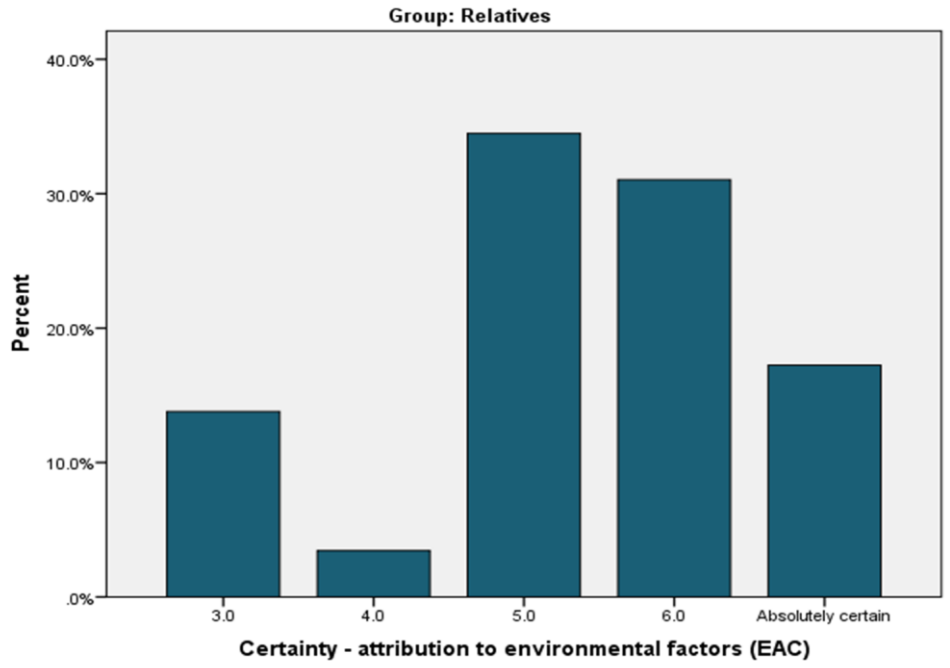


Figure 39: Frequency diagram showing relatives' certainty regarding attribution of the mental illness to environmental factors (EAC)



Perceptions of aetiology – statistical analyses

Spearman' rank correlation coefficient

Spearman's rank correlation was used to test for associations between illness attribution variables (GA; EA) and illness attribution certainty variables (GAC; EAC).

Correlations are shown in tables 14 and 15.

The correlation between certainty regarding attribution of the mental illness to genetic factors (GAC) and certainty regarding attribution of the mental illness to environmental factors (EAC) was significant and positive for both affected individuals (r_S=.389, p=0.003) and relatives (r_S=.797** , p<0.001). Thus, greater certainty regarding attribution of the mental illness to genetic factors was associated with greater certainty regarding attribution to environmental factors.

For affected individuals, the correlation between attribution of the mental illness to environmental factors (EA) and certainty regarding attribution of the mental illness to environmental factors (EAC) significantly and positive (r_S=.475**, p<.001). Thus, greater attribution of the mental illness to environmental factors was associated with greater certainty regarding attribution of the mental illness to environmental factors.

No other significant associations were detected.

Table 14: Spearman's rank correlations showing associations between illness attribution variables (GA, EA) and illness attribution certainty variables (GAC, EAC) for affected individuals

	GA	**GAC**	**EA**	**EAC**
GA	1.000	.075	-.200	-.015
GAC	.075	1.000	.103	.389**
EA	-.200	.103	1.000	.475**
EAC	-.015	.389**	.475**	1.000

*Note: GA = attribution of mental illness to genetic factors, GC = Certainty regarding attribution of mental illness to genetic factors, EA = attribution of mental illness to environmental factors, EC = Certainty regarding attribution of mental illness to environmental factors. *p <.05, **p <.01*

Table 15: Spearman's rank correlations showing associations between illness attribution variables (GA, EA) and illness attribution certainty variables (GAC, EAC) for relatives

	GA	**GAC**	**EA**	**EAC**
GA	1.000	.210	-.276	.043
GAC	.210	1.000	.155	.797**
EA	-.276	.155	1.000	.277
EAC	.043	.797**	.277	1.000

*Note: GA = attribution of mental illness to genetic factors, GC = Certainty regarding attribution of mental illness to genetic factors, EA = attribution of mental illness to environmental factors, EC = Certainty regarding attribution of mental illness to environmental factors. *p <.05, **p <.01*

Mann-Whitney U-Test

To test for differences between illness attribution variables (GA; EA) and illness attribution certainty variables (GAC; EAC) between groups of respondents (i.e. affected individuals compared to relatives; males compared to females), Mann-Whitney U-Test was applied.

Comparing relatives and affected individuals, attribution to environmental factors (EA) was significantly greater for affected individuals (mean = 5.018, mean rank = 47.64, median = 5.00) than for relatives (mean = 4.310, mean rank = 35.36, median = 5, U=590.500 ,Z= -2.233, p= 0.026, r = -.241). This result was therefore significant but the difference between the groups was relatively small, indicating the result is relatively non-substantive.

Comparing genders, there were no significant differences between illness attribution variables or illness attribution certainty variables between males and females.

Summary of findings

The majority of respondents attributed mental illness to both genetic and non-genetic factors. Very few respondents indicated they believed genetics had no role to play whatsoever, and only two respondents believed genetics had contributed entirely to the mental illness (i.e. played a causal role).

Attribution to environmental factors was significantly greater amongst affected individuals than relatives, but there were no other significant differences between affected individuals and relatives, nor between males and females.

Degree of certainty regarding respondents' attribution to both genetic and environmental factors was relatively high, with over 65% of respondents indicating they were somewhat to extremely confident in the answers they provided regarding their attribution of the mental illness to both genetic factors and environmental factors. A proportion of relatives (17%) indicated complete uncertainty about the role of genetic factors in pathogenesis, however.

For all respondents greater certainty regarding attribution to genetic factors was significantly associated with greater certainty regarding attribution to environmental factors.

For affected individuals, greater attribution to environmental factors was significantly associated with greater certainty regarding attribution to environmental factors.

No other significant associations were detected.

3.3 Familial risk - perceptions and implications

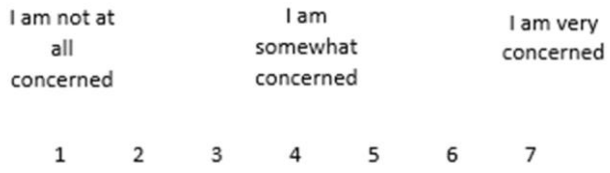
The section of this study explored perceptions and implications of familial risk amongst respondents by enquiring respondents':

i. Degree of concern for other relatives becoming ill, queried using a 7-point Likert-type response item.
ii. Perceptions of quantitive risk to first degree relatives, assessed by obtaining respondents' estimates of risk to offspring and sibling,
iii. Impact of mental illness on family-planning

i. Concern for other relatives developing mental illness

Respondent's concern for other relatives developing mental illness was queried using a 7-point Likert-type response item (1= not at all concerned; 4= somewhat concerned; 7= very concerned; see figure 40.

Figure 40: Likert-type response item to assess respondent's concern for other relatives developing mental illness.



Concern for other relatives developing mental illness – results

Of the 86 respondents that answered this question, 84% (n=48) of affected individuals and 55% (n=16) of relatives reported they were somewhat to extremely concerned about other relatives also developing mental illness. Almost quarter of affected individuals (n=13, 23%) reported being 'very concerned' about the risk to other relatives.

Conversely, 7% of affected individuals (n=4) and 14% of relatives (n=4) reported not being at all concerned about other relatives becoming affected.

All frequencies are shown in table 16 and figs. 41-42.

Table 16: Frequency table showing respondents' concern for other relatives developing mental illness

Group	*Concern*							**TOTAL**
	1	**2**	**3**	**4**	**5**	**6**	**7**	
Affected individuals	4	2	3	9	18	8	13	57
Relatives	4	4	5	4	6	5	1	29

Note: 1 = 'Not at all concerned'; 4 ='Somewhat concerned'; 7 =' Very concerned.'
Responses interpreted as indicating being 'somewhat' concerned or more are highlighted in yellow.

Figure 41: Frequency diagrams showing affected individuals' concern for other relatives developing mental illness.

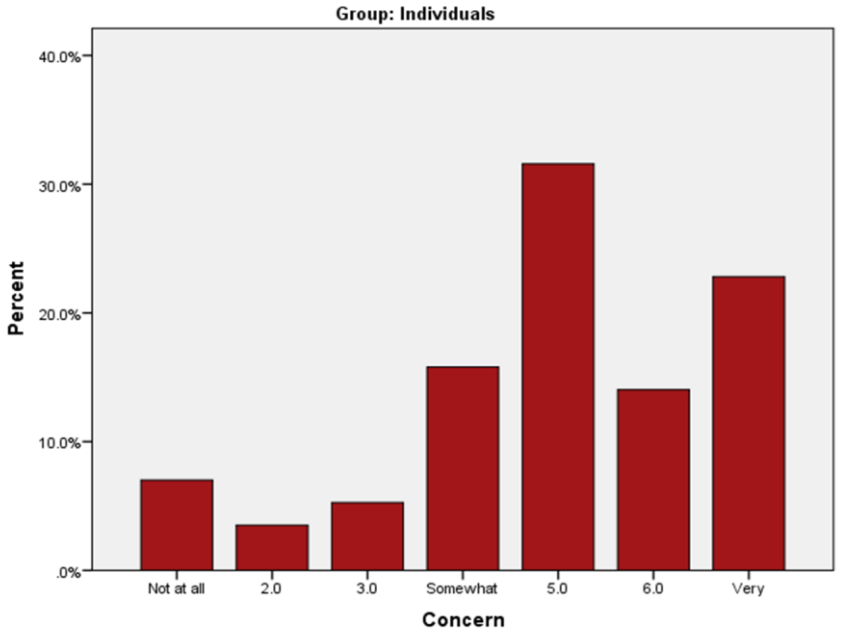
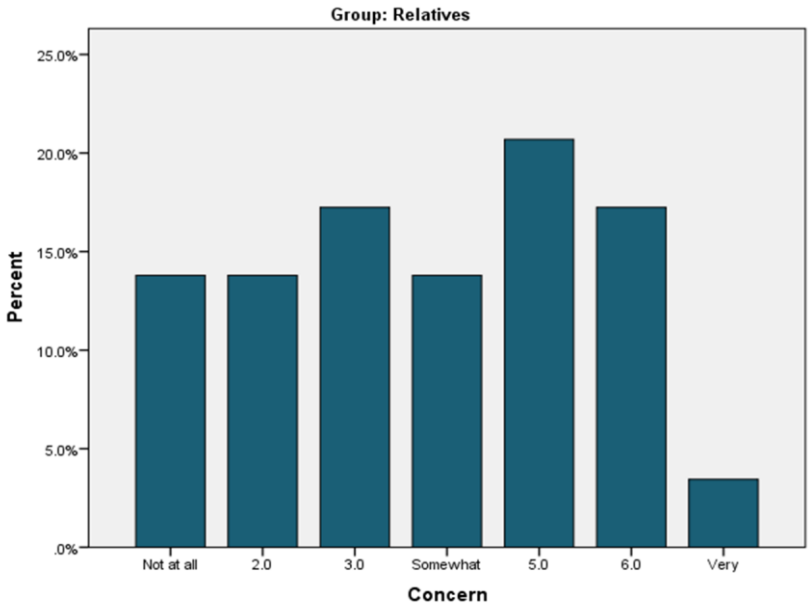


Figure 42: Frequency diagram showing relatives' concern for other relatives developing mental illness.



Concern: statistical analyses

Mann Whitney U-Test

Mann-Whitney U Test was applied to test for differences in concern between groups of respondents (i.e. affected individuals compared to relatives; males compared to females).

Comparing affected individuals and relatives, concern was significantly greater amongst affected individuals (mean rank = 48.67, median =5.00) than relatives (MDN= = 4.00, U= 532.000; Z= -2.738*, p*=0.006). Effect size value size (r = -0.295) suggest this approached moderate practical significance.

Gender had no significant effect on concern when analysing all respondents together (U= 752.500, Z = -.701, p>0.05). However, analysing relatives and affected individuals separately revealed that concern was significantly greater amongst female affected individuals (mean rank = 32.11 , median = 5.00, mean =5.219) than male affected individuals (median = 5.00, mean rank = 23.69, mean = 4.500, U = 268.500, Z = -1.962, p = .05). Effect size value (r = -.262) suggests a small to moderate practical significance.

Spearman's rank correlation

In order to identify potential variables influencing concern spearman's rank correlation was used to test for associations between concern and i) illness attribution variables (GA; EA) and ii) illness attribution certainty variables (GAC; EAC).

All correlations are shown in table 17.

The only testable variable significantly associated with concern was attribution of mental illness to genetic factors (GA). This correlation was significant and positive, for both affected individuals (r_s=.324, p=0.014) and relatives (r_s =.558, p=0.002).

Thus, for all respondents, greater concern for relatives becoming ill was associated with greater attribution of the mental illness to genetic factors (GA).

Table 17: Spearman's rank correlations between concern and attribution and attribution certainty variables for affected individuals and relatives

Concern (group)	***Testable variable***			
	GA	GAC	EA	EAC
Concern (Affected individuals)	.324**	.187	.005	.009
Concern (Relatives)	.558**	-.063	-.308	-.158

*Note: GA = attribution of mental illness to genetic factors, GC = Certainty regarding attribution of mental illness to genetic factors, EA = attribution of mental illness to environmental factors, EC = Certainty regarding attribution of mental illness to environmental attribution. *p <.05, **p <.01*

ii. Perceptions of familial risk (Risk estimation)

To explore accuracy of perceptions of quantitative risk amongst respondents, respondents were asked to estimate the chances of i) the offspring and ii) the sibling of an individual *with their or their relative's mental illness* also developing mental illness.

Anchored options provided were: 1% (representing an approximation of population base rate) 10% (representing an approximation of actual first-degree relative risk), 25%, 50%, 100%. (three higher than actual risks*) "Don't know" and "Other" were two alternative options provided.

**For depression, estimations of 50% or 100% were considered 'higher than actual' risks, as age-adjusted risk to first-degree relatives is 5-30%.*

Perceptions of familial risk (Risk estimation) - Results

Risk to offspring

For all diagnoses, 16% of affected individuals (n=9) and 19% of relatives (n=5) reported that they did not know the risk of recurrence to offspring.

Of the 70 respondents that provided actual estimates to offspring, 63% of affected individuals (n=30) and 82% of relatives (n=18) overestimated risk. 30% of respondents (n=21) correctly estimated risk to offspring. Only one respondent, a relative, underestimated risk to offspring.

All data are shown in tables 18-19 (following page)

Table 18: Frequency table showing affected individuals' estimation of risk to offspring.

Psychiatric Diagnosis	Risk estimation to offspring – affected individuals						TOTAL
	1%	10%	25%	50%	100%	I don't know	
BPD	0	11[a]	8	14	1	4	38
Psychosis	0	1	0	0	1	3	5
Depression	0		2	2	0	2	6
Anxiety disorders	0	3	1	2[b]	0	0	6
Other	0	1	0	1	0	0	2

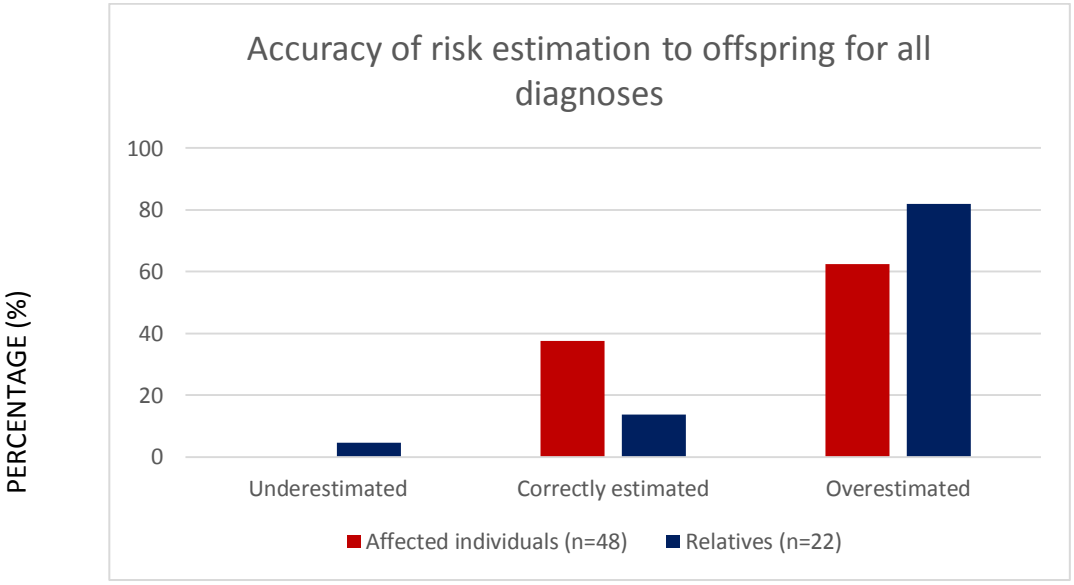
Note: BPD = Bipolar disorder 1, Bipolar disorder 2, rapid cycling. 'Psychosis' = schizophrenia, schizoaffective disorder, psychotic depression. Depression = recurrent depression, clinical depression. Anxiety disorders = anxiety, anxiety with depression, OCD, PTSD including with depression and/or anxiety. Diagnoses grouped according to ICD-10 classification. 'Other' = anorexia. Overestimations are highlighted in yellow; responses reporting uncertainty of risk are highlighted in turquoise. a = 15% estimate included, b= 75% estimate included. n=57.

Table 19: Frequency table showing relatives' risk estimation to offspring.

Psychiatric Diagnosis	Risk estimation to offspring – relatives						TOTAL
	1%	10%	25%	50%	100%	I don't know	
BPD	0	1	2	4	1	2	10
Psychosis	1	1	1	4	0	1	8
Depression	0	0	0	1	0	0	1
Anxiety disorders	0	1	1	4	0	1	7
Other	0	0	0	0	0	1	1

Note: BPD = Bipolar disorder 1, Bipolar disorder 2, rapid cycling. 'Psychosis' = schizophrenia, schizoaffective disorder, psychotic depression. Depression = recurrent depression, clinical depression. Anxiety disorders = anxiety, anxiety with depression, OCD, PTSD including with depression and/or anxiety. Diagnoses grouped according to ICD-10 classification. 'Other' = anorexia. Overestimations are highlighted in yellow; responses reporting uncertainty of risk are highlighted in turquoise. n=27.

Figure 43: Bar chart showing accuracy (%) of respondents' risk estimation to offspring



Note: Represents respondents providing an estimate of risk; respondents that indicated they did not know risk were excluded. For all diagnoses. n=70.

Risk to sibling

Of all respondents that answered this question (n=82), across all diagnoses, 11% of affected individuals (n=6) and 19% relatives (n=5) reported that they did not know the risk to sibling.

Of the respondents that provided an estimate of risk (n=71), 28% of affected individuals (n=14) and 29% of relatives (n=6) correctly estimated risk. Conversely, 60% affected individuals (n=30) and 48% of relatives (n=10) overestimated risk. 28% (n=14) of affected individuals and 28% (n=6) relatives correctly estimated risk. 12% affected individuals (n=6) and 24% relatives (n=5) underestimated risk.

28% of affected individuals (n=14) and 29% of relatives (n=6) correctly estimated risk. 12% of affected individuals and 24% relatives underestimated risk.

All data are shown in tables 20-21 and fig. 44.

Table 20: Frequency table showing affected individuals' risk estimation to sibling.

Psychiatric Diagnosis	Risk estimation to siblings – affected individuals						TOTAL
	1%	10%	25%	50%	100%	I don't know	
BPD	4	10	14	4	1	4	37
Psychosis	0	1	1	2	0	1	5
Depression	0	1	0	4	0	1	6
Anxiety disorders	2	1	1	2[c]	0	0	6
Other	0	1	0	1	0	0	2

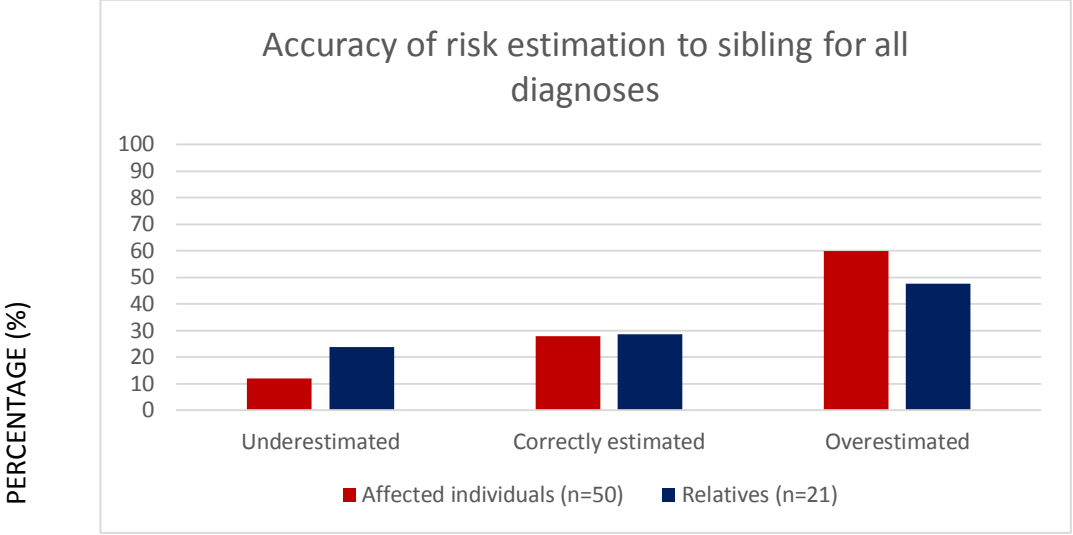
Note: BPD = Bipolar disorder 1, Bipolar disorder 2, rapid cycling. 'Psychosis' = schizophrenia, schizoaffective disorder, psychotic depression. Depression = recurrent depression, clinical depression. Anxiety disorders = anxiety, anxiety with depression, OCD, PTSD including with depression and/or anxiety. Diagnoses grouped according to ICD-10 classification. Overestimations are highlighted in yellow; responses reporting uncertainty of risk are highlighted in turquoise. C = 75% risk estimate included. n=56.

Table 21: Frequency table showing relatives' risk estimation to sibling.

Psychiatric diagnosis	Risk estimation to sibling – relatives						TOTAL
	1%	10%	25%	50%	100%	I don't know	
BPD	1	3	2	2	0	2	10
Psychosis	3	1	2	0	0	1	7
Depression	0	0	0	1	0	0	1
Anxiety disorders	1	2	2	1	0	1	7
Other	0	0	0	0	0	1	1

Note: BPD = Bipolar disorder 1, Bipolar disorder 2, rapid cycling. 'Psychosis' = schizophrenia, schizoaffective disorder, psychotic depression. Depression = recurrent depression, clinical depression. Anxiety disorders = anxiety, anxiety with depression, OCD, PTSD including with depression and/or anxiety. Diagnoses grouped according to ICD-10 classification.. Overestimations are highlighted in yellow; responses reporting uncertainty of risk are highlighted in turquoise. C = 75% risk estimate included. n=26.

Figure 44: Bar chart showing accuracy of respondents' risk estimations to sibling



Note: Represents respondents providing an estimate of risk; respondents that indicated they did not know risk were excluded. For all diagnoses. n=71

Perceptions of familial risk (Risk estimation) - Statistical analyses

Mann Whitney U-Test

To attempt to identify factors influencing accuracy of risk estimation, Mann-Whitney U-Test was used to enable comparison between respondents who overestimated risk to offspring and those who did not overestimate risk to offspring in regards to i) illness attribution variables (GA; EA) ii) illness attribution certainty variables (GAC; EAC) and ii) concern for other relatives becoming ill (C).

Concern for relatives becoming ill was significantly greater amongst respondents that overestimated risk (MDN = 5, mean rank = 38.86) than those who did not overestimate risk (MDN = 4, mean rank = 28.16, U = 366.500, Z = -2.094, p = 0.036).

Analysing affected individuals and relatives separately, concern for relatives becoming ill was significantly greater amongst affected individuals that overestimated risk (MDN = 5.5, mean rank =28.05) than those that did not overestimate risk (MDN = 4.5, mean rank = 12.39,U = 163.500, Z = -2.355, p = .019).

No other significant differences were found.

iii. Effect of mental illness on family planning decisions

To assess impact on family-planning, respondents were presented with the following question:

'Has your diagnosis of mental illness affected your decision-making regarding family planning, or do you think it may in the future?

Anchored responses provided were 'Yes,' 'No' or 'other' in which respondents were provided with a free-form response to write their own answer.

Respondents that selected 'Yes' were also asked to describe how the mental illness had impacted their family-planning decisions.

Anchored responses provided with 'A decision to have more children,' 'A decision to have less children,' 'A decision to have no children,' or 'Other' in which respondents were provided with a free-form response to write their own answer.

Effect on family-planning decisions - Results

Of the 55 affected individuals that provided answers for this question, almost half (n=27, 49%) reported that their mental illness had influenced their family-planning decisions. Of these respondents, 68% (n=19) reported it had favoured a decision to have fewer or no children.

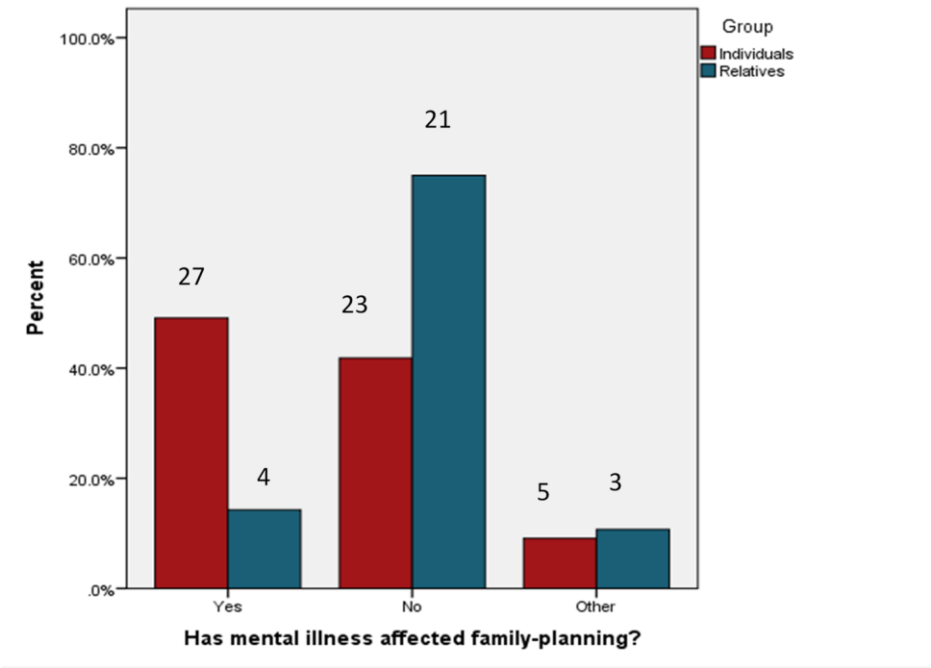
Fewer relatives (14%, n=4) indicated that their family history of mental illness had influenced their family-planning decisions, and only 2 relatives (7%) reported it had resulted in a decision to have fewer or no children.

All frequencies are shown in tables 22-23 and fig. 45.

Table 22: Frequency table showing impact of mental illness on family-planning decisions for affected individuals and relatives.

Group	Affected family-planning?			**TOTAL**
	Yes	No	Other	
Affected individuals	27	23	5	55
Relatives	4	21	3	28

Figure 45: Frequency diagram showing impact of mental illness on family-planning decisions for respondents.



Significantly more affected individuals (red bars) reported effects on family-planning than relatives (blue bars)

Table 23: Frequency table describing effect of mental illness on family-planning decisions

Group	Effect on family-planning				**TOTAL**
	More children	Less children	No children	Other	
Affected individuals	0	4	15	8	27
Relatives	0	0	2	2	4

Effect on family planning – statistical analyses

Chi-square test

Chi-square test revealed a significant difference on impact on family planning between affected individuals and relatives (χ^2 (1, n=75) = 9.925, *p*= 0.002). Odds ratio = 6.163.

Using the equation

$$RR = \frac{OR}{(1 - P_{ref}) + (P_{ref} * OR)}$$

Whereby RR = risk ratio; OR = odds ratio
P_{ref} = Prevalence of the outcome in the reference group

Gives RR as 3.375

This seems to represent that the chances of affected individuals reporting effect on family planning is 3.375 times higher than relatives.

Comparing genders, there was no significant difference between males and females when all respondents were analysed together (χ^2 (1, n=74) = .323, *p>.05*).

However, analysing affected individuals and relatives separately, there was a significant difference between female affected individuals and male affected individuals in regards to family-planning (χ^2 (1, n=49) = 4.426, p =.046). Odds ratio = 3.529.

Using the equation

$$RR = \frac{OR}{(1 - P_{ref}) + (P_{ref} * OR)}$$

Whereby RR = risk ratio; OR = odds ratio
P_{ref} = Prevalence of the outcome in the reference group

Gives RR as 2.156

This seems to represent that, based on the odds ratio, the chances of female affected individuals reporting effects on family planning is 2.156 times higher than male affected individuals.

Mann-Whitney U-test

To attempt to identify factors influencing family-planning decisions, Mann-Whitney U-Test was applied to compare differences between respondents who reported effects on family-planning, and those who reported no effect on family-planning, in regards to i) illness attribution variables (GA; EA) ii) illness attribution certainty variables (GAC; EAC) and ii) concern for other relatives becoming ill (C).

Analysing all respondents together, attribution of mental illness to genetic factors (GA) was significantly greater amongst respondents whom reported effects on family planning (median=4.00) than those who did not (median = 3.50, U = 437.500, Z = -2.692, p = 0.007). Effect size value (r=-.311) suggests moderate practical significance, indicating this difference is both significant and substantive.

Additionally, concern for other relatives becoming ill (C) was significantly greater amongst respondents whom reported effects on family planning (median = 5.00) than those who did not (MDN =4.500, U = 431.500, Z = -2.746, p = 0.006, Effect size value (r=-.317) suggests moderate practical significance, indicating this difference is both significant and substantive.

Analysing only affected individuals, attribution of mental illness to genetic factors (GA) was significantly greater amongst affected individuals whom reported effects on family planning (median = 4.00) than those who did not (median = 3.00, U = 164.500, Z = -2.921, p = 0.003, r=-.413); the medium effect size indicates this difference is both significant and substantive.

Additionally, certainty regarding the attribution of illness to genetic factors (GAC) was significantly greater amongst affected individuals whom reported effects on family planning (median= 6.00) than those who did not (median = 5.00, U =202.500, Z= -1.970, p= 0.049, r=-.281).

No other significant differences were found between respondents.

Summary of findings

Across all diagnostic categories, the majority of respondents quantitatively overestimated risk to both sibling and offspring of somebody with their or their relatives' mental illness. Of all respondents providing estimates, almost half believed the risk to offspring to be 50% or higher. Overall, there was greater accuracy in regards to risk estimation to sibling in comparison to risk estimation to offspring.

A notable proportion of respondents also indicated complete uncertainty regarding risk to first-degree relatives, with almost 20% (n=14) respondents reporting that they did not know the risk to offspring and almost 15% (n=11) that they did not know the risk to sibling.

Concern about familial risk was high amongst respondents, with almost three quarters of respondents (n=64) reporting being concerned about familial risk, and almost a third (n=27) indicating very high degrees of concern. Concern was significantly greater amongst affected individuals in comparison to relatives, and affected individuals that were female in comparison to affected individuals that were male, indicating both being female and having a mental illness is associated with greater concern over familial risk.

Additionally, for all respondents, concern was significantly and positively associated with attribution of mental illness to genetic factors, indicating greater endorsement of a genetic explanation of illness is associated with greater concern over familial risk.

In regards to family-planning, almost half of affected individuals (n=27) reported that their mental illness had affected their family planning decisions, and of these respondents the majority (n=19) stated this resulted in decisions favouring having fewer or no children. Conversely, only 4 relatives reported that their relatives' mental illness had influenced their family-planning decisions.

For all respondents, attribution to genetic factors was significantly greater amongst respondents that reported effects on family-planning than those that did not.

3.4 Awareness and perceptions of GC and PGC

This section examined respondents' awareness of genetic counselling prior to taking part in the study, and qualitatively assessed respondents' perceptions of GC and quantitatively assessed perceptions of PGC, prior to respondents receiving information about genetic counselling.

i. Awareness of GC

To assess prior awareness of GC, respondents were presented with the question: "Had you heard of the term "genetic counselling" before participating in this study? You do not have to know what it is." Anchored responses provided were 'yes' or 'no.'

Respondents who identified they **had** previously heard of GC were then asked to identify how they had previously come across the service.

Respondents were presented with anchored responses, listed in table 24, and asked to select the most relevant answer. Respondents were also provided with the alternative option 'other,' which provided a drop-down free-entry box in which they could write their own answer.

Awareness of GC - Results

Of the 84 respondents that answered this question, the majority of affected individuals reported no prior awareness of GC, with (62%, n=34) having not heard of GC prior to participating in this study. Conversely the majority of relatives (66%, n= 19) reported they had heard of GC prior to participation.

All data are given in table 24.

Table 24: Frequency table showing awareness of GC prior to partaking in study

Group	Prior awareness of GC?		**TOTAL**
	Yes	No	
Affected individuals	21	34	55
Relatives	19	10	29

Chi-square test showed significantly greater awareness amongst relatives than affected individuals (χ^2 (1, n=84) = 5.688,*p*= 0.022). Odds ratio = 3.076.

Using the equation:

RR= (Probability exposed)/(probability non-exposed)

Gives the equation: $RR = \frac{OR}{(1 - P_{ref}) + (P_{ref} * OR)} = 1.716$

Whereby RR = risk ratio; OR = odds ratio
P_{ref} = Prevalence of the outcome in the reference group

This seems to represent that relatives were 1.716 times more likely to have heard of GC as affected individuals.

There was no significant difference in awareness between males and females (χ^2 (1, n=83) =.829 , *p>.05).*

ii. Sources of exposure to GC

Respondents whom stated they **had** heard of GC prior to taking part in the study (n=40) were asked to indicate how they had come across GC and were provided with anchored responses: *1)Have received PGC 2) Have received GC for another health condition 3) Relative/friend has received GC 4) News 5) Internet 6) TV/Film 7) School/college/university 8) Am/ am training to become a GC 9) Through my job 10) Other* (see table 25 and figure 27b), as well as the option to write their own alternative response.

Results are given on the following pages

Sources of exposure to GC - Results

Over half of respondents (58%, n=23) that reported awareness of GC reported exposure via media/communication sources (i.e. internet, TV and newspapers, see figure 27b). 10% respondents (n=4) had come across GC through their education (e.g. school/college/university), whilst only 8% respondents (n=3) were qualified or training to become genetic counsellors. Only 5% of respondents (n=2, both relatives) had actually received GC for another health condition, whilst only one respondent (~3%) reported that a friend or relative had received GC.

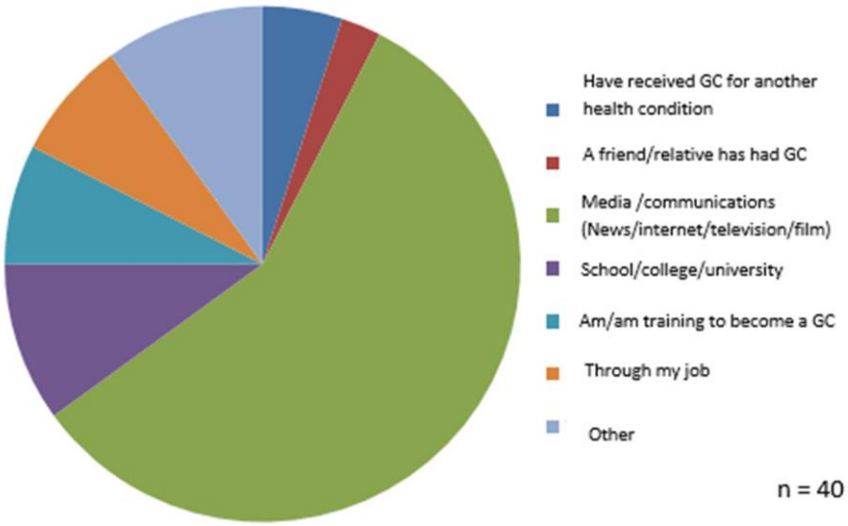
'Other' responses that were listed by relatives included having a relative that was interested in GC (n=1) and through NHS-related courses (n=1). Full data are shown in table 25 and figures 46-48.

Table 25: Frequency table showing sources of exposure to GC for respondents that reported prior awareness

Group	Source of exposure										TOTAL
	1	**2**	**3**	**4**	**5**	**6**	**7**	**8**	**9**	**10**	
Affected individuals	0	0	1	5	7	3	2	1	2	0	21
Relatives	0	2	0	4	2	2	2	2	1	4	19

Note: 1)Have received PGC 2) Have received GC for another health condition 3) Relative/friend has received GC 4) News 5) Internet 6) TV/Film 7) School/college/university 8) Am/ am training to become a GC 9) Through my job 10) Other

Figure 46: Pie chart showing sources of exposure to GC reported by respondents



The majority of respondents (58%, n=23) had come across GC through media and communication mediums such as news, TV, internet and films, represented collectively by the green segment of the pie chart.

Figure 47: Frequency diagram showing sources of exposure to GC for affected individuals

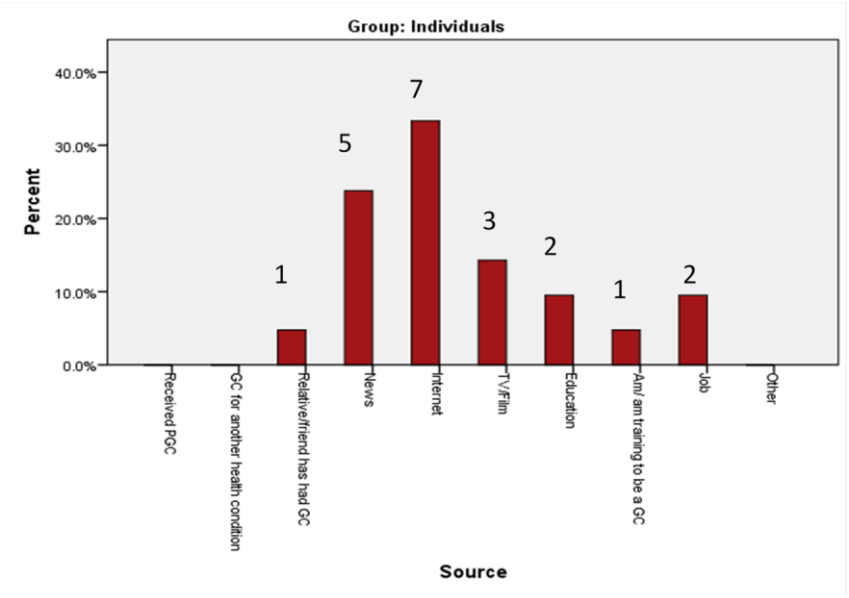
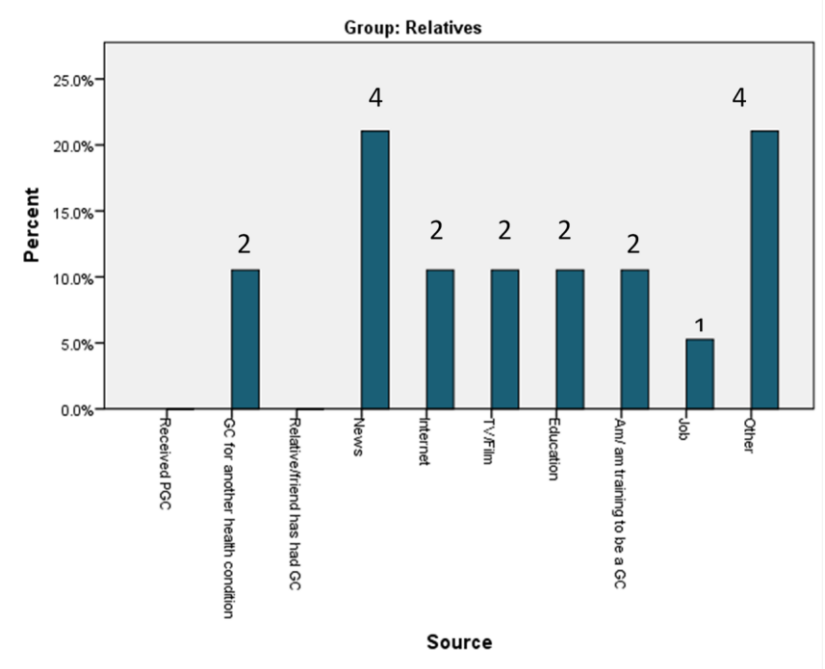


Figure 48: Frequency diagram showing sources of exposure to GC for relatives



iii. Perceptions of genetic counselling (qualitative analysis)

To explore perceptions of genetic counselling prior to receiving any information about the service, all respondents were presented with the open-ended question:

> *'What is the first thing that comes to mind when you hear the term 'genetic counselling'?'*

and were invited to write their own response.

Results are presented below.

Perceptions of genetic counselling (qualitative analysis) – Results

Because of the differential awareness of GC between affected individuals and relatives, responses were analysed separately; however, the same major themes were identified between the two groups and so were pooled for means of reporting.

Table 26 (on next page) shows views expressed according to group along with frequencies of each theme, to enable comparisons

Table 26: Themes, subthemes and examples for perceptions of GC

Table 26: Themes, subthemes and examples for perceptions of GC	**TOTAL Responses** (n=80;100%)	**AFFECTED INDIVIDUALS Responses** (n=53;100%)	**RELATIVES Responses** (n=27;100%)
Uncertainty about purpose – What is genetic counselling? *"What is it?"; "Can't see the connection between genetics and counselling."*	8 (10%)	7 (13%)	1 (4%)
Statements conceptualising GC as "just therapy" i) Psychotherapeutic counselling ii) Family therapy iii) Therapy for relatives of ill affected individuals *"Prevents distress"; "Just counselling."; "Therapy for related affected individuals."; "Therapy for family members of the sick person."*	9 (11%)	7 (13%)	2 (7%)
Statements associating GC with disease – Genetic counselling and disease i)Genetic conditions ii) iv) Non-genetic contributions to disease iii) Family history of mental illness *'Family history of illness'; 'Inherited problems'; 'Nature and nurture.'*	46 (58%)	28 (53%)	18 (67%)
Statements associating GC with concepts of familial risk – "Genetic counselling is about genetic risk" i) To offspring ii) To self and relatives iii) Genomic technologies for risk assessment (i.e. genetic testing) *"Risk of children developing the disorder"; "Chances of (children) inheriting the disorder"; "Isolating the risk gene and discussing chances of it being passed on."*	18 (23%)	9 (17%)	9 (33%)
Statements conceptualising GC as a medically therapeutic/supportive intervention" i) Coping/therapy/support in regards to having/at risk of having an illness ii) Facilitates psychological adjustment to the illness iii) Reduces self-blame *"Therapeutic counselling for people affected by genetic conditions." ;"Coming to terms with illness/risk ."""Reinforce the belief that it's not the individual's fault they developed this illness."*	11 (14%)	6 (11%)	5 (19%)
Statements associating GC with decision-making in regards to having/being at risk of having illness i) Advice to base decisions on ii) Understanding treatment options iii) Risk-reduction strategies iv) Reproductive decision-making *"Advice regarding choices":"Taking action before illness manifests"; "Information about genetic conditions when deciding to start a family."*	7 (9%)	4 (8%)	3 (11%)
Statements associating GC with ethical/moral issues i) Eugenic-type values/directive counselling ***"Being told not to have children so you don't pass on your defective genes.";"To remove abnormality from the gene pool."*** ii) May cause psychological distress *"Gives people options but also dilemmas." "Might cause worry/anxiety."*	9 (11%) *3 (4%)* *6 (8%)*	8 (15%) 3 (6%) 5 (9%)	1 (4%) 0 1 (4%)

"What is genetic counselling?"

One relative and several affected individuals stated that they had not heard of genetic counselling before taking parting in the study and were uncertain about what it meant, or what it would involve and/or what its purpose may be.

> "What is it?" (Relative);

> "Can't see the link between genetics+ counselling." (Affected individual)

GC is "just therapy"

A number of affected individuals and two relatives conceptualised GC as a therapeutic intervention but not specifically in relation to having or being at risk of having genetic conditions:

> "Preventing distress"; (Affected individual)

> "Just counselling, nothing too specialised." (Relative)

A few affected individuals specifically indicated they believed GC may be a form of therapy for related affected individuals:

> "councelling(sic) around people genetically related."

> "Bringing the whole family together to talk about any mental ill health…"

Additionally, a small proportion of respondents conceptualised GC as counselling specifically for relatives of affected individuals affected by medical conditions:

> "Talking with and supporting the relatives of the person with the illness." (Affected individual)

> "Counselling provided for family of a mentally ill person, to try and prevent them also suffering the same condition." (Relative)

Genetic counselling and disease"

The majority of respondents correctly conceptualised GC as a medical intervention regarding disease; and most affected individuals and relatives associated GC with genetic conditions and/or family history of a medical disorder.

> "…. I'd guess it's about inherited problems" (Affected individual)

> "Someone seeking genetic counselling may be concerned about hereditary illness in their family." (Relative)

Several respondents named specific genetic conditions including Huntingdon's, muscular dystrophy, cystic fibrosis, and cancer:

> "Someone seeking genetic counselling may be concerned about hereditary illness in their family e.g. heart conditions, cancer etc." (Relative)

> "That it is to do with passing on faulty genes, for example Tay Sach's disease." (Individual)

Only one respondent indicated that GC may also involve discussions non-genetic contributions to disease:

> "Counselling about Nature N(sic) Nurture" (affected individual).

A proportion of respondents discussed GC in relation to genetic contributions mental illness specifically:

> "The professionals would look at the biological workings of the family as a whole to identify any trends or similarities which may lead to a conclusion that mental health issues are possibly hereditary…" (Relative)

> "Counselling about the genetic factors affecting mental illness…" (Individual)

“GC involves information about familial risk”

A proportion of respondents associated GC with concepts regarding familial risk of genetic disease. Respondents typically discussed how GC might involve information or advice regarding risk to offspring or future descendants, as well as to themselves and other family members:

> “Talking through how mental illness may affect children by passing on the illness through the blood line.” (Relative)

> “An examination regarding the breakdown of inherited genetic traits (with regards to mental health or otherwise) and potential risk factors for subsequent descendants.” (Affected individual)

> “Advice given in regard to a parent to be or a family on the likelihood of genetics effecting their offspring, or indeed current family members in the future.” (Relative)

> “…discussing the likelihood of that gene affecting future or existing members of the family.” (Individual)

In addition a small number of respondents specifically alluded to concepts related to genomic technologies (e.g. testing) for medical conditions, in order to obtain risk assessments:

> “The analysis of ones genes to determine those which carry the propensity for certain conditions.” (Relative)

> “Isolating the gene which causes the illness and discussing the likelihood of that gene affecting future or existing members of the family.” (affected individual)

“GC may be supportive/therapeutic for those with a medical condition”

Several respondents conceptualised GC as helpful, supportive, and facilitating coping for those with/at risk of a genetic/medical condition, including alluding that it may facilitate psychological acceptance to illness or risk:

> “Therapy for people’s who’s(sic) genes show potential to have an illness.” (Relative)

> “Counselling for coping with a congenital(sic) illness” (affected Individual)

> “Counselling regarding the genetic history of a disorder and how to come to terms with it.”(affected individual)

> “…If hereditary, the counselling aspect would be to help those come to terms with the possibility that they may ‘inherit’ a relative’s mental health condition or one similar.” (Relative)

Additionally one affected individual indicated that GC may reduce feelings of self-blame around illness causation by alleviating feelings of self-blame regarding the illness:

> “It makes me think that a sufferer of a particular mental illness could be counselled about the likelihood that their disability has been passed onto them and therefore hopefully re-inforce the belief that it is not their own fault that they have developed such a disability.”

“GC may help with decision-making”

A few respondents conceptualised GC as an information-provision service that may provide advice that may facilitate decision-making relation to having an illness or being at risk of having an illness, that may reduce risk of illness or facilitate better management, including better understanding available treatment options and risk reduction strategies: and some reflected on their own experiences of GC:

> “Have had genetic counselling in the past as have a family history of muscular dystrophy. Was visited by a specialist health visitor and advised regarding choices.” (Relative)

> “Finding the suitable genetic precipitant for a particular health issue and how to digest information with regards to future decisions/treatments.” (affected individual)

> “…Being given advice that it may be in my interests to take action before a disease/illness manifests, e.g. pre-emptive surgery to prevent cancer.” (affected individual)

A small proportion of respondents associated GC with reproductive settings and/or reproductive decision-making:

> “I think it means being advised about genetic disorders when deciding to have a family…” (affected individual).

> “It makes me think of a couple hoping to start a family but are concerned about the risk factor of having a child who may develop a mental illness…” (Relative)

Ethical/moral issues

Several respondents raised ethical and moral issues that may be associated with providing GC.

A few affected individuals associated GC with eugenic-type values, with some conceptualising it as a directive approach that may be used to influence individual's reproductive decisions, to reduce the presence of certain genes/characteristics within the population:

> "There is the fear that it has the potential to slip into eugenic thinking." (affected individual)

> "The idea that certain characteristics have been deemed as undesirable by others and people are given counselling to irradiate(sic) the abnormality from the gene pool. It feels cold, unfeeling, unhelpful, disrespectful… and is extremely worrying." (Individual)

> "Being told not to have a kid because U(sic) may pass on your defective genes." (Individual)

Additionally, a small number of affected individuals and one relative made statements associating GC with psychological distress. A proportion (n=2) proposed that the information received may increase anxiety and induce worry and/or fear in affected individuals, including themselves personally, particularly in relation to concern regarding familial risk, alluding to concepts of genetic determinism:

> "Trying to put a mentally ill person into a position where they are worrying about their children being ill like themselves…" (Individual)

> "It would be horrible for me to find out I have given this illness to my children. And they might give it to my grandchildren." (Individual)

Whilst another individual simply stated:

> "Being given bad news."

A few other respondents alluded that the information gained may present cognitive burden by inducing decision-making uncertainty or presenting new psychological and/or ethical considerations for people, and some specifically due to the uncertain nature of genetic information:

> "Would give people options but also dilemmas…" (Relative)

> "…I wonder if the suggestion of counselling for a problem that might or might not occur could cause more anguish and problems than it might alleviate." (Individual)

iv. Perceptions of Psychiatric genetic counselling (quantitative analysis)

To explore baseline beliefs and perceptions regarding the process and purpose of GC specifically for psychiatric conditions, respondents were presented with statements about PGC (listed in figs 49-52), prior to watching the informational video, and asked to indicate whether they believed them to be true or false.

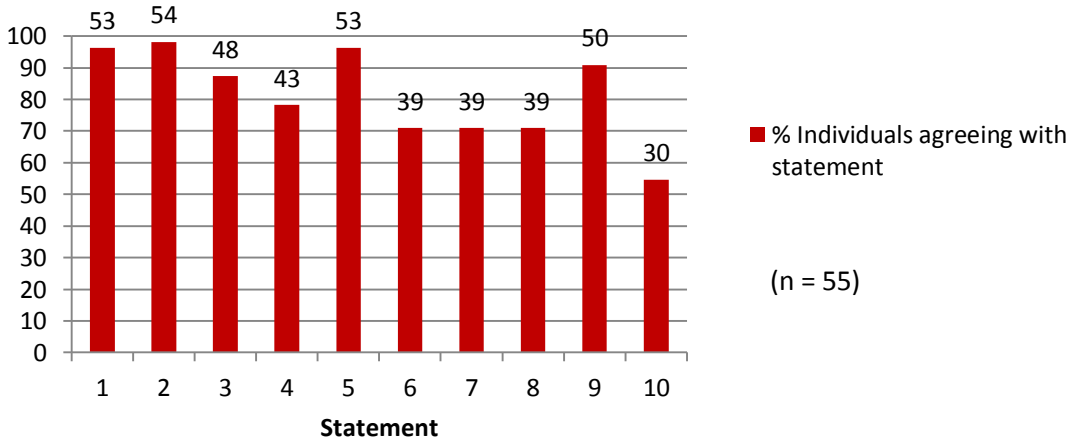
Perceptions of Psychiatric Genetic Counselling (quantitative analysis) – Results

Of the 82 respondents that answered this question, over 96% of affected individuals (n=53) and 100% (n=55) of relatives believed that a GC would provide information about the genetic contributions to mental illness. 89% respondents (n=73) correctly believed that a genetic counsellor would provide information about the chances of other children becoming ill.

However fewer affected individuals (71%, n=39) and relatives (74%, n=20) believed that GC could provide information about non-genetic factors involved in mental illness pathogenesis. Comparatively fewer respondents (74%, n=61) also believed a GC would provide information about protecting mental health. Additionally, only 55% of affected individuals (n=30) and 70% of relatives (n=19) believed a GC would provide emotional support.

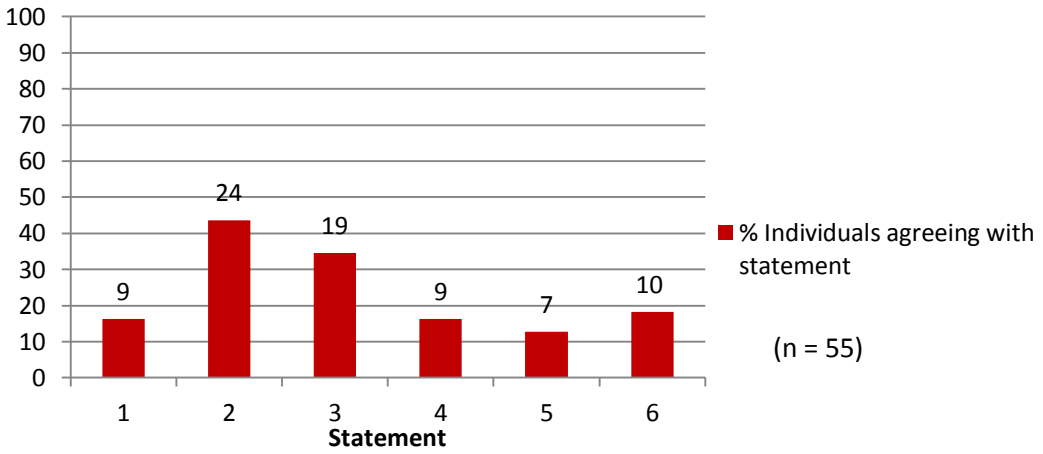
Almost half of affected individuals (n = 24) and 52% of relatives incorrectly believed that a genetic counsellor could arrange genetic tests to diagnose mental illness in themselves or their relatives, and 35% of affected individuals (n=19) and 30% of relatives (n=8) incorrectly believed pre-natal testing for mental illness could be arranged through a genetic counsellor. Almost 20% of affected individuals (n=9) believed a genetic counsellor would advise them whether or not to have children, and 15% of relatives (n=4) reported that they believed genetic counsellor would advise their affected relative whether or not have children.
Full data are given in figs. 49-52.

Figure 49: Perceptions of PGC - Affected individuals: Agreement with correct statements



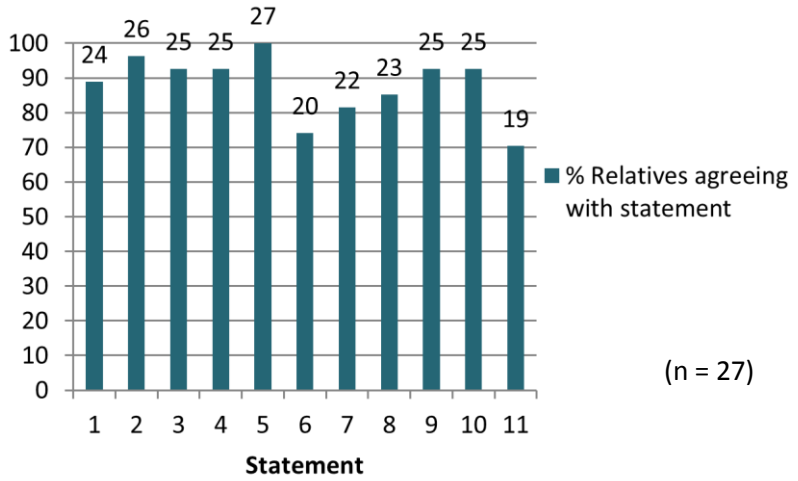
Key: 1)Gather information about my family's medical history 2)Gather information about my family's history of mental illness 3)Provide information about the chances of my children (including form future pregnancies) also becoming ill 4)Provide information about the chances of other relatives also becoming ill 5)Provide information about the genetic contributions in mental illness 6)Provide information about the non-genetic factors in mental illness 7)Discuss ways I can protect my mental health 8)Discuss ways my relatives can protect their mental health 9)Provide referrals and information to other services that may be relevant to me 10)Provide emotional support

Figure 50: Perceptions of PGC - Affected individuals: Agreement with incorrect statements



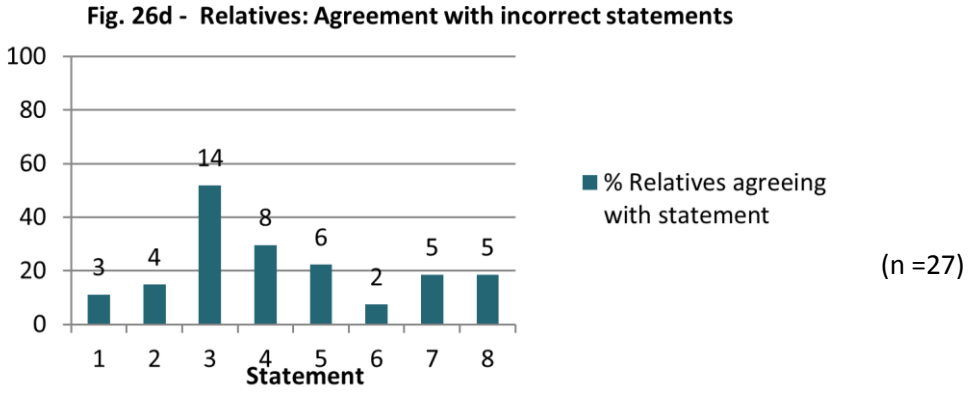
Key: 1) Advise me whether or not to have children 2) Arrange genetic tests to diagnose mental illness in myself or my relatives 3)Arrange genetic tests to test for mental illness in future pregnancies 4)Prevent future children from having mental illness 5)Arrange gene therapy to cure mental illness 6)Decide what medications I should take for my mental illness

Figure 51: Perceptions of PGC – Relatives: Agreement with correct statements



KEY: 1)Gather information about my family's medical history 2)Gather information about my family's history of mental illness 3)Provide information about the chances of my children (including form future pregnancies) also becoming ill 4)Provide information about the chances of other relatives also becoming ill 5)Provide information about the genetic contributions in mental illness 6)Provide information about the non-genetic factors in mental illness 7)Discuss ways I can protect my mental health 8)Discuss ways my relative with a mental illness can protect their mental health 9)Provide referrals and information to other services that may be relevant to my relative with a mental illness 10)Provide referrals and information to other services that may be relevant to me 11)Provide emotional support

Figure 52: Perceptions of PGC – Relatives: Agreement with incorrect statements



KEY: 1) Advise me whether or not to have children 2) Advise my relative with a mental illness whether or not to have children 3) Arrange genetic tests to diagnose mental illness in myself or my relatives 4) Arrange genetic tests to test for mental illness in future pregnancies 5)Prevent future children from having mental illness 6)Arrange gene therapy to cure mental illness 7)Decide what medications my relative should take for their mental illness 8)Tell me what medications I can take to prevent myself from developing mental illness

Summary of findings

Collectively less than half of respondents reported having heard of GC prior to participation, and awareness was significantly lower amongst affected individuals in comparison to relatives. Of the respondents that had heard of GC almost 60% stated awareness was via media and communication sources such as the television, films and internet. A small proportion reported being GC's themselves.

To a degree respondents indicated an understanding of the process and goals of both traditional GC (assessed qualitatively) and GC specifically within psychiatry (assessed quantitatively).

For example, the majority of respondents conceptualised GC, qualitatively, as a medical intervention for affected individuals with or at risk of having a genetic condition; that it often involves learning about genetic risk to family members, and some identified that it may help with decision-making, typically in regards to reducing genetic risk. A number of respondents also discussed how GC may have therapeutic values, predominantly conceptualised as enabling psychological or emotional acceptance of the condition or risk (e.g. "coming to terms with the condition.").

In regards to perceptions of GC specifically for psychiatric conditions, almost all respondents correctly identified that a GC would gather family history of mental illness, discuss information about the genetic contributions to mental illness, and provide estimates for the risk of a child developing mental illness, for example.

However respondents also demonstrated some limited comprehension and misconceptions regarding the practice and purpose of both GC and PGC.

For example, in regards to perceptions of GC, respondents often conceptualised the patient's role as passive, e.g. the patient was given information, or being told what action to take, in contrast to GC being a two-way, dynamic exchange between patient and counsellor. Additionally, other psychotherapeutic outcomes of GC, such as reducing guilt and self-blame, alleviating anxiety, and addressing shame and stigma were not discussed by the majority of respondents, with only one proposing that GC may reduce 'self-blame.' Furthermore, whilst the majority of respondents discussed concepts relating to risk communication and genetics, only one

respondent indicated that information about aetiology may incorporate information about non-genetic factors (i.e. “nature v. nurture”).

Notably, a few affected individuals also associated GC with eugenic-type values, conceptualising the role of the GC as directive, with the GC influencing the patient’s decision-making in regards to family-planning – as one respondent put it: “counselling to irradiate (sic) abnormalities from the gene pool.”

In regards to GC specifically for psychiatric conditions, comparatively fewer respondents correctly identified a GC would provide emotional support, discuss the contribution of genetic factors in mental illness pathogenesis, and provide information about protecting mental health, which are in practice integral to PGC interventions. Additionally, almost half of respondents incorrectly believed that a GC would provide diagnostic testing and prenatal testing, which is not currently clinically available for psychiatric conditions, and almost 20% believed a GC would influence reproductive decision-making and advise an affected individual whether or not to have children, rather than promoting autonomous decision-making.

3.5 Interest in receiving PGC

Interest in receiving PGC was initially assessed before respondents had received information about GC and PGC.

Respondents that indicated they would not like to have PGC, or were unsure about whether they would like to have PGC, were invited to indicate why they might not wish to receive PGC, and provided with anchored responses, listed in figures 27a and 27b.

Respondents then watched the informational video which provided information about PGC and clarified some common misconceptions about the service.

Following the informational video, respondents' interest in receiving PGC was then assessed again, using a Likert-type response item.

Respondents' perceived usefulness of the service, also using a Likert-type response item, was also assessed following the informational video.

Results are given in the following pages

i. Interest in PGC prior to watching informational video

Prior to watching the informational video, respondents were asked 'Would you like to have genetic counselling regarding your mental illness (for affected individuals)/the mental illness in your family (for relatives)'.

Anchored responses were 'Yes,' 'No,' or 'Not sure.'

Interest in PGC prior to watching informational video - results

Of the 83 respondents that answered this question, 45% affected individuals (n=25) and 46% of relatives (n=13) indicated they would like to receive PGC. 24% (n=13) of affected individuals and 29% (n=8) of relatives indicated they would not like to receive PGC. 31% (n=17) affected individuals and 25% (n=7) relatives indicated they were uncertain about whether they would like to receive PGC.

Chi square analysis on interest in receiving PGC did not differ significantly between affected individuals and relatives (χ^2 (1, N=59) = .089, p>.05), or between males and females χ^2 (1, N=58) = 4.230, p=.075).

Frequencies are shown in table 27.

Table 27: Frequency table showing demand for PGC prior to watching informational video

Group	Like to receive PGC?			**TOTAL**
	Yes	No	Not sure	
Affected individuals	25	13	17	55
Relatives	13	8	7	28

ii. Reasons for not wanting PGC prior to watching informational video

Respondents that selected ‘No’ or ‘Not sure’ in response to the question ‘would you like to have PGC’ were invited to explain why they may not wish to receive the service.

Respondents were provided with a list of statements (see figures 53-54) and asked to select any answers any which applied to them.

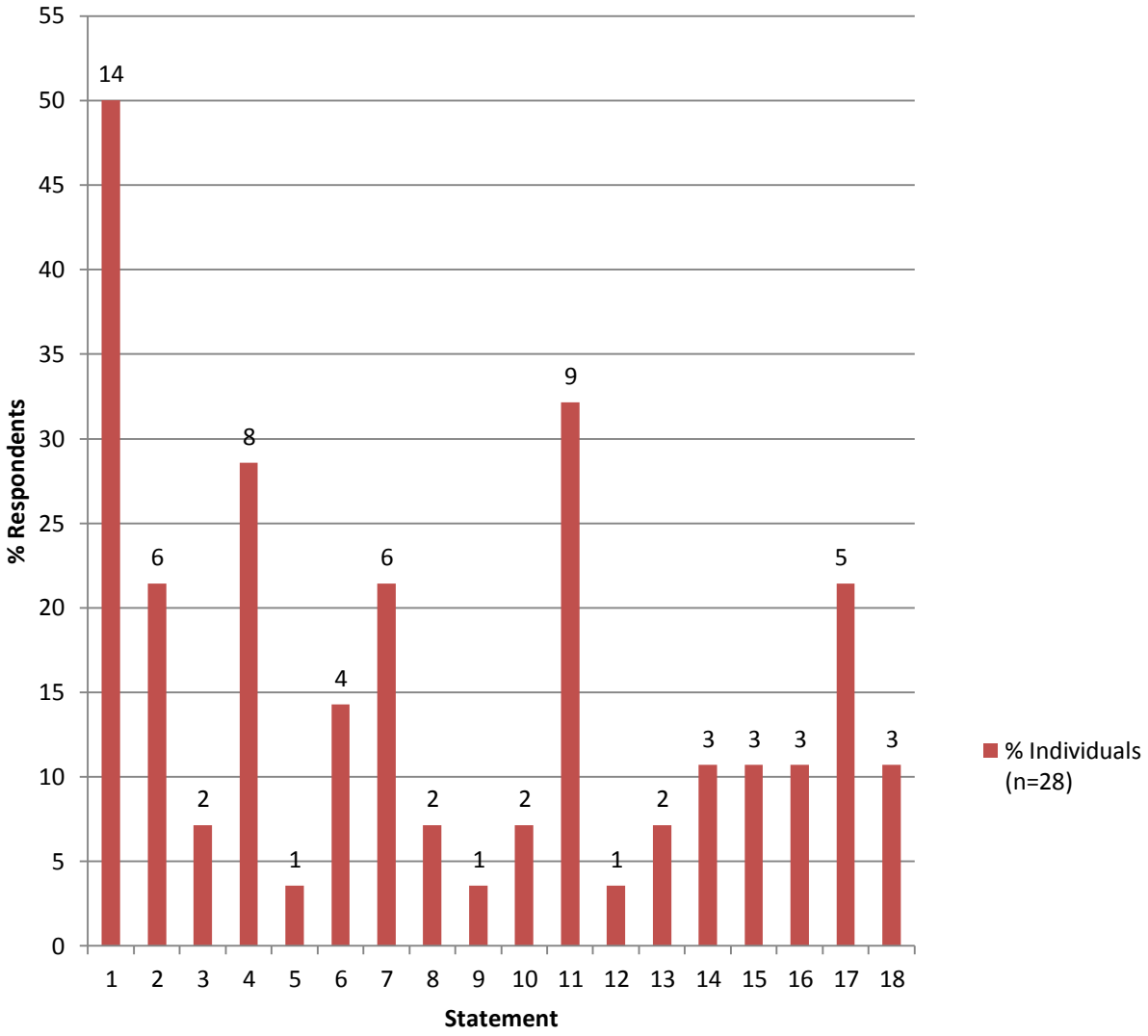
Reasons for not wanting PGC prior to watching informational video - results

Of the 43 respondents that answered this question, half of affected individuals (n=14) and 47% of relatives (n=7) identified that they would not wish to receive PGC because they did not know enough about the service. Almost a third of affected individuals (n=9) reported being worried that they would find out things they wish they hadn’t; 20% of relatives (n=3) reported this a possible deterrant. A proportion of respondents (21%, n=9) believed they would not be able to afford an appointment with a GC.

Several affected individuals (21%, n=6) reported that they would not wish to receive PGC because they did not have or did not want to have children, however no relatives reported this as a potential reason. Conversely, a number of relatives (n=4,26%) but only one affected individual reported not knowing their family history of mental illness as a potential barrier.

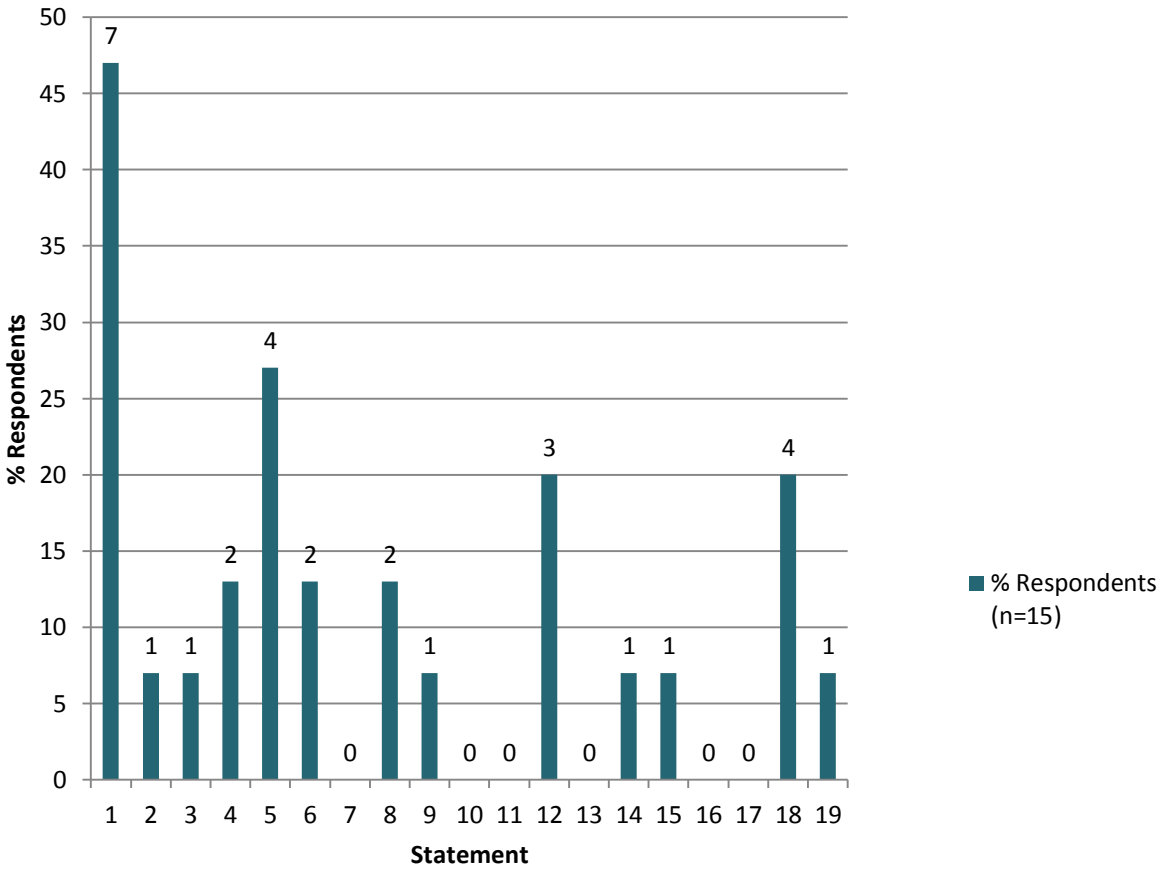
Full data for all items are shown in figures 53-54 (following page).

Figure 53: Reasons for not wanting PGC prior to watching informational video (Affected individuals)



Key: 1) I do not know enough about psychiatric genetic counselling 2) There is no role/only a small role for genetics in mental illness 3) MY mental illness is not genetic - no other affected individuals in my family have this mental illness 4) Scientists still don't know what gene(s) cause mental illness 5) I don't know my family history of mental illness 6) I don't want to have genetic testing 7) I do not have children or do not want to have children 8) I am worried the genetic counsellor might tell me not to have children 9) I don't want to know the chances of me or my relatives developing mental illness 10) The people I care most about are past the age at which they'd develop mental illness 11) I am worried I will find out things I wish I hadn't 12) I am worried the genetic counsellor will tell me my mental illness is my fault 13) I am worried the genetic counsellor might tell me there is nothing I can do about my mental illness 14) I am not currently unwell 15)There is not a lot that can be done to prevent mental illness 16) I do not have the time 17) I do not think I can afford an appointment with a genetic counsellor 18) I am worried it will affect my insurance or privacy

Figure 54: Reasons for not wanting PGC prior to watching informational video (Relatives)

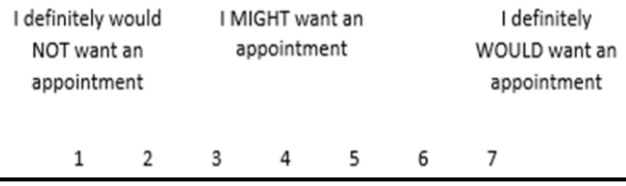


Key: 1) I do not know enough about psychiatric genetic counselling 2) There is no role/only a small role for genetics in mental illness 3) MY RELATIVE'S mental illness is not genetic - no other affected individuals in my family have this mental illness 4) Scientists still don't know what gene(s) cause mental illness 5) I don't know my family history of mental illness 6) I don't want to have genetic testing 7) I do not have children or do not want to have children 8) I am worried the genetic counsellor might tell me not to have children 9) I don't want to know the chances of me or my relatives developing mental illness 10) I am too old to develop mental illness 11) The people I care most about are past the age at which they'd develop mental illness 12) I am worried I will find out things I wish I hadn't 13) I am worried the genetic counsellor will tell me my relative's mental illness is my fault 14) I am worried the genetic counsellor might tell me there is nothing I can do about my relative's mental illness 15) My relative is not currently unwell 16)There is not a lot that can be done to prevent mental illness 17) I do not have the time 18) I do not think I can afford an appointment with a genetic counsellor 19) I am worried it will affect my insurance or privacy

iii. Interest in receiving PGC following informational video

After watching the informational video, respondents' interest in receiving PGC was queried using a 7-point Likert-type response item (1= I definitely would not like to have PGC; 4 = I might like to have PGC, 7=I definitely would like to have PGC; see figure 55).

Figure 55: Likert-type response to assess interest in PGC following informational video



Interest in receiving PGC following informational video – results

Of the 50 affected individuals and 24 relatives that answered this question, 78% of affected individuals (n=39) and 79% of relatives (n=19) indicated they might or would definitely like to receive GC if it were available. 52% of affected individuals (n=26) and 46% of relatives (n=11) indicated high levels of interest in receiving PGC.

Conversely, 18% of affected individuals (n=9), and only 1 relative (4%) indicated that they definitely would not want to receive PGC.

All data items are shown in table 28.

Table 28: Frequency table showing respondents' interest in receiving PGC following informational video

Group	*Like to receive PGC?*							**TOTAL**
	1	**2**	**3**	**4**	**5**	**6**	**7**	
Affected individuals	9	2	0	7	6	6	20	50
Relatives	1	2	2	3	5	6	5	24

Note: 1 = I definitely would NOT want to have PGC, 4 = I might like to have PGC 7= I definitely WOULD want to have PGC.

Responses interpreted as indicating somewhat to definite interest in receiving PGC are highlighted in yellow.

Interest in receiving PGC following informational video: statistical analysis

Mann-Whitney U-Test

To test for differences in interest in receiving PGC between groups of respondents (affected individuals compared to relatives, and males compared to females) Mann Whitney-U Test was applied.

There was no significant difference in interest between affected individuals (MDN =6.00) and relatives (MDN =5.00), U=557,000 Z = -.510, *p*>.05*;* or between males (MDN = 5.00) and females (MDN = 6.00, U – 527.000, Z = -.872, *p* >.05).

Analysing affected individuals separately, there was no significant difference in interest in PGC between males (MDN = 5.00) and females (MDN = 6.5), U = 260.00, Z = =.716, *p >.05);* or between male relatives (MDN = 4) and female relatives (MDN = 5.5), U = 27.00, Z = -1.025, *p >.05).*

Thus, it appears that neither gender nor having a mental illness significantly influenced interest in receiving PGC.

Spearman's rank correlation

To attempt to identify variables influencing interest Spearman's rank correlation was used to test for association between interest in receiving PGC and i) illness attribution variables (GA, EA); ii) illness attribution certainty variables (GAC, EAC); and iii)concern for other relatives becoming ill (C).

All correlations are shown in table 29.

Table 29: Spearman's rank correlations showing association between respondents' interest in receiving PGC; illness attribution variables; illness attribution certainty variables; and concern for other relatives becoming ill.

Group	*Testable variable*				
	GA	*GAC*	*EA*	*EAC*	*C*
Affected individuals	.382**	-290*	-135	-115	.411**
Relatives	-.195	.074	.156	.025	.174

*Note: GA = attribution of mental illness to genetic factors, GC = Certainty regarding attribution of mental illness to genetic factors, EA = attribution of mental illness to environmental factors, EC = Certainty regarding attribution of mental illness to environmental factors. *p <.05, **p <.01*

Amongst affected individuals, there was a significant and positive association between interest in receiving PGC and attribution of the mental illness to genetic factors (GA) (r_s = .382, p=0.006).

There was also significant and positive association between interest in receiving PGC and concern for other relatives also becoming ill (C) (r_s = .411, p=0.003).

Additionally, there was a significant and negative association between interest in receiving PGC and certainty regarding attribution of mental illness to genetic factors (GAC) (r_s=-.290, p = .043).

Thus, for affected individuals, greater interest in was associated with greater attribution to genetics in mental illness causation; greater concern for other relatives becoming il; and greater uncertainty regarding role of genetics in pathogenesis.

No significant associations were found for relatives.

iv. Perceived usefulness of PGC following informational video – Quantitative analysis

After watching the informational video respondents' perceived usefulness of PGC was queried using two 7-point Likert-type response items (1= not at all useful; 4 = somewhat useful, 7=extremely useful; see figures 56a-d).

Affected individuals were invited to report their perceived usefulness of PGC to both themselves and their family members.

Relatives were invited to report their perceived usefulness of PGC to both themselves and their affected relative.

All data are given in tables 30-31 and figs. 57-60

Figures 56 a-d:Likert-type response items to assess perceived usefulness of PGC

Fig. 56a) Likert-type response item to assess affected individuals' perceived usefulness of PGC to self

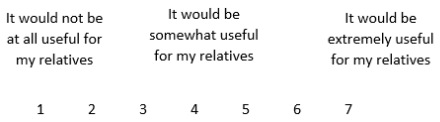


Fig. 56b) Likert-type response item to assess affected individuals' perceived usefulness o

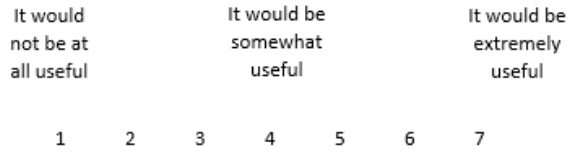


Fig. 56c) Likert-type response item to assess relatives' perceived usefulness of PGC to self

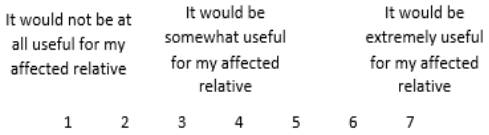
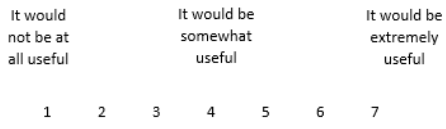


Fig. 56d) Likert-type response item to assess relatives' perceived usefulness of PGC to their affected relative



Perceived usefulness of PGC following informational video - Results

Perceived usefulness to *self*

Of the 51 affected individuals and 24 relatives that answered this question, 75% of affected individuals (n=38) and 79% of relatives (n=19) perceived PGC as somewhat to extremely useful to *themselves.* Almost half of respondents (45%, n=34) selected '6' or '7' on the Likert-scale, indicating that they believed PGC would be highly useful to *themselves.*

Conversely, 12% of *affected individuals* (n=6) and 13% of relatives (n=3) indicated they did not believe PGC would be at all useful for *themselves.*

Perceived usefulness to *others*

Affected individuals:

66% of affected individuals (n=33) perceived PGC as potentially somewhat to extremely useful to their *family members.*

However, 22% (n=11) of affected individuals believed PGC would be not at all useful to their *family members.*

Relatives:

92% of relatives (n=22) perceived PGC as somewhat to extremely useful to their *affected relative*. No relatives perceived PGC to be not at all useful to their *affected relative.*

All data are given in tables 30-31

Table 30: Frequency table showing affected individuals' perceived usefulness of PGC

Perceived usefulness	*Perceived usefulness*							*TOTAL*
	1	**2**	**3**	**4**	**5**	**6**	**7**	
For self	6	2	5	9	6	7	16	51
For family members	11	5	1	9	8	7	9	50

Note: 1 = "Not at all", 4 = 'Somewhat", 7 = "Extremely". Responses interpreted as PGC considered as somewhat to very useful are highlighted in yellow.

Table 31: Frequency table showing relatives' perceived usefulness of PGC

Perceived usefulness	*Perceived usefulness*							*TOTAL*
	1	**2**	**3**	**4**	**5**	**6**	**7**	
For self	3	1	1	5	3	4	7	24
For affected relative	0	1	1	2	4	6	10	24

Note: 1 = "Not at all" Somewhat",, 7 = "Extremely". Responses interpreted as PGC considered as somewhat to very useful are highlighted in yellow.

Figure 57: Frequency diagrams showing affected individuals' perceived usefulness of PGC to self

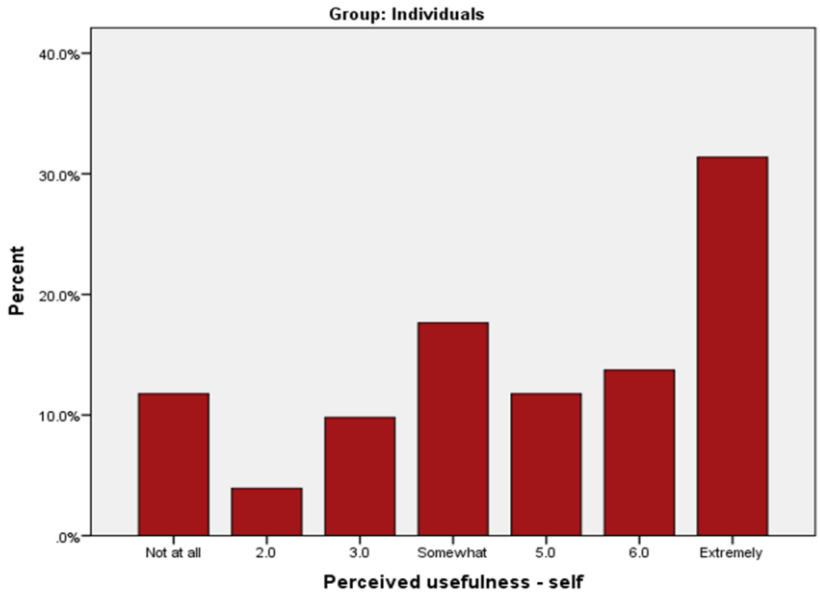


Figure 58: Frequency diagram showing affected individuals' perceived usefulness of PGC to family

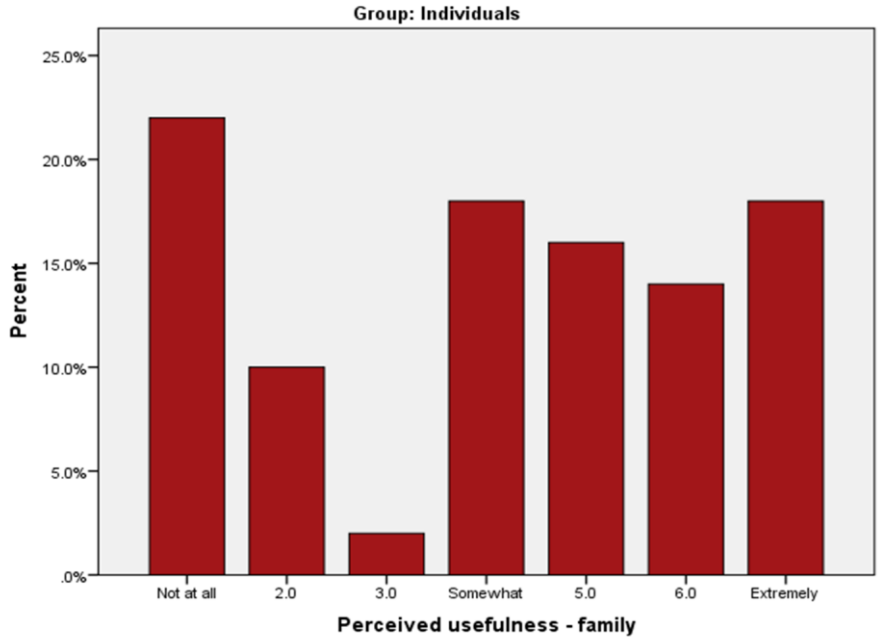


Figure 59: Frequency diagram showing relatives' perceived usefulness of PGC to self

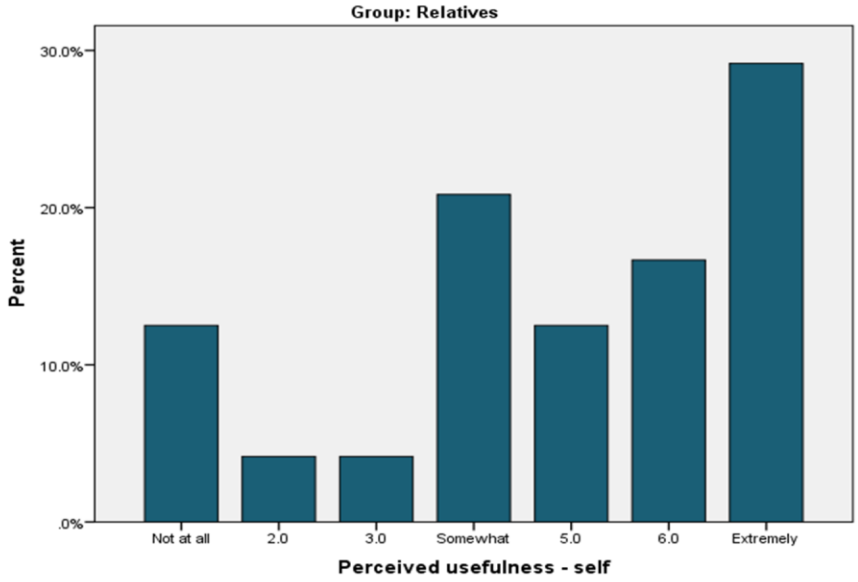
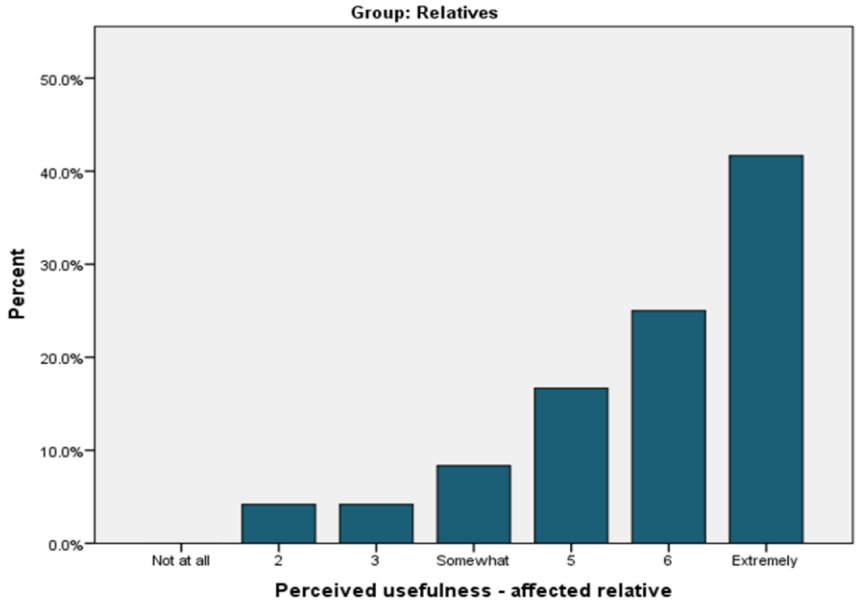


Figure 60: Frequency diagram showing relatives' perceived usefulness of PGC to their affected relative



Perceived usefulness of PGC following informational video: Statistical analyses

Mann Whitney U-Test

Mann Whitney U-Test showed no significant difference in perceived usefulness for self between affected individuals (MDN = 5.000) and relatives (MDN = 5.000, U=608 Z=-.046, *p >.05)*; or between males and females (MDN = 5.000, U = 552.500, Z = -.701, p >.05).

Thus neither gender nor having a mental illness influenced perceived usefulness of PGC.

Spearman's rank correlation coefficient

In order to identify potential variables influencing respondents' perceived usefulness of PGC, Spearman's rank correlation was used to test for associations between perceived usefulness of PGC and i) illnes attribution variables (GA, EA) ii) illness attribution certainty variables (GAC, EAC) and iiI) concern for other relatives becoming ill (C)

All correlations are shown in tables 32 and 33.

Table 32: Spearman's rank correlations between affected individual's perceived usefulness of PGC and illness attribution variables (GA, EA); illness attribution certainty variables (GAC, EAC); and concern

Affected individuals' perceived usefulness of PGC	*Testable Variable*				
	GA	*GAC*	*EA*	*EAC*	*C*
For self	*.291**	*-.244*	*-.090*	*-.087*	*.405***
For relatives	*.309**	*-.120*	*-.045*	*-.034*	*.454***

*Note: GA = attribution of mental illness to genetic factors, GC = Certainty regarding attribution of mental illness to genetic factors, EA = attribution of mental illness to environmental factors, EC = Certainty regarding attribution of mental illness to environmental factors. *p <.05, **p <.01*

Table 33: Spearman's rank correlations between relatives' perceived usefulness of PGC and attribution variables; attribution certainty variables; and concern.

Relatives' perceived usefulness of PGC	*Testable Variable*				
	GA	*GAC*	*EA*	*EAC*	*C*
For self	*-.194*	*-.005*	*.169*	*.019*	*.249*
For affected relative	*-.059*	*.386*	*-.218*	*.435**	*.271*

*Note: GA = attribution of mental illness to genetic factors, GC = Certainty regarding attribution of mental illness to genetic factors, EA = attribution of mental illness to environmental factors, EC = Certainty regarding attribution of mental illness to environmental factors. *p <.05, **p <.01*

Amongst affected individuals, there was a significant and positive association between perceived usefulness of PGC *for self* and attribution of the mental illness to genetic factors (GA) (r_s= .291, p=0.038).

Additionally, there was a significant and positive association between perceived usefulness of PGC for *family members* and attribution of the mental illness to genetic factors (GA) (r_s= .309, p= 0.029).

There was also a significant and positive association between perceived usefulness of PGC *for self* and concern for other relatives becoming ill (r_s= .405, p=0.003).

Additionally, there was a significant and positive association between perceived usefulness of PGC for *family members* and concern for other relatives becoming ill (r_s= .454, p=0.001).

Thus, perceived usefulness of PGC to both self and to family members was associated with greater attribution of the mental illness to genetics factors, and greater concern for relatives becoming ill.

Amongst relatives, none of the tested variables reached or approached significant significance in association with perceived usefulness to self.

Perceived usefulness of PGC for their affected relatives was significantly associated with certainty regarding attribution to genetic factors (GAC)(r_s=.435, p=0.034).

No other significant correlations were found.

Summary of findings

Prior to watching the informational video interest in PGC was relatively low amongst both affected individuals and their relatives, with less than half of all respondents reporting that they would wish to receive PGC. Additionally, there was high degrees of uncertainty regarding the service, with almost a third of respondents (29%, n= 24) reported they did not know whether they would like to have PGC.

The major reasons for not wishing to receive PGC reported by respondents was not knowing enough about the service, reported by almost half of respondents, and concern that they may find out information they wish they hadn't, reported by almost a third of respondents. Over a fifth of respondents also believed that they would not be able to afford an appointment, when PGC would, in fact, be free of charge if provided on the NHS. For affected individuals, not having or not wanting children, and scientists not knowing which genes cause mental illness, were other frequent reasons given for hypothetical decline. Conversely, for relatives, not knowing their family history of mental illness was cited as a reason by a number of respondents.

After watching the informational video, interest is PGC was much higher, with 78% of respondents (n=58) reported they might or would like to receive PGC. Additionally, 76% of respondents (n=57) believed PGC would be useful to themselves; 92% (n=22) of relatives believed PGC would be useful to their affected relation; and 66% (n=33) of affected individuals believed PGC would be useful to their family members, providing further indications that PGC was favourably viewed.

For affected individuals, greater interest in PGC was significantly associated with greater attribution to genetics in mental illness causation; greater uncertainty regarding the role of genetics in illness causation; and greater concern for other relatives becoming ill.

Similarly, for affected individuals, greater perceived usefulness of PGC following the informational video was significantly associated with greater attribution of the mental illness to genetic factors and greater concern for other relatives becoming ill.

No significant associations between interest in, nor perceived usefulness of, PGC amongst relatives were detected.

3.6 PGC – Perceptions of value (qualitative analysis)

To explore perceptions regarding the value of PGC, respondents' were invited to write what aspects of PGC they considered useful to themselves, their family members (for affected individuals), and their affected relative (for relatives), after watching the informational video.

PGC – Perceptions of value (qualitative analysis) – Results

The same major themes were identified between affected individuals' and relatives' responses: increasing understanding about mental illness, increasing understanding about familial risk, management (protective/coping strategies), and psychotherapeutic values. Therefore respondents' answers are pooled for means of reporting, however contextual differences between groups of respondents are highlighted in the text.

Tables 34-37 reports theme and sub-themes identified, and separately reports i) affected individuals responses and ii) relatives' responses

Better understanding mental illness

Almost all respondents reported that increased understanding of the mental illness would be a useful aspect of PGC.
The majority of respondents identified specifically that understanding of causal factors in mental illness pathogenesis, and especially gaining better understanding of interaction between genetic and environmental factors, would be valuable:

> "I think being able to understand mental illness better whilst looking at what may contribute to mental illness…" (relative)

> "Understanding the illness: especially relationship between environment and genes." (affected individual)

> "Gain more understanding of genetic research and links between genetics and mental health conditions." (affected individual)

Several respondents discussed how they would particularly value this information in the context of the individual, e.g. discussing their/their relatives own life experiences and their personal family history of mental illness, and how this may have contributed to illness onset or risk of illness:

> "To have some kind of more definite answer as to how much my experiences(sic) have been a physiological problem." (affected individual)

> "Understanding more specifically about why the mental illness has affected my brother." (relative)

> "The chance to talk through your own individual circumstances." (relative)

A few affected individuals also discussed how, through gaining increased insight into the mental illness, they believed PGC may help their relatives to better understand their symptoms and/or their behaviour:

> "Understanding the complex nature of mood swings and how I cannot always recognise the state I am in until after the event which leaves my family completely at a loss as to how to deal with me." (affected individual)

> "…It might also help them to understand why I'm such a giant pain in the arse at times." (affected individual)

Managing mental illness

The majority of respondents discussed that a valuable aspect of PGC may be facilitating better management of mental health.

Respondents identified that discussion of and identification of strategies that both promote mental well-being in both themselves and their other family members, and coping strategies for affected individuals with regards to managing their symptoms, would be helpful:

> "…It would also be useful to learn ways of improving and maintaining good mental health and wellbeing." (relative)

> "Ways to protect mine and my relative's mental health." (affected individual)
> "Helping my son to understand more about his illness and how to manage it…" (relative)

> "…the use of different strategies to manage my mood swings." (affected individual)

Specifically, a number of respondents specifically identified that the opportunity for discussions on an individual basis would be especially valuable:

> "Treating people as affected individuals so as to work out what will be suitable for them as regards managing their mental health." (relative)

> "helping him (affected family member) cope with his mental illness better… talking about what does and doesn't work for him." (relative)

> "…advice that is personal to us(family)…" (relative)

Additionally, a few affected individuals discussed how PGC may facilitate better management of mental illness on a *psychological level*, through empowering their sense of control of the condition:

> "..With greater insight into our weaknesses are we able to better combat them, and I feel the more I know about the matter the more effective I will be in attaining a close-to-normal level of functionality." (affected individual)

> "Understanding the cause and contributing factors. Strategies to deal and heal from this awareness – not to just deal with guilt/shame/powerlessness but to feel more 'empowered' and in control of my thoughts/behaviours and life going forward." (affected individual)

> "…How to better self-manage and make decisions based on positive rational thoughts…" (affected individual)

A small proportion of relatives expressed their hopes that, through increasing understanding of the biological contributions to mental illness, PGC may encourage treatment adherence amongst their affected family member:

> "…He (affected relative) might also be more open to medication if he understands how it might help him."

> "…Helping him (affected relative) understand why medication can help manage his symptoms (i.e. that a genetic basis means a biological basis)"

> "Finding ways to explain to my brother…why it's important for him to take his medication as he does not want to do this."

Shame, stigma, self-blame , guilt and anxiety

Many of the respondents providing answers discussed how, through increasing understanding of causation and addressing current beliefs and attitudes regarding causation, PGC may have psychotherapeutic values for both affected individuals and their relatives.

A number of respondents identified that this increased understanding may help with guilt-reduction. For example, several affected individuals explored how it may reduce their or their parents' sense of responsibility regarding onset of the mental illness:

> "Unburdening the guilt that it was my fault or my parents' guilt that it was their fault." (affected individual)

> "My mother feels particularly guilty, I hate that. She is incredible and I want her to realise it's not her fault." (affected individual)

> "I think it would be extremely useful for my parents to know that my upbringing is not what brought this all on." (affected individual)

> "My mum – there's been a lot of guilt there, that's what's been hardest for her I think. She focuses on small events that happened in life and blames them/her role in them. I think talking about biology/genetics part to play in it would really help." (affected individual)

Whilst some respondents expressed that PGC may help reduce affected individuals' sense of self-blame and guilt regarding the illness, and especially how the mental illness had impacted the family:

> "Finding ways to explain to my brother why his diagnosis isn't his fault…" (relative)

> “Showing her it’s not her (affected relative’s) fault. She blames herself for her way of thinking” (relative)

> “Having someone to work through the guilt and regret I have about things which I have done whilst very sick.” (affected individual)

> “Looking at coping with guilt surrounding the impact my illness has had on my family.” (affected individual)

A number of relatives and one affected individual indicated that PGC may help alleviate affected individuals’ sense of shame regarding the mental illness – and therefore possibly reducing stigma - through increasing understanding of causation to subsequently ‘normalise’ the mental illness:

> “To be assured it’s a "normal disease" and nothing to be ashamed of. And even if there is something in my family history I couldn't have avoided it or changed it.” (affected individual)

> “Being able to accept that he is not ‘biologically weird’ compared to his friends. For him the shame about the mental illness is the hardest thing and I think this might be really useful in changing his thoughts around this…”

> “Helping him to understand that his diagnosis is… not due to him being weak.”

Additionally, a small number of respondents indicated belief that PGC may help facilitate psychological acceptance of their illness, including through appearing their attributional search regarding explanations for their mental illness:

> “Counselling that might answer for me ‘at last’ the reasons why I developed (mental illness)…” (affected individual)

> “Understanding why I am why I am.” (affected individual)

> “To find out why him.”(relative)

> "Helping my son to understand more about his illness… also about the potential causes. Also to come to terms with his illness and to feel less ashamed." (relative)

One affected individual also indicated PGC it may be useful in reducing anxiety over genetic risk within the family through addressing fatalistic ideas about genetic determinism:

> "They (relatives) have a better understanding about the illness and what "not causes it. Its not the family history and doesn't have to affect everyone now or in the future"

Familial risk

Several affected individuals and a few relatives identified that an assessment of risk for their children developing psychiatric conditions, including current children and those born in the future, would be useful to them:

> "A realistic assessment of the risks that my son faces (affected individual)

> "Talking about the risks of my children developing mental illness, and what I could do as a parent to help minimise this risk." (affected individual)

> "…Interested in causes and risk of passing on condition to next generation." (relative)

> "Providing information on the likelihood of having passed on the mental disorder to our child." (relative)

A small number of respondents conceptualised the value as gaining a greater understanding of the implications of genetic contributions to mental illness rather than retrieving a *specific* risk assessment:

"… Its not the family history and doesn't have to affect everyone now or in the future."

"Explain things to relatives about any things that happen to some people (sic) and not others.."

Only two respondents discussed how increased understanding of familial risk may be useful specifically in regards to discussions about family-planning:

"I think having a discussion about mental illness in my family… and my husband's (mental illness) would be useful although its not likely that it would change our decision to have children. We're in our mid thirties, so this is very much an issue for us." (affected individual)

"My partner in the future - might not have experience of mental illness so might be a scary concept for them. I'd imagine if it came to discussions about starting a family etc this service would be useful to them." (relative)

Summary of findings

Both affected individuals and relatives identified valuable elements of PGC, and although there were contextual differences the same major themes of value were reported by the two groups of respondents.

The majority of respondents discussed that increased understanding of mental illness and especially of contributing factors, ascertained through both the provision of aetiological information and addressing pre-existing beliefs, would be a valued aspect of PGC.

Many respondents also reported that PGC may facilitate better management of mental health for both unaffected and affected individuals within the family. Respondents discussed how this may be obtained on both a practical-basis (e.g. through identifying effective strategies) and also on a psychological-basis (e.g. by increasing sense of control over mental well-being and subsequently facilitating protective behaviours and attitudes). A number of relatives also proposed that they believed PGC may encourage treatment adherence amongst their affected family-member.

Several respondents specifically identified that discussions regarding both aetiology and management of mental health that were *individualised* to the patient (e.g. incorporating own family history, own mental health history, discussing effective and ineffective protective strategies) would be an especially valued aspect of PGC.

Respondents also discussed how, largely through increasing understanding of aetiology and exploring and challenging currently held beliefs and attitudes regarding the mental illness, PGC may have psychotherapeutic values for both affected individuals and their relatives including reducing guilt and self-blame, shame, and anxiety over genetic risk.

Better understanding familial risk, and better understanding the implications of genetic contributions to mental illness, especially for children, was also identified as a useful aspect of PGC by a small number of respondents. Only two respondents conceptualised this specifically in regards to family-planning decision-making and discussions

3.8 Reasons for not wanting PGC after watching informational video (Qualitative analysis).

Respondents that selected an answer of ‘4’ or lower in regards to whether they would like to have PGC were invited to list any reasons they might not wish to have PGC in a free-form entry response.

Reasons for not wanting PGC after watching informational video (Qualitative analysis) – Results

Several themes were identified by respondents (see table 38). For relatives, only four respondents’ answered this section and so answers have been pooled to be reported in the text.

Table 34: Reasons for not wanting PGC (post-informational video)

Themes and subthemes - Reasons for not wanting PGC	AFFECTED INDIVIDUALS Responses (n=17;100%)	RELATIVES Responses (n=5; 100%)
It would not be useful to me/my family personally	9 (53%)	1 (20%)
i) I am happy with my current coping strategies/am currently mentally stable	5 (29%)	1 (20%)
ii) It would not be helpful for my recovery	5 (29%)	0
PGC may cause psychological distress	6 (35%)	1 (20%)
i) I might hear things I don't want to hear regarding the risk to my children	2 (12%)	0
ii) It might affect my decision to have children	0	1 (20%)
iii) The new information might compromise my mental well-being	3 (18%)	0
iv) It might increase stigma by endorsing genetic contributions to mental illness	1 (6%)	0
I am not worried about familial risk	3 (18%)	1 (20%)
i) I do not wish to have children	1 (6%)	0
ii) I am confident that my children have not inherited this illness/are well	2 (12%)	1 (20%)
I support alternative approaches in psychiatry	3 (18%)	0
i) Greater emphasis should be given to environmental factors in causation of mental illness	2 (12%)	0
ii) The role of genetics in causation is contestable	2 (12%)	0

It would not be useful to me/my family

A number of respondents identified that they did not believe PGC would be helpful to them. A few affected individuals (n=5) and one relative reported that they would not wish to receive PGC as they were currently stable in terms of their mental health or were currently coping well in terms of management of their condition, or supporting their relative's condition::

> "I am very happy with my coping strategies and acceptance…" (Affected Individual)

> …had extensive therapy am stable+have been for a very long time+understand all I need to so the resource best used for someone struggling with these issues as I'm not." (Affected individual)

> "…suspect the opportunities for it might be difficult to provide for many people due to cost and I (along with my family) am at a stage in life where we have learned to cope with my (partners) condition…" (Relative)

Similarly several affected individuals (n=5) reported that they believed it would be ineffective to them *personally,* including because they felt it was too late on from their diagnosis to be of benefit and/or it would not change the fact that they have mental health problems:

> "it is irrelevant for my recovery"

> "Dubious as to how effective or useful it would be to me."

> "I feel it would be futile as it is too late to do much about my mental illness"

> "It's not going to make any difference to reality"

I am worried PGC may cause psychological distress.

Several affected individuals (n=6) and one relative identified that they may not want to receive PGC as they were concerned they may learn new information that may cause them, or others, psychological distress including anxiety, stress and/or worry. For a number of affected individuals this concern especially centered around genetic risk information, and learning the potential implications for their children:

> "…It could unearth some bits or get me worrying about stuff too much when an important time expecting my 1st child" (affected individual)

> "It might stir things up and I might hear things I don't want to hear e.g. that my son might have bipolar." (affected indidivual)

Similarly, one relative reported that they were worried the new information may induce psychological distress and new dilemmas, and influence decision-making especially in relation to family-planning:

> "…I just don't want to know right now. I don't want it to affect my decision in having children, raising children, etc." (Relative)

Other affected individuals (n=3) identified that the PGC process, and especially new information learned through it, may compromise their current stability in terms of mental health:

> "I have always found in the past when I have been unwell or vulnerable that counselling might make me look inwards too much and might increase my depressive state." (affected individual)

> "I don't want to open a can of worms. I am in control of my condition, my children are both well (ish), I fear that genetic councilling (sci)would put that at risk" (Individual)

In addition, another affected individual identified that attributing psychiatric illness to genetics may have negative psychological impacts generally, potentially by increasing stigma and/or resulting in deterministic attitudes:

> "…could actively harm those with a diagnosed mental health issue just by reinforcing idea that genes are a key factor (just by being called 'genetic counselling')." (affected individual)

I am not worried about familial risk

A number of affected individuals (n=3) and one relative identified that they would not be interested in receiving PGC because they were not concerned about risk to other relatives, specifically either because they did not have children, or were not worried that their children would develop mental health problems:

> "…I have no wish to have children." (Individual)

> "…I am confident that my children… will not develop serious mental health problems." (Individual)

> "I am convinced my child has not inherited this mental disorder." (relative

I support alternative approaches in psychiatry

Several affected individuals (n=3) identified that they would not like to receive PGC because they contested the role of genetics in causation and/or believed greater emphasis should be given in both research and clinical practice, of environmental factors in psychiatry:

> "I think psychiatry needs to address the imbalance of the belief of the environment and genetic causation of mental distress to work more on the environment. Knowing inequality, social issues, poverty, etc as influences, I want to see more emphasis on changing that." (affected individual)

> "…the 'genetic' focus on this pre-supposes the role of importance genetics in 'mental illness' which is contestable. Yes, we all have genes and there's bound to be a genetic influence somewhere .. but we know a lot more about the role of trauma and adverse life experiences in psychosis and it feels lots more helpful to focus on creating safer and more supportive families and communities than getting sucked into genes…" (affected individual)

> "do not believe in the neuron/gene/DNA science-ification of consciousness, which is very unfortunately the scientific hegemony of this age and is giving birth, especially in the neuroscience community, to a form of scientific fundementalism… Which from 'the research', isn't conclusive." (affected individual).

Summary of findings

Respondents reported several reasons for which they may not want to receive PGC.

A number of respondents discussed how GC, as a genetics-based approach, did not fit within their own beliefs about the disease construct, or their own personal philosophies in regards to approaches to psychiatry and psychiatric healthcare.

A few respondents also reported that they did not believe PGC would be helpful to them, either because they were currently stable with their own coping strategies, or did not believe it would be helpful or meaningful for them personally.

Some respondents also expressed concerns that PGC may induce psychological distress. Specifically this was in regards to the new information and the potential impact this could have on them, and often in regards to concerns for their children.

4. Evaluation of findings

4.1 Discussion

This study highlights some important findings relevant for consideration regarding the application of PGC within the UK.

Perceptions of aetiology

The majority of respondents attributed the mental illness to both genetic and environmental factors indicating a multifactorial explanation for mental illness is endorsed. This is consistent with previous studies that have explored aetiological perceptions amongst affected individuals and their relatives. (Gamm et al. 2004, Meiser et al. 2005, Meiser et al. 2007, Peay et al. 2008, Baines and Wittkowski 2013). The potential clinical implication of this finding is that it indicates lay beliefs about causation are in line with scientific explanations of illness and also the aetiological models that would be discussed in PGC, and that genetic education, such as that provided by PGC, would be more readily and positively received.

That the majority of respondents reported relative certainty about their attribution of the mental illness to both genetic and environmental factors in psychiatric pathogenesis is surprising and in contrast to the hypothesis that respondents would be uncertain about aetiology, based on studies that have reported that there is uncertainty and that misconceptions exist amongst affected individuals and families regarding psychiatric aetiology (Hodgkinson et al. 2001, Holzinger et al. 2003, Austin and Honer 2005, Costain and Bassett 2012, Costain et al. 2014b).

In direct contrast, however, when respondents' perceived value of PGC was explored using qualitative analysis the majority reported that they would still value information and discussions about contributions to mental illness and especially the relative contributions between genetic and non-genetic factors in pathogenesis. Furthermore, respondents' answers revealed – including amongst those who had reported high levels of certainty about their understanding quantitatively- that there was a tendency towards the adoption of oversimplified ideas about causation amongst affected individuals and their

family members, such as affected individuals considering the mental illness being due to them ‘being weak’, parents experiencing profound guilt owing to their feelings of responsibility for the onset of their child’s illness, and affected individuals feeling ashamed of the mental illness due to beliefs that the mental illness makes one “biologically weird” or not “normal”.

To this degree, the findings highlight the strength of using mixed-methods in this study. Collecting both quantitative and qualitative data enabled deeper exploration of respondents’ perceptions of aetiology, as well as insight into the emotional and psychological elements associated with beliefs about origins, which resultantly revealed that there were uncertainties, oversimplified ideas, and gaps in knowledge regarding the nature and origins of the mental illness, consistent with study hypothesis and fitting with previous literature.

The implication of these findings is that PGC - a major goal which is to facilitate comprehensive understanding of aetiology whilst also providing supportive psychotherapeutic counselling around these concepts- may thus be clinically valuable to the UK population.
Given that outcomes studies of practice have produced some encouraging data indicating value of PGC in terms of increasing perceived aetiological understanding and reducing misconceptions (Austin and Honer 2008, Costain et al. 2014a, Costain et al. 2014b), and furthermore the additional potential psychosocial outcomes this increased knowledge may bring for service-users including reduced self-blame (Costain et al. 2014a), stigma (Costain et al. 2014a) and increased hope and empowerment (Austin and Honer 2008, Inglis et al. 2014) , the findings of this study therefore justify further research regarding their perceptions and understanding of aetiology and pathology amongst the UK population, and the potential value of PGC in this regard.

Familial risk

This study explored several aspects relating to familial risk amongst respondents. It found misconceptions about risk of familial recurrence, with a tendency to overestimate degree of familial risk of recurrence; high degree of concern over such risk; and also, for affected individuals, that family-planning decisions may be influenced by the presence of mental illness, with a tendency towards decisions favouring having fewer or no children. Furthermore, in regards to perceived value of PGC respondents themselves identified that they would value an increased comprehension of familial risk and also the implications of genetic contributions to mental illness. These findings raise the possibility that PGC, a major goal of which is to increase understanding of familial risk, may thus be potentially helpful for the UK population.

In line with study hypothesis and previous studies exploring attitudes and beliefs about familial risk amongst affected individuals and their relatives (Austin and Honer 2008, Peay et al. 2008), the majority of respondents reported being concerned about the risk of family members developing a mental illness, and moreover, a large number reported *very high l*evels of concern. Furthermore, statistical analysis showed concern was significantly and positively associated with attribution to genetic models, indicating endorsement of a genetic model may be associated with greater levels of concern over familial risk.

Furthermore perceived familial recurrence risk was overestimated, and often dramatically, by the majority of respondents. This was both consistent with hypothesis and in line with the majority of studies that have previously explored perceptions of genetic risk for psychiatric conditions (SCZ and BPD) amongst non-UK populations (Targum et al. 1981, Schulz et al. 1982, Trippitelli et al. 1998, Quaid et al. 2001, Austin et al. 2006, Costain et al. 2014a, Costain et al. 2014b). Notably there was a tendency to overestimate risk across the diagnostic boundaries, not only for diagnoses which are typically considered more 'serious' (i.e. SCZ, psychosis) or have a higher heritability (i.e. SCZ, BPD in comparison to anxiety-related disorders or depression). To best knowledge there is little available published evidence (if any) exploring perceptions of risk regarding anxiety disorders e.g. OCD,

PTSD, and limited literature for depression. This finding therefore indicates a need for further research into risk estimations amongst the population for other, more common, psychiatric diagnoses.

Impact on family planning, typically favouring decisions towards having fewer or no children, was also reported by the majority of affected individuals, consistent with findings of previous studies (Austin et al. 2006, Meiser et al. 2007), and also consistent with study hypothesis. Furthermore, there was a significant and positive association between attribution of the mental illness to genetic factors and impact on family-planning, supporting previous findings that a genetic model of explanation may negatively influence reproductive decisions (Meiser et al. 2007).
Conversely, contrary to study hypothesis, very few relatives reported that the mental illness had impacted their family-planning decisions. This is interesting as it highlights potential differences in regards to information needs and concerns, and therefore practice of GC, between affected individuals and relatives. This is a concept that been previously discussed (Austin et al. 2006, Austin and Honer 2008), and will be explored later (see 'avenues for future research').

Given the encouraging data reporting a reduction in concern over familial risk (Austin and Honer 2008), facilitation of better understanding of the true empiric risk estimate amongst patients following PGC (Costain et al. 2014a, Costain et al. 2014b), these findings thus provide tentative indications that PGC could be helpful to this population. Further research regarding perceptions and implications of risk amongst the UK population, and the potential value of PGC in this regard, is thus warranted.

On a final note however it must also be considered that, whilst the quantitative findings of this study, explored above, thus provide indications that provision of familial risk information may be helpful to respondents, increased understanding of familial risk was in fact reported by only a small number of respondents as a valuable aspect of PGC after watching the informational video. Furthermore, whilst interpretation of the quantitative data provides some indications that facilitating decision-making with family-planning would be an *especially* helpful aspect of PGC, owing to the high reported impact on

family-planning decisions amongst affected individuals, as few as 2 respondents actually discussed value of PGC specifically in regards to family-planning decision-making.
Conversely, other aspects of PGC such as increased aetiological understanding, facilitating management of mental health, and psychotherapeutic values of PGC, were discussed to a greater extent by respondents.
These contrasts between interpretations derived from the quantitative and qualitative data in regards to familial risk are interesting, and further emphasise the value of using mixed-methods approach in the approach to this study.

Indeed it has been previously explored, to a degree that, in the literature that although GC is very typically associated with risk communication, in terms of practice of PGC provision of a specific risk assessment may not actually be required by the patient, especially once aetiology has been discussed, indeed even if obtaining a risk assessment was the presenting reason for attending the session (Austin and Honer 2007, Morris 2015, pers comms, 10 January, Austin 2015, pers comms, 16 February). It may be that discussing aetiology may be perceived to have the desired outcomes for the patient, e.g. increasing sense of control, realising genes are not 'destiny,' reducing anxiety, shame and guilt – to the extent that the risk estimate and/or providing accurate genetic information thus becomes less relevant, or even irrelevant, to the patient – even if it was the reason for them presenting for PGC. From a philosophical approach, in terms of approaches to practice, this approach would be supported by the counselling model, i.e. that education is not an overall goal but rather a means to facilitating other, important goals (Resta 2006). This may therefore partially explain the inconsistencies between interpretations from the quantitative and qualitative data regarding familial risk collected in this study.

Whilst no clinical implications regarding this could be drawn from a sample of such small scale, it does highlight that concepts relating to patient *desires and wishes* in regards to communication about risk information, that extends beyond focusing on their apparent 'needs,' identified from exploring their current comprehension (or miscomprehension) of familial risk or aetiology, are

also helpful and important to consider in regards to considering goals of interventions, such as PGC.

Awareness and perceptions of GC and PGC

The findings of the study collectively indicate that, in line with hypothesis, awareness and comprehension of GC is relatively low amongst the UK population. This is consistent with evidence from other studies that have explored awareness and perceptions of GC amongst the public (Hallowell et al. 1997, Bernhardt et al. 2000, Metcalfe et al. 2007, Lyus et al. 2007, Maio et al. 2013). As the majority of respondents were of the information-seeking population – and indeed some respondents were trained GC's themselves - it is likely that awareness and understanding is much less for the general population.

Although awareness was higher than has been reported by previous studies (Lyus 2007, Maio et al. 2013), it remained that less than half of respondents had previously heard of GC. Furthermore that awareness was significantly lower amongst affected individuals in comparison to relatives, consistent with previous studies exploring awareness of GC amongst the population (Lyus et al. 2007) is interesting, and likely reflects increased exposure of relatives due to a number of factors, including that they are more proactive in information-seeking; have increased access to mediums through which they come across genetic counselling (e.g. work and media) which their relatives do not due to their incapitation; and/or are perceived to have differential information needs by healthcare practitioners (Lyus et al. 2007). In regards to future implementation, this may mean it would be especially important to provide supporting information to affected individuals that are referred for PGC.

In terms of preconceptions of GC and PGC, although to a degree respondents showed good comprehension of GC (assessed qualitatively) and PGC (assessed quantitately) there were also some limitations in their understanding and some profound misconceptions about the practice and purpose of GC.

This included almost 50% respondents incorrectly believing PGC would involve diagnostic or prenatal testing, which is not currently clinically available and would not routinely be offered at GC. This finding is also consistent with that of a previous study exploring perceptions of PGC amongst the American population in which a

large proportion of respondents believed testing would be available for psychiatric conditions (Lyus 2007), and provides indications that testing may be expected at the point of referrals in the future.

Furthermore, fewer respondents also identified that GC for psychiatric conditions would involve the provision of emotional support, as has been reported by other study groups exploring perceptions of GC for physical illnesses (Bernhardt et al. 2000) and psychiatric illness (Lyus 2007). This is particularly interesting given that i) available literature regarding goals of PGC have emphasised the importance of psychosocial aspects of the intervention and ii) in this study, a large majority of the respondents identified the emotional aspects of PGC as being particularly valuable in regards to their perceptions of the service, following information about the service.

Similarly, in regards to perceptions of *traditional* GC, assessed qualitatively, whilst a small number of respondents identified that GC may have therapeutic benefits, this was predominantly limited to facilitating acceptance of risk, whilst alleviating guilt, shame, stigma, self-blame and anxiety, which are other, commonly reported and well-established, psychosocial outcomes of GC practice (Biesecker 2001, McCarthy Veach et al. 2007) were not typically discussed. Additionally, in respondents' answers there was a notable emphasis on concepts relating to genetic risk, even in regards to decision-making aspects of GC (i.e. 'reducing risk') and psychological and emotional aspects of GC (i.e. 'coming to terms with risk'). In practice, GC for multifactorial disorders has a much wider scope and purpose than risk communication (McAllister et al. 2011, Austin et al. 2014).

Awareness and perceptions – practical implications

Low awareness and comprehension have potentially important *practical* considerations, because a growing body of evidence this could have a major influence on future delivery of the service.

For example. lack of awareness and comprehension of purpose of PGC may influence service *uptake and engagement*, because those who are not aware of the service will be less likely to access it (Metcalfe et al. 2007, Maio 2013), and additionally those who are less aware of how it may be relevant and valuable to

them personally will also be less likely to engage, even if they are referred (Maio et al. 2013).

Secondly awareness and comprehension may influence *patient outcomes,* for example through increasing anxiety, reducing the patients' ability to effectively prepare in advance for the session and therefore maximising the utility of the session, and potentially influencing patient's expectations and therefore potentially satisfaction with the service (Bernhardt et al. 2000, Davey et al. 2001, Metcalfe et al. 2007); as is supported by a growing body of GC process and outcomes studies for physical illnesses (Davey et al. 2005, Hallowell et al. 1997, Brown et al. 1999, Metcalfe et al. 2007, Maio et al. 2013).

Thus, considering both the findings presented here and previously published literature, this highlights the need for more research pertaining awareness and perceptions of both GC and PGC amongst the lay public in regards to future application of PGC.

Awareness and perceptions – ethical considerations

On a further note, participants' perceptions of GC and PGC also raise issues of an *ethica*l nature, which are important for consideration.

For example, respondents expressed concern that both GC and PGC may cause psychological distress, with this being given as a key reason given for respondents not wanting to have PGC. Indeed for some respondents this concern remained even after following information about the service, stating their concerns that the information may cause them worry, potentially even jeopardising their current mental state.

Indeed literature regarding provision of GC more generally has also discussed the psychological and emotional impact of receiving genetic information regarding any genetic condition (Bisecker 2001, Davey et al. 2005, Resta 2006). Furthermore, that PGC may cause psychological distress, especially due to the uncertain nature of the aetiology of psychiatric conditions, has been reported and explored as a concern amongst genetic counsellors in regards to providing PGC (Monaco et al. 2010, Hippmann et al. 2013). Additionally, in regards for GC within psychiatry it has been asserted that the psychological state of the patient should be assessed by the clinician, to ensure that they are well enough to receive such information (Tsuang 1994, Papadimitriou and Dikeos 2003, Austin and Honer 2007).

These negative outcomes would however be the antithesis of goals of contemporary practice, which aims to minimise distress, empower individuals and facilitate adaptation to the illness over time (Resta 2006, Resta et al. 2006, McCarthy-Veach et al. 2007, McAllister et al. 2011) Furthermore, available evidence that has assessed interventions of PGC specifically has reported that PGC is associated with positive outcomes, including increased sense of empowerment (Inglis et al. 2014), reduction of guilt, and also that the intervention did not increase psychological distress (Costain et al. 2014a, Costain et al. 2014b).

Thus, respondents' concerns that PGC may cause psychological distress highlight a need for further research pertaining this, and especially support previous assertions emphasising the need to further develop an evidence base for PGC. This would help ensure optimal practice and also address any controversies pertaining to such concerns and potential issues.

Other ethical considerations in regards to the provision of PGC that were highlighted in this study was the association of both GC and PGC with eugenic-type values, with respondents, in qualitative analysis, conceptualising its practice as directive, with the genetic counsellor influencing reproductive decision-making to reduce the presence of certain genes within the population.
Consistent with these beliefs, in quantitative analysis, a relatively large proportion of respondents believed, specifically in regards to providing GC for psychiatric conditions, that a genetic counsellor would advise affected individuals whether or not to have children, which has been previously reported by studies that have explored perceptions of GC amongst the general public (Maio et al. 2013).
Additionally, that a proportion of respondents also believed a goal of PGC would be to 'prevent' mental illness in children could also be considered, arguably, to be associated with eugenic values (Maio et al. 2013).
Fundamentally, these beliefs about the purpose and practice of GC is in contrast to the non-directive approach of modern day practice of genetic counselling, which strives to promote autonomous decision-making (Resta 2006) - a viewpoint which is considered to be the direct opposite of a eugenics-based approach (Gottesmann and Shields 1982, Maio et al. 2013).

Psychiatric genetics has a troubled and ugly history, with deep associations with the eugenics movement of the early 20th Century. Through policies, the foundations of

which were based on flawed genetic theorums, the reproductive rights of tens of thousands of society's most stigmatised individuals – including those with mental and learning difficulties - across America and Europe were entirely diminished. Consequently widespread segregation, mass involuntary sterilisation and institutionalisation was conducted on a huge scale (Nuffield Council on Bioethics 2002, Brüne 2007).
In Nazi Germany hundreds of thousands of mentally ill individuals were murdered through 'racial hygiene' policies inspired by the works of leading figures in the field of psychiatric geneticists, such as Ernst Rüdin (Ritter and Roelcke 2005, Roelcke 2007). providing further associations between genetic approaches within psychiatry and the eugenics movement. Indeed, even the practice of early GC itself, with its wider, 'public-health' centered approach which meant it had a directive, and in turn, sometimes discriminatory and prejudiced approach against those considered genetically inferior in society, including mentally ill individuals (Resta 2006).

Although these eugenic policies no longer drive modern genetic approaches in science and healthcare policy (Nuffield Council on Bioethics 2002), the findings presented here indicate that past eugenic ideas and practices may potentially still, for some, be associated with *contemporary* genetic approaches within psychiatry, including, in the case of this study, in regards to the provision of GC for psychiatric conditions.

Whilst the small sample size of this study limits the generalisability of this finding to the wider UK population, it does raise important questions for future research focus, as it may indicate a potential need to raise the profile of not only genetic counselling, but also genetic approaches within psychiatry more widely.

Thus, the findings presented in this study that there are, for some potential service-users, concerns regarding psychological distress, and that there are associations of its practice with eugenic-type values, further emphasises the need for more research into awareness and perceptions of PGC in regards to future implementation of PGC within the UK.

Interest in receiving PGC

Prior to this study there was no published data evidencing interest or demand regarding PGC. Overall the findings of this study demonstrate that PGC was favourably viewed amongst respondents, and that there was an interest in receiving PGC; however that initial interest prior to receiving information about the service was very low. The results presented here overall provide tentative indications that, whilst PGC may be welcomed by service-users in the future, efforts to raise the profile of GC, and especially within psychiatry, may be important in future efforts to implement PGC within the UK.

Initially, before receiving information about PGC, respondents' interest in receiving PGC, with only 46% positively identifying that they would wish to receive PGC, was much lower in comparison to studies conducted amongst other populations, which have typically reported interest rates around 65-80% amongst respondents (Quaid 2001,DeLisi and Bertisch 2006, Lyus 2007). Uncertainty regarding the service was also high, with some ~30% respondents indicating they were unsure as to whether they would wish to have PGC.

Conversely, after watching the informational video about the service, interest was much higher, with 75% respondents indicating they would wish to receive PGC. Similarly the majority of respondents believed PGC would be 'useful,' with 92% of relatives reporting they believed it would be useful to their affected relative. Whilst perceived usefulness is not a valid indicator of utility of a healthcare service, it could reasonably be interpreted as further evidence of a keeness in receiving PGC amongst respondents.

The initial lack of interest in PGC is an interesting finding. A likely factor is respondents' lack of awareness and comprehension of the service, and this can be supported by several bodies of evidence from the study.

First and foremost, respondents in this study were deliberately provided with little information about GC and PGC prior to participation in order to obtain base-line rates of interest. In contrast, it is likely that other studies exploring interest, that reported higher rates of interest (Quaid 2001,DeLisi and Bertisch 2006, Lyus 2007) provided respondents with more information about the service e.g. in participant information sheets, directly through the researcher or clinician, especially those studies conducted in clinical research settings, and also by presenting participants with a definition of the service during the study. The implication is that these

situations may not reflect real-life situations in which patients may be offered GC, e.g. if the referring clinician has limited comprehension of the service, and thus, without adequate information provision, the findings of this study provide indications that interest rates may in fact be lower.

Furthermore, the reasons given by respondents for not initially wishing to receive PGC also support the assertion that low comprehension about the service may have influenced low interest rates. For example, not knowing enough about the service was the major reason given by respondents, as well as concerns that they would not be able to afford an appointment (when in fact it would be free on the NHS), and that they did not know the family history of mental illness and/or did not want to have children (when in fact the scope of GC goes beyond that of risk communication, and these would not be caveats to accessing PGC).

Finally, that interest was much higher following the informational video provides further support. The video not only provided information about the service but also addressed key misconceptions, such as belief that the major focus of PGC is discussing risk to offspring, or involves genetic testing. Although due to the differential methods of analysis used, differences in interest prior and following the informational video could not be statistically tested, it was clear that interest rates were much higher following the video.

Collectively, therefore, the study of these findings provides supporting evidence that misconceptions and lack of comprehension regarding the service may impact engagement and behavioural responses to being offered PGC in the future, consistent with available literature regarding PGC (Hunter 2010) and GC more generally (Maio et al. 2013).

In regards to perceived value of PGC, respondents' expressed beliefs that PGC may help with facilitating better understanding of aetiology; identification and comprehension of protective factors and management strategies; better understanding of genetic risk; increased sense of empowerment; and psychotherapeutic values, especially reducing guilt, shame, blame.

Notably these are commonly reported goals and outcomes of non-psychiatric GC and, further, are in line with the core goals of the model of empowerment and thus the GCOS-24 scale (McAllister et al. 2011). These goals are also consistent with findings from previous groups from other countries that have both hypothesised the

potential value of PGC (Hodgkinson et al. 2001, Austin and Honer 2005, Hill and Sahaar 2006, Finn and Smoller 2006, Austin and Honer 2007, Peay et al. 2008) and also study groups that have provided actual outcomes data for PGC (Austin and Honer 2008, Costain et al. 2014a, Costain et al. 2014b, Inglis et al. 2014), indicating agreement between goals of approaches described by clinicians and researcher in previous, non-UK groups (Tsuang 1994, Hodgkinson et al. 2001, Papadimitriou and Dikeos 2003, Austin and Honer 2007) and perspectives of UK respondents – potential future service-users - in this study.

This thus provides further, qualitative data supporting interest and keeness, measured objectively, in receiving PGC amongst respondents following provision of information.

Therefore the findings presented in this study collectively demonstrate that, overall, PGC was favourably viewed amongst members of the UK population; however they also indicate a potential need to increase awareness and address misconceptions around GC, and especially its role within psychiatry. This is worthy of future consideration in regards to exploring the application of PGC within the UK.

4.2 Avenues for future research.

This study has highlighted several areas for future investigation that will help guide implementation efforts within the UK and wider. Fundamentally, outcomes data, obtained through the provision of GC within the UK, is critically needed. This would provide data regarding uptake when offered, which is especially important in psychiatry in which engagement with medical services is traditionally low. Indeed, research groups that have provided PGC have demonstrated that rates of actual uptake has been lower than rates of interest (Austin and Honer 2008, Costain et al. 2014a). This knowledge would thus provide greater insight into demand for PGC, and will also identify additional barriers to its delivery.

In addition, outcomes data would enable assessment of PGC by exploring client outcomes and satisfaction with the service. This would therefore add to the accumulating data evidencing positive outcomes of PGC (Inglis et al. 2014), to justify making the service more routinely available within the UK. This is especially fundamental in healthcare, in which any new intervention, and expenditure of money

and time, must be justified by rigorous outcomes data (Costain et al. 2014b); and would be especially important within the UK, as the austerity forces the need for evidence-based practice and efficacious healthcare interventions. Such assessment will also be critical in guiding delivery in clinical settings within the UK to ensure client's needs in relation to GC are being met. This information would also be particularly valuable given that client's perceived informational needs may differ pre- and post-intervention; and therefore it would be optimal to guide practice based on data derived from actual practice rather than perceptions prior to receiving the service.

Specifically, a number of relatives indicated that PGC may assist with medication adherence by their affected family member, largely, it seemed, due to their relative understanding more about the biology and thus having more biological and psychological faith in medications and the idea of being able to better manage their symptoms. Consistent with this, research from health psychology and GC has consistently asserted that knowledge enhancement and consequential alteration of the disease construct can have a psychological impact on influencing health-related behaviours through empowering affected individuals and increasing their perceived personal control, and this is supported by a body of outcomes data. This would be a particularly valuable area of research as, in psychiatry, engagement and compliance with health advice, and especially in relation to medication, is often low.

Future studies involving larger sample sizes would also be a valuable area of research and would provide greater insight that may guide optimal practice in the UK. Specifically, this study reported findings from a broad range of relatives, and therefore further investigation amongst subgroups of relatives would be valuable as it is likely that the informational needs and perceptions of PGC differ (Austin et al. 2006). For example, siblings of affected individuals in their childbearing ages may have differential information needs than parents of affected individuals (Austin and Honer 2008). Future studies may help identify and ultimately address the issues faced by, and the informational needs of, these groups. Similarly, whilst there is now a firm body of literature in relation to provision of GC for psychotic disorders (Austin et al. 2006, Lyus et al. 2007, Austin and Honer 2008, Costain et al. 2014a, Costain et al. 2014b), there is much less for other, more common disorders, such as OCD and MDD; and it is likely that affected individuals with these diagnoses may face different issues and have differential information needs. The accrual of larger sample sizes of different subgroups of service-users would enable identification of

similarities and differences in terms of factors influencing interest, perceived value, and engagement with the service. This information would help inform delivery of PGC more widely, to ensure the best outcomes for all patients and optimal delivery.

There is no data available amongst psychiatric healthcare practitioners in relation to provision of PGC, and therefore research exploring their perceptions would be valuable. Fundamentally, healthcare practitioner's perceptions of value for affected individuals and relatives may differ from that of patients', and their insight and views may provide additional insight and help identify additional potential goals of PGC, especially within the UK, and may identify additional potential barriers to delivery efforts. In addition, research into health care practitioners knowledge of psychiatric genetics, and the attitudes towards the provision of psychiatric genetic information, may identify potential training needs of clinicians within the UK – as has been identified as a need by previous groups (Martin et al. 2012), and has been asserted as fundamental by other groups in regards to strategies to provide PGC (Costain and Bassett 2012, Costain et al. 2014b)

Finally, this research focussed on perceived value of PGC. Whilst aspects that were considered unhelpful were not considered, as they are beyond the scope of this research project, such information may also be useful in helping guide optimal practice. In this regard, outcomes data (i.e. post-intervention) would be preferential as again perceptions are likely to differ pre- and post-intervention, and post-intervention views would be most reflective of actual practice of PGC.

4.3 Limitations

There are some inherent limitations to the study.

Firstly, the survey design creates some limitations. Although the mixed-methods approach allowed for capture of a broad range of views and beliefs; for data collected qualitatively, statistically valid generalisations cannot be undertaken and causal relationships between certain variables cannot be determined.

In addition, the collection of attitudinal data could potentially result in inherent bias. First and foremost, although the surveys were completed anonymously, awareness of the researcher's background may have skewed results. Specifically, out of

awareness of the researcher's associations with genetic counselling and psychiatric genetic research, respondents may have selected against giving an obviously undesirable or response. This is particularly notable given the use of the researcher's twitter account to publically promote the study as well as their other research interests. Resultantly respondent's answers may not reflect their true opinions. Secondly, participants might not gave provide all the details that shaped their thinking at the time of answering the question, meaning their answers give a limited scope in regards to their true perceptions and attitudes. This is particularly notable as the qualitative questions featured towards the end of the questionnaire, where participants may have been tired. Similarly, the use of a survey provided less qualitative data than other qualitative approaches, such as focus groups or interviews, which also likely also limited the scope of answers in regards to reflecting participants' full perceptions, and also made interpretation of certain elements or themes more challenging.

A further limitation is the relatively small sample size of the study, which may result in sample bias. Despite recruitment through a diversity of sources, the views of those who chose to participate in the study may not be generalizable to that of the wider UK population. For example, those who chose to participate may be more proactive in their or their relative's mental health; in their information-seeking activities; and/or have greater insight into their or their relative's personal recovery. This is particularly relevant to consider for those participants recruited from websites and self-help groups. Conversely, the study did not include hospitalised patients who may have different insight and perceptions pertaining to the questions investigated. Further, the study also excluded non-English speaking participants, which may result in the exclusion of certain populations from this research. This is important as mental illness may be perceived differently amongst different cultures, and similarly approaches to healthcare and/or treatment may differ.

Another important point of consideration to this degree is that respondents who chose to participate in the study may have a natural interest in psychiatric genetics, perhaps owing to a family history of mental illness for example; or conversely, they may be strongly opposed to genetic attributions to mental illness and wish to voice their rejection of healthcare services based on such models. Consequently their evaluations may not reflect those of the general population. This issue is of particular importance because participants' pre-conceptions of chance for other relatives to develop mental illness, and attributional theories to mental illness, could

impact their perception of value of psychiatric genetic counselling, even after receiving an explanation of the service.

The coding process is a further limitation of the study design as it opens the possibility for subjectivity by the researcher. In the coding process, there is a level of researcher imposition, in which the researcher is making their own decisions and assumptions as to what is and is not important in regards to the respondents' answers. Furthermore, the researcher interprets participants' responses, and these interpretations could not be verified due to anonymity of data collection. Additionally, the wording of some of the questions assumed a reasonable degree of education; some responses indicated the respondent had not fully comprehended the question but, again, their answer could not be verified or clarified due to anonymity of data collection.

Further, the description of genetic counselling provided in the survey will also be a critical factor influencing participants' responses. The video content was based on the definition of genetic counselling provided by the NSGC, and was formulated and presented by Dr. Jehannine Austin, president of the and founder of the NSGC. However, certain components of the video may have influenced participants' responses. For example, some participants indicated suggestions of ways to improve their mental health as not helpful or even patronising; whilst others reported this would be a particularly useful part of genetic counselling. Affected individuals' reactions to such information will be personal to them and dependent on their previous experiences; and reactions to such specific details may influence participant's overall perceptions and responses to genetic counselling on the whole. A further technical limitation pertaining to the use of the video is that one respondent was unable to watch the video on their mobile device. No other respondents indicated they had encountered this problem but it is a possibility. Likewise, some respondents' may have missed out on certain parts of the audio, which was not possible to investigate. This is a further factor that must be considered.

In addition, specifically, the issue of differential and/or multiple diagnoses for respondents meant that it was difficult to statistically analyse familial risk data. Although familial risk was overestimated by the majority of respondents, this limits generalisability of this finding to the whole of the UK population. Future research methods exploring this for the UK would be needed.

5 Conclusion

This study is, to best knowledge, the first of its kind to explore the application of PGC within the UK.

It provides evidence of an interest and keenness in receiving PGC amongst this population. It also demonstrates that uncertainties, misconceptions and concerns exist amongst affected individuals and their relatives regarding aetiology of psychiatric conditions and concepts relating to familial risk, providing tentative indications that PGC may be clinically helpful to this population.

However the findings presented here also indicate that awareness of genetic counselling is low amongst the UK population, especially amongst affected individuals. The findings also provide evidence of limited comprehension, and some profound misconceptions, about the purpose and process of genetic counselling, including its role specifically within psychiatry. This includes association of its practice with eugenic type values and also beliefs and anxieties that PGC may cause psychological distress, which collectively raises concerns of an ethical nature. These results highlight a potential need to raise the profile of genetic counselling, and especially its place within the field of psychiatry.

With no other published data currently available regarding the application of PGC within the UK, further exploration of the findings presented here, in larger sample sizes, are thus needed to provide recommendations regarding the future of PGC within the UK.

None the less, the results highlight some interesting and potentially important concepts in regards to the application of PGC that are deserved of such further investigation.

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APPENDICES

Appendix A - Survey – Affected individuals.

Psychiatric Genetic Counselling in the UK

1. Demographic and diagnostic data

1. What is your age? Please tick your answer.

- ❍ 18-24
- ❍ 25-30
- ❍ 31-35
- ❍ 36-40
- ❍ 41-45
- ❍ 46-50
- ❍ 51-55
- ❍ 56-60
- ❍ 61-65
- ❍ 66+

2. What is your gender? Please tick your answer.

- ❍ Male
- ❍ Female
- ❍ Would prefer not say

3. What is the highest degree or level of schooling you have completed? Please tick your answer.

- ❍ Would prefer not to say
- ❍ No schooling completed
- ❍ Secondary school without GCSE's/O-levels
- ❍ GCSE's/O-levels
- ❍ A-levels or equivalent (e.g. BTEC)
- ❍ Bachelor's degree (
- ❍ Higher degree (e.g. master's degree, doctorate - please decribe in the space provided):

4. What is your current employment status? Please tick your answer.

- ❍ In employment (full or part-time)
- ❍ Self-employed
- ❍ Not currently working
- ❍ In full time education
- ❍ Retired
- ❍ Unable to work
- ❍ Would prefer not to say

5. What is your ethnic group? Please tick your answer.

- ❍ White (British)
- ❍ Irish traveller
- ❍ Black or Black British - Caribbean
- ❍ Black or Black British - African
- ❍ Asian or Asian British - Indian
- ❍ Asian or Asian British - Pakistani
- ❍ Asian or Asian British - Bangladeshi
- ❍ Chinese
- ❍ Mixed - White and Black Caribbean
- ❍ Mixed - White and Black African
- ❍ Mixed - White and Asian
- ❍ Other ethnic background (please describe)

- ❍ Would prefer not to say

6. How would you describe your nationality? Please tick your answer.

- ❍ English
- ❍ Welsh
- ❍ Scottish
- ❍ Northern Irish
- ❍ British
- ❍ Other (please describe below)

7. Is the UK your country of permanent residence?

- ❍ Yes
- ❍ No (please describe below)

8. What is your psychiatric diagnosis? Please tick your answer

- ❍ Bipolar disorder 1
- ❍ Bipolar disorder 2
- ❍ Schizophrenia
- ❍ Schizoaffective disorder)
- ❍ Other (please describe):

Q9. How many years ago were you first diagnosed? Please write your answer in the space provided:

Section A

1. Please circle a number between 1 and 7 according to how much you think **your** mental illness was caused by genetic factors. For example, circling 1 would indicate that you believe that genetic factors did not contribute at all to your mental illness, circling 4 would indicate that you believe that genetic factors contributed somewhat, and circling 7 would indicate that you believe your mental illness was caused entirely by genetic factors.

Genetics did not contribute at all		Genetics contributed somewhat		Genetics alone caused my mental illness
1	2	3	4	5	6	7

2. Now, please circle a number between 1 and 7 according to how confident or sure you are of the answer you provided above. For example, circling 1 would indicate that you are not at all sure that the answer you provided is correct, and circling 7 would indicate that you are absolutely certain that the answer provided above is correct.

I am not sure at all				I am completely confident
1	2	3	4	5	6	7

3. Please circle a number between 1 and 7 according to how much you think **your** mental illness was caused by your life experiences. (You can think about this as things that have happened to you, or environmental factors, such as where you have lived). For example, circling 1 would indicate that you believe that your experiences did not contribute at all to your mental illness, circling 4 would indicate that you believe that your experiences contributed somewhat, and circling 7 would indicate that you believe your mental illness was caused entirely by your experiences.

1 2 3 4 5 6 7

My experiences did not contribute at all

My experiences contributed somewhat

My experiences alone caused my mental illness

4. Now, please circle a number between 1 and 7 according to how confident or sure you are of the answer you provided above. For example, circling 1 would indicate that you are not at all sure that the answer you provided is correct, and circling 7 would indicate that you are absolutely certain that the answer provided above is correct.

I am not sure at all

I am completely confident

1 2 3 4 5 6 7

Section B

1.Please circle a number between 1 and 7 according to how concerned you are about other family members developing the same mental illness as you have.

For example circling 1 would indicate that you are not at all concerned, circling 4 would indicate that you are somewhat concerned, and circling 7 would indicate that you are extremely concerned.

I am not at all concerned	I am somewhat concerned	I am extremely concerned

1 2 3 4 5 6 7

2. For someone with the same mental illness as you, how likely do you think it is that their **child**, who is not currently unwell and has never been, will also develop the mental illness in the future? Please tick the answer that you think is the most accurate.

- ❍ 1%
- ❍ 10%
- ❍ 25%
- ❍ 50%
- ❍ 100%
- ❍ Not sure
- ❍ Other (please state in the space provided)

3. For someone with the same mental illness as you, how likely do you think it is that their **sibling** (i.e. brother or sister), who is not currently unwell and has never been, will also develop this mental illness in the future? Please choose the answer that you think is the most accurate.

- ❍ 1%
- ❍ 10%
- ❍ 25%
- ❍ 50%
- ❍ 100%
- ❍ Not sure
- ❍ Other (please state in the space provided)

4. Has your diagnosis of mental illness affected your decisions about having a family, or do you think it may in the future?

- ❍ Not sure
- ❍ No
- ❍ Yes – please describe how:
  - □ A decision to have more children
  - □ A decision to have less children
  - □ A decision to have no children
  - □ Other (please explain below)

Section C

1. What is the first thing that comes to mind when you hear the term ‘genetic counselling’?
Please write your answer in the space provided below:

2. Had you heard of "genetic counselling" before participating in this study? You do not have to know what it is.

- ❍ Yes
- ❍ No

3. Have you received genetic counselling regarding your mental illness?

- ❍ No *Please move onto **question four** (question below)*
- ❍ Yes *Please move onto **SECTION D***

4. How did you come across genetic counselling before today? Please tick the answer which is most relevant to you

- ❍ I have been referred to or have seen a genetic counsellor regarding another health condition
- ❍ A relative or friend has seen a genetic counsellor
- ❍ I have read about genetic counselling in the news
- ❍ I have read about genetic counselling on the internet
- ❍ I have seen a movie/ TV programme in which a genetic counselling was mentioned or portrayed
- ❍ I learned about genetic counselling at school/college/university
- ❍ I am a genetic counsellor/ I am studying to become a genetic counsellor

- ❍ I have contact with genetic counsellors through my job
- ❍ Other (please give more information in the space provided below

5. What do you think a genetic counsellor might do, if anything, in relation to your mental illness? Please indicate your answer to each statement by ticking either **YES** or **NO**.

	YES	NO
Gather information about my family's medical history (1)	❍	❍
Gather information about my family's history of mental illness (2)	❍	❍
Provide information about the chances of my children (including form future pregnancies) also becoming ill (3)	❍	❍
Provide information about the chances of other relatives also becoming ill (4)	❍	❍
Provide information about the genetic contributions in mental illness (5)	❍	❍
Provide information about the non-genetic factors in mental illness (6)	❍	❍
Advise me whether or not to have children (7)	❍	❍
Arrange genetic tests to diagnose mental illness in myself or my relatives (8)	❍	❍
Arrange genetic tests to test for mental illness in future pregnancies	❍	❍

(9)		
Prevent future children from having mental illness (10)	❍	❍
Arrange gene therapy to cure mental illness (11)	❍	❍
Discuss ways I can protect my mental health (12)	❍	❍
Discuss ways my relatives can protect their mental health (13)	❍	❍
Provide referrals and information to other services that may be relevant to me (14)	❍	❍
Provide emotional support (15)	❍	❍
Decide what medications I should take for my mental illness (16)	❍	❍
Other (please give more information in the space provided below) (17)	❍	❍

6. Would you like to have genetic counselling regarding your mental illness?

- ❍ Yes *Please move onto question 8*
- ❍ No *Please move onto question 7 (below)*
- ❍ Not sure *Please move onto question 7 (below)*

7. Why might you not want to have genetic counselling regarding your mental illness? Please tick **any answers** that apply

- ❑ I do not know enough about psychiatric genetic counselling (1)
- ❑ There is no role/only a small role for genetics in mental illness (2)
- ❑ MY mental illness is not genetic - no other affected individuals in my family have this mental illness (3)
- ❑ Scientists still don't know what gene(s) cause mental illness (4)
- ❑ I don't know my family history of mental illness (5)
- ❑ I don't want to have genetic testing (6)
- ❑ I do not have children or do not want to have children (7)
- ❑ I am worried the genetic counsellor might tell me not to have children (8)
- ❑ I don't want to know the chances of me or my relatives developing mental illness (9)
- ❑ The people I care most about are past the age at which they'd develop mental illness (10)
- ❑ I am worried I will find out things I wish I hadn't (11)
- ❑ I am worried the genetic counsellor will tell me my mental illness is my fault (12)
- ❑ I am worried the genetic counsellor might tell me there is nothing I can do about my mental illness (13)
- ❑ I am not currently unwell (14)
- ❑ There is not a lot that can be done to prevent mental illness (15)
- ❑ I do not have the time (16)
- ❑ I do not think I can afford an appointment with a genetic counsellor (17)
- ❑ I am worried it will affect my insurance or privacy (18)
- ❑ Other (Please give more information in the space provided): (19)

  _____

8. The researcher will now show you a video. Please take your time to watch it, and then answer the questions below.

- Now that you have watched a film about what psychiatric genetic counselling is, please circle a number showing how useful you think it would be for you.

For example, circling 1 would indicate that you would not find it at all useful, circling 4 would indicate that you would find it somewhat useful, and circling 7 would indicate that you would find it extremely useful.

It would not be at all useful						It would be extremely useful
1	2	3	4	5	6	7

9. Which aspects of psychiatric genetic counselling that you heard about in the film, if any, do you think would be particularly useful to you? Please write your answer in the space provided below:

10. Please circle a number showing how useful you think psychiatric genetic counselling would be for your **close relatives** (for example, your parents, your brothers or sisters, or your children)?

For example, circling 1 would indicate that you think they would not find it at all useful, circling 4 would indicate that you think they would find it somewhat useful, and circling 7 would indicate that you think they would find it extremely useful.

It would not be at all useful for my relatives						It would be extremely useful for my relatives
1	2	3	4	5	6	7

11. Which aspects of psychiatric genetic counselling that you heard about in the film, if any, do you think would be particularly useful to your close relatives? Please write your answer in the space provided below:

12. If psychiatric genetic counselling was offered to you, would you want an appointment? Please circle a number between 1 and 7 to show your answer.

For example, circling 1 would indicate that you definitely would NOT want to have an appointment, circling 4 would indicate that you might want to have an appointment, and circling 7 would indicate that you definitely WOULD want to have an appointment.

I definitely would NOT want an appointment						I definitely WOULD want an appointment
1	2	3	4	5	6	7

13. Please explain any reasons why you **might NOT** want to have a psychiatric genetic counselling appointment in the space below:

END OF SURVEY for affected individuals that had NOT received PGC.

Section E

1. Who provided you with the psychiatric genetic counselling? Please tick your answer.

- ❍ A person specially qualified as a genetic counsellor
- ❍ A psychiatrist
- ❍ A nurse
- ❍ My GP
- ❍ I'm not sure
- ❍ Other (please give more information in the space provided)

2. How did you get to have the psychiatric genetic counselling session? Please tick your answer

- ❍ I referred myself (self-referral)
- ❍ A healthcare worker (e.g. doctor/nurse) referred me
- ❍ The session was part of an appointment with a healthcare worker
- ❍ Other (please give more information in the space provided)

3. Please circle a number between 1 and 7 showing how useful you found the psychiatric genetic counselling session.

For example, circling 1 would indicate that you did not find it at all useful, circling 4 would indicate that you found it somewhat useful, and circling 7 would indicate that you found it extremely useful.

Not at all useful						Extremely useful
1	2	3	4	5	6	7

4. What topics were covered during the psychiatric genetic counselling session? Please show your answer to each statement by ticking either **YES** or **NO**

	YES	NO
Discussed my family history of mental illness with me (1)	❑	❑
Discussed information about the genetic contrinbutions to mental illness with me (2)	❑	❑
Discussed information about the non-genetic factors to mental illness with me (3)	❑	❑
Discussed family planning decisions in relation to my mental illness (4)	❑	❑
Discussed ways to protect my own	❑	❑

mental health (5)		
Discussed ways to protect my relatives' mental health (6)	❑	❑
Discussed strategies to help me cope better with my mental illness (7)	❑	❑
Discussed genetic testing regarding mental illness (8)	❑	❑
Provided information about the chances of my relatives also developing mental illness (9)	❑	❑
Provided information about the chances of children in my family (including from future pregnancies) also developing mental illness (10)	❑	❑
Provided referrals and information to other services that may be relevant to me (e.g. support groups/referral to psychiatrist/psychotherapy) (11)	❑	❑
Provided emotional support (12)	❑	❑
Other (please give more information in the space provided): (13)	❑	❑

5. Were there any aspects of the psychiatric genetic counselling that you found particularly useful or helpful? If yes, what were they? Please write your answer in the space provided below:

6. Were there any topics that you would have liked more information about, or that were not covered during the session that you would have liked to have been? Please write your answer in the space provided below:

7. Were there any aspects of this service that you did not find useful or helpful, or felt uncomfortable talking about? Please write your answer in the space provided below:

8. Did you share any information you from your genetic counselling session with your relatives or friends? If yes, what did you discuss, and with who? Please write you answer in the space provided below:

9. Are there any ways in which your psychiatric genetic counselling session could be improved to make it more useful or helpful to you? Please write you suggestions in the space provided below:

10. Please choose a number showing to how useful you think psychiatric genetic counselling could be for your **close relatives** (for example your parents, your brothers or sisters, or your children).For example, circling 1 would indicate that you think it would not be at all useful to them; circling 4 would indicate that you think it would be somewhat useful to them; and circling 7 would indicate that you think it would be extremely useful to them.

Not at all useful						Extremely useful
1	2	3	4	5	6	7

11. What aspects of psychiatric genetic counselling, if any, do you think would be particularly helpful or useful to your close relatives? Please write your answer in the space provided below:

END OF SURVEY

Appendix B – Survey – Relatives.

1. Demographic and Diagnostic data

1. What is your age? Please tick your answer.

- ❍ 18-24
- ❍ 25-30
- ❍ 31-35
- ❍ 36-40
- ❍ 41-45
- ❍ 46-50
- ❍ 51-55
- ❍ 56-60
- ❍ 61-65
- ❍ 66+

2. What is your gender? Please tick your answer.

- ❍ Male
- ❍ Female
- ❍ Would prefer not say

3. What is the highest degree or level of schooling you have completed? Please tick your answer.

- ❍ Would prefer not to say
- ❍ No schooling completed
- ❍ Secondary school without GCSE's/O-levels
- ❍ GCSE's/O-levels
- ❍ A-levels or equivalent (e.g. BTEC)
- ❍ Bachelor's degree (
- ❍ Higher degree (e.g. master's degree, doctorate - please decribe in the space provided):

4. What is your current employment status? Please tick your answer.

- ❍ In employment (full or part-time)
- ❍ Self-employed
- ❍ Not currently working
- ❍ In full time education
- ❍ Retired
- ❍ Unable to work
- ❍ Would prefer not to say

5. What is your ethnic group? Please tick your answer.

- ❍ White (British)
- ❍ Irish traveller
- ❍ Black or Black British - Caribbean
- ❍ Black or Black British - African
- ❍ Asian or Asian British - Indian
- ❍ Asian or Asian British - Pakistani
- ❍ Asian or Asian British - Bangladeshi
- ❍ Chinese
- ❍ Mixed - White and Black Caribbean
- ❍ Mixed - White and Black African
- ❍ Mixed - White and Asian
- ❍ Other ethnic background (please describe)

- ❍ Would prefer not to say

6. How would you describe your nationality? Please tick your answer.

- ❍ English
- ❍ Welsh
- ❍ Scottish
- ❍ Northern Irish
- ❍ British
- ❍ Other (please describe below)

7. Is the UK your country of permanent residence?

❍ Yes

❍ No (please describe below)

8. What is your relative's psychiatric diagnosis? Please tick your answer

❍ Bipolar disorder 1

❍ Bipolar disorder 2

❍ Schizophrenia

❍ Schizoaffective disorder)

❍ Other (please describe):

Q9. How many years ago was your relative first diagnosed? Please write your answer in the space provided:

Section A

1. Please circle a number between 1 and 7 according to how much you think **your relative's** mental illness was caused by genetic factors. For example, circling 1 would indicate that you believe that genetic factors did not contribute at all to your relative's mental illness, circling 4 would indicate that you believe that genetic factors contributed somewhat, and circling 7 would indicate that you believe your relative's mental illness was caused entirely by genetic factors.

Genetics did not contribute at all			Genetics contributed somewhat			Genetics alone caused my relative's mental illness
1	2	3	4	5	6	7

2. Now, please circle a number between 1 and 7 according to how confident or sure you are of the answer you provided above. For example, circling 1 would indicate that you are not at all sure that the answer you provided is correct, and circling 7 would indicate that you are absolutely certain that the answer provided above is correct.

I am not sure at all						I am completely confident
1	2	3	4	5	6	7

3. Please circle a number between 1 and 7 according to how much you think **your relative's** mental illness was caused by their life experiences. (You can think about this as things that happened to them, or environmental factors, such as where they have lived). For example, circling 1 would indicate that you believe that your relative's experiences did not contribute at all to their mental illness, circling 4 would indicate that you believe that your relative's experiences contributed somewhat, and circling 7 would indicate that you believe your relative's mental illness was caused entirely by their experiences.

My relative's experiences did not contribute at all	My relative's experiences contributed somewhat	My relative's experiences alone caused their mental illness

1 2 3 4 5 6 7

4. Now, please circle a number between 1 and 7 according to how confident or sure you are of the answer you provided above. For example, circling 1 would indicate that you are not at all sure that the answer you provided is correct, and circling 7 would indicate that you are absolutely certain that the answer provided above is correct.

I am not sure at all		I am completely confident

1 2 3 4 5 6 7

Section B

1. Please circle a number between 1 and 7 according to how concerned you are about other family members also becoming ill with your relative's mental illness.

For example circling 1 would indicate that you are not at all concerned, circling 4 would indicate that you are somewhat concerned, and circling 7 would indicate that you are very concerned.

I am not at all concerned			I am somewhat concerned			I am very concerned
1	2	3	4	5	6	7

2. For someone with the same mental illness as your relative, how likely do you think it is that their **child**, who is not currently unwell and has never been, will also develop the mental illness in the future? Please tick the answer that you think is the most accurate.

- ❍ 1%
- ❍ 10%
- ❍ 25%
- ❍ 50%
- ❍ 100%
- ❍ Not sure
- ❍ Other (please state in the space provided)

3. For someone with the same mental illness as your relative, how likely do you think it is that their **sibling** (i.e. brother or sister), who is not currently unwell and has never been, will also develop this mental illness in the future? Please tick the answer that you think is the most accurate.

- ❍ 1%
- ❍ 10%
- ❍ 25%
- ❍ 50%
- ❍ 100%
- ❍ Not sure
- ❍ Other (please state in the space provided)

4. Has the mental illness in your family affected your decisions about having a family, or do you think it could in the future?

- ❍ Not sure
- ❍ No
- ❍ Yes – please describe how:
  - □ A decision to have more children
  - □ A decision to have less children
  - □ A decision to have no children
  - □ Other (please explain below)

Section C

1. What is the first thing that comes to mind when you hear the term ‘genetic counselling’?
Please write your answer in the space provided below:

2. Had you heard of "genetic counselling" before participating in this study? You do not have to know what it is.

- ❍ Yes
- ❍ No

3. Have you received genetic counselling regarding your relative’s mental illness?

- ❍ No *Please move onto **question four** of this section (below)*
- ❍ Yes *Please move onto **SECTION E***

4. How did you come across genetic counselling before today? Please tick the answer which is most relevant to you

- ❍ I have been referred to or have seen a genetic counsellor regarding another health condition
- ❍ A relative or friend has seen a genetic counsellor
- ❍ I have read about genetic counselling in the news
- ❍ I have read about genetic counselling on the internet
- ❍ I have seen a movie/ TV programme in which a genetic counselling was mentioned or portrayed
- ❍ I learned about genetic counselling at school/college/university
- ❍ I am a genetic counsellor/ I am studying to become a genetic counsellor
- ❍ I have contact with genetic counsellors through my job

❍ Other (please give more information in the space provided below

5. What do you think a genetic counsellor might do, if anything, in relation to the mental illness in your family? Please show your answer to each statement by ticking either YES or NO (see next page)

	YES	NO
Gather information about my family's medical history (1)	❍	❍
Gather information about my family's history of mental illness (2)	❍	❍
Provide information about the chances of children in my family (including from future pregnancies) also becoming ill (3)	❍	❍
Provide information about the chances of other relatives also becoming ill (4)	❍	❍
Provide information about the genetic contributions in mental illness (5)	❍	❍
Provide information about the non-genetic factors in mental illness (6)	❍	❍
Advise me whether or not to have children (7)	❍	❍
Advise my relative with a mental illness whether or not to have children (8)	❍	❍
Arrange genetic tests to diagnose mental illness in myself or my relatives (9)	❍	❍

Arrange genetic tests to test for mental illness in future pregnancies (10)	❍	❍
Prevent future children from having mental illness (11)	❍	❍
Arrange gene therapy to cure mental illness (12)	❍	❍
Discuss ways my relative with a mental illness can protect their mental health (14)	❍	❍
Discuss ways I can protect my own mental health (13)	❍	❍
Provide referrals and information to other services that may be relevant to my relative with a mental illness (15)	❍	❍
Provide referrals and information to other services that may be relevant to me (16)	❍	❍
Provide emotional support (17)	❍	❍
Decide what medications my relative should take for their mental illness (18)	❍	❍
Tell me what medications I can take to prevent myself from developing mental illness (19)	❍	❍
Other (please give more information in the space provided on the next page) (20)	❍	❍

6. Would you like to have genetic counselling regarding the mental illness in your family?

- ❍ Yes *Please move onto question 8*
- ❍ No *Please move onto question 7*
- ❍ Not sure *Please move onto question 7*

7. Why might you not want to have genetic counselling regarding the mental illness in your family? Please tick **any answers** that apply

- ❑ I do not know enough about psychiatric genetic counselling (1)
- ❑ There is no role/only a small role for genetics in mental illness (2)
- ❑ MY RELATIVE'S mental illness is not genetic - no other affected individuals in my family have this mental illness (3)
- ❑ Scientists still don't know what gene(s) cause mental illness (4)
- ❑ I don't know my family history of mental illness (5)
- ❑ I don't want to have genetic testing (6)
- ❑ I do not have children or do not want to have children (7)
- ❑ I am worried the genetic counsellor might tell me not to have children (8)
- ❑ I don't want to know the chances of me or my relatives developing mental illness (9)
- ❑ I am too old to develop mental illness (10)
- ❑ The people I care most about are past the age at which they'd develop mental illness (11)

- ❑ I am worried I will find out things I wish I hadn't (12)
- ❑ I am worried the genetic counsellor will tell me my relative's mental illness is my fault (13)
- ❑ I am worried the genetic counsellor might tell me there is nothing I can do about my relative's mental illness (14)
- ❑ My relative is not currently unwell (15)
- ❑ There is not a lot that can be done to prevent mental illness (16)
- ❑ I do not have the time (17)
- ❑ I do not think I can afford an appointment with a genetic counsellor (18)
- ❑ I am worried it will affect my insurance or privacy (19)
- ❑ Other (Please give more information in the space provided): (20)

  _____

8. The researcher will now show you a video. Please take your time to watch it, and then answer the questions below.

- Now that you have watched a film about what psychiatric genetic counselling is, please circle a number showing how useful you think it would be for you.

For example, circling 1 would indicate that you would not find it at all useful, circling 4 would indicate that you would find it somewhat useful, and circling 7 would indicate that you would find it extremely useful.

It would not be at all useful						It would be extremely useful
1	2	3	4	5	6	7

9. Which aspects of psychiatric genetic counselling that you heard about in the film, if any, do you think would be particularly useful to you? Please write your answer in the space provided below:

10. Please circle a number showing how useful you think psychiatric genetic counselling would be for your **relative with a mental illness?**

For example, circling 1 would indicate that you do not think it would be at all useful for them, circling 4 would indicate that you think they would find it somewhat useful, and circling 7 would indicate that you think they would find it extremely useful.

It would not be at all useful for my relative						It would be extremely useful for my relative
1	2	3	4	5	6	7

11. Which aspects of psychiatric genetic counselling that you heard about in the film, if any, do you think would be particularly useful to your **relative with a mental illness**? Please write your answer in the space provided below:

12. If psychiatric genetic counselling was offered to you, would you want an appointment? Please circle a number between 1 and 7 to show your answer.

For example, circling 1 would indicate that you definitely would NOT want to have an appointment, circling 4 would indicate that you might want to have an appointment, and circling 7 would indicate that you definitely WOULD want to have an appointment.

I definitely would NOT want an appointment						I definitely WOULD want an appointment
1	2	3	4	5	6	7

13. Please explain any reasons why you might NOT want to have a psychiatric genetic counselling appointment in the space below:

END OF SURVEY for respondents that had NOT received PGC

Section D.

1. Who provided you with the psychiatric genetic counselling? Please tick your answer.

- ❍ A person specially qualified as a genetic counsellor
- ❍ A psychiatrist
- ❍ A nurse
- ❍ My GP
- ❍ I'm not sure
- ❍ Other (please give more information in the space provided)

2. How did you get to have the psychiatric genetic counselling session? Please tick your answer

- ❍ I referred myself (self-referral)
- ❍ A healthcare worker (e.g. doctor/nurse) referred me
- ❍ The session was part of an appointment with a healthcare worker
- ❍ Other (please give more information in the space provided)

3. Please circle a number between 1 and 7 showing how useful you found the psychiatric genetic counselling session.

For example, circling 1 would indicate that you did not find it at all useful, circling 4 would indicate that you found it somewhat useful, and circling 7 would indicate that you found it extremely useful.

Not at all useful						Extremely useful
1	2	3	4	5	6	7

4. What topics were covered during the psychiatric genetic counselling session?
Please show your answer to each statement by ticking either **YES** or **NO**

	YES (1)	NO (2)
Discussed my family history of mental illness with me (1)	❑	❑
Discussed information about the genetic contrinbutions to mental illness with me (2)	❑	❑
Discussed information about the non-genetic factors to mental illness with me (3)	❑	❑
Discussed family planning decisions in relation to my mental illness (4)	❑	❑
Discussed ways my relative with a mental illness can protect their mental health (6)	❑	❑
Discussed ways to protect my own mental health (5)	❑	❑
Discussed genetic testing regarding mental illness (7)	❑	❑
Provided information about the chances of my relatives also developing mental illness (8)	❑	❑
Provided information about the chances of children in my family (including from future pregnancies) also developing mental illness (9)	❑	❑
Provided referrals and information to other services that may be relevant to me or my	❑	❑

relative (e.g. support groups/referral to psychiatrist/psychotherapy) (10)		
Provided emotional support (11)	❑	❑
Other (please give more information in the space provided below): (12)	❑	❑

5. Were there any aspects of the psychiatric genetic counselling that you found particularly useful or helpful? If yes, what were they? Please write your answer in the space provided below:

6. Were there any topics that you would have liked more information about, or that were not covered during the session that you would have liked to have been? Please write your answer in the space provided below:

7. Were there any aspects of this service that you did not find useful or helpful, or felt uncomfortable talking about? Please write your answer in the space provided below:

8. Did you share any information you from your genetic counselling session with your relatives or friends? If yes, what did you discuss, and with who? Please write you answer in the space provided below:

9. Are there any ways in which your psychiatric genetic counselling session could be improved to make it more useful or helpful to you? Please write you suggestions in the space provided below:

10. Please circle a number between 1 and 7 showing how useful you think psychiatric genetic counselling would be for your **relative with a mental illness.**

For example, selecting 1 would indicate that you do not think it would be at all useful for them, selecting 4 would indicate that you think they it would be somewhat useful, and selecting 7 would indicate that you think it would be extremely useful for them.

Not at all useful						Extremely useful
1	2	3	4	5	6	7

11. What aspects of psychiatric genetic counselling, if any, do you think would be particularly helpful or useful to your relative with a mental illness? Please write your answer in the space provided below:

END OF SURVEY

Appendix C – Research Ethics Approval

Bournemouth University

Research Ethics Checklist

Reference Id	6669
Status	Approved
Date Approved	11/03/2015

Researcher Details

Name	Rosa Spencer-Tansley
School	Faculty of Science & Technology
Status	Postgraduate Research (PhD, MPhil, DProf, DEng)
Course	Postgraduate Research
Have you received external funding to support this research project?	No
Please list any persons or institutions that you will be conducting joint research with, both internal to BU as well as external collaborators.	Dr Kevin McGhee, BSc, PhD, Bournemouth University. Dr Johannine Austin, PhD, MSc (Genetic Counselling) CCGC/CGC, University of British Columbia

Project Details

Title	Psychiatric Genetic Counselling: Addressing the needs of the UK population
Proposed Start Date	15/02/2015
Proposed End Date	30/09/2015

Summary (including detail on background methodology, sample, outcomes, etc.)
Please see attached document B- 'Supporting Summary'

External Ethics Review

Does your research require external review through the NHS National Research Ethics Service (NRES) or through another external Ethics Committee?	No

Research Literature

Is your research solely literature based?	No

Human Participants

Will your research project involve interaction with human participants as primary sources of data (e.g. interview, observation, original survey)?	Yes
Does your research specifically involve participants who are considered vulnerable (i.e. children, those with cognitive impairment, those in unequal relationships—such as your own students, prison inmates, etc.)?	Yes
Is a DBS check check required?	No
Does the study involve participants age 16 or over who are unable to give informed consent (i.e. people with learning disabilities)? NOTE: All research that falls under the auspices of the Mental Capacity Act 2005 must be reviewed by NHS NRES.	No
Will the study require the co-operation of a gatekeeper for initial access to the groups or individuals to be recruited? (i.e. students at school, members of self-help group, residents of Nursing home?)	Yes
Will it be necessary for participants to take part in your study without their knowledge and consent at the time (i.e. covert observation of people in non-public places)?	No
Will the study involve discussion of sensitive topics (i.e. sexual activity, drug use, criminal activity)?	Yes

Are drugs, placebos or other substances (i.e. food substances, vitamins) to be administered to the study participants or will the study involve invasive, intrusive or potentially harmful procedures of any kind?	No

Will tissue samples (including blood) be obtained from participants? Note: If the answer to this question is 'yes' you will need to be aware of obligations under the Human Tissue Act 2004.	No

Could your research induce psychological stress or anxiety, cause harm or have negative consequences for the participant or researcher (beyond the risks encountered in normal life)?	Yes
Will your research involve prolonged or repetitive testing?	No

Will the research involve the collection of audio materials?	No
Will your research involve the collection of photographic or video materials?	No
Will financial or other inducements (other than reasonable expenses and compensation for time) be offered to participants?	No

Please explain below why your research project involves the above mentioned criteria (be sure to explain why the sensitive criterion is essential to your project's success). Give a summary of the ethical issues and any action that will be taken to address these. Explain how you will obtain informed consent (and from whom) and how you will inform the participant(s) about the research project (i.e. participant information sheet). A sample consent form and participant information sheet can be found on the Research Ethics website.
•Your research involves participants who are considered vulnerableInvolvement of participants who are considered vulnerableThis research project aims to explore the understanding and perceptions of mental illness and genetic counselling amongst individuals that have a mental illness (CONDITIONS INCLUDED IN THE RESEARCH STUDY: bipolar disorder, schizophrenia, schizoaffective disorder). As such, it is necessary to interview individuals that have a mental illness in order to include them as participants in this study.Importantly, all participants in this study that have a mental illness will be living at home, not currently experiencing a psychotic episode, and able to give informed consent (i.e. not under the auspices of the Mental Capacity Act 2005).Please see Document B.4 which provides further explanation and evidence of consideration of this criteriaInformed consentIn order to ensure informed consent is obtained, all study participants will be given participant information sheets and consent forms to read and, should they decide to take part in the study, to sign – a signed copy will be kept by them, and a signed copy kept by the research team. The participant will also keep a copy of the participant information sheet.Please see document B.4 and Document D (Participant Information sheet) and Document E (Consent Form)GatekeepersParticipants will be recruited via UK mental health charities including 'Rethink' and 'Hafal' and 'Mind.' This study will not gather personal contact information of potential participants from individual charities. Rather, the charities will promote this study through social media, their literature and associated support groups. No participant data will be exchanged between them and us.Briefing sheets will be sent to the gatekeepers of these charities (copies of the briefing sheets have been uploaded as part of this ethics submission). The charities will promote the research through their literature, social media, and support groups. Please see document B (4.1) and Document C (Briefing Statement) as supporting evidence of our consideration of this criteriaDiscussion of sensitive topicsAlthough no direct risks have been identified to participants that take part in this study, the study does involve discussion of topics that may be sensitive to some individuals. These topics include: causes of mental illness; genetic risk in mental illness; reproductive decisions including in relation to mental illness; family history of mental illness; genetic testing and genetic counselling. For some individuals this may mean coming across new information related to mental illness which may raise questions or concerns they had not previously considered; for others this may remind them of issues that are already of a sensitive nature to them. This may cause anxiety, worry or stress. In anticipation of this, we have stated in the participant information sheet (Document D) that individuals should contact their GP should they have any concerns/questions related to information or topics discussed in the study. Participants will also have the research team's contact details; should they contact us in relation to such concerns, we would also recommend that they visit their GP.

Final Review

Will you have access to personal data that allows you to identify individuals OR access to confidential corporate or company data (that is not covered by confidentiality terms within an agreement or by a	Yes

separate confidentiality agreement)?	

Please explain below why your research requires the collection of personal data. Describe how you will anonymize the personal data (if applicable). Describe how you will collect, manage and store the personal data (taking into consideration the Data Protection Act and the 8 Data Protection Principles). Explain how you will obtain informed consent (and from whom) and how you will inform the participant about the research project (i.e. participant information sheet).
This research will involve the collection of demographic data (age, occupation, nationality). This may be useful to us when it comes to analysing the data, for example to identify relationships between certain responses and ages of participants.Contact details of participants are no longer necessary as we have amended the terms to withdrawing data up to the point of anonymisation following Panel's recommendation (see document 'Response to Panel's recommendations') This research team respects the importance of participant's confidentiality and it is a matter that we take very seriously. All the information that we collect about participants during the course of the research will be kept strictly confidential. Any information regarding individual's participation in this research study will not be disclosed to anyone outside the stated research team. The research data will be processed and stored securely at Bournemouth University, in a locked drawer in a locked room.The survey forms (for paper questionnaires) will include only the participant's unique study ID number, and no personal identifiers or any information that may be used to identify the individual (such as name, initials, date of birth, or contact information). Similarly, the online survey response forms will contain no personal identifiers or any information that may be used to identify the individual (such as name, initials, date of birth). Participants will not be able to be identified in any reports or publications that use the results of the research study (i.e. for a subsequent PhD project)To ensure informed consent, All participants will read and sign copy the participant information sheet and consent form, and will be given a copy of these to keep for their own purposes. A copy of these has been uploaded as part of this ethics application (Document D and E)

Will your research involve experimentation on any of the following: animals, animal tissue, genetically modified organisms?	No
Will your research take place outside the UK (including any and all stages of research: collection, storage, analysis, etc.)?	No

Please use the below text box to highlight any other ethical concerns or risks that may arise during your research that have not been covered in this form.

Appendix D – Cover letter

Faculty of Science and Technology
Translational Genetics Research Group
Department of Life and Environmental Sciences

Dear participant,

Thank you for your interest in our study.

You will find attached a participant information sheet which gives more detailed information on the study.

However in brief, we wanted to highlight the main reason for undertaking this research:

- We are always looking for ways to improve mental health services within the UK.
- We still have a lot to learn about how our brains work and how biology, psychology and environment interact to produce human behaviour.
- The biggest problem we face today is finding effective ways of translating research into something beneficial for patients and their families.
- We think that Psychiatric Genetic Counselling will be an important tool in achieving this goal.
- We want to obtain your views around this, so we would be really grateful if you could take part in this study.

The questionnaire should take you around 15 minutes to complete and is online.

Please click here to take part.

Thank you for your time.

Yours sincerely

Rosa Spencer-Tansley
Postgraduate Researcher

Dr Kevin McGhee
Research Supervisor
01202 968189

If the link to the questionnaire above doesn't work please use this web address:
https://surveys.qualtrics.com/jfe/form/SV_3NO0xWiTaPwFLhz

If you want to find out more about our group's research click the following link:
http://kmcghee4.wix.com/translational

Appendix E – Participant information form

Participant Information Sheet

Genetic counselling for psychiatric illness.

We would like to invite you to take part in our research project. Before you decide whether or not you want to take part it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully and discuss it with others if you wish. Ask us if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part.

Purpose: While genetic counselling has been around since the 1960s, specialised genetic counselling to individuals with a mental illness and their family members is not currently routinely offered in the UK. We would like to explore whether genetic counselling may be beneficial for UK individuals with mental illness and their relatives, and whether there is interest in receiving genetic counselling amongst this population.

Why have I been invited?
You are being invited to participate in this research study because you have been diagnosed with a mental illness; or you have a relative that has been diagnosed with a mental illness. Your perceptions of mental illness and genetic counselling may be helpful in indicating the suitability of psychiatric genetic counselling in the UK. A total of 200 participants are to be recruited from the UK.

Do I have to take part?
Your participation is entirely voluntary, so it is up to you to decide whether or not to take part to take part in this study.
If you do decide to take part you will be given this information sheet to keep and you will be asked to sign a consent form. If you do decide to take part, you can still withdraw yourself and your results up to the point of anonymisation. You do not have to give a reason for your withdrawal and it will involve no penalty or loss of benefits you or your relative are entitled to, nor will it affect the care you or your relative may currently be receiving in the NHS or privately.

If you do not wish to participate, you do not have to provide any reason for your reason not to participate. Your decision not to participate will involve no penalty or loss of benefits to which you or your relative are entitled to in any way, nor will it affect the care you or your relative may currently be receiving in the NHS or privately.

What will the study involve?
If you decide to participate in the study, you will be asked to complete a short questionnaire. This will either be completed online, or paper copies may be administered by the study's researcher. You will be asked to provide basic demographic information, including your age, gender and nationality. You will also be asked to provide basic clinical information including your (or your relative's) clinical psychiatric diagnosis, and age (or your relative's age) at time of diagnosis.
Many of the questions on the questionnaire will ask about your perceptions of the causes of mental illness, your perceptions of genetic risk in mental illness, and your opinions regarding genetic counselling.
As part of the study you will also watch a short video about psychiatric genetic counselling. You will then complete some questions about your opinions of topics raised in the video. The video will be approximately 1 minute long.

Particpant information form (ctd).

The questionnaire should take around 20-25 minutes in total. Should you not wish to answer any particular question(s) you are free to decline and there will be no negative consequences. You will be expected to give full concentration to the experiment. You are not expected to do any preparation in advance of the research study.

Potential benefits:
Whilst there are no immediate benefits for those people participating in the project, it is hoped that this research will indicate whether psychiatric genetic counselling may be useful to the UK population. The findings may also indicate ways in which current mental health services in the UK could be improved.

Compensation and payments:
You will not receive any monetary rewards or compensation for taking part in this study. However through participating you will be contributing towards important scientific research that we hope may help improve clinical services for individuals and families affected by mental illness in the future.

Potential risks:
Through participating in this study it you may come across new information related to mental illness which may raise questions or concerns that you had not previously considered. Should this happen, you should seek advice from your G.P who will be able to refer you to the appropriate specialist services.

Confidentiality:
Your confidentiality will be respected. All the information that we collect about you during the course of the research will be kept strictly confidential. Any information regarding your participation in this research study will not be disclosed to anyone outside the stated research team.

The survey forms will include only your unique study ID number, and no personal identifiers or any information that may be used to identify you (such as your name, initials, date of birth, or contact information). The research data will be processed and stored securely at Bournemouth University, in a locked drawer in a locked room.

The results will be used to produce a thesis; participants are welcome to request a copy of this once it has been published. The intention is also to publish the results in open-access medical journals relevant to psychiatric genetics and genetic counselling. You will not be able to be identified in any reports or publications.
The data collected during the course of the project might be used for additional or subsequent research. For example, the data may be used for a PhD project following on from this study.

Who has reviewed this study?
This study has been reviewed and approved in line with Bournemouth University's Research Ethics Code of Practice.

Particpant information form (ctd.)

Contact for information about the study
If you have any questions or desire further information regarding this study you may contact the researcher whose contact information is available on this sheet. Any complaints are to be directed to Professor Matt Bentley, Deputy Dean – Research and Professional Practice – whose contact details can be found at the bottom of this sheet.

Please note that this sheet is for participants to keep. Should you choose to participate you will be asked to sign a consent form, a copy of which you will also keep.

Thank you very much for taking the time to read through the information provided.

Kind regards

Rosa Spencer-Tansley, BSc

Contact information

Researcher
Rosa Spencer-Tansley, Post-graduate Research Student
rspencertansley@bournemouth.ac.uk

Supervisor
Dr. Kevin McGhee, PhD. Senior lecturer in Health Sciences at Bournemouth University
kmcghee@bournemouth.ac.uk

Faculty of Science and Technology,
Christchurch House, Bournemouth University,
Fern Barrow, Talbot Campus, Poole, Dorset
BH12 5BB

01202 968189

Professor Matt Bentley
Deputy Dean – Research and Professional Practice, Bournemouth University
Christchurch House C227, Talbot Campus, Fern Barrow, Poole, BH12 5BB
01202 962203

This research forms part of the Bournemouth University Translation Genetics Research Group led by Dr. Kevin McGhee BSc, PhD. The project is running in collaboration with the Psychiatric Genetic Counselling Clinic, British Columbia, Canada, led by Dr. Jehannine Austin, PhD, MSc (Genetic Counselling) CCGC/CGC

Appendix F – Consent form

Consent Form

Evaluating the application of Psychiatric Genetic Counselling in the UK.

Researcher:
Rosa Spencer-Tansley, Post-graduate research student
rspencertansley@bournemouth.ac.uk

Supervisor:
Dr. Kevin McGhee, Senior lecturer in Health Sciences at Bournemouth University
kmcghee@bournemouth.ac.uk

	Please initial here
I confirm that I have read and understood the participant information sheet and consent form for the above research project	
I have had the opportunity to consider the information and ask any questions I have, and that these have been answered satisfactorily.	
I understand that my participation is voluntary and that I am free to withdraw up to the point of anonymisation, without giving reason and without there being any negative consequences. In addition, should I not wish to answer any particular question(s), I am free to decline.	
I give permission for members of the research team to have access to my anonymised responses. I understand that my name will not be linked with the research materials, and I will not be identified or identifiable in the report or reports that result from the research.	
I understand that the data collected during the course of the project might be used for additional or subsequent research, such as a subsequent PhD project.	
I understand that there is no guarantee that this study will provide any benefits to me	
I agree to take part in the above research project.	

Printed name of Participant	Date	Signature
Printed name of Researcher	Date	Signature

Appendix G – Video script

"Psychiatric genetic counselling is something that's often quite misunderstood I think. People have some misperceptions about what it might be. So specifically, when people hear the phrase psychiatric genetic counselling they tend to think about pregnancy, childbearing decisions and conversations about "What are the chances that my son or daughter might have a psychiatric illness?"

Those are absolutely things we can discuss in the context of psychiatric genetic counselling, but we can also do way more than that.

For example people who have experiences themselves of psychiatric problems often feel really guilty or ashamed about having that illness. They will often feel that perhaps they have done something themselves that caused the experience that they have, the illness that they have, and feel ashamed about that. Parents of people with psychiatric problems will worry that perhaps there was something that they did that caused their child to become sick, or they might wonder if there was something they could have done to prevent it.

Those sorts of guilt, shame and stigma things can be really problematic for people and those are things that we can address really helpfully in psychiatric genetic counselling

Fundamentally psychiatric genetic counselling is about helping people to better understand what we know from research about the causes of psychiatric disorders - about how genes and environment can work together to contribute to the development of these conditions. And it's about providing people with support and counselling around that to address any guilt or shame or stigma they might be feeling.

But in addition what we can also do is talk with people about strategies they might be able to use to protect mental health going forward. So for some people that might involve things like meditation, for other people perhaps spending time with a pet, going for walk with dog, or so on. We help people to find on an individual basis things that will work for them to help protect their mental health.

So these are all things that genetic counsellors can do."

Dr. J. Austin, PhD, CCGC. February 2015.

AVAILABLE FROM: https://www.youtube.com/watch?v=PqnxqMnPk_g

Appendix H: Causal Attribution Questionnaire (Clinical tool)

Received from: (Dr. J. Austin, pers comms., 26 November 2014. © J. Austin, 2014).

Please circle a number between 1 and 9 according to how much you think YOUR mental illness was caused by genetic factors. For example, circling 1 would indicate that you believe that genetic factors did not contribute at all to your mental illness, circling 5 would indicate that you believe that genetic factors contributed a moderate amount, and circling 9 would indicate that you believe your mental illness was caused *entirely* by genetic factors.

Genetics did not contribute at all		Genetics contributed moderately					Genetics alone caused my illness	
1	2	3	4	5	6	7	8	9

Now, please circle a number between 1 and 9 according to how confident or sure you are of the answer you provided above. For example, circling 1 would indicate that you are not at all sure that the answer you provided is correct, and circling 9 would indicate that you are absolutely certain that the answer provided above is correct.

I am not sure at all			I feel somewhat uncertain				I am completely confident	
1	2	3	4	5	6	7	8	9

Please circle a number between 1 and 9 according to how much you think YOUR mental illness was caused by your experiences (you can also think about this as things that happened to you, or environmental factors). For example, circling 1 would indicate that you believe that your experiences did not contribute at all to your mental illness, circling 5 would indicate that you believe that your experiences contributed a moderate amount, and circling 9 would indicate that you believe your mental illness was caused *entirely* by your experiences.

My experiences did not contribute at all		My experiences contributed moderately					My experiences alone caused my illness
1	2	3	4	5	6	7	8	9

Now, please circle a number between 1 and 9 according to how confident or sure you are of the answer you provided above. For example, circling 1 would indicate that you are not at all sure that the answer you provided is correct, and circling 9 would indicate that you are absolutely certain that the answer provided above is correct.

I am not sure at all		I feel somewhat uncertain					I am completely confident
1	2	3	4	5	6	7	8	9