

Running Head: CHARACTERISING DEVELOPMENTAL PROSOPAGNOSIA

The Definition and Diagnosis of Developmental Prosopagnosia

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Abstract

Over the last 20 years much attention in the field of face recognition has been directed towards the study of developmental prosopagnosia (DP), with some authors investigating the behavioural characteristics of the condition and many others using these individuals to further our theoretical understanding of the typical face-processing system. It is broadly agreed that the term “DP” refers to people who have failed to develop the ability to recognize faces in the absence of neurological illness or injury, yet more precise terminology in relation to potential subtypes of the population are yet to be confirmed. Further, specific diagnostic techniques and inclusion and exclusion criteria have yet to be uniformly accepted across the field, making cross-paper comparisons and meta-analyses very difficult. This paper presents an overview of the current challenges that face research into DP, and introduces a series of papers that attempt to further our understanding of the condition’s characteristics. It is hoped that that this special issue will provide a springboard for further research addressing these issues, improving the current state of the art by ensuring the quality of theoretical investigations into DP, and by posing advances that will assist those who have the condition.

Keywords: Developmental prosopagnosia; face recognition; subtypes; diagnosis.

The Definition and Identification of Developmental Prosopagnosia

Prosopagnosia is a cognitive condition characterised by a severe impairment in face recognition. While it has traditionally been reported in a relatively small number of individuals following neurological illness or trauma (e.g. Bate et al., 2015; Damasio, Damasio, & Van Hoesen, 1982; Young & Ellis, 1999), the last 20 years has seen a sharp rise in reports of people who experience the same difficulties but in the absence of brain injury (e.g. Bate, Haslam, Jansari, & Hodgson, 2009; Bate & Cook, 2012; Bennetts, Butcher, Lander, Udale & Bate, 2015; Duchaine & Nakayama, 2006; Jones & Tranel, 2001). This form of the condition is typically referred to as “developmental” or “congenital” prosopagnosia, with the latter term reflecting observations that the condition can sometimes run in families (e.g. Duchaine, Germine & Nakayama, 2007; Lee, Duchaine, Wilson & Nakayama, 2010). While prevalence studies estimate that developmental prosopagnosia (DP) affects approximately two per cent of the population (e.g. Bowles et al., 2009; Kennerknecht et al., 2006), there has been increasing debate about the manner in which DP is identified, drawing the findings of some studies into question. Moreover, the evidence thus far suggests that the DP population is by no means homogenous (for reviews see Bate & Bennetts, 2014; Susilo & Duchaine, 2013), and as a consequence clarity on testing procedures and classification is sorely warranted. Finally, and perhaps most fundamental, ongoing debates about the precise nature of DP and its diagnosis raise potential issues for the plethora of theoretical studies that have investigated the condition as a means to make inferences about the typical face-processing system.

DP: HISTORICAL BACKGROUND

It was initially thought that DP was an extremely rare disorder (e.g. de Haan, 1999; Jones & Tranel, 2001). In 1963, Bornstein briefly described some cases of prosopagnosia that appeared to be developmental in origin, but McConachie (1976) was the first to publish a formal case study. She described the case of AB, an intelligent 12-year-old girl who reported severe difficulties in the recognition of unfamiliar faces and those of her peers. AB's mother also reported problems with face recognition. De Haan and Campbell (1991) performed a follow-up study with AB 15 years after the original report, and found that, as an adult, AB still presented with obvious difficulties in face recognition, and also with the recognition of emotional expression and within-class objects. A handful of other cases of DP were also reported in the 1990s (e.g. Bentin, Deouell & Soroker, 1999; Kracke, 1994; Temple, 1992). However, at the beginning of the 21st century it became apparent that DP is not as rare as originally thought. Wide availability of the Internet and flurries of international media coverage led to the uncovering of many more individuals who appear to have the condition, and there are now well over 100 reports in the published literature.

Given the large numbers of people who have made themselves known to researchers, some authors have attempted to estimate the prevalence of the condition. In an initial study, Kennerknecht et al. (2006) screened 689 German secondary school pupils and medical students for DP, using self-report measures and semi-structured interviews. They concluded that 17 members of their sample had DP, corresponding to a prevalence rate of 2.47%. In a later paper, the same group performed a similar survey in 533 medical students at the University of Hong Kong, finding a prevalence rate of 1.9% (Kennerknecht, Yee-Ho & Wong, 2008). However, the use of self-report measures alone for the diagnosis of DP has been contentiously debated (e.g. Duchaine, 2008; Tree, 2011), limiting the validity of these prevalence estimates. Most work examining the typical population suggests that people have

very little insight into their face recognition skills, reporting only weak-to-moderate correlations between subjective ratings and scores on objective tests (Bindemann, Attard & Johnston, 2014; Bowles et al., 2009; Rotshtein, Geng, Driver & Dolan, 2007; McGugin, Richler, Herzmann, Speegle & Gauthier, 2012). A similar lack of insight has not been formally reported in DP, yet would be unsurprising given many people do not realise that their face recognition skills differ from typical perceivers until they reach mid- to late-adulthood. Further, many people with DP can often “get by” through the use of elaborate compensatory strategies that are successful at least some of the time (Bate & Bennetts, 2014), perhaps obscuring insight into the severity of their condition. Indeed, unlike people with acquired prosopagnosia, those with DP have never had normal face recognition skills and therefore have no baseline for comparison.

One recent study developed a 20 item questionnaire and reported a strong correlation between scores on the objective Cambridge Face Memory Test (CFMT; Duchaine & Nakayama, 2006) and questionnaire scores (Shah, Gaule, Sowden, Bird & Cook, 2015), yet the authors combined their typical participants and those with DP into one group for analysis. It is therefore unclear whether the effect sizes are driven by only one group, particularly because the participants with DP had already self-referred to the authors’ laboratory for assessment. The conclusions that can be drawn from this study with regard to the reliability of self-report measures are therefore limited, and it may be that the questionnaire was simply distinguishing those who already suspected they had DP from the typical population without revealing trends within each group. Nevertheless, Shah et al.’s paper has resurrected the self-report debate.

The only prevalence study reported to date that used objective measures is that of Bowles et al. (2009). This investigation screened 241 Australian adults using the CFMT – a computerized test that has been thoroughly validated and standardized, and has been found to

reliably diagnose both acquired and developmental cases of prosopagnosia (e.g. Bate et al., 2014; Bate, Haslam, Tree & Hodgson, 2008; Duchaine et al., 2007; Rezlescu, Pitcher & Duchaine, 2012). These authors used the standard neuropsychological procedure of taking two standard deviations from the control mean to calculate a cut-off score that signifies impaired performance, resulting in a prevalence rate of between 2.0 and 2.9 per cent. However, one could claim that this statistical procedure inevitably results in such a prevalence estimate, given it typically identifies the bottom two per cent of the population in a normally distributed sample. Although Bowles et al. (2009) make the case that their participants who were candidates for DP also performed very poorly on a famous face test, it is unclear whether at least some people who are thought to have DP are simply those at the “bottom end of normal”. Evidence supporting this perspective comes from findings that the face recognition continuum is broader than originally thought, with substantial variation in performance within the typical population (e.g. Bate, Parris, Haslam & Kay, 2010; Wang, Li, Fang, Tian & Liu, 2012). Further, some work has identified individuals at the opposite extreme to those with DP – so-called “super-recognizers”, who have extraordinarily good face recognition skills (e.g. Bobak, Bennetts, Parris & Bate, in press; Bobak, Dowsett & Bate, 2016; Bobak, Hancock & Bate, 2016; Russell, Duchaine & Nakayama, 2009). If DPs and super-recognizers simply represent the top and bottom end of the typical population, it may be the case that their performance merely reflects a quantitative rather than qualitative difference to that of the majority. Alternatively, if actual differences in processing strategy or biological markers of DP and super recognition are observed, this would be stronger evidence that the two terms refer to qualitatively different groups of people. There is some evidence to support this in individuals with DP, given recent neurological findings that suggest structural atypicalities within critical regions implicated in face-processing (e.g. Behrmann, Avidan,

Gao & Black, 2007; Burns, Tree & Weidemann, 2014; Garrido et al., 2009; Song et al., 2015).

An alternative approach to addressing this issue is to examine whether DP candidate cases are impaired on multiple tests that assess the same process – in effect, the judgment is based on a *convergence* of evidence, rather than that of a single test. For instance, while the CFMT is probably the most well-used test of unfamiliar face recognition, some researchers assess their participants using additional versions of the same task (e.g. McKone et al., 2011) or alternative paradigms (e.g. Lee et al., 2010). Further, most researchers use tests that assess familiar face recognition, typically using the faces of celebrities (e.g. Bate et al., 2014; Duchaine & Nakayama 2006; Lee et al., 2010). Consistently poor performance on a number of tests provides more convincing evidence that a person is clinically poor at face recognition, even if (perhaps by chance) they score within the normal or borderline range on a single task. Likewise, an individual without everyday face recognition difficulties may perform within the impaired range on a single attempt at a particular test, and a multi-test approach would prevent them from inappropriately being categorized as having prosopagnosia.

The tests discussed so far assess the hallmark symptom of prosopagnosia – that is, severe difficulties in facial identity recognition. However, face-processing skills can also be evaluated by examining performance on the *perception* of facial stimuli. In this case, testing places no demands on face memory; participants typically make a series of perceptual judgments (same/different) about simultaneously-presented faces. In the acquired prosopagnosia literature there have been reports of individuals with intact (e.g. de Haan, Young & Newcombe, 1987, 1991; Barton, Press, Keenan & O'Connor, 2002) and impaired (e.g. Young, Hellowell & de Haan, 1988; Barton et al., 2002) face perception performance, motivating a claim for different subtypes of the condition (sometimes also referred to as

apperceptive and *associative* variants: De Renzi, Faglioni, Grossi & Nichelli, 1991).

However, other authors claim that all people with prosopagnosia show at least *some* deficits in face perception when appropriately tested, indicating that face perception deficits may reside on a continuum of impairment (Farah, 1990). For instance, individuals who achieve apparently normal scores on face matching tasks may take an unusually long period of time to make their responses, indicating the use of atypical strategies despite apparently normal accuracy performance. In the DP literature, there have also been reports of individuals who achieve normal (e.g. Bate et al., 2009; Lee et al., 2009; McKone et al., 2011) and impaired (e.g. Bate et al., 2009; Chatterjee & Nakayama, 2012; Duchaine et al., 2007) scores on tests of face perception, typically using the Cambridge Face Perception Test (CFPT; Duchaine et al., 2007) which requires participants to sort faces in order of their similarity to a target face. As a consequence, there remains a similar issue in the DP literature as to whether evidence of impaired/intact perceptual performance warrants the claim for two *subtypes* of this population – or whether (to echo Farah, 1990) face perception performance in the DP population is continuum in nature.

Clearly, further work is required to confirm the precise nature of DP and to provide a firm set of qualifying criteria. This is particularly reflected in the interchangeable use of the terminology that is used to refer to the condition, with some authors using the terms “congenital” or “hereditary” prosopagnosia to refer to individuals who would be classified by others as “DP”. This confusion in terminology comes from both confirmed (e.g. Duchaine et al., 2007; Lee et al., 2010) and anecdotal (e.g. Kennerknecht et al., 2006) reports suggesting that DP runs in families, raising the possibility that the condition may have a genetic basis. However, many other cases fail to report family members who are also suspected to have the condition (e.g. Bate et al., 2009; Duchaine, 2008), and no gene for the condition has yet been found. The latter is perhaps unlikely given the apparent cognitive heterogeneity in the

presentation of DP, implying that the condition may have different subtypes and result from a potentially large variation of genetic and/or developmental atypicalities (Bate & Bennetts, 2014). Given the possibility that even apparent familial cases may not necessarily reflect genetic influence, other authors prefer to use the more conservative term “DP” for all cases of prosopagnosia with an absence of brain injury (e.g. Bate et al., 2014; Duchaine et al., 2007; Susilo & Duchaine, 2013; Susilo, Wright, Tree & Duchaine, 2015).

An absence of neurological injury or illness is not the sole exclusion criteria for DP classification: most researchers will also exclude individuals with low-level perceptual, cognitive or intellectual difficulties and those with a history of psychiatric or other developmental conditions. The most contentious of these exclusion criteria relates to concurrent socio-emotional developmental conditions; in particular autism spectrum disorder (ASD). Face-processing difficulties can also occur in ASD, although the extent to which identity recognition is affected does seem to substantially vary between individuals (e.g. Barton et al., 2004; Boucher, Lewis & Collis, 1998; Weigelt, Koldewyn & Kanwisher, 2012). Nevertheless, a tightly-locked developmental link between face-processing difficulties and social dysfunction has been suggested, raising the possibility that the two abnormalities inevitably co-occur from an early point in development (Schultz, 2005). However, evidence of DPs without socio-emotional symptoms provide a strong counter to this hypothesis (e.g. Duchaine, Murray, Turner, White, & Garrido, 2009), thus implying that DP should be viewed as an independent condition to ASD.

Importantly, hypotheses about the developmental course of DP and its link to other developmental influences are probably best tested in the context of childhood studies. Such work has the potential not only to investigate early influences on the development of face recognition difficulties, but can also answer other key theoretical questions about the development of the typical adult face-processing system. For instance, many authors accept

that there are well specified cognitive and neural processes that are dedicated to face-processing over and above other classes of object (Duchaine & Yovel, 2015; Kanwisher, McDermott & Chun, 1997; Yin, 1969; Yovel & Kanwisher, 2004). It is possible that these systems are more generalised at birth and go through a process of refinement that makes them specialised for face recognition (e.g. Nelson, 2001). If this is the case, a key question concerns the age at which DP might emerge, and particularly the age at which it becomes face-specific. Of course, many people with both acquired (e.g. Damasio, Damasio & Van Hoesen, 1982; Gauthier, Behrmann & Tarr, 1999) and developmental (e.g. Behrmann, Avidan, Marotta & Kimchi, 2005; Garrido, Duchaine & Nakayama, 2008) prosopagnosia also present with at least some deficits in object-processing, although it is recommended that these individuals are nevertheless classed as prosopagnosic when the over-riding difficulty is with faces (Dalrymple & Palermo, 2016). The same line of reasoning can be applied to the issue surrounding facial identity perception – some recent work examining DP in children found deficits in this process in their entire sample, but only in some adults with the condition (Dalrymple, Garrido & Duchaine, 2014). Again, it may be that early deficits in face perception can in some cases be spontaneously rectified during the course of development, presenting important implications for developmental theories of face-processing. While there are relatively few reports of DP in childhood compared to those in adults, further investigation of cases that are early in development and longitudinally monitoring their progression is fundamental for progress in the field.

CONTENTS OF THIS SPECIAL ISSUE

This special issue brings together a variety of novel investigations that provide insights into the nature of DP, and have important implications for its definition and classification. First, Bobak and colleagues describe findings from an eye-movement investigation involving

individuals with DP, super-recognizers and matched control groups. While the authors found that DPs spend less time examining the eyes and more time examining the mouth than controls, super-recognizers spent more focusing on the nose. Pertinently, the latter finding positively correlated with face recognition skills in the control group, indicating that while the super-recognizers are perhaps at the top end of “normal”, the DPs are using strategies that are qualitatively different to those of the typical population. This work raises the possibility that DP may be detected by such a measure, and supports the case that these individuals are not simply those who are quantitatively at the bottom end of the normal distribution.

In the second paper, Palermo and colleagues address the issue of self-report in the diagnosis of DP. Across a number of studies conducted at different universities, the authors screened the face recognition skills of a large number of typical participants and a group of DPs using a self-assessment questionnaire and objective tests of face recognition. They found little evidence of an association between objective and subjective measures in typical participants, and although the DP group did rate their face recognition skills as significantly lower than the typical participants, their self-ratings were not predictive of the severity of their prosopagnosia. These findings indicate that self-report questionnaires may be a useful pre-screen to identify candidate individuals for prosopagnosia, but do not provide reliable insights into the severity of face recognition deficits.

Third, Bennetts and colleagues report the first prevalence estimate of DP in childhood, testing nearly 600 children on a variety of tasks that assessed the perception and recognition of faces and objects. While 1.2-5.2% of children showed difficulties with face recognition, these were restricted to faces over objects in 1.2-4% of the sample. The authors raise important questions about the age-appropriateness of the tests that are currently available for diagnosing DP in children, and in particular the age at which impaired scores can be used as reliable indicators of the condition. In addition, they highlight the possibility

that the prevalence of developmental face recognition difficulties may be higher in childhood, and that, for some individuals, normal face recognition performance may just be developmentally delayed.

Dalrymple, Elison and Duchaine continue the developmental theme of this special issue by examining object-processing skills in six children with DP, aged 5-12 years. Four of the children, including the youngest, displayed impairments that were limited to faces. These findings do not only suggest that face-specific cases of DP can be identified as young as five years of age, but also add weight to the hypothesis that face and object processing are underpinned by independent systems that diverge early in development (for reviews see McKone, Crookes, Jeffery & Dilks, 2012; Weigelt et al., 2014).

The domain-specificity of DP is further assessed by Rivolta and colleagues in the fifth paper. These authors assessed body perception skills in a group of 11 adults DPs, and found that although the participants achieved normal scores, their response times were slower than those of typical participants. This work converges with reports that suggest similar holistic mechanisms may be used for face and body recognition (Minnebusch, Suchan & Daum, 2009; Reed, Stone, Bozova & Tanaka, 2003), raising the possibility that, at least in some cases, face recognition impairments in DP may be underpinned by more general impairments in holistic processing.

The issue of impaired face perception in adults with DP is addressed by two papers. White and colleagues tested the ability of adult DPs to make same/different identity judgments for pairs of faces. In an initial experiment, DPs were found to perform at similar levels to controls when matching pairs of unfamiliar faces where the photos of repeated individuals were taken in the same studio on the same day. However, on a more challenging task where the images showed more variability in their appearance (i.e. photographs of the same person were taken many weeks apart), the performance of DPs was impaired on trials

involving both familiar and unfamiliar faces. In particular, DPs were poor at matched rather than mismatched trials. Ulrich and colleagues assessed the face perception skills of a group of 11 DPs using a variety of tests, and did not find evidence of perceptual deficits in seven cases. While Ulrich et al.'s paper suggests that, akin to acquired prosopagnosia, deficits in facial identity perception are not necessary for a diagnosis of DP and indeed may be indicative of different subtypes, the findings of White and colleagues demonstrate the importance of task design and difficulty when exploring this issue.

Finally, Towler and colleagues present a review of investigations in the DP literature that employed event-related brain potential measures of face perception and recognition. Pertinently, this paper informs the behavioural findings presented by Ulrich, White and colleagues with regard to the impairment or preservation of the perception of facial identity. Towler et al. conclude that the speed and efficiency of information propagation through the cortical face network is altered in DP, and note specific perceptual difficulties in processing spatial and contrast deviations from canonical upright visual perceptual face templates, together with atypical visual experience with the eye region (converging with the findings of Bobak and colleagues, discussed above).

CONCLUSION

In sum, it can be seen that urgent attention needs to be directed to the definition and characterization of DP. This is of course important from a clinical perspective given the large numbers of people who potentially have the condition and are seeking diagnosis and potential remediation. However, it is also important in the context of theory testing that participants do indeed meet a universally accepted set of criteria of classification – otherwise comparisons across studies will be difficult. The fact that most case reports have focused on adults (and particularly older adults) is striking, given some of the most powerful insights into DP itself

and the development of the typical face-processing system may actually be gleaned from studies involving children. Future work therefore needs to clarify the inclusion and exclusion criteria for DP across development, and to identify potential phenotypes. This can only be achieved via the development of appropriate tests that thoroughly and reliably assess different processes, and permit cross-comparison between papers that currently reflect a variety of diagnostic practices. Clearly, these aims must be achieved in order for remediation strategies to reach their full potential (for a review see Bate & Bennetts, 2014). This special issue attempts to further the current state of the art by beginning to address these issues surrounding the diagnosis and characterisation of DP. We hope that the progress of future work will benefit from our attempt to highlight current challenges that underpin face recognition research in this field.

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