

# Knowledge and attitudes towards Sickle Cell Disease Screening: A study of members of the UK Sickle Cell Society.

Jenny Millan<sup>1</sup>  
Edwin van Teijlingen<sup>1,2</sup>  
Winifred Eboh<sup>3</sup>

1. Department of Public Health, University of Aberdeen, Scotland

2. Dugald Baird Centre for Research on Women's Health, University of Aberdeen, Scotland

3. School of Nursing & Midwifery, The Robert Gordon University, Aberdeen, Scotland

## **Introduction**

Over the past fifty years there has been an increase in the number of people in the UK who have a Sickle Cell Disorder (SCD) or are carriers. This increase has led to an expansion in research in this area; however one particular area that has not been researched rigorously is people's knowledge, attitudes and behaviour towards SCD and genetic screening. Members of the Sickle Cell Society provided an ideal population for this exploratory study looking at people's knowledge, attitudes and behaviours towards the SCD and genetic screening. This paper reports on the process and findings of this postal study.

## **Background**

Thalassaemia and SCD are autosomal recessive disorders affecting haemoglobin, a crucial component of the red blood cell. These two disorders commonly known as haemoglobinopathies affect people with origins in malarial endemic parts of the world, such as Africa, Asia, and Middle East. Carriers of the sickle, thalassaemia genes are said to have protection against malaria, but paradoxically those with the disorder are more susceptible to this parasitical condition and death from cerebral malaria being one of the most severe complications (Serjeant & Serjeant, 2001).

SCD, a condition that has variable clinical manifestations, is characterised by (1) severe obstruction of blood vessels by sickled cells preventing oxygen supply reaching tissues and organs; and (2) haemolytic anaemia where the body breaks down damaged blood cells, which have become permanently sickled (Frewin & Provan, 1997).

### **Screening for SCD**

Carriers of SCD are asymptomatic and often unaware that they carry the gene (Eboh & van den Akker, 1994). If made aware of this carrier status before they contemplate having children they could make an informed choice not to go ahead with a pregnancy if at risk of having an affected child (Laird et al., 1996). This depends on good screening and counselling services being in place. However, even in areas of the UK with a high prevalence of relevant ethnic minority groups, haemoglobinopathy screening and counselling services have been criticised for being fragmented (Anionwu & Atkin, 2001; Atkin & Ahmad, 1998).

## **Methods**

The researchers approached the Sickle Cell Society to conduct a study of its members. The Sickle Cell Society kindly agreed to post 200 questionnaires to its members. The questionnaire was designed specifically for this study using some questions designed by Dyson (1997). The importance of piloting the questionnaire was recognised and questionnaire was piloted in a small group of people with SCD in their family in both Manchester and Aberdeen. This pilot of the questionnaire aimed to test the wording, the order of questions, the range of possible answers, the clarity of the instructions for the whole questionnaire as well as those for the individual questions (Van Teijlingen & Hundley 2005: 219).

The questionnaire and a pre-paid enveloped (addressed to the University of Aberdeen) was posted by the Sickle Cell Society to 200 of its members. Following the Data Protection Act the authors did not have access to any personal details of the Sickle Cell Society members, as no names or addresses were provided to the University of Aberdeen. The completed questions were entered

on an electronic database and analysed using a statistical software package. Due to the low number of participants only descriptive statistics are reported. A thematic analysis was used for the open-ended questions in the questionnaire (Mason 2002; Forrest Keenan 2005). To illustrate the themes raised, quotes are presented within the text below, “indented from the main text and with single line spacing” (Pitchforth *et al.* 2005: 133).

## **Results**

A total of 61 questionnaires were returned to the university. In total 43 women and 16 men responded whilst two did not write down their gender. Nearly 75% defined themselves as Christians, another 20% stated they had no religion, two were of Muslim or Hindu faiths and three did not answer the question. Of the 24 people who were married, the overwhelming majority (77.2%) had married partners from within their own ethnic group.

### **Knowledge of the Haemoglobin Disorders**

The second section of the questionnaire asked six multiple-choice questions on the members' knowledge of haemoglobin disorders. Table 1 lists the six statements, each with the correct answer for the knowledge questions underlined, and for each the percentage of respondents who answered the statement correctly. The question that was answered correctly by the largest group of respondents was the first question, which was a basic knowledge question. The last two questions, based on genetic inheritance (questions 5 and 6), were answered poorest, with only just over half of respondents giving the correct answer.

Table 1 Replies to knowledge statement on Sickle Cell & Thalassaemia

**1. The sickle cell diseases and thalassaemias are disorders affecting; (Tick one box only)**

|                 |                              |
|-----------------|------------------------------|
| Red Blood Cells | 96.7% answered<br>correctly. |
| Ability to walk |                              |
| The Lungs       |                              |
| Don't know      |                              |

**2. How do people get the sickle cell diseases or the thalassaemias? (Tick one box only)**

|   |                |
|---|----------------|
| They are infectious; they can be caught like coughs and colds | 95.1% correct. |
| They are inherited, passed on to children from their parents  |                |
| Nobody knows the reason                                       |                |
| Don't know  |                |

**3. What is sickle cell trait or ? thalassaemia trait? (Tick one box only)**

|   |                   |
|---|-------------------|
| When someone is a carrier for sickle cell/ ? thalassaemia and could perhaps pass it on their children | 73.8%<br>correct. |
| They are a less severe form of sickle cell anaemia/ ? thalassaemia                                    |                   |
| Where one half of the family has sickle cell anaemia/ ? thalassaemia and one half does not            |                   |
| Don't know  |                   |

**4. How can you tell if you have the sickle cell/? thalassaemia trait? (Tick one box only)**

|                               |                           |
|-------------------------------|---------------------------|
| By having a simple urine test | 96.1% answered correctly. |
| It is impossible to find out  |                           |
| By having a simple blood test |                           |
| Don't know                    |                           |

**5. In cases where one parent carries sickle cell/ ? thalassaemia trait and the other parent does not carry the trait, what are the chances for each pregnancy that the parents will have a child with sickle cell anaemia/ ? thalassaemia? (Tick one box only)**

|  |                |
|--|----------------|
| All the children will have sickle cell anaemia/ ? thalassaemia               | 57.4% correct. |
| None of the children will have sickle cell anaemia/ ? thalassaemia           |                |
| One in 4 (25%) of the children will have sickle cell anaemia/ ? thalassaemia |                |
| ? thalassaemia   |                |
| Don't know   |                |

**6. In cases where both parents carry the sickle trait/? thalassaemia trait, what are the chances for each pregnancy that the parents will have a child with sickle cell anaemia/ ? thalassaemia? (Tick one box only)**

|   |  |               |
|---|--|---------------|
| All of the children will have sickle cell anaemia/?           |  |               |
| thalassaemia  |  |               |
| 1 in 2 (50%) of the children will have sickle cell anaemia/ ? |  |               |
| thalassaemia  |  |               |
| 1 in 4 (25%) of the children will have sickle cell anaemia/ ? |  | 52.5% correct |
| thalassaemia  |  |               |
| Don't know  |  |               |

Figure 1 shows the number of questions that each individual answered correctly. Five respondents failed to answer one or more questions. The assumption was made that the participants did not know the correct answers for these questions. Out of the 61 replies 40.9% (n=25) answered all six questions correctly, and 75.4% (n = 46) answered over 50% of the questions correctly.

**Figure 1: Number of knowledge questions that each participant got correct.**



#### Attitudes towards screening

Of the 43 people who had been screened, 7 % was screened because they were trying for a child, 41.3% because the trait or disorder were found in the family and 51.2% stated other reasons and 18 either had not been screened or did not answer the question.

Of the 39 who would undergo antenatal screening for SCD, seven stated that they would terminate the pregnancy. Interestingly enough, five of these seven quoted having a previous child with Sickle Cell Anaemia as the main reason.

Only 25 people identified any disadvantages of volunteering for screening. The most common reason was problems associated with insurance; 12 people identified this as a problem. For example, one participant stated:

*“If the data was used by insurance and mortgage companies to increase the cost of premiums.”*

Another disadvantage identified was the emotional impact of waiting for the results and the actual receiving of the results. One participant expressed this as follows:

*“Some people could be highly affected emotionally. Where without the information they would cope and live a full uninhibited life not being labelled disabled.”*

The third disadvantage identified was that of stigma and being labeled, as suggested in the quote above and highlighted specifically in the quote below from another respondent:

*“Only certain groups of people can have sickle cell diseases, and speaking from personal experience they can be stigmatised against and can be excluded.”*

### Influence of religion

On the question of antenatal screening one respondent stated:

*“The baby has already been conceived, so why terminate it, leave it to God to take control.”*

All respondents, except one, who had previously said ‘no’ to an antenatal test due to religious reasons, were willing to undertake a neonatal test. Only one respondent was not willing to undertake a neonatal test. They stated,

*“ I will leave it with God to take control.”*

### Universal screening test

One participant who did not know whether universal screening for neonates was a good idea stated:

*“Neonatal screening should be universal. Screening in later life should be available on request and offered in certain circumstances e. g. when considering starting a family.”*

None of the respondents who thought that neonatal screening was a bad idea gave a particular reason for this opinion.

## **Discussion**

### Knowledge

No significant difference in knowledge was found between males and females. Overall the knowledge questions were answered well with 75.4% of respondents answering four or more knowledge question correctly. However, a lack of knowledge in inheritance patterns was identified. These findings are compatible with a study by Dyson (1997), which found that patterns of inheritance and the range of ethnic groups who carry the sickle cell trait were the knowledge questions that were least understood. It is thought that this lack of knowledge is one reason why people do not go for genetic screening. However, lack of understanding of genetic inheritance is not peculiar to this group alone Eboh (2004) in her study of health professionals knowledge of the haemoglobinopathies found this is a poorly understood area

### Attitudes-

Considering the proportion of respondents who stated that they were religious, it was not surprising that their religious beliefs played a role in the answers they gave. However, religious views towards termination appeared to change if the participant had a previous child with a SCD, especially Sickle Cell Anaemia. This is an area that requires further research as many parents of affected children although they see the suffering that Sickle Cell Anaemia causes would not be without their child with this condition.

There was an overall lack of awareness of the screening process, which could be linked to the lack of knowledge in certain areas. Although a majority had been screened, over half of these people had been screened when it was not their main intention, for example in preparation for an operation. This shows a lack of knowledge and awareness on the subject of who can be carriers amongst health professionals (Dyson & Kirkham, 1996).

Many disadvantages to screening were identified, the main being financial and emotional implications. Individuals should be made aware that the results of a predictive test might have a serious impact upon their future chance of getting insurance and (certain types of) employment (Morrison *et al.* 2001; Wilson *et al.* 2004).

Overall the consensus towards universal screening was positive, although in some cases it was clear that the term 'universal screening' was not understood. This highlights one of the disadvantages of postal screening of not being able to clarify unclear ideas. Since the study has been conducted the NHS has introduced universal screening for the SCD in England (Department of Health 2000).

### Behaviour-

Apart from the participant's religious views, the other reason identified that lead people not to undertake antenatal testing was the increase in the chance of a miscarriage. Although many people stated that they would not be prepared to have the antenatal screening test at present. However, a significant number of the survey were interested and would attend genetic counselling about this particular disorder. From this they were prepared to envisage the possibility that they may take up antenatal or neonatal screening, as they would have more knowledge and understanding of the situation.

A large majority of respondents would take up neonatal screening, largely so that the child would receive the best possible treatment.

### Limitations of the study

The sample was small and highly selected. In this exploratory study the respondents were a selective groups of people, namely those members of the UK Sickle Cell Society who responded to our invitation to participate in a questionnaire study. Thus the relative high levels of knowledge are likely to be higher than the general population at risk of SCD. In addition the authors fully appreciate that this group are probably the most over surveyed within the affected population, giving rise to a relatively low response rate.

The overwhelming majority of respondents were women. This was not unexpected as significantly more women than men come forward for genetic counselling and predictive testing; it has been suggested that experiences of genetic risk, particularly in relation to disclosure, may be highly gendered (d'Agincourt-Canning 2001).

### Conclusion

This study aimed to look at people's knowledge, attitudes and behaviour towards genetic screening for haemoglobin disorders. It is clear that there is a lack of knowledge in certain areas of the topic, the most evident being the inheritance patterns of the disorders (Dyson, 1997). There is an overall lack of awareness of the screening process. It is important to increase people's knowledge in these areas, either through counselling or education; the Sickle Cell Society has played a major role to here but there is need for better co-ordinated NHS for these disorders.

The key factors affecting attitudes towards screening are the problems of stigma against ethnic minority groups and religious views (Wyke 2004). Universal screening has been identified as a way to increase awareness and decrease stigma. However, even with a small sample it provided evidence of why (NHS Plan suggests that screening should be offered to all parents. This has been shown to be cost effective in areas with high at risk groups (Griffiths et al, 1989; Cronin et al, 1998) universal screening may not be cost effective. This is of course very much dependent on how the screening programme is run and the kind of pre-screening counselling provisions made available as part of the programme.

The advantages of neonatal screening seem to have been identified, as a large majority would have their babies screened. Counselling and support have been identified as a potential factor in influencing the behaviour of people.

## **Conclusion**

Our study of member of the UK Sickle Cell Society suggests several interesting aspects of screening. Many had found out their sickle cell status not through targeted antenatal screening, but through other routes. For example, some members had found out their sickle cell status when being prepared for an operation, others had been screened after a miscarriage.

Members recognised that they lacked knowledge of the inheritance patterns of the disease and how to best avoid it. It was identified that screening had several negative social connotations: (1) it was associated with abortion;(2) it had financial (insurance) implications; and (3) it was associated with an increased risk of miscarriage. It appeared that religious beliefs played a large role in members' attitudes towards testing. However, those members who had close experience of sickle cell anaemia, for example one of their children was affected, were more likely to go for testing and more likely to consider termination of pregnancy.

## **Recommendations**

We would like to make some recommendations for practice on the basis of this exploratory study. First, counselling has been identified as a key factor affecting behaviour; it should be strongly recommended for all pregnant women in a high-risk group to see a trained counsellor. This will help women to make an informed choice. The way these women are identified in a culturally sensitive manner remains a contentious issue.

Secondly, any education in this area should focus on the importance of being screened in order to



be able to make an informed choice. Education should also be given to health care workers especially those who provide counselling as it is seen as a key factor influencing behaviour. Education could be used to increase awareness of the disorders, and, hopefully, help reduce the stigma attached to the disorders.

Thirdly, there is a need for a more rigorous and larger-scale study to be carried out. The results largely provided information on people suffering from SCD. To provide more conclusive results the sample should be larger and comparisons made with other groups affected by similar genetic disorders, such as, for example, cystic fibrosis. Further research should focus on some of the issues identified in this study. For example, a semi-structured interview should be used as it can explore relevant topics but at the same time can clarify and explain answers. There is a need to conduct more research, focusing on the attitudes and views of women of childbearing age.

Last, but not least, it is also important in terms of both research and practice that the participants or patients are able to discuss issues with someone who understands their cultural beliefs and speaks their language fluently.

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