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# Living with Sickle Cell or Beta Thalassaemia Trait: Implications for Identity and Social Life

SUMMARY OF RESEARCH FINDINGS FOR HEALTH AND SOCIAL CARE PROFESSIONALS

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## INTRODUCTION

Carriers of recessive gene disorders are increasingly a focus of public health interventions, facilitated by the growth and greater social acceptance of new genetic and reproductive technologies. The NHS-coordinated twin antenatal and neonatal screening programme for sickle cell and thalassaemia disorders, formalised in 2002 in England, identifies thousands of 'healthy carriers' (adult and children) each year. Current estimates suggest that there are 240,000 sickle cell carriers (predominantly among people of African and African-Caribbean origins) and 214,000 carriers of thalassaemia disorders (largely of Greek, Turkish, Cypriot, South and South-East Asian, and Arab origins).

While newborn screening helps early therapeutic intervention for babies affected by sickle cell disorders, it also raises significant social, ethical, psychological and medical issues by inadvertently picking up some type of carriers (Laird, Dezateux and Anionwu, 1996; Oliver et al., 2009). (The current heel prick test for newborn babies does not identify thalassaemia disorders) There are currently no consistent guidelines supporting how parents are informed about their baby's carrier status and the level of counselling and support offered to them to address any potential concerns (Lempert 2004). Interestingly, carrier screening of babies/children sits uncomfortably with the cautious approach recommended by the Human Genetics Commission in (2010), since it has no therapeutic benefit and potentially compromises their right to reproductive choices as an adult.

Antenatal screening for sickle cell and thalassaemia variants, in contrast, whilst simultaneously connected to individual choice and preference, can be viewed as representing a more 'preventative' approach. The stated policy aim is:

*... to facilitate informed choices in screening, identify women/couples at risk of pregnancy with sickle cell or thalassaemia disorders and provide appropriate referral and care for prenatal diagnosis, so that women/couples can make informed choices as to whether to continue the pregnancy or request a termination (Harcombe and Armstrong, 2008:582).*

As is clear from similar policy documents, antenatal carrier screening presupposes and supports the notion of reproductive choice. However, paradoxically, its success is closely associated with earlier and/higher uptake of prenatal diagnosis aiming to reduce the incidence of live births of babies affected by these variable, though life-threatening conditions (Greengross et al., 1999: 3; Williams, Alderson and Farsides, 2002); measured in 'incremental cost effectiveness ratios for affected live births prevented' (Zeuner et al, 1999:79). It is hardly surprising that public health policies supporting prenatal diagnosis are bound to be contested in relation to wider social values underpinning disability/impairment (Kerr, Cunningham-Burley and Amos, 1998; Shakespeare, 1999). Equally importantly, antenatal screening brings the intimate sphere of reproduction and family/kinship life of individuals/couples irrespective of their ethnic background (cf. Down's syndrome) under clinical and state surveillance. Carrier screening for haemoglobinopathies, in particular, focuses attention on culture, religion and genetic literacy of 'at risk communities'. Recent policy move towards pre-conceptual screening, for example, assumes that individuals from 'high risk communities' want to know their carrier status and will/should make 'rational' choices about not having children with another carrier.

Historically in the UK, sickle cell and thalassaemia have been associated with homogenised views of minority ethnic groups, often marginalized as a 'minority' health issue. With shifts in family formation and plural, ethnic identities located transnationally, old labels denoting 'ethnic origin' may no longer be a reliable predictor of who might carry a particular trait (Dyson 2005) or the presence of different traits within the same family over a period of time. Equally, the identification of carriers from majority white ethnic backgrounds prompts us to think more critically about the assumed links between ethnicity, race and genetics being straight forward and predictable (Carter and Dyson, 2011).

## OUR RESEARCH

Previous policy oriented and sociological research on sickle cell and thalassaemia disorders has tended to focus on how carrier status impacts on reproductive choices or decisions related to antenatal screening. We know little about how being a 'healthy carrier' impacts on people's ideas about health and illness or, more broadly, their sense of social relationships and identity at different phases of the life-course (Kerr, 2005).

The main aims of our two year research, funded by the Economic and Social Science Research Council (January 2012 to December 2013), were to:

- understand how individuals from diverse ethnic backgrounds make sense of being a sickle cell or thalassaemia carrier at different phases of the life-course;
- analyse whether being identified as a carrier is associated with notions of health and illness for one's self and significant relationships;
- explain how these understandings about being a carrier might/might not be related to one's ethnic identity and shift over time (for individuals and communities);
- explore whether 'being a carrier' influences significant decisions about seeking partners and reproductive choices within extended families in relation to current and potential screening pathways; and
- disseminate findings with potential significance for policy and practice for both professionals well as members of the communities.

### Methods used

The research using qualitative methods was carried out in two overlapping phases. We first conducted four workshops (PW) with professionals (n=26), focusing on their perceptions and experiences of providing support to carriers from diverse ethnic backgrounds. The participants ranged from a paediatric haematologist, haemoglobinopathy specialist nurses and counsellors, midwives, community support workers from voluntary sector organisations, centre managers, a young people's nurse for long term conditions, and outreach and communication officers working across the NHS and the voluntary sectors. Participants also had an opportunity to comment on the new suite of carrier leaflets introduced on request from

the Outreach Office of the National Screening Programme for Sickle Cell and Thalassaemia. This helped further focus our discussion on issues of information and support. We also held four focus group discussions (FFFG) with family members and friends (n= 29) who knew a carrier (but were not a carrier themselves), to understand what they thought about the experiences of carriers within the communities.

The second phase involved in-depth interviews with a theoretical sample of 57 participants (33 women, 24 men) between 17-70 years of age, recorded digitally and translated (n=6)/transcribed. Each participant was interviewed once, and 25 of the 57 interviews involved a range of family relationships (eg parent/ child, husband/wife, uncle/niece, cousins). The age range allowed us to explore the different policy pathways and personal circumstances leading to the identification of being a carrier, covering different phases of the life-course across the dataset. The participants represented five broad ethnic backgrounds African, African-Caribbean; South Asian (Indian, Pakistani); Greek, Cypriot, Turkish and white or plural heritage with complex trans-national family histories. Only six participants chose to speak in a language other than English (Urdu/Hindu/Punjabi), and their interviews were translated into English.

A *thematic guide* was used for conducting the interviews drawn from relevant literature (screening policy, health service research, sociology and anthropology), and the focus group discussions with family, friends and professionals. For the purposes of this summary, we used thematic analysis based on biographical case studies to provide an overview of the similarities and differences within and across families of carriers. Each case study focused on the personal context within which the meaning and implications of carrier status are located, whilst relating it to the wider social and policy context. Moving from the individual to the wider social and structural factors (including the impact of policy or interaction with professionals) avoids the danger of predefining ethnicity as the only/main factor underpinning the experience of carriers.

### Presenting the findings

A summary of the main findings is presented below which, we hope, policy makers and service providers across the NHS and voluntary sectors will find useful. It is not our intention to provide

a critique of services or a defence of ‘cultural understandings’ of genetics rather to translate ideas across different and complex social and experiential frameworks within which people make sense of the ‘diagnosis’ of being a carrier. In doing so, we highlight various tensions which have the potential of undermining the stated aims of the screening policy and practice. This is not to deny the examples of good practice, highlighting the sensitive and appropriate support provided by professionals to carriers and their families, in the face of emotionally challenging situations. However, our concern is to build on previous debates and further encourage reflexive practice, based on a wider understanding of the history, aims and social implications of carrier screening on one hand; and a better understanding of the personal context within which individuals across (majority and minority) ethnic groups interpret carrier ‘diagnosis’ and make moral choices related to genetic risk on the other.

In focusing on the tensions evident in current policy, we want to specifically support a move away from the ‘deficit model’ of science/genetics (and public health), wherein minority ethnic communities are often blamed for a ‘lack’ of knowledge/understanding of genetics and genetic risk. The ‘deficit model’ treats health information within a rationalist model of autonomous behaviour (see Gregory and Miller 2000). The assumption being that more extensive dissemination of information to ‘at risk’ communities can lead to ‘rational’ individual life-choices and, thereby, prevention of births of children with these disorders. Here, ‘lay people’ are perceived as passive recipients of knowledge, while their individual interests are assumed as being similar to those of the community ‘at risk’.

We argue that in promoting public engagement with genetic disorders, with a view to bridging the gap between professional/scientific and lay perspectives, it is important to recognise and address the significant personal, social and ethical issues surrounding carrier screening that are often left unaddressed within policy guidance and literature. A shift from the ‘deficit model’ is crucial for making public engagement more inclusive and equitable by involving different communities and forms of knowledge (policy, lay as well as professional), and broadening the debate on carrier screening before any future policy initiatives and/or any changes to current policy are undertaken.

We now turn to our main findings, presented in two

sections. The first section summarises four main intersecting themes discussed in the professional workshops. These relate to communicating test results and providing support; the conundrum of ‘healthy carriers’; focus on ethnicity and ‘lack of understanding’; and balancing care with screening (prevention). The next section provides a bird’s-eye view of the main themes generated by the in-depth interviews with carriers. These themes focus on: the salience of a trait and engagements with genetic risk; the context of testing and quality of support; challenging stigma within competing sources of knowledge; responses to hypothetical and real scenarios of screening/testing; and what is wrong with the consanguinity argument? We conclude with a few recommendations for policy and practice.

## THE MAIN FINDINGS: PROFESSIONAL WORKSHOPS

### Communicating test results, counselling and support

We begin by connecting the practical implications of disclosing test results with broader philosophical discussions about the meaning of diagnosis. Discussions in our workshops with practitioners suggested that there is no consensus on how the results of new born carrier status are disclosed in practice across geographical sites (Ulph et al, 2014). Parents might be told by letter, in person or on the phone by a receptionist that their baby is a carrier, raising concerns about the quality of such communication and lack of follow up, as reflected below:

*I’m always concerned about the result of ... the trait carrier, because ...even if they are informed about the baby’s result, they’ll be told not to worry because the trait doesn’t cause problem. ... by the time the baby, the child gets to teenage the mum will have forgotten what she was told.... So when the child gets into their teens, she or he doesn’t know whether she has the trait or whatever .... (PW4)*

Contrary to policy guidelines (NHS, 2011:4) and assumptions of most parents in our sample, professionals were not sure whether GPs always receive information about new born carrier results:

*... one of the problems is the IT (...) it would be wonderful if this research would kind of endorse this point ... that the result to be in the child's record with the GP and for there to be some specific intervention that the GP should do to just remind the child, and at the moment that's kind of, that's definitely not (happening) ....(PW4)*

One example of good practice was shared from a London borough where a photocopy of the new born test result is sent to the health visitor, parents and the GP simultaneously. But perhaps not surprisingly, the specialist nurses and counsellors reflected a lack of confidence in non-specialist colleagues and GPs disclosing carrier results to adults and providing required counselling:

*... with all due respect, I mean in general practice (laughs)... I find mostly a lot of GPs don't really understand the differences between a trait and a (condition). And sometimes, they have been telling people they have the condition when they don't, and there are times when they do have the condition and they are telling them that they are carriers.... (PW4).*

The overlap between the two categories - also reflected in the 'family and friend' focus group discussions and in-depth interviews - impinges on a fundamental semantic issue of whether or not a trait constitutes a diagnosis in itself and the implications thereof.

### 'Healthy carriers': a Pandora's box?

The International Classification of Diseases (ICD-10 version 2010) subsumes traits within the diagnostic categories (D56.3 for thalassaemias and D57.3 for sickle cell disorders). However, in practice, heterozygotes (carriers) seem to receive a quasi- diagnosis of having a 'trait' that may not have any implications for their own health. Even though clinicians recognise that Beta thalassaemia carriers might have anaemia due to their red blood cells being slightly smaller, this is generally not acknowledged in the information leaflets endorsed by the NHS. The leaflet for Hb AS mentions that 'in very rare situations' carriers might face possible health issues and provides appropriate advice ([www.sct.screening.nhs](http://www.sct.screening.nhs)). On the whole, the main implication and thrust of the information is on

explaining the risk of passing on the gene/disorder to the children if the partner also is a carrier.

Several professional participants raised the issue of carriers complaining of symptoms, especially those with Hb AS, that might be disregarded by their GPs, raising doubts about the idea of a 'healthy carrier'. The following observation by a voluntary sector professional well summarises the point:

*...they (...) get the results, and then it's, 'You're healthy, go on along your way'. ... we're hearing it a lot more where traits are saying, "You know what, I'm getting the symptoms". ... I'm not a medical person, none of us here (organisation) are ..., so we do say again, 'Go back to your GP'. But I think it's kind of ingrained in the GPs to say, 'Well actually, no, you're a healthy carrier'. That's the definition almost, it's not just a carrier, you are a 'healthy carrier', and if you're getting the aches in your joints, whatever, well you're just getting old, aren't you? I do think we need to do something about it.*

Clinically oriented participants did think that sickle cell carriers, in particular, might have symptoms even though their status did not trigger a crisis. This acknowledgement was often backed by research in the US (given the racialised history of sickle cell and the politics of health insurance, clinical opinion is divided on the potential risk of pain and sudden death among carriers):

*... (laughs)... basically you have all those red blood cells that could potentially sickle as well, but wouldn't warrant a crisis. Because ... if we look on American studies and research we can see that there's loads of research done for carriers, because in America I think a lot of people who are carriers have some form of conditions and very open to say, 'I have this condition, in relation to being a trait' (PW2).*

The acknowledgement that carriers might experience symptoms depending on their haemoglobin type composition came with a caution that the diagnostic label might be appropriated by 'patients' to 'get round the system'. Diagnostic labels generate symbolic meaning and in this context, the semantic association between the trait and the condition creates room for doubt, 'confusion' and alternate explanations of the meaning of 'being a carrier'. At the same time, a

focus on ethnicity, culture and 'community at risk' rather than genetic transmission *per se* generates a critique of the underlying policy intentions within the broader racialised history of health for particular minority ethnic groups (discussed on page 11).

### A focus on ethnicity, 'lack of understanding' and informed choice

Often professionals found it hard to understand why despite counselling and co-ordinated information disseminated within relevant minority ethnic communities, and women and their partners during antenatal period in particular, they were not able to grasp the implications of their carrier status for passing on the trait/condition to their children. Sounding exasperated, one of them commented:

*... we have very good screening coordinators who do a lot of counselling and that's great at the time, but when I follow them up neonatal ... with a (baby) carrier, they're shocked. Now, whether they lose the information somewhere between the ten weeks of screening antenatal and the five or eight days postnatal, I don't know. But they're often really shocked ....(later) ...they've misinterpreted what they were told, forgotten ... or they're so tired postnatally that they hadn't quite taken on board that we were screening for this.... (PW2)*

Others were more forthright about the 'need' for counselling to be more 'targeted':

*...when you give the information it's reinforcing, and sometimes even your tone of voice has to change. You've given them the information but when it comes to saying, "Now are, are you taking this on board?" ... you know, it's all body language isn't it? The look, the direct look, the, you know, and you're actually pointing it out slowly, precisely, clearly, and then when you've finished .... So you're making sure that they're making an informed choice if it comes to the antenatal screening .... (PW2)*

The notion of 'informed choice' often reflected default cultural values of risk and rationality; challenged by perceived 'deviant' cultural practices of minority ethnic communities defined as ignorant and lacking in knowledge and understanding. However, such criticisms (or indeed counter-criticisms) were not confined to professionals of a particular majority or minority background. The

following reference to consanguineous marriages among families of Pakistani origin by a (white) participant provides only one example of a long standing thorny issue that continues to divide public opinion, defining the limits of 'rational' choice:

*... we can have young adults with the condition and we find that the, you know, they're still marrying within the family and they're still getting people over from Pakistan and they've seen, you know, the sibling and all the things that have happened and yet they will still bring somebody in .....*  
(PW2)

However, another (white) participant (from a different site within the same group, PW2) cautioned:

*'It's not really consanguinity. It's the gene. You've got to make sure (to emphasise), it's the gene that you've got to be careful of'.*

This prompted what might seem like the obvious conclusion from another participant:

*'That's why we never ... link cousin marriage to the counselling, because ... people can come up with the impression that, as long as you don't marry your cousin, it's fine, and that's totally not the case' (PW2).*

Finally, some participants reflected more critically on the need for professionals to challenge the taken for granted 'matching' of haemoglobinopathies to particular ethnic groups:

*I had a real horrible case a couple of years ago where there was a girl who was Jamaican Indian ... and she was a beta thal trait, and her partner was Caribbean, he was sickle cell trait. They'd gone to the antenatal clinic early, from about seven weeks ... it was a newly qualified midwife, didn't send a referral through to the sickle cell centre, cos just assumed thalassaemia and sickle didn't cause a problem. By the time I got this referral, the girl was thirty-eight weeks' gestation. After I gave her all the information, she, she just got so upset, she delivered two days later.... And it was just mistake after mistake after mistake. And it was really, really sad because ... you know, between the cultures, 'it needs to be black on black or Asian on Asian'.... ..., I think last year alone, we saw about eight beta thal girls, white, white girls, white British (PW1).*

The uncritical acceptance of this dominant perception of sickle cell and thalassaemia disorders being an attribute of particular ethnic/racial groups as mutually exclusive categories can result in significant emotional disruption and mistrust within relationships. The following example shared by one of the professionals captures well, what sociologists describe as, geneticisation of kinship (Finkler et al, 2003):

*... and the sister was tested and found to have a sickle cell trait. And she said, 'I went to my mother, ..., sat my mother down. I said, 'Could you tell me what happened thirty-four years ago (laughter) that you didn't tell me about?'' And the poor woman said "What are you talking about?" She said "Who's my father?" And she said "I beg your pardon!" She says, "Well I've just been told I've got sickle cell trait and there's only black people who have sickle cell trait, so who's my father?" (PW4)*

Despite a partial recognition of the presence of white carriers reflecting genetic variance within broad ethnic groups, rather than simply a result of 'mixed' ethnic relationships, the dominant health promotion literature continues to portray these conditions as an attribute of minority ethnic groups. Given the long history of racialization of health of minorities in the UK, this often results in reinforcement of stereotypes; whilst creating internal differences along the fault lines of 'genetic responsibility' and responsible citizenship (ie preventing births of children with these conditions). As explained later, this can lead to a culture of community groups perpetuating this dominant view within which having a child with the condition can be stigmatised, raising serious ethical issues about the rights of the disabled within our society and how we make judgements about lives 'worthy of living' (Shakespeare, 1999).

### Balancing care with a focus on screening

Most of the information and informal counselling to carriers was being provided by specialist services and community workers within voluntary sector organisations. Both sectors have suffered significant shortfalls in funding, affecting the networks of support and services within the community, which have often being built up over many years and much struggle. Some professional participants suggested that screening in the UK currently receives more funds, at the expense of

haemoglobinopathy services, which are believed to be patchy.

*... but here we are more screening driven and that in the expense of ... the full blown conditions are quite left behind. Because most ... if we look on the different services, I think the different services are commissioned with the screening rather than care ... and if we have care, they put the element of screening as well. So it, the screening takes more of the service nationally than the care itself (PW1).*

These findings resonate closely with the experiences of carriers identified through different pathways (at times, outside the UK) and having very different experiences in relation to the twin screening policy introduced in 2002. It is to these experiences we now turn.

## THE LONG TERM IMPLICATIONS OF BEING IDENTIFIED AS A CARRIER

### Salience of a trait and engagements with genetic risk

Interviews with carriers highlighted the significance of the biographical context, of which ethnicity is only one dimension, underpinning the meaning and importance of having a trait/being a carrier. A trait (for a recessive gene disorder) can remain *invisible* or *unknown* in a family for generations; and once disclosed, its meaning shifts over time with changes in the life-course of individuals within the context of significant relationships (family and friends). Hence, *biographical risk* unfolds in time and might be at odds with the notion of *genetic risk* being static and predefined for an individual (as represented in the inheritance diagrams used in the information leaflets and genetic counselling). Consider the following example from one of the interviews capturing moments across three time periods (T1, T2 and T3).

Anne (not her real name), a woman of Greek –Cypriot origin in her 50, had always 'known' since she was a teenager that she was a beta thalassaemia carrier. However, she said that it was 'verified and clarified' only when she was pregnant with her first child:



(T1) ... I remember the doctor coming back then (teenage) and saying, 'Everything's fine..., you've got that thalassaemia thing (trait) but that's quite common in Greek people,' and that was it. I paid that much attention to it because he was very dismissive, and I never ever questioned it or researched it or thought anything more but it was always at the back of my mind.

(T2) Anne's husband had been tested as a child in Cyprus and the family assumed that he was not a carrier (though it transpired that the results of siblings had been mixed up, at the time). By the time Anne married, she knew two cousins living in Cyprus, diagnosed with beta thalassaemia major and had heard of stories about an aunt and a cousin losing babies. The significance of the trait now focused on the risk of having a child with the condition, since thalassaemia seemed pervasive in different sides of the family and 'rife' within the community:

...she (sister) had the trait, her husband has the trait, my husband's sister has the trait, my husband's sister's husband has the trait. So they'd both had ... (to make difficult decisions surrounding prenatal diagnosis (PND)). ... I was mature by then. I was in my twenties (but) had no concern because I knew from (in-laws)... that my husband wasn't a carrier. But I went to hospital, received a phone call and they said to me, 'You've been found to carry... beta-thalassaemia trait, and you need to send your husband in for a blood test' .... I think, I was about four months (pregnant) by then, and they rang me and I... fell to pieces obviously and they said, 'You're both carriers ....'

(T3) Knowing a close friend whose child had beta thalassaemia, Anne felt 'lucky' that neither of her children were affected by the condition. Her concern for her son as a carrier only surfaced once he grew up and started dating. Yet, she would not expect him ever to start a relationship by asking his girlfriend, 'Do you carry thal?' With arranged marriages (families introducing the couple to each other) declining as a social practice, she did not imagine a couple 'standing at a bar and asking each other whether they were carriers'. Besides, raising the issue, in itself, assumes that both are committed to a long term relationship with an intention to have children. Hence, people leave it

until later. As she put it, young love is about, '... you hold hands, skip down the road and face it together' .... I don't know if it's enough to break people up, it certainly didn't occur to me to .... even if I'd found out he was a carrier beforehand I would have stayed with him and thought, 'Right, we're going to get through this together'.

Hence, what might seem like an 'irrational' choice or a 'risk minimising' strategy from a clinical perspective, especially where both partners know each other's carrier status, often signals a test of a relationship and moral credibility of self. It is, therefore, important to understand genetic risk within the social domain of significant relationships. Such *risk minimising* rather than *risk averting* strategies are not confined to people from less educated, traditional, religious or minority ethnic backgrounds, as we know from literature on other genetic disorders such as cystic fibrosis. Different families or members of the same extended family might choose either of these strategies. For example, Jacob, a man of African-Caribbean heritage in his 40s, perceived his relationship with a white woman as averting the risk of having a child with sickle cell anaemia, since, to him, 'Caucasian' and 'English people don't get it'. In contrast, his brother, also a carrier, said that he would never date a white woman since he could not identify with them. Jacob had always known that he was a carrier and had lost two close relatives to sickle cell anaemia. He said:

... that's also part of your build-up as you're growing ..., because I did know ... the chances of me having children, full-blown sickle cell by (...) one of my own origin ... I didn't want that.... I don't think it'd been fair to bring a child into the world knowing its chances of reaching twenty-one are slim.

An acceptance of such an essentialised view of sickle cell disorder being an attribute of African/ black 'origin' or heritage can hardly be attributed to the realm of 'folk' or lay knowledge ('misinformation'). Rather it reflects a long history of sickle cell being the example of a racialised disease within medicine and policies related to public health in the UK and the US (Duster, 2003). At the same time, a positive affirmation of sickle cell being part of a black identity has long been part of the struggle for seeking citizenship rights in the UK (Nelson 2011; Anionwu and Atkin, 2001),

the US (Wailoo, 2001) as well as parts of Africa (Fulwille, 2011). Stories of the infamous Tuskegee Syphilis trials (conducted by the US Public Health Department between 1932- 1972) morphed into transnational rumours about sickle cell anaemia being a product of a post-colonial conspiracy to wipe out the African race. According to John;

*'... well my theory is..., well back in the like World War I's and II's there was a lot of genetic testing and germ warfare.... ..so I, me personally, I think it was manufactured to kill off a race or kill off a nation or whatever and then it got out of hand, you know what I mean?'*

Four participants narrated different versions of this story. Even though such rumours may not be widespread, they manage to generate mistrust in the system and might serve as post facto explanations for being treated differently by healthcare professionals.

Further, higher incidence of mixed ethnic marriages/relationships (one in ten according to a recent analysis) and serial monogamy pose a significant challenge to the bureaucratic ways of identifying 'family origin'. A greater proportion of individuals will have more than one partner over their life-course; and children/step children in a family might inherit different traits. Further, this results in discontinuities in family health histories and (genetic) narratives of inheritance, in contrast with the fixed notions of ethnicity and of bilateral inheritance (from fathers and mother's sides) assumed within clinical genetics and the inheritance diagrams.

### The context and pathways of testing and finding the 'wrong trait'

The level of information and support provided to carriers varied significantly with how and when a person was identified. Women and men who were identified through the antenatal screening pathway generally received more information (face-to-face and written). Yet, contrary to policy guidelines, only couples identified as potentially 'at risk' or who, in fact, had a baby with a condition received formal counselling and support. Needless to mention, men whose partners were not carriers 'fell through' the net of antenatal carrier testing pathway, unless their carrier status was already known or disclosed following the birth of a child who was a sickle cell

carrier. Women who were identified as having a Hb variant but whose partners were not carriers were often left to seek further information on their own, especially where the trait did not match their perceived ethnic/racial profile. For example, Nora, a woman of Black-British heritage, in her 30s, was sent for sickle cell screening along with her partner when she was pregnant (about eight years ago). She recalled:

*... to get tested and he came back clear and I... and I came back with thalassaemia trait. From there I went to the doctor and ... the doctors didn't really know much more, to be fair. I think it was me, I'd gone on, online and found out what it was and thought it was more to the Indian side of my (father's side) family where it came from and...*

Nora felt that once they found that she and her husband did not have a sickle cell trait, there was no further discussion:

*It was just kind of ... it wasn't as important as the sickle cell. 'The main issue here, what you've been tested for is sickle cell, because if you're both a carrier then these are the implications. But you haven't got that, you're fine'. You've got thalassaemia trait .... I mean they didn't say, 'It's all right', but it ... wasn't made an issue of.*

Returning to the issue later, she complained:

*But again, when you find out if you've been diagnosed with a condition, so to speak, I think you should be fully informed. ... I mean the whole point was to test that the baby was OK, but at the same time I'm, I'm the mother so a bit more information should be given to me... how's it going to affect me? But I think being pregnant at the time, my main concern was that the baby's fine, so I didn't think much more of it. But thinking back on it now ... if anyone's told, you've got any type of condition, you should be sat down and maybe counselled slightly just to say how it could possibly affect yah.*

Nobody had explained the possibility that her child might also be a carrier. Two women of Indian origin, who were thalassaemia carriers and whose proficiency in English was limited, thought that they had beta thalassaemia major (condition). One of them, Tejinder, who was in her 20s and had moved

to the UK following her marriage, was a mother of two small children under five years of age. She said that even though she was healthy apart from being anaemic and breathless at times, she worried about the impact thalassaemia might have on her health in future. She was particularly concerned about potential health implications for her children since she had breast fed both and wanted to find out whether they needed any special diet to keep them healthy. Having been identified during antenatal care, she had received a haemoglobinopathy card specifying beta thalassaemia trait. She had also been given an information leaflet in Punjabi. However, she had no experiential knowledge about thalassaemia and, since her husband was not a carrier, there was no further discussion on the issue with the antenatal nurse or others involved in her care. Even though her GP spoke Urdu/Punjabi, she had not raised these concerns with him, reconciling to the fact that she had an 'illness'. Rather than dismiss it as a 'cultural' issue or a result of her 'lack of English' and 'lack of understanding' about the condition (the deficit model), Tejinder's experience reiterates the need to focus on the language of genetics and to tailor the information to the circumstances of an individual/couple. Receiving information, face to face or in the form of a leaflet/ other resources, is a process rather than a discrete, one off activity or event that happens at a point of time. Hence, it is important to enable the person to relate the technical information within the context of their own life, and to signpost carriers to the right professionals in case they have doubts or any concerns in future.

Men and women, especially those who were older and found out their carrier status inadvertently, outside the antenatal and new-born screening pathways, had received minimal information with little support to address any potential anxieties related to health. Some of them were not sure why they had been given a haemoglobinopathy card and whether they could donate blood (since they had a blood disorder). For example, one of the older participants Mary, of Ghanaian origin and in her 60s, had found out that she had a beta thalassaemia trait when she was in her 50s, during a hospitalisation. Nobody had bothered explaining what that meant. She had known a relative and a close neighbour affected by sickle cell anaemia and, not knowing anything about thalassaemia, having a 'blood disorder' conjured images of a painful illness and a fear of death. Her GP dismissed her fears, leading her to find out more through her network of friends

at church and a voluntary sector organisation. Interestingly, rather than worry about the carrier status or health of her children, her concerns focused on the health of her grandchildren. They seemed healthy but since they did not know the 'symptoms' of thalassaemia, she thought, they might miss it and the condition might resurface in the future generations. Given that she herself did not know where she got it from, the idea of genes 'skipping a generation' fitted well within her own narrative, as it did for those who tried making sense of the elusive category of a trait and an inherited blood disorder.

A trait recedes into the background or might be treated as an insignificant detail of one's health history where the individual or couple have moved away from the phase of childbearing either because of their age or by choice. James, who was in his 50s (Jamaican family and professional background), already had a daughter from a previous relationship, as did his second wife. Both had decided against having any more children. Being a carrier had never been an issue in his life and he did not remember having concerns about his daughter being a carrier or having been tested. Instead, he used it as a positive sign of his identity (distinguishing him from the white majority) in following terms:

*I know it sounds silly but it, it's, it's one of these things you sort of live with. I never really consider that, 'Oh! Sickle cell ... is a problem so I need to be considering this and I need to be considering that. I see sickle cell ... a positive note, as a natural defence for malaria ....*

Interestingly, the only white participant (in her 50s), Jackie, had been diagnosed with beta thalassaemia trait in her 40s, when she already had teenaged children. She had a lingering concern about having passed it on to them (in case they ever got together with a man from an 'ethnic' background who happened to be a carrier). Being in the health profession herself, she felt that they should make their own choice about carrier testing as an adult. One of them decided to have a test while the other did not want to know.

Where a child was known to have a blood disorder in a family other children/siblings were more likely to have been tested. This was not the same in the case of a parent/s or a child being a carrier (where no close family member was affected by

the condition). We did not include any parents/ grandparents who themselves had the condition; except for one man of Nigerian origin in his 50s, Osman. He had been diagnosed as a carrier (AC) in Nigeria, in his 30s, and had suffered from persistent painful crises as a child. Following his move to the UK, he survived serious health crises events including a diagnosis of sarcoidosis and pulmonary embolism, and was eventually re-diagnosed as having a complex haemoglobinopathy based on a DNA analysis. He had three children from two previous relationships, and each had been tested. Osman felt a responsibility to share his knowledge about sickle cell with his extended family in Nigeria and set up a local support group for families affected by the condition in his town in England. He believed that the diagnosis saved his life since he was able to understand and manage his illness better with treatment. It is hardly surprising that he was more sympathetic towards carriers who complain of having painful crises and believe that they might develop symptoms as they get older. While he differentiated between being a carrier and having the condition in terms of severity, for him, these belonged on a continuum rather than as types or an absence/presence of a disorder.

Carriers who experience significant symptoms, especially those who have Hb AS and complain of feeling very cold, achy and easily tired believed that carriers can have ‘mini crises’ in extreme weather conditions or under social and emotional stress – challenging the notion of a ‘healthy carrier’ as being misleading. Such responses can suggest potential confusion over the idea of a ‘carrier diagnosis’, as identified by some practitioners. In an extreme case, Linda, a woman of African-Caribbean origin in her late 50s, strongly felt that all this ‘misinformation out there that sickle cell trait is not significant and does not cause any health issues’ needs to be addressed. She had been complaining to her GP about headaches, muscular pains, memory and concentration issues and the GP dismissed her symptoms as being psychosomatic. Linda said:

*In my experience, my Ex GP Dr. .... tried to relay them as being psychosomatic. I believe that he thought that I was imagining the pain or that it was all in my head. I could not understand how I could have such power and why, of all the wonderful things to imagine, I would imagine a pain.*

Linda (like others in her situation), acknowledged

the role of extreme weather, dehydration, stress as well as difficult personal circumstances as contributing to her symptoms. Her perseverance led the GP to refer her for an MRI scan that did not show any lesions and she was told that she had cluster headaches and myalgia. She then searched for information online and found ‘scientific papers’ (journal articles) suggesting that sickle cell carriers can experience muscular aches or a sickling crisis if they are dehydrated; could have headaches, silent strokes and memory issues. She set up a blog and initiated an online support group with membership across UK and the US, where carriers share their experiences and concerns. The important issue here is not whether Linda’s symptoms were clinically ‘real’ or imaginary. Rather, a lack of recognition of her symptoms led her to seek information and support online, diminishing her faith in professional expertise and support within the NHS. This is not to deny that others, especially parents and siblings, who knew someone close with the condition, disagreed with their clinician’s suggestion that their symptoms might be caused by their carrier status.

### Challenging stigma within competing and contested sources of knowledge

Literature suggested that carriers of sickle cell disorders might face discrimination in seeking employment or insurance due to controversial evidence surrounding the likelihood of sudden death under extreme circumstances. However, this seems to reflect the specific economic and political history of sickle cell anaemia in the US (see Franklin and Atkin, 1986). Discrimination against employees, servicemen and sportspersons who were carriers resulted in the 2008 Genetic Information Non-discrimination Act, prohibiting employers from discriminating on the basis of genetic information, and against mandatory genetic testing.

None of the participants in our sample reported facing discrimination on the basis of their carrier status. This largely reflected the fact that they did not consider this information to be relevant for inclusion in forms for employment, insurance or mortgage, unless a form specifically asked whether the individual suffered from ‘any blood disorders’. In one family, a young person who was a beta thalassaemia carrier had had to disclose her carrier status but had a positive outcome for her application to join the Forces. While none reported discrimination, two participants shared some concerns. One was a mother who had a child

with sickle cell anaemia and thought disclosing her carrier status might push up her insurance for air travel. The other, Anina, a woman of Gujarati-Indian origin in her 20s, felt that it was unfair that her fertility treatment had been withheld until her husband had also been tested. She observed:

*Yeah they explained about the one in four chance of the actual thalassaemia rather than the trait, yes so I think we understood that. But then since then, I wanted to go on... my fertility treatment and because of both of us having thalassaemia (trait), they said there was a possibility that we wouldn't get any Clomid and then they referred us to a geneticist.*

*(Why did they say that?)*

*Well I think my GP said she had never come across a couple who both had thalassaemia trait and she wasn't sure that we could (have fertility treatment)..., I think it was a mix up. I'm not very happy with it... but that is another story.*

The British Committee for Standards in Haematology recommends screening women/ couples being considered for infertility and those having assisted conception (NHS Screening Programmes, *Information for healthcare professionals: 9*). However, in the above case, the GP had not explained why they had to wait for treatment until her husband was tested. Further, she and a friend (another participant) were given the impression that if both partners were carriers, they had to have pre-implantation genetic diagnosis of embryos alongside IVF. Contrarily, some of the professionals suggested that the private fertility clinics may not follow the same guidelines, reiterating the need for national guidelines on the rationale and ethics of carrier testing/not testing within the context of fertility treatment.

Even though participants often differentiated between having the condition and being a carrier/ having a trait, a conflation between the two terms might lead to culturally specific stigma associated with blood/genetic disorders, especially for those who have no experiential knowledge of these conditions. Hence, the terms 'thalassaemia major' for the condition and 'minor' for a trait can be confusing for those who are not familiar with the condition. The historically and culturally specific association of SCD with 'full blown' and 'blood disorder' (terms commonly used by professionals

and community members) leads to images of HIV/ AIDS and sexually transmitted diseases, resulting in an extension of stigma for some younger men of African origin.

Given its potential discontinuities in family histories and the invisibility of the trait, despite the high carrier incidence in the minority ethnic communities represented in our sample and stories about stigma (narrated by professionals and corroborated by family members and friends) across broader ethnic groups, a majority of participants had little experiential knowledge about the condition. Equally, this marks a dissonance between the links between collective/community history, knowledge about the condition and the family/individual histories. This seemed like a surprising finding that partly reflects our sampling strategy and might be helpful in reorienting the emphasis away from simplified, bureaucratic ideas of 'community genetics' and ethnicity currently informing policy and practice (for a review and ethnographic case studies see Raz, 2010; Shaw, 2011). As pointed out by Alex, a young professional in his 20s from a Greek-Cypriot background, '... if you target a community, it puts a stamp on it to say there is something wrong with ... this community... and officially it is so serious, we need to ... root them out .... I think, ... where stigma could come from'. Whilst at the Greek school that he attended, and within his own extended family, nobody mentioned thalassaemia in derogatory terms, he had noticed that in the wider community parents who had a child with thalassaemia were blamed.

Anne, given her wider experience, was more critical of the ethical issues underpinning such an attitude within her community. She remarked (with reference to the Greek screening policy):

*I think what's happened over the years is ..., hardly any babies are born now in the Greek community because of the CVS.... And, and I have heard it said that those who do carry thalassaemia feel almost like they're the forgotten cause. Because, because it's an illness that people are trying to eradicate it's almost, horrible as it is to say, it's almost as if, 'Oh, once we get rid of this batch we can actually eradicate this....'*

*... that's how people that carry thalassaemia feel, that I've spoken to..., if we've got a thousand, which there isn't, but if we've got a thousand, once they're gone there won't be a thousand to replace them because we've used the CVS and we're slowly eradicating.... ..., now my friend (has a child with thalassaemia) has often said to me, you know, 'They don't care about us... ... you really have to push (for support), you have to really insist, yeah'.*

This is a poignant reminder that we need a wider ethical debate on the role of genetic technologies (antenatal/prenatal/preimplantation/preconceptual) and collective attitudes to disability and difference underpinning state policies supporting an expanding continuum of 'reproductive choice'. At the same time routinisation of technology leads to shifts in attitudes and expectations and any change in current policy must follow on from a proper public consultation on improving and widening access to services. This demands a critical engagement with public response as an outcome of these consultations which, as suggested by the scenarios below, cannot be taken at its face value for justifying 'giving them what they want', particularly since current screening policies are often defended on the basis of public demand and the offering of choice.

### Scenarios of screening/testing

Out of the various real and hypothetical scenarios of carrier testing presented to the participants, there was a surprisingly overwhelming support for offering a test to teenagers/young adults before leaving school/sixth form. Those who endorsed mandatory screening in schools (wrongly) subsumed it within the category of vaccinations that they thought children 'had to have' (without parental consent). Others believed that early testing (conflating carrier status with the condition) must be good since it would improve treatment. However, as suggested by their own experiences, and reflections on the impact of such a policy on actions/reproductive decisions, they did not think knowing their carrier status would impact on who they might have a relationship/children with.

We knew only one couple who had separated under family pressure but eventually got back together. We can extend the logic to pre-conceptual testing and why it may still not lead to the expected

outcomes. This option also raises ethical concerns about our attitudes to 'imperfect' births and the potential stigma being generated against carriers within a new form of *genetic exogamy*, the rule of exclusion in seeking partners who might be genetically 'affected'. In communities with a longstanding tradition of endogamy, carrier status can have serious repercussions for the moral identities of individuals, especially women, as 'damaged goods' and the social standing of families who will not be able to fulfil the obligation of reciprocity of which consanguineous marriages is only a part (Rozario, 2013; Shaw, 2011). The final section below summarises the internal contestation and a critique of the focus on consanguinity by professionals within practice.

### What is wrong with the consanguinity argument?

Most importantly, a focus on consanguinity locates risk within 'relatedness' as marrying within cousins rather than the potential carrier status of a partner. This is a long standing problem (Atkin and Ahmad, 1997). Such a notion of relatedness is complex where the structural principle of both parallel and cross cousin marriages results in more distant 'cousins' related on either side marrying. The pragmatic response of 'not marrying a cousin' does not exclude the possibility of marrying a carrier and having a child with thalassaemia, thus resulting in mistrust for the biomedical explanation. Second, it is not validated by an experiential framework where parallel and cross-cousin marriages might have been followed within an extended kin group for generations without any visible signs of an inherited blood disorder. Hence, it does not explain why other couples of the same generation who are cousins have perfectly healthy children and other ethnic groups who do not follow this tradition, such as Greek, Cypriot Turkish or Gujarati-Hindus or whites still have children with thalassaemia. Third, genetic risk implies uncertainty and the doctors cannot be certain about the risk for each couple in each pregnancy. This creates a space for alternate explanations, risk minimising strategies alongside a critique of the biomedical framework on genetic risk. At the same time, such a critique of medical framework does not exclude the possibility of self-reflection and change.

Most couples of Pakistani origin had experienced the (potential) birth of a child with thalassaemia within a broader critique of professional attitudes towards

consanguinity that made them feel responsible for their child's condition rather than simply an assault on their cultural heritage. While doubting the logic of biomedical explanation that consanguinity *caused* their child's illness for reasons explained above, their own experience of caring had prompted a move for change within the extended family. There was greater sharing of information in close extended families affected by the condition, prompted by prospective marriage arrangement of younger siblings, often transnationally. This shift in cultural practice appears in conjunction with a scathing critique of professionals who openly blame the birth of a child with thalassaemia in families of Pakistani origin to consanguinity. The logical message for professionals from Sakina, and other parents caring for a child with thalassaemia, was to tell everybody, 'No matter who you marry, get tested'. She was particularly upset since her parents had gone out of their way to listen to their GP's advice and married her to someone who was neither a cousin nor Pakistani.

## IMPLICATIONS FOR POLICY AND PRACTICE AND RECOMMENDATIONS

Our summary captured some of the complexity of biographical contexts within which individuals are identified as carriers and negotiate notions of risk. As we have seen, ethnicity was frequently evoked but was not the only consideration when explaining how people made sense of their carrier status. We now outline the broad implications of our findings for policy and practice alongside specific recommendations. In doing so, we specifically challenge the ideas which associate health information with a rationalist model of autonomous behaviour; and equate 'at risk' individuals with 'at risk communities'. We argue that policy needs to move away from its current emphasis on instrumental rationality underpinning screening and ideas of reproductive choice, to a broader focus on the significant personal, social and ethical issues generated by identifying carriers of a recessive gene disorders.

### Implications for policy and practice

- A trait can remain invisible in families for generations. Hence, despite the higher incidence of sickle cell disorders or thalassaemias in particular ethnic groups, individuals/families might have little or no experiential knowledge

about the condition. Nor can assumptions about community awareness, in which knowledge about sickle cell and thalassaemia is seen as taken-for granted as collective resource, be guaranteed.

- Individuals across ethnic groups experience personal (biographical) genetic risk as unfolding and changing over time, within the context of significant relationships, experiential knowledge, professional communication as well as historical and cultural factors. This contrasts with the static notion of genetic risk as given and unchanging over the course of an individual's life (as represented in the genetic diagrams explaining inheritance).
- Despite policy guidelines, communication of carrier results and the level of information and support are variable, depending largely on the pathway through which carriers are identified as well as the geographical location of the service. This leads to inconsistencies in the experiences of individuals within and across families.
- Provision of appropriate counselling remains largely confined to antenatal care for women/couples identified as being 'at risk' for having a baby with a haemoglobinopathy. This leaves many carriers to fend for information on their own, with no support.
- Much one-to-one information and support for carriers who are anxious about different aspects of carrier status is provided by the local voluntary sector organisations (working closely with the statutory sector/ NHS). The role of voluntary and community organisations in providing information and personalised support better suited to the individual needs, beyond the remit of 'reproductive choice', cannot be overstated. Third sector organisations have an excellent track record of listening to and engaging with local communities. Their community workers are easily accessible, well known and trusted in the communities they serve. However, this support is currently threatened by significant reductions in funds available to them. This is despite NHS changes highlighting the value of involving voluntary organisations in Clinical Commissioning Groups as well as Specialist Commissioning.
- Professionals as well as community members reflect a lack of confidence in General Practitioners in explaining the implications

of carrier results and addressing particular concerns of carriers about potential symptoms and prospective health issues. This is a long standing problem.

- The idea of a ‘healthy carrier’ is a caricature that fails to take into account the potential emotional, physical and social impact of a ‘diagnosis’ and the likelihood that some carriers might experience symptoms. Equally, it fails to recognise the anxiety some parents might face whose baby/child has been identified as a carrier.
- Professional perceptions of sickle cell disorders and thalassaemia being an attribute of minority ethnic groups/culture (rather than a genetic condition per se) can reinforce stigma for carriers and undermine their confidence in biomedical explanations as well as the credibility of healthcare professionals.
- A policy focus on ‘at risk communities’ (rather than individuals) is similarly unhelpful and misleading. While aiming at challenging stigma and promoting choice, it can inadvertently strengthen misperceptions and stereotypes such as the association of sickle cell anaemia with AIDS in some communities of African origin; or the attribution of thalassaemia to consanguinity among Muslims of South Asian origin (thereby reducing genetics to culture).
- The use of language and terms by healthcare professionals, such as ‘full blown’ for sickle cell anaemia and ‘major’/‘minor’ for beta thalassaemia and trait respectively, only seeks to perpetuate a conflation between trait and the condition and the specific health implications of being a carrier.
- With around 1 in 10 couples in England living in mixed ethnic relationships and an increasing proportion likely to marry/ have children with multiple partners over a life-course, parents and (half) siblings within the same family can inherit different traits or complex haemoglobin variants. Identifying ethnicity/ family origin as a marker of ‘at risk’ individuals in practice will increasingly be less reliable.

## Recommendations for policymakers

1. Given the pivotal role of local voluntary sector organisations in providing information and support to carriers and families with carrier children and/or siblings with a disorder, we

recommend ring-fencing of relevant funds by local Commissioning Groups to ensure continued support for carriers in the long term. Current shortfalls in funding are undermining the networks of support and services within the community, which have often been built up over many years of long struggle. Once they are gone, they will be difficult to re-invent and a valuable resource central to care in the community will be lost.

- 1.1 There is an urgent need for revising and implementing current policy guidelines regarding carrier screening and testing across various pathways of care (both private and public sectors), with a view to improving access to information and counselling. In addition, documentation of test results in the health record of individuals in primary care needs to be standardised.
- 1.2 The national haemoglobinopathy cards are a useful *aide de memoir* for individuals and families in constructing their genetic histories over the long term. We recommend their use be standardised across adult and paediatric services, with the type of haemoglobinopathy variant clearly mentioned, as advised by the British Society for Haematology (Ryan, Bain, Worthington et al., 2010: 47).
- 1.3 With demographic shifts in mixed heritage relationships and greater national and international geographical mobility, the Family Origin Questionnaire may not necessarily be an appropriate indicator of the risk profile of individuals. Local Commissioning Groups need to budget for appropriate training of professionals enabling them to understand and appreciate the relevance of ethnic origin, without relying on visible difference (skin colour) as a marker of genetic risk.
- 1.4 We recommend a wider recognition in policy guidelines of the complex emotional, social and ethical dilemmas thrown up by identifying recessive gene disorders. Such recognition should also inform the training and practice of professionals. Carrying a trait, for example, has implications beyond reproductive choices and can assume meaning at different points of time across



the life course. While this is acknowledged in some policy documents, it is not reflected in the information leaflets or support provided to a majority of carriers.

- 1.5 Given the twin screening policy, it is a State responsibility to conduct and/or synthesise evidence from clinical trials on potential symptoms or health implications for 'healthy carriers'. Sociological research prompts us to recognise that a diagnosis in itself might result in subjective experience of symptoms and significant health concerns (for self or close relatives including children and grandchildren) over time. This needs to be recognised and addressed by professionals, with appropriate signposting for further information and support.
2. We recommend wider and more inclusive community engagement and debate on the benefits as well as the social and ethical dilemmas posed by identifying infant carriers as an unintended consequence of the newborn screening policy. This is in tune with the advice of the British Society for Human Genetics (2010), and (previously) Human Genetics Commission (2006). Apart from the complexity of informed consent and parental rights, this involves reproductive rights and the right 'not to know' for the child as a future adult. Any change in policy must follow a proper consultation with a cross section of stakeholders across communities, without assuming an easy fit between individual and community interests.
  - 2.1 We recommend clear policy guidelines for recording and following up the carrier status, once the cohort of babies/children identified as carriers through newborn screening reach the age of maturity. While most parents accept that passing this information on to their children is a parental responsibility to be shared with their GP, there is little confidence in how well these records are being maintained or updated. Current consent procedures might also need to change, as neo-natal screening is presented to parents as identifying those with the condition rather than carriers of the trait.
3. Professionals and counsellors need to identify alternate frameworks to 'the deficit model' when addressing lay/professional differences

in explanations and understandings of genetic risk. It is important to recognise that carriers (like patients) are not passive recipients of information on genetic risk, and will often seek plural sources to validate their own experience and interpretation depending on their life situation. Acknowledging this can benefit practitioners in addressing the individual context within which the implications of carrier status, genetic risk and reproductive choice are understood and negotiated.

- 3.1 We strongly recommend changes to the current information leaflets available to carriers on potential health implications of being a carrier. The thrust of the current information leaflets endorsed by the NHS Screening Programme is on the importance of being tested with a view to avoiding/preventing a potential birth of a child with the condition. The idea of a 'healthy carrier' is, at best, an unhelpful caricature that fails to acknowledge the range of other concerns carriers might have/develop over the course of time.
- 3.2 The booklet 'Screening Tests for you and your baby', in particular, provides a sketchy and one-sided overview of why screening for sickle cell and thalassaemia is being offered to mothers and fathers (giving the impression that the test in itself is important for the baby's health: p.8). There are no positive messages about the level of support and care available for children with these variable conditions. Importantly, there is no acknowledgement of the emotional and ethical issues raised by the process of confronting a decision to have a PND which, as we know, can be emotionally distressing for both women and their partners. Clearly, there is a need to balance an emphasis on screening/testing with positive messages of supportive care for potential babies born with these conditions, as outlined in the section on Down's syndrome in the same booklet.
4. Whilst there might be support, at a hypothetical level, for pre-conceptual testing within communities (at birth, in schools or as adults), there is no simple co-relation between knowing one's carrier status and seeking a non-carrier partner. Indeed, these responses are often

sensitive to personal and social context. Hence, any policy support for pre-conceptual testing must be informed by wider public consultation ensuring choice - rather than essentially handing out screening in the guise of testing.

4.1 We recommend wider public consultation across majority/minority ethnic groups on the complex social and ethical issues underpinning carrier screening for recessive gene disorders, especially within the context of the international convention on the rights of disabled people. The model followed by the Nuffield Council on Bioethics for open consultancy/deliberative workshops (eg, *Children and clinical research* and *Donor conception: ethical aspects of information sharing*) is a potentially good one to follow. Widening such consultation across and beyond minority ethnic groups will help unpick policy responses that have so far been located in a racialized history of haemoglobinopathies and politics of marginalisation of the health of minority ethnic communities.

## REFERENCES

- Anionwu, E.N. and Atkin, K. (2001) *The Politics Of Sickle Cell And Thalassaemia* Buckingham: Open University Press.
- Atkin, K., Ahmad, W.I.U. (1997) Genetic screening and haemoglobinopathies: ethics, politics and practice, *Social Science and Medicine*, 46 (3), 445-58
- British Society of Human Genetics (2010), *Report on the Genetic testing of Children*, Birmingham, <http://www.bshg.org.uk>.
- Carter, B. and Dyson, S.M. (2011) Territory, ancestry and descent: the politics of sickle cell disease. *Sociology* 45(6): 963-976.
- Duster, T. (2003) *Backdoor to Eugenics*. Second Edition. New York/London: Routledge.
- Dyson, S.M. (2005) *Ethnicity and Screening for Sickle Cell/Thalassaemia* Oxford: Churchill Livingstone.
- Finkler K, Skrzynia C, and Evans, JP (2003) The new genetics and its consequences for family, kinship, medicine and medical genetics. *Social Science and Medicine* 57(3): 403-12.
- Franklin, I.M, Atkin, K. (1986) Employment of persons with sickle-cell disease and sickle-cell trait, *Journal of Social Occupational Medicine* 36: 76-9.
- Fulwilley, D. (2011) *The Enculturated Gene: Sickle Cell Health Politics and Biological Difference in West Africa* Princeton, NJ: Princeton University Press.
- Greengross, P, Hickman, M, Gill, M, Dugan, B and Davies, S. (1999) 'Outcomes of universal antenatal screening for haemoglobinopathies', *Journal of Medical Screening*, 6: 3-10.
- Gregory, J and Miller, S. (2000) *Science in Public: communication, culture and credibility*, London: Pegasus.
- Harcombe, J. and Armstrong, V. (2008) Antenatal Screening, The UK NHS Antenatal Screening Programmes: Policy and Practice, *InnovAiT*, Vol. 1 (8): 579-588.
- Human Genetics Commission, 2006. *Making Babies: Reproductive decisions and reproductive technologies*, [www.hgc.gov.uk](http://www.hgc.gov.uk).
- Kai, J, Ulph, F, Cullinan, T and Qureshi, N. (2009) *Communication of carrier status information following universal newborn screening for sickle cell disorders and cystic fibrosis: qualitative study of experience and practice*, *Health Technology Assessment* Vol. 13: No. 57.
- Kerr, A. (2005) 'Understanding genetic disease in a social-historical context: a case-study of cystic fibrosis', *Sociology of Health and Illness*, 27(7), 873-896.
- Kerr, A, Cunningham-Burley, S and Amos, S. (1998) Eugenics and the new genetics in Britain: examining contemporary professionals' accounts, *Science Technology and Human values*, 23, 2: 175-98.
- Laird, L, Dezateux, C and Anionwu, E. (1996) Fortnightly review: neonatal screening for sickle cell disorders: what about the carrier infants? *BMJ* 313 (7054): 407-411.
- Lempert, T, Oliver, S, Stewart, R and Dezateux, C. (2004) Communication of carrier results following newborn blood spot screening: parents' and health professionals' experiences. Social Science Research Unit, Institute of Education: University of London, Skirton.
- Nelson, A. (2011) *Body and Soul: The Black Panther Party and the Fight against Medical Discrimination*. Minneapolis, MN: University of Minnesota Press.
- NHS Sickle Cell and Thalassaemia Screening Programme Standards. (Oct 2011) available online at <http://sct.screening.nhs.uk/publications>
- Oliver, S, Dezateux, C, Kavanagh, J, Lempert, T and Stewart, R. (2009) *Disclosing to parents newborn carrier status identified by routine blood spot screening (Review)*. The Cochrane Collaboration, <http://www.thecochranelibrary.com>.
- Raz, E A. (2010) *Community Genetics and Genetic Alliances: Eugenics, carrier testing and networks of risk*. London/New York: Routledge.
- Rozario, S. (2013) *Genetic Disorders and Islamic Identity Among British Bangladeshis*. Carolina Academic Press, Durham, North
- Ryan, K, Bain, B.J, Worthington, D. et al. (2010) 'Significant haemoglobinopathies: guidelines for screening and diagnosis', *British Journal of Haematology*, 149: 35-49.

Shakespeare, T. (1999) 'Losing the plot'? Medical and activist discourses of contemporary genetics and disability', *Sociology of Health and Illness*, vol. 21 (5): 669–688.

Shaw, A. (2011) 'Risk and reproductive decisions: British Pakistani couples' responses to genetic counselling. *Social Science and Medicine*, 73: 111–120.

Wailoo, K. (2001) *Dying In The City Of The Blues: Sickle Cell Anaemia And The Politics of Race And Health*. Chapel Hill: University of North Carolina Press.

Ulph, F, Cullinan, T, Qureshi, N and Kai, J. (2014) 'Parents' responses to receiving sickle cell or cystic fibrosis carrier results for their child following newborn screening', *European Journal of Human Genetics*, 1–7.

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