‘I don’t see any point in telling them’: attitudes to sharing genetic information in the family and carrier testing of relatives among British Pakistani adults referred to a genetics clinic

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(Received 16 October 2007; final version received 18 March 2008)

The sharing of genetic information following the diagnosis of a genetic condition can be important for managing familial risks for genetic conditions. This paper explores factors that impede or facilitate the sharing of genetic information within a sample of British Pakistani families. It draws from research investigating understandings of genetics and inheritance, attitudes to prenatal diagnosis and risk communication in the family that used methods of participant observation and interview with adults from 66 families of Pakistani origin referred to a genetics clinic in southern England. We found a lack of English often restricted one partner’s access to genetic information and partners fluent in English sometimes withheld information to protect a partner (usually the wife) from blame, stigma or feelings of marital insecurity. Many couples felt genetic information was private to them as a couple and were unwilling to share it with the wider family, commenting on its potentially stigmatizing and emotionally and socially disruptive effects on themselves, their child and their marriage, as well as on the marriage prospects of other family members. Those who sought carrier testing because of a family history did so when considering their own marriage or parenting, sometimes on the insistence of an affected relative, but did not readily discuss carrier testing with other relatives. Despite the complex consanguinity in some families, a family-based approach to risk management is not necessarily any easier among British Pakistanis than other ethnic groups.

Keywords: British Pakistanis; carrier testing; disclosure; genetic testing; family communication; families

Introduction

A genetic diagnosis uniquely implicates the affected person’s close biological relatives as at risk of developing the condition or having affected children because of their shared DNA (deoxyribonucleic acid). It thus raises questions about risk management in the wider family. Depending on the inheritance pattern of the condition, certain kin may be at particular risk. For an increasing number of genetic conditions, carrier tests can be offered to relatives to confirm or rule out these risks. Although risks to other family members may be discussed in genetics consultations, the emphasis given to familial risks varies within current UK clinical genetics practice.
and discussion may focus more on an individual or couple’s own risk and reproductive decision-making than on risk communication within the family. Following the principle of non-directiveness (Wertz and Fletcher 2004), clinicians do not insist that their patients ‘disclose’ genetic information to their relatives. Nonetheless, clinicians routinely send their patients a letter containing information about the condition and advice for at-risk kin that can be shown to relatives. It is usually assumed that patients do inform their relatives of the diagnosis and its implications for other family members, unless they state explicitly that they do not want relatives to know. The assumptions relating to disclosure and non-disclosure such as the implied responsibility to share genetic information with relatives are, however, problematic (D’Agincourt-Canning 2001). Even where a patient foresees no difficulties with disclosing genetic diagnostic and risk information, relatively little is known about processes of family communication of genetic information and the accuracy of what is told, to whom, by whom and when.

Patients’ sharing of genetic diagnostic and risk information with relatives may be influenced by the nature of the condition, the potential stigma attached to it, or to a genetic diagnosis generally, and by their beliefs about the etiology and inheritance of the condition (Weil 2000, Meiser et al. 2001). Patients may find it easier to share information about common treatable conditions than information about severe or stigmatizing ones such as sex-linked or recessive conditions (Peterson et al. 2003, Wilson et al. 2004). Patients’ everyday theories about a condition’s etiology and inheritance may be based on patterns in the observed family history rather than on genetic theory. Everyday theories can influence who is told about genetic risk, who is believed to be at risk and who is watched for signs of the condition and may suggest risk where, according to genetic theory, none exists, or overlook it where it is present (Davison 1996, Richards and Ponder 1996, Featherstone et al. 2006). Similarly, theories of inheritance, which vary cross-culturally, may provide models for understanding genetic risk at odds with genetic theory: for instance, the patrilineal kinship systems of South and South East Asia that prioritize links though men may provide a model for understanding biological inheritance ‘unilaterally’ (Richards 1996, Meiser et al. 2001). Some of the Pakistani-origin adults attending an UK genetics clinic considered risk for recessive conditions to be greater for people married to patrilateral rather than matrilateral kin, thus underestimating the risk of recessive conditions in the wider family (Shaw and Hurst 2008).

Communication about genetic risk is also influenced by the structure of the family, patterns of authority, gender norms, interpersonal interaction and relationships with ‘significant others’ (Kenen et al. 2004b), all of which may vary with ethnicity and culture. Among White British users of general genetic services, ‘popular psychology’ informs an individual or couple’s assessments of how different family members will cope with the burden of genetic risk information, influences whether, when and to whom risk information is disclosed, and may result in risk information being withheld from particular kin considered to be at risk (Featherstone et al. 2006). Those with whom genetic information is shared may be those most likely to reciprocate by offering support in coping with risk information; thus, women may talk to female rather than male kin about hereditary breast and ovarian cancer (Kenen et al. 2004a). Women may be more likely to act as ‘gatekeepers’ in communicating risk information within the family, at least with respect to breast cancer risk (Hallowell 1999, Richards 1996), and may provide
care-based ethical justifications for disclosing genetic information to kin (Hallowell et al. 2003). On the other hand, within patriarchal extended families, senior males may take on this role (Meiser et al. 2001).

Understanding the processes affecting genetic risk communication in the wider family is particularly relevant within populations favoring consanguineous marriage, because such marriage confers an elevated risk of recessive conditions, which are those caused by inheriting two copies, one from each parent, of a mutation that in a single copy confers no significant health risk. Consanguineous marriage increases the chance that marriage partners will be unaffected carriers of a mutation inherited from a common ancestor (Bittles 2001). Consanguineous communities are found in many regions of the Middle East, Asia and Africa and among migrants from these regions to North America, Western Europe and Australia (Bittles 2001, Port et al. 2005). In principle, depending on the particular genetic structure of the community and the distribution of recessive alleles within it, a proportion of this elevated risk can be managed by premarital or preconception carrier testing of relatives (in addition to prenatal diagnosis) because carrier tests are available for some known recessive conditions. A 'positive' carrier test result (showing the presence of the mutation) in one partner and a negative result (indicating that absence the mutation) in the other would rule out the risk of affected children for that couple. Alternatively, a test showing both partners are carriers would give a 25% risk of having an affected child; see Figure 1. In such a case, a proposed marriage might not go ahead, or a married couple might be offered preconception or prenatal advice about their reproductive options, including genetic prenatal diagnosis.

Communicating genetic risk information within consanguineous families may, however, pose particular challenges. Where marriages are both arranged and

![Figure 1. Autosomal recessive inheritance.](image-url)
consanguineous, as among the Muslim Bedouin of Israel, a diagnosis may be potentially stigmatizing particularly of women but also of the entire family (Raz et al. 2003). Individuals or couples may be reluctant to discuss a diagnosis that may adversely affect their own or other family members’ marriage prospects. Personal or religious beliefs and local understandings of illness causality may also mean that genetic problems are viewed as matters of fate, destiny or God’s will, as among Muslims in Saudi Arabia, rather than as risks to be managed (Panter-Brick 1991).

Some national strategies and ‘carrier matching’ schemes have reduced the births of children with particular recessive conditions. In Palestine, obligatory premarital testing for beta thalassaemia has produced a gradual reduction in the percentage of carrier couples deciding to marry and the number of affected children (Tarazi et al. 2007). In Cyprus, the Greek Orthodox Church will not marry a couple unless their beta thalassaemia carrier status is known, while social consensus and medical infrastructure supports thalassaemia carrier screening, prenatal diagnosis and selective abortion (Richards 1996, p. 263). In Iran, premarital screening and counseling combined with the option of prenatal diagnosis and selective abortion has reduced the birth incidence of thalassaemia (Samavat and Modell 2004). However, premarital carrier testing potentially stigmatizes those identified as carriers, while prenatal diagnosis raises the possibility of abortion which is unacceptable within some communities. Orthodox New York Hasidic Jews have avoided these problems by storing individual test results in a database available only to marriage brokers, who avoid arranging marriages of carriers (Merz 1987). Similarly, global orthodox Ashkenazi Jews operate a scheme called Dor Yesharim offering carrier tests for Tay-Sachs disease (and other recessive conditions, including cystic fibrosis): participants receive a code number instead of test results; two people contemplating marriage can then phone a hot line to learn of their genetic ‘compatibility’ (neither or only one partner is a carrier) or ‘incompatibility’ (both partners are carriers) (Broide et al. 1993, Abeliovich et al. 1996, Ekstein and Katzenstein 2001). Among the Muslim Bedouin of the Negrev region of Israel, a similar scheme is expected to reduce births of children with thalassaemia and congenital hearing loss (Raz et al. 2003, Raz and Atar 2004, p. 50).

This paper focuses on British Pakistanis, a population of relatively recent migrant origin, comprising 1.245% of Britain’s 60 million people and 43% of Britain’s 1.6 million Muslims (UK 2001 Census). Among British Pakistanis, rates of consanguineous and particularly first cousin marriage are comparatively high (Shaw 2001) and the risk of having a child with a serious genetic problem (6%) is approximately double that of the general UK population (3%), and tripled (9%) when there is complex consanguinity (Bundey and Alam 1993). Since the 1980s, professional debate has focused on how best to manage this elevated risk of recessive disorders. Some local public education programs, such as in Birmingham, UK have sought to discourage cousin marriage (Haslam 2001), but it has also been asserted, without empirical substantiation, that the complex networks of consanguinity and affinity within Pakistani extended families are a ‘positive resource’ likely to facilitate the communication of risk for recessive conditions via cascade genetic screening (Darr 1997).

There is a dearth of research to inform these debates. In areas of substantial Pakistani settlement such as the Midlands, referral rates of Pakistanis to UK genetics clinics are lower than expected from the prevalence of recessive conditions in
this population (Roberts et al. 1996). Clinical experience also indicates a low uptake of genetic counseling by close relatives of carrier couples with affected children (Canham et al. 2002). While relatives of carrier couples might know they could be carriers but choose to forego genetic counseling and carrier testing, one study shows that parents and relatives of children with thalassaemia, which is relatively common among British Pakistanis, had poor knowledge of recessive inheritance and less than half of the relatives had undergone carrier testing (Ahmed et al. 2002). For some British Pakistanis, as with other relatively new migrant populations, access to and disclosure of genetic information may also be impeded by patients’ lack of knowledge about prenatal services and problems with language and literacy (Meiser et al. 2001, Shaw and Ahmed 2004). General practitioners (GPs) or other specialists may fail to make appropriate referrals for genetic screening; a recent confidential enquiry showed that clinicians sometimes withhold the offer of carrier testing for thalassaemia, for instance, assuming that Muslims are not interested in carrier testing, on religious grounds (Modell et al. 2000).

This paper draws from a fieldwork-based study of the impact of genetic risk information on families of Pakistani origin referred to a UK general genetics clinic, funded by the Wellcome Trust UK (GR063078MA) and approved by the local research ethics committee. The study found that patients may draw from a range of everyday theories about the causality of problems in a pregnancy, infant, child or adult, such as accidents, childhood immunizations, medical negligence, eclipses and the influence of malicious spirits. It also identified a range of ideas about the inheritance of personal characteristics and medical problems across the generations, demonstrating parallels with White British clients’ understandings of inheritance, noting gendered ideas about inheritance and illness causality and showing how some of these ideas can be at odds with genetic theory in clinically significant ways (Shaw and Hurst 2008). Here, we focus on attitudes to sharing genetic information in the family and carrier testing of relatives, with a view to identifying factors that facilitate or impede such communication.

Methods

Given the dearth of information on this topic, the research used qualitative methods that enable a focus on clients’ understandings; these methods are summarized below because a fuller account is provided elsewhere (Shaw and Hurst 2008). Alison Shaw (A.S.) conducted the fieldwork in Urdu and English, according to the preferences of the participants, since she speaks both languages. A.S. does not speak or fully understand Mirpuri dialect and this restricted her communication with some of the older, pioneer-generation migrants, but most clinic referrals concerned younger adults (in connection with a problem in a pregnancy, infant or child) including Kashmiri-origin and Pakistan-raised adults who were generally pleased and able to converse with her in Urdu. The fieldwork included participant observation of genetics consultations and one or more semi-structured interviews in respondents’ homes before and after the genetics consultations. A.S. also facilitated some respondents’ access to genetic information by interpreting for them during their clinical consultations.

Participants were recruited from all the Pakistani referrals to the genetics clinic in a general hospital, their ethnicity inferred from their names. The hospital serves a
district with the second-largest Pakistani Muslim population of Southern England by comparison of UK 2001 Census data. The district’s Pakistani population shares many of the demographic and socio-economic features of British Pakistanis generally. It now comprises two- or three-generational families, descended from the pioneer-migrants of the 1950s and 1960s, who are originally from rural Mirpur in Azad Kashmir and from various parts of the Punjab (Shaw 2000, 2001). Many of the young second-generation adults among our study participants had spouses from Pakistan and most couples lived with kin or had extended family nearby.

Potential participants received information about the project and a consent form in Urdu and English. They were informed that the fieldworker is a social anthropologist, not a health-care professional, and were reassured about confidentiality. All but two invitees consented to be part of the study. Through the genetics clinic, we recruited 55 adults representing a referral concerning a problem in a couple’s pregnancy, infant or child, and six adults (four married, one engaged, one unmarried), from six different families, who were explicitly seeking risk information and carrier testing because of a known family history. Through local community contacts, A.S. also recruited five more people representing five families with an adult or older child with medical or intellectual problems; these families, it emerged, had previously attended the general genetics clinic or (in one case) the thalassaemia service. These participants also received written information about the project and gave written consent to participate.

The conditions observed reflected the background risks for genetic conditions shared with the general population and the elevated risk of recessive conditions associated with parental consanguinity. Most referrals (61/66) involved some degree of consanguinity. In 46/59 cases, the parents of the index case were reported as related as first cousins, though often with added consanguineous connections in the family pedigree, and in four of these instances they were related as double first cousins; in 15/59 cases, parents were reported as related as either second cousins, first cousins once removed, or more distantly. Five couples were unrelated. In 66 families, 69 conditions were observed, most of them (44/69) rare recessive disorders (including dysmorphic and lethal dysmorphic syndromes as well as metabolic, neurological, hematological, skeletal, renal, hormonal and dermatological conditions), nine autosomal dominant conditions, four chromosomal conditions, nine unknown, and three non-genetic conditions. Thalassaemia cases were underrepresented, as these are seen by the thalassaemia service. Recessive conditions were more common among, but not exclusive to, consanguineous families: one unrelated couple had children with a recessive condition but both parents had a family history of the same condition found in their children, while six consanguineous couples had children with dominant conditions.

The focus in this paper is mainly on cases involving conditions for which a conclusive genetic diagnosis was reached and where, from the clinical viewpoint, the risk implications for other close biological kin were straightforward. The firmly diagnosed conditions accounted for a majority (33/44) of the recessive conditions. A carrier test was potentially available to about half of the families with a recessive condition; diagnosis was inconclusive for a significant minority (11/44) of the ‘probably or possibly’ recessive conditions. In such cases, clinicians may not raise the issue of disclosure within the wider family unless asked, because the risk manage-
ment they could offer would be speculative, conditional upon firmer clinical knowledge or genetic research.

Analysis for this paper involved identifying themes relating to sharing genetic diagnostic and risk information with relatives – spouses, siblings and parents. Some themes were present in participants’ explicit statements about whether or not they would inform their relatives about a risk for a particular condition. Others only emerged from reading and re-reading fieldwork notes and considering the significance of casual remarks or less obvious themes, such as expressions of ambivalence about genetics and comments illustrating a desire to conceal facts indicative of a genetic problem or compromised fertility. In most cases, research participants represented couples referred in connection with a problem in a pregnancy, infant or child. A minority of referrals were motivated by an individual or couple’s desire to understand genetic risks for children they might have and/or explore the possibilities of carrier testing because of a known family history of a genetic condition in a sibling or the child or children of a sibling. We refer to our research participants using pseudonyms.

Results

Risk communication between spouses

Both partners in a couple usually attended referrals concerning a pregnancy, infant or child, but were often not equally engaged in the genetics consultation. A lack of English combined with the gender and power dynamics of the marital relationship frequently limited one partner’s access to genetic information, particularly in the case of women with little English who were married to UK-raised men. For instance, Mr K, British-raised and fluent in English, said initially that his wife did not need an interpreter as he would explain everything later. The purpose of the consultation was to consider whether Mrs K should be tested to confirm her carrier status. At first, Mrs K sat quietly during the consultation, but as it progressed, Mr K asked A.S. to explain the diagnosis and its implications to his wife in Urdu. It emerged that although the problem had been diagnosed nine months earlier, Mrs K knew nothing of its genetic aspects, and she welcomed the opportunity to put her questions to J.H. via A.S. who translated for her. Conversely, wives with fluent English sometimes had much more engagement with the genetics consultation than their husbands, particularly if their husbands had poor English. In these cases, the wives might say they did not need an interpreter, as they would explain everything to their husbands later. In an interview at home after their genetics appointment, Mrs N, whose niece and nephew have a recessive condition, admitted she did not ‘go into it in detail’ with her husband, although they had both consented to give blood for carrier testing.

Protecting a spouse from worry or blame

Mrs N’s understanding of the inheritance of the condition had identified her husband’s family as the source of the problem, erroneously, as it happened (Shaw and Hurst 2008). Because of this understanding, which their genetics consultation had failed to challenge:
we just waited for the results ... It was in my husband’s family because this is where the problems were ... [So I] did not see any point in making him feel bad by discussing it all again afterwards.

Similarly, some men interviewed at home after their clinical appointments said they had not communicated details of a clinical consultation to wives with poor English because they did not want to worry them. After the consultation in which Mr K’s wife learnt, through an interpreter, that her son’s condition implies she is a carrier, Mr K explained that he already knew this, but ‘I did not go into it all with her before (i.e., when the condition was first diagnosed) as I did not want to worry her’. As during Mr and Mrs K’s appointment, so during some other consultations, husbands or other male kin informed in English about the diagnosis of a genetic condition affecting their child, and its prognosis, inheritance and reproductive risks, turned to ask A.S. to ‘break the news’ or ‘explain’ the diagnosis to wives whose knowledge of English was poor. One husband left the consulting room to renew a car-parking ticket while this discussion took place. During a few home-based interviews, husbands asked A.S. to explain to wives with poor knowledge of English aspects of the inheritance or risk of a particular condition that had previously been discussed in the clinic.

Some men were explicit that, besides not wanting to worry their wives, they were uncomfortable about effectively having to take on a clinical role, feeling that they would be unable to answer the questions their wives might ask. This was less a simple matter of translation and more about enabling wives or, in one case, a fiancée, to have direct access to the clinical information. Mr A, a university graduate and unaffected carrier of a chromosomal condition, attended his first appointment alone but requested a second appointment to which he could bring his fiancée, a young British-raised graduate whom he had met while at university. As he said, ‘I am only just taking it in myself, and I want her to hear it straight and be able to ask you herself, as she is the sort of person who is going to ask all sorts of questions’.

Protecting spouse or self from marital insecurity

Some men elaborated on their concerns to protect wives from worry by saying they did not ‘go into the details’ fear that this would mean that their wives would be blamed for the condition and would consequently feel insecure about their marriages. During one home-based visit, a father who has lost three infants to a fatal recessive condition suggested A.S. talk privately with his wife in Urdu: she had attended the clinic appointments but understood little of what was discussed about the inheritance and associated reproductive risks:

You can talk to her, but you will see, I didn’t go into the details, as I did not want to her to think she might be blamed and I didn’t want her to think I might want to get another wife. It would make her insecure. But as you will see, we have different ways of understanding and coping.

After a genetics consultation in which the reproductive risks associated with an infant’s recessive metabolic syndrome were discussed in English and translated into Urdu for the infant’s mother, who became tearful, the father said he did not want anyone to talk further with his wife:
I don’t want anyone talking to her about the genetics because she gets very upset if she thinks there is anything wrong with him.

At a subsequent appointment, this man said he believed his son’s developmental delay and obesity was not genetic but caused by his wife’s overfeeding their child, whose care he had how taken time off work to oversee. Mr Q, another UK-raised husband with a wife from Pakistan, commented that, if he had been living in Pakistan, he might not have informed his wife of all the details of his child’s uncertain diagnosis:

The doctors in Pakistan, they say don’t tell the ladies, the ladies don’t need to know, the men make the decision. If we were in Pakistan, I would not tell her [my wife] everything. I might tell my Dad, but not my Mum, and I would not tell my brothers.

The expectation of patriarchal control and its consequences for women also meant that women with access to medical information might sometimes keep it to themselves. As Mr Q also observed, ‘A woman would not even tell her husband in Pakistan, in case he will remarry – she has to be careful, because her husband might then take another wife’.

Compromised fertility, genetic problems in children or evidence of risk of genetic problems in children can, in principle, be used justify divorce and a second marriage – even though divorce and remarriage is relatively rare, given the complex ties of affinity and consanguinity in the wider family. Mrs P, a British-raised woman with mild learning problems, specifically requested that she be informed first, verbally and without her husband’s knowledge, of the results of genetic tests on blood samples from herself, her husband and their child, rather than in a letter addressed to herself and her husband or communicated to him in Urdu (which she did not speak) via an interpreter because she feared her husband, a first cousin from Pakistan, would use this information as a reason for ending their marriage. The husband told A.S. he was not fully aware of his wife’s problems before his marriage and his friends were now advising him to leave his wife, but he felt loyalty to her, as his first cousin, and to their child. As it turned out, the genetic test results provided no evidence of a genetic problem but this husband did eventually leave his wife.

In the study sample, there was one other case of separation following the diagnosis of a genetic problem in a child: a woman left her husband and returned to her parents. In three other cases, relatives raised the possibility of divorce, in two cases jokingly. Mrs R has a sister, who is also her husband’s brother’s wife, and a sister-in-law, who is also her brother’s wife, each of whom is childless and has lost more than one pregnancy. Mrs R told me,

Sometimes I tell them ‘the husbands should try to have a baby with someone else, and then the baby would be okay’ . . . but we only say it as a joke. In our culture, in our religion, it is allowed, but never in our family has someone taken a second wife. One of our relatives, he did not have a child for 8 years, but he did not take another wife.

In the third case, Mrs L told A.S. that her husband’s parents had suggested their son remarry after he and his wife had lost a second infant to a recessive condition. She thought that family-based genetic counseling would not help dispel the blame being put on her (since a recessive diagnosis establishes both partners as carriers of the mutation) but would only worsen the situation: ‘If the mutation comes from both of
us, then they just have to change the wife’. At a later clinical appointment, Mr and Mrs L consented to mutation testing in a research laboratory but not to results being made available to the wider family.

**Risk communication in the wider family**

Individuals and couples may be concerned to withhold information about inheritance and risk from relatives in order to minimize interference and pressure, including pressure on a man to remarry. Mr Q, quoted above as saying if he was in Pakistan, he would withhold clinical details from his wife, saw the genetics referral as something that concerned himself and his wife but not his parents or wider family:

Here, it involves both of us, but we keep it to ourselves – not even my Mum and Dad, no-one [else in the family] understands what we went through with our daughter.

In part, this is to protect the couple from the harmful consequences of gossip about the nature and possible cause of an infant’s problem. As one father put it, ‘You know how Asian families are like; it’s like Chinese whispers’.

Some conditions are particularly stigmatizing. Mrs N has an older sister with two children with a recessive hormonal condition that includes genital ambiguity in female infants. When a younger sister gave birth to a girl with this condition, Mrs N said this younger sister was so ‘gutted’ that she told no one apart from two sisters and her affected niece, despite the family history of the condition:

Perhaps if I was in the same situation, I would do the same, but it is more of a shame factor – she is afraid to tell anyone. The trouble is, we won’t accept an imperfection. Our kids can be nothing less than perfect. It is so difficult to accept they might not be.

It seemed, too, that history was being repeated in this younger sister’s attempts to keep the condition a secret:

My sister is now behaving just like my other [older] sister ... She does not want anyone in the family to know about her child’s condition. The other day she said, ‘What if people find out about this condition? What if they notice something?’ I told her, ‘All you have to say is it’s the condition our niece and nephew have. That’s all you need to say, and as far as anyone else is concerned it’s just a matter of taking a couple of tablets’ ... She won’t take the baby to see our grandmother in Pakistan. She hasn’t even told our Mum. Her husband knows all about the condition; his brother is their father [father of the affected niece and nephew], but he, too, is saying nothing ... My husband does not even know about the new baby’s condition, and I find this very hard, because when I am upset I cannot talk to him because I have promised my sister I won’t.

**Protecting parents and in-laws**

The ‘shame factor’ is often mixed with concern to avoid causing distress to others. Mrs Z kept the fact of her stillborn baby’s genital ambiguity, noted at the birth, to herself and her husband and reported to her family that the baby was a boy:

If it was up to me, I would have kept it all to myself, but my family and his family knew I was pregnant. Most of my family knows what happened because they found out in the end – not because I told them – but they did not find out about the sex. It is not
something you can discuss even with your mother or your sisters . . . at least, not for me . . . the only person I discussed this with was my husband. Gradually my Mum found out that I was going through something, because it was obvious to her I was in a state, and eventually I talked to her. She was very shocked to hear the baby was not properly formed. She didn’t think that would happen in our family. You don’t think it will happen to you . . . But only my husband and myself know the baby had no sex.

I did not tell my parents, because they would have taken it very badly. It’s not that we’re ashamed, my husband and I, but my in-laws [in Pakistan] took it badly when they heard about the baby – my father-in-law was really ill when he heard the baby was born dead. To hear also that it had no sex – that would have been too much for him.

Protecting the child

Secrecy about a child’s condition may also reflect a desire to protect a living child from the remarks of relatives and neighbors. As Mrs N said of her sister:

She is afraid that people will notice that her baby is different; she is worried about anyone else changing the baby’s nappy. She wants her to have the operation so people don’t stare, so that she can be as normal as possible.

The father of a boy with a condition involving genital ambiguity said of his relatives:

They know he had an operation, but they don’t know the details. They don’t need to know. It would be bad for him. They would pick him out.

Protecting children’s marriage prospects

Mr Q felt that premarital carrier testing would damage people’s marriage prospects. In the quotation below, rishta, which literally means ‘connection’ or ‘relation’, refers to a potential marriage partner:

I do not agree with the idea of carrier testing. If you know you were carrying thalassaemia, if I knew I was a carrier, and the family knew, they would not want anyone else to know – they would not want it to get out. If my Mum was looking for a rishta for me, she is not going to tell them that I am a thalassaemia carrier. It will be the family’s well-kept secret. If I had blood cancer, my parents would not tell anyone, because if the other people where you are looking for a rishta know beforehand that you have a problem, they will say ‘no’ and bang, there goes the rishta.

Mrs U’s teenage son has a recessive metabolic disorder associated with learning difficulties and severe behavioral problems, for which a carrier test is available. Mrs U had a prenatal genetic test for this condition in a subsequent pregnancy, but said she had not raised the issue of carrier testing with any relatives and considered it inappropriate to raise it now, even with her eldest daughter who is studying medicine. Mrs S’s third child has learning difficulties associated with a recessive metabolic condition for which the causative mutation was identified during the fieldwork. Mrs S was interested in having a prenatal test in another pregnancy because she did not want to have another affected child but did not want any other family members, such as her brothers and sisters, to know that a carrier test was available. At first she was also uncomfortable with the idea that their GP (the family doctor) would be informed that a carrier test is available for her unaffected children:
I do not want my children to know ... Well, perhaps when they are older, if they ask ... and then the information is with the GP, but I would not want to talk to them about it.

Such concerns may also reflect a lack of understanding or confidence in genetic theory, particularly about recessive inheritance. Miss I, who has thalassaemia, said that when her condition was first diagnosed over 20 years ago, her parents understood their reproductive risks and were not unduly concerned as they had completed their family, but they did not understand the possibility that their older unaffected children might be carriers. They therefore said nothing about genetic risk when arranging their eldest son’s marriage to his father’s brother’s daughter, when their younger son married his mother’s sister’s daughter, and when their other daughter married her mother’s sister’s son.

Editing out the ‘genetic’ aspects

Although parents cannot keep secret a child’s obvious physical abnormalities or sudden death, they may, in addition to concealing shameful’ details from relatives, ‘edit out’ the specifically genetic aspects of the information shared within the family. Mr and Mrs D have lost two children to recessive conditions – the first, to a kidney problem, and the second to a cardiac condition. When the second child died:

We said bluntly [to family] that it is when the heart swells up and stops. And with our daughter, we said about the polycystic kidney that the kidneys did not develop. But I don’t think they know that it is a genetic thing. I don’t see any point in telling them’.

When pressed about the fact that a genetic diagnosis has implications for others because of the shared DNA, Mr Q, who ‘did not agree’ with the idea of premarital carrier testing, said:

Genetic? Well, if they say you have got this thing and we have to check your brother, I would not want to agree unless it is something they can cure. If I am going to die, what difference would it make? I would not even want her [his wife] to know. I would not want her wailing and being upset.

Protecting siblings’ marriages and keeping ‘good relations’

During clinical consultations, clinicians may also raise the possibility, depending on the inheritance of the condition, that relatives such as siblings are carriers at risk of having affected children. Following a diagnosis that raised the possibility that not just his wife but his wife’s sisters are unaffected carriers, Mr K was clear that discussing this possibility with his wife’s relatives was out of the question:

I could not tell her sister, for example, that she might be a carrier. They would think, ‘why is he trying to do this to me?’ It would cause divisions in the family. You can’t raise these sorts of things. You have to keep good relations.

Besides the need to ‘keep good relations’ and avoid divisions in the family, Mr K added that his wife’s sisters did not need to know about their possible carrier status because:
They’ve all had their children and all are okay, so there is no point telling them. If one of them had a child with this problem, then perhaps, yes, I would give them some advice, or if they had not got children yet, then perhaps . . .

The diagnosis of an autosomal recessive condition implies that the siblings of both parents might also be carriers. This is a significant risk where one parent’s sibling is married to a sibling of the other parent. Mr and Mrs D, who are first cousins, are carriers of two recessive conditions. Mrs D’s brother is married to Mr D’s sister, which makes them possible carriers of one or two recessive conditions. Yet on being informed of the risk implications for her siblings, Mrs D’s initial response was to say that no one in the family apart from her husband’s brother knows about ‘the genetic thing’, and she herself was not convinced that the cause was genetic and not simply ‘bad luck’. ‘It is all theory’, she said, ‘the theory is that we lost him because of genetics, and so we have a theoretical risk’.

Mrs D then said she would tell her siblings about genetic risk if they asked directly, but not otherwise:

I don’t think they need to know . . . I don’t think they have to worry about something that doesn’t happen . . . and probably won’t happen. I don’t see any point in telling them. It has got nothing to do with them anyway, unless they ask. The trouble is, once you have got into it – this high technology medicine and into thinking about risk and the options – it is hard to get out of it. My husband says I should just put it out of my mind.

Reasons given for not telling siblings they might be carriers of a recessive mutation included their having completed their childbearing, their not having any affected children, or their being married ‘out of the family’. Mrs D said:

My oldest brother has finished his family, all are healthy, so there’s no point telling him. My oldest sister has healthy children, all girls, and she is widowed and not going to have any more children so there is no point telling her. My youngest brother is married out of the family so I don’t think he needs to know. My middle brother is married to my husband’s sister. So they do have the same risk as us, but they have two healthy children – so why worry them? I do have another sister, but she is disowned by the family because she divorced my husband’s brother. She has healthy children from her first marriage and two children from her second marriage, but there is not point telling her, since her husband is not even Pakistani.

Marriages ‘outside the family’ thus represent a reduced risk of recessive disorders; indeed, such a marriage may represent a means of risk management that avoids or minimizes the harm to family relationships associated with raising the topic of genetic risk or suggesting that a prospective spouse seek a carrier test. Miss W, who has a recessive dermatological condition, is unmarried and wanted to know her reproductive risks should she marry. On learning that marrying a carrier of the same condition would give her a 50% chance of having an affected child, Miss W said she could not discuss genetic risk and carrier testing with her immediate family: her parents would not understand and would worry about offending their relatives with the suggestion that a son should have a carrier test. ‘In any case’, she added, ‘there is no one of a suitable age among my cousins’. She was therefore pinning her hopes on a marriage outside the family. Mr A, another unmarried adult referred by his GP for genetic advice was also intending to marry ‘outside the family’.
Closeness to an affected sibling or other relative

Having an affected sibling, niece or nephew can provide the impetus for someone seeking a genetics referral and having a carrier test. Occasionally, diagnosis and carrier testing can occur almost simultaneously. Mr H sought a carrier test immediately on learning that his sister, married to his wife’s brother, had given birth to a baby with severe, fatal, metabolic problems. Closely involved in liaising with medical staff when his sister’s baby was born, he was deeply affected by his sister’s predicament, having been responsible for arranging her marriage, and his wife was in the early stages of pregnancy.

Miss W’s older sister and her husband, who are first cousins, sought genetic advice shortly after their marriage, but Miss W’s younger sister and her husband, first cousins who married some years later, had not sought any genetic advice. Miss W said this was because her younger sister did not know very much about Miss W’s condition whereas her older sister had often accompanied her to medical appointments and as a result knew much more about the condition.

Considerable time may elapse between diagnosis of a condition in a child and carrier testing of a relative such as sibling. Even then, other family members including the affected person (in the case of lifelong medical conditions) may not know that a close relative has sought genetic advice. Rather than discuss thalassaemia risk with his parents, his siblings or his unaffected sister, Miss I’s eldest brother talked to his GP soon after he married a first cousin, had a carrier test, learnt he was not a carrier and decided not to inform anyone in the family that he had been tested. Some years later, Miss I’s newly married younger brother also sought advice from his G.P when his wife, also a first cousin, became pregnant. As Miss I reported:

What made him get the test is that he is very close to me. He has seen all that I have to go through. He could not take the thought that one of his children might have the same problems as me. They gave my brother all this information, and he then explained it for everyone. After that my sister got tested, and her husband – though they don’t have any children. It was only then that our parents understood. Up to then, I had gone for treatment without understanding that this was a genetic thing, with implications for other relatives.

Miss I also thought her eldest brother had not sought a carrier test and was, like his parents, skeptical that thalassaemia has a genetic cause and confers familial risks, but in a later interview said that during further, recent, discussion of the causality and inheritance of thalassaemia in her family she learnt, to her surprise, that her eldest brother had in fact sought a genetic test years ago, soon after getting married.

Concern and insistence of affected relative

An affected person who, like Miss I, now fully understands the inheritance and risks for their condition may be very concerned to share this knowledge with close kin, to enable them to consider risk-management options such as – depending on the condition – pre or postnatal therapy, in order to protect the well-being and quality of life of the close sibling and their unborn child. It was only on the insistence of her affected niece, now a biology student, that Mrs N requested a risk assessment after marrying a first cousin:
Actually, when we married, I didn’t really understand that the risk was quite high. I always knew that my niece and nephew took medication and had some condition, but as child I never thought anything of it. It was just a way of life for us. No one said this is it, [names the condition], and this is how it happens, and this is the risk. But when I wanted a baby I spoke to my niece because I am close to her in age – there are only 5 years between us. She told me about the risk and she was quite insistent that we should get tested before we started planning for a baby. Yes, it was my niece, not her Mum, who talked about it. My sister has never spoken about it, never. But then I found I was already pregnant. Looking back, I can see she [my niece] was quite upset that we had not been tested first.

Their daughter was about six months old when Mr and Mrs N were eventually tested and learnt Mr N is a carrier and Mrs N is not. Mrs N now wants her daughter to have the carrier test when she is older:

My husband says she should have the test and if [it] shows she is a carrier, then she should not be married in the family, or if she is, then the boy should be tested. But that’s down to [my niece] opening my eyes – she was the driving force behind me getting tested.

Discussion

Whether patients or clinicians should communicate genetic information to patients’ relatives has generated ethical debate over who has ownership of and responsibility for genetic information (D’Agincourt-Canning 2001). Currently, the onus is on clients to ‘disclose’ genetic information to their biological kin. This paper has described some of the difficulties that British Pakistanis face in sharing genetic information with relatives. It shows that these difficulties can begin within the married couple. For the parents of an infant or child with a newly diagnosed condition, a diagnosis implies compromised reproductive potential that can generate marital insecurity. For men, discussing genetic information with wives with limited knowledge of English can be particularly difficult. They may feel insecure in their understanding of the information and ability to translate it, be concerned with gendered propriety in discussing details of conception and inheritance, or not want the emotional burden of sharing potentially distressing or stigmatizing information about their own or their spouse’s carrier status. Although everyday ideas about biological inheritance among British Pakistanis may sometimes imply a larger generative contribution from the father, via the blood of the patriline, women tended to be blamed or to blame themselves for a problem in a child (Shaw and Hurst 2008). This tendency for women to be blamed or blame themselves has been observed for other patient groups particularly in connection with the diagnosis of X-linked conditions (James et al. 2006), but in this study was independent of the clinically established inheritance of the condition, as has also been observed for British Bangladeshi users of genetic services (Rozario 2007). Some of the situations reported in this paper suggest that, in contrast to what is reported of breast cancer risk communication (Richards 1996, Hallowell 1999), men rather than women may act as ‘gatekeepers’ in relation to the disclosure of genetic information to spouses and to people within the wider family. On the other hand, the paper also indicates that some men would welcome the opportunity for information to be discussed with
their wives by a bilingual professional, in this research by the fieldworker acting as translator.

That the fieldworker at times effectively had the role of translator or genetic counselor raised some interesting ethical issues. A.S. felt that to refuse to translate or seek answers to patients’ questions on clinical matters would have been unethical; indeed, doing this offered insights into patients’ understandings of genetic concepts. However, A.S. checked any clinical information she was unsure of with J.H., alerted clinical staff to some patients’ needs for additional genetic counseling, and received honorary clinical contracts from two hospitals in recognition of her role in facilitating patients’ access to clinical information. The fact that this role was necessary raises issues for genetic counseling service provision and practice that we discuss elsewhere (Shaw and Hurst 2008).

**Privacy of couples within extended families**

Many couples had regular contact with extended family members, yet our data suggest that medical or genetic information is not readily or easily shared within the wider family. Individuals and couples in general welcomed genetic information and counseling but felt strongly that genetic information was private to them as a couple. Respondents repeatedly commented on the potentially stigmatizing effects of genetic information for those identified as carriers, for mothers of affected children, for the parents’ marriage, for other relatives’ marriages and for wider family relationships. Couples were concerned to protect not just their own marriages but the marriages and marriage prospects of their siblings and other kin. Our data indicate that the very idea of suggesting, for instance, that one’s brother or sister might ‘carry’ a genetic condition might be viewed by relatives as a willful attempt to create rifts in the family by implying something is ‘wrong’ with someone or that their marriage is ‘at risk’. A desire for privacy may be particularly acute within a tight-knit community where social norms require relatives and neighbors to participate in many aspects of family life, and where marital success is judged largely by reproductive success, as in many parts of South Asia (Shaw 2000, Baradwaj 2003).

The desire to withhold genetic information from at-risk kin was expressed through the use of what Katie Featherstone and colleagues have described for White British users of genetic services as popular psychology and practical ethics (Richards 1996, Hallowell 1999, Featherstone et al. 2006). Similarly, our respondents felt that certain relatives ‘do not need to know’, because, for instance they do not have an affected child, and it would therefore be wrong to worry them. As we have reported elsewhere, our respondents’ beliefs about the causality of their child or relative’s condition indicated a general reluctance to accept a diagnosis as genetic, relatively poor understandings of medical genetics, and skepticism of the discourse of risk associated with consanguineous marriage. Thus, as one respondent put it, even if they were to inform their relatives ‘they would not believe it anyway’.

**Contexts of risk communication**

The paper highlights the importance of social context, particularly position in the life course, marriage and childbearing, as well as the nature of the condition that is in the family, in shaping responses to genetic risk and its implications for relatives. For
the parents of children with newly diagnosed conditions, the implications for unaffected children or siblings may seem remote or irrelevant in comparison with the child’s prognosis and the parents’ reproductive risks. While the significance of a diagnosis for other relatives may be immediately apparent, for instance if a parent’s sibling is closely involved in a child’s medical care, usually considerable time elapses before those aware of a problem ‘in the family’ choose to confront their own risk, often only when marriage or children become real, immediate concerns. Similarly, White British people aware of their risk for dominant conditions such as Huntington’s disease may live with uncertainty for years, only seeking testing when they reach the age at which symptoms of this late-onset condition usually appear (Madigan 1996, p. 24).

Experience of a genetic condition via an affected sibling (or other close relative) may have an important influence over individuals’ perceptions of their risk of being carriers (Fanos and Johnson 1995). Nonetheless, our findings suggest that, as has been shown for other ethnic groups, being sufficiently motivated to ask a GP for a referral for carrier testing does not necessarily indicate willingness to disclose the experience of testing or test results with relatives. Only about 18% of people with a parent with Huntington’s disease actually seek testing, despite earlier estimates that perhaps 80% of such people would do so (Harper et al. 2000, Creighton et al. 2003); moreover, those who have been tested may be reluctant to communicate test results to kin because of the emotional and psychological burden this carries (Wright 1996, p. 7). Those with negative results may feel guilty about being clear when other relatives may be carriers, or fear that sharing results will divide the family, especially if some relatives are ambivalent about testing (Madigan 1996, pp. 10, 18, 21, and 26). As a result, clients of genetic services may underestimate their siblings’ or other relatives’ interest in testing or degree of concern about risk. Some relatives may only seek carrier testing because of the insistence of an adult relative who has a genetic condition; such adults may— in some families, and in relation to some genetic conditions— prove key to facilitating the seeking of risk information and carrier testing.

Recessive conditions, indicating something ‘in the family’, may be particularly difficult to discuss with kin (along with other stigmatizing conditions that may or may not be recessively inherited) because of the wider discourse of disapproval associated with consanguineous marriage in contemporary Europe and America (Shaw 2006). Confronting genetic risk may thus be easier in relation to chromosomal or other conditions not associated with parental consanguinity. It may also be easier for unrelated individuals contemplating marriage to discuss genetic risk than for consanguineously related people, particularly if their marriage is being arranged by parents or other kin, because in the latter case there are many prior social and emotional investments in the success of the relationship (Shaw and Charsley 2006).

Policy implications

In the UK, the elevated risk of recessive disorders associated with consanguineous marriage is primarily managed by giving risk information and allowing individual reproductive choice, although some local public health campaigns have sought to reduce this risk by discouraging consanguineous marriage. We note elsewhere how the discourse of genetic risk associated with consanguineous marriage is potentially
alienating of Pakistani Muslim users of UK genetic services (Shaw and Hurst 2008). In some parts of the world where consanguineous marriage is more common, risks for recessive conditions such as beta thalassaemia, congenital deafness and Tay-Sachs disease are managed through policies of premarital carrier testing, supported by prenatal diagnosis and abortion, or, where the latter is unacceptable, by ethnic group-specific ‘carrier matching’ schemes for preventing the marriage of carriers. Such approaches are generally considered more effective than ‘preaching against consanguinity’ (Raz and Atar 2004, p. 50). The situation for British Pakistanis differs in several important respects. Currently, there is no evidence that one or more recessive conditions prevail among British Pakistanis at frequencies to justify selective carrier testing on grounds of either ethnicity or parental consanguinity, apart from thalassaemia, for which there is now (since 2007) universal prenatal and neonatal screening in the UK. Secondly, British Pakistani Muslims, unlike, say, orthodox Ashkenazi Jews or the Bedouin Muslims of the Negrev region of Israel, are heterogeneous in their social characteristics and religious beliefs, including in the acceptability of ending a pregnancy. Thirdly, while arranged consanguineous marriage is often preferred, it is not the only form of marriage that occurs, the process of arranged marriage is itself changing and young adults are increasingly aware of the elevated genetic risks associated with parental consanguinity (Shaw 2006). Thus, a strategy such as ‘carrier matching’ is unlikely to be appropriate for all.

Since the elevated risk is mostly for very rare recessives and is therefore particular to individual families, the individual or family case-based approach to genetic risk management, as currently offered to all users of clinical genetic services, therefore seems more appropriate, even though, as we have shown, it also presents many challenges. The argument has been made, perhaps in order to counter the negative association of arranged consanguineous marriage within dominant UK public discourse, that the links of affinity and consanguinity that characterize Pakistani families offer a ‘positive resource’ that will facilitate cascade screening in the family following the diagnosis of a recessive condition (Darr 1997). Yet the efficacy of cascade screening in the general population is not necessarily higher than population-wide screening (Krawszak et al. 2001). In pointing to the contexts in which genetic information may be either withheld or shared, the evidence presented here suggests that we cannot assume that a family-based approach to genetic risk management will necessarily be easier among British Pakistanis than among other ethnic groups.

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