ORIGINAL ARTICLE

Attitudes towards prenatal testing and termination of pregnancy in British Pakistani parents and relatives of children with recessive conditions in the UK

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ABSTRACT

Objective To compare British Pakistani parents' and their relatives' attitudes to prenatal testing (PND) and termination of pregnancy (TOP) for a range of conditions.

Method A total of 222 British Pakistani participants: 117 parents of children with a child with a genetic condition (52 fathers and 65 mothers) and 103 of their relatives (51 males and 52 females) completed a structured questionnaire about their attitudes toward PND and TOP for 30 different conditions.

Results Parents were more accepting of PND ($P < 0.001$) and TOP ($P < 0.001$) than their relatives for most of the conditions. Male relatives were consistently least interested in PND and TOP, except for conditions at the serious end of the continuum, where over 90% would opt for PND for quadriplegia and anencephaly, and over 60% would opt for TOP for these conditions.

Conclusion The lower level of interest in PND and TOP in relatives, particularly men, may be due to lack of information disseminated by parents about their child's recessive inheritance and its implications for relatives, resulting in poor understanding of genetic risk. These findings highlight the need for the provision of proactive genetic counselling to raise awareness of genetic risk and facilitate informed reproductive decision-making in at-risk relatives. © 2012 John Wiley & Sons, Ltd.

Funding sources: National Institute of Health Research, Department of Health.
Conflicts of interest: None declared

INTRODUCTION

Research shows that British Pakistanis account for 3.1% of all births in the UK, but constitute 30% of all British children with recessive conditions.[1] Approximately one third of these children die before the age of five years and many of the survivors have chronic disabilities and are cared for by community or paediatric services. In 2008, the Bradford Paediatric Department reported seeing around 150 different autosomal recessive conditions.[2] Moreover, the Bradford District Infant Mortality Commission reported that the infant mortality rate for babies born to mothers of Pakistani origin was twice that of babies born to White British mothers.[3] The Bradford District Infant Mortality Commission suggested that this was partly due to congenital anomalies, and although the risk of being affected by a recessive condition was relatively small, these conditions were 'significantly more likely' to be the cause of death in infants of Pakistani origin compared with British White infants.[3] The comparatively high incidence of autosomal recessive conditions among British Pakistanis is considered a public health issue in the UK. The increase in prevalence of recessive conditions is not limited to the Pakistani population in the UK. Other European countries have observed similar increases in minority ethnic populations where consanguineous marriages are customary, including France,[4] Denmark,[5] and Norway.[6,7]

Advances in genetic analysis, now and in the future will allow relatively cheap and rapid carrier testing for rare recessive conditions in consanguineous families with affected children.[8] Such technology will allow affected families and their relatives to make informed reproductive choices about their family condition. Given the importance of cultural factors in planning prenatal screening programmes,[9] the introduction of such technology into service delivery should be based on the target population's attitudes toward prenatal diagnosis (PND) and termination of an affected pregnancy (TOP). However, there is currently limited research about the acceptability of such technology within Pakistani families in the UK who have a child with a recessive condition. For example, a qualitative

study with British White and Pakistani mothers of affected children in the UK, found that both groups' attitudes toward PND and TOP for a range of conditions were based on their perceptions of the quality of life for the child.[10] Similar to other studies,[11,12] these mothers would consider whether the child would 'suffer' or could lead a 'normal' life and were likely to accept PND and TOP for conditions resulting in severe mental or physical disabilities or death in childhood.

Comparatively, there is much research on attitudes toward PND and TOP in Western populations at increased risk of specific genetic conditions,[13–17] which shows that women tend to hold favourable views about screening and that screening uptake rates are high.[18] There is also much research on such attitudes in Muslim populations, but most of this is based in non-Western countries, such as, Lebanon,[19] Pakistan[20] and Saudi Arabia.[21] This literature suggests that PND and TOP are more acceptable in Muslim populations when delivered with a Fatwa (Islamic ruling), stating that termination of pregnancy is permissible if the fetus is affected with a serious condition and termination is within 120 days of gestation. However, such findings need to be interpreted with caution when generalising to the UK populations because of the potential impact of acculturation.[22] Research shows that while religion is important in decision-making, the perceived severity of the condition and the implications of the condition for the child's quality of life can be more important.[23]

Although there is some research on British Pakistani populations' attitudes toward PND and TOP, it is mainly qualitative in nature, focuses on a particular condition, and/or is with populations that are not affected by a genetic condition.[10,24,25] Furthermore, there is no equivalent published research with Pakistani men or Pakistani fathers of an affected child, in the UK or elsewhere in the world. Understanding men's attitudes is important because research shows that many women would prefer to make decisions about prenatal testing with their partner.[26]

In addition, people may be more accepting of PND and TOP after having a child with a genetic condition,[27,28] suggesting that there are differences in attitudes between people who have a child with a genetic condition and those who do not. Relatives of children with genetic conditions can also be considered to have experience of the implications of that condition. To our knowledge there is no published research comparing parents and relatives attitudes toward PND and TOP for a range of conditions, in a Pakistani population based in a Western country. Therefore, the aim of the present study was to compare attitudes towards, hence the acceptability of, PND and TOP in British Pakistani parents and relatives of children with a recessive condition for a range of conditions.

METHODS

Questionnaire

A self-completion postal questionnaire developed by Hewison *et al.* and available in English and in Urdu was used.[25] The questionnaire contained descriptions of 30 real conditions. The conditions were not named to minimise the influence of preconceived ideas and because participants may not be familiar with all the names. Participants were asked to respond to two questions for each condition: whether or not they would want a prenatal test and whether or not they would terminate an affected pregnancy. Participants could respond 'no', 'not sure', or 'yes' to these questions (the questionnaire is described in, and appended to, Hewison *et al.*). To complete the questionnaire, participants were asked to assume a hypothetical scenario in which testing would be carried out early in pregnancy, with routinely collected blood, and would provide a diagnostic result.

Participants and recruitment procedure

The study was approved by the appropriate NHS Local Research Ethics Committees. During January 2008 and December 2009, genetic counsellors contacted Pakistani parents who already had a child with a recessive condition and who had attended genetic counselling within the last 3 years. The questionnaire, information sheet and consent form were posted both in English and Urdu to interested participants, with extra copies for them to pass these on to their relatives for completion. Relatives were described as the participants'/parent's siblings (aunts and uncles of the affected children) and adult siblings of the affected child (18 years old and over). All participants were asked to complete the questionnaire independently and return it separately in the stamped address envelopes provided.

RESULTS

Data were managed in SPSS Version 11.[29]

Sample characteristics

Two hundred and sixty-seven parents of a child with a recessive condition were contacted and all agreed to complete the questionnaire. The final sample consisted of 220 participants: 117 parents (52 fathers and 65 mothers, not all parents were couples) representing 70 families (47 couples, 18 mother only and 5 father only), and 103 relatives (51 males and 52 females). All participants described their ethnic origin as Pakistani and their religion as Islam. See Table 1 for sample characteristics.

Attitudes to prenatal testing and termination of pregnancy

For brevity, names of the 30 conditions are used from here on instead of the scenario descriptions.

Figures 1 and 2 show the percentage of parents and relatives, divided by gender, who answered 'yes' they would want a prenatal test and 'yes' they would terminate an affected pregnancy, respectively. For purposes of comparison with the Hewison *et al.*,[25] the conditions have been ordered by the percentage of participants saying 'yes' to prenatal testing in their article, and this order is the same in Figures 1 and 2 below.

Figure 1 shows that participants from all the subgroups were interested in prenatal testing. The greatest difference in attitudes toward prenatal testing was between the fathers and male relatives, where the level of interest was highest in the fathers and lowest in the male relatives for most of the conditions. Male relatives were consistently least interested in

Table 1 **Demographics characteristics of participants ($n = 220$)**

		Parents		Relatives	
		Male $n = 52$ *N* (%)	Female $n = 65$ *N* (%)	Male $n = 51$ *N* (%)	Female $N = 52$ *N* (%)
Education	Up to GCSE level	31 (60)	38 (59)	30 (59)	31 (60)
	Above GCSE level	21 (40)	27 (41)	21 (41)	21 (40)
Employment status	Employed	43 (83)	11 (17)	38 (74)	11 (21)
	Unemployed	7 (13)	1 (1)	2 (4)	9 (17)
	Student	—	2 (3)	11 (22)	10 (19)
	Looking after home	2 (4)	51 (79)	—	22 (43)
Language in which questionnaire was completed	English	19 (37)	21 (32)	10 (20)	8 (15)
	Urdu	33 (63)	44 (67)	41 (80)	44 (85)
Age in years	Mean (SD)	35 (6.3)	33 (6.9)	29 (6.3)	28 (7.6)

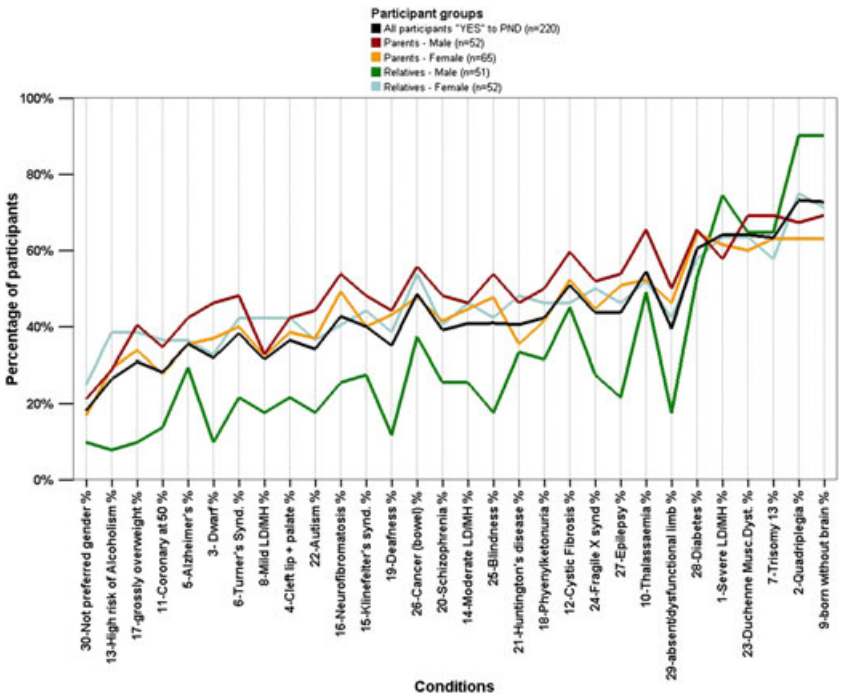

Figure 1 **Participants who said YES to prenatal diagnosis in each group, and in the full sample. Conditions are ordered by the percentage of participants saying YES to prenatal diagnosis in the work of Hewison *et al.*[25]**

prenatal testing, except for conditions at the serious end of the continuum, where over 70% wanted prenatal testing for severe learning difficulties, and over 90% for quadriplegia and for anencephaly.

As indicated in Figure 1, Cochran's Q tests for differences between correlated proportions, for participants saying 'yes' to prenatal testing for each condition, confirmed that each of the four subgroups had significantly different attitudes toward prenatal testing for the different conditions ($p < 0.001$).

Figure 2 shows that both mothers and fathers of a child with a genetic condition were more accepting of termination of an affected pregnancy for most of the conditions. All the subgroups were interested in termination for the more severe conditions, including diabetes, severe learning difficulties, Duchenne muscular dystrophy, trisomy 13, quadriplegia and anencephaly. Less than 30% of the participants would accept termination for the remaining 26 conditions. Male relatives were least accepting of termination for most of the conditions, where none of them would terminate for 11 of the 30 conditions, and 2% would terminate for nine conditions. Conversely, male relatives had the highest level of interest in termination for the two most serious conditions on the continuum, where over 60% and 70% would opt for termination for quadriplegia and anencephaly, respectively.

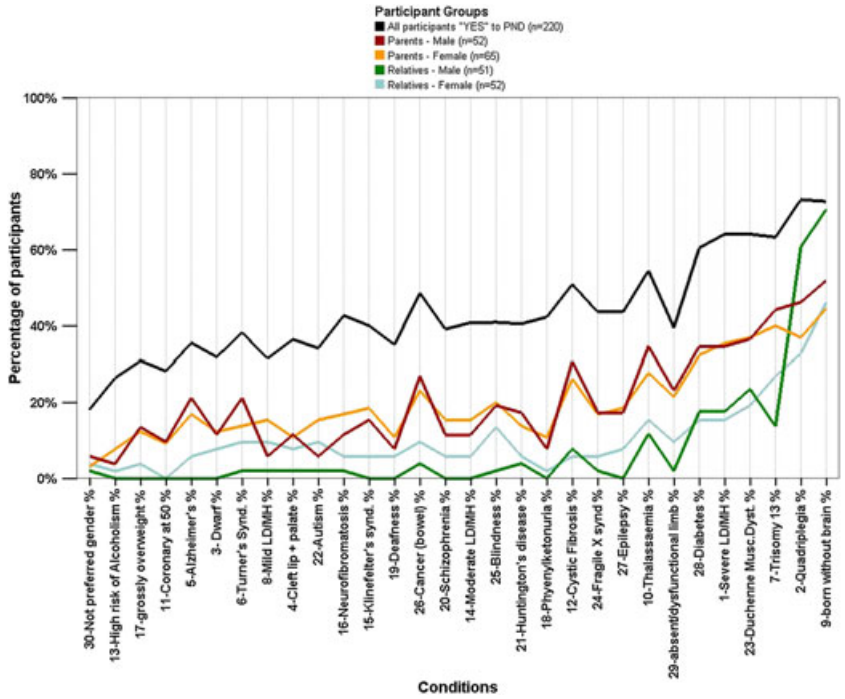

Figure 2 Participants who said YES to termination of pregnancy in each group and YES to prenatal diagnosis in the full sample. Conditions are in the same order as in Figure 1

Again, Cochran's Q tests confirmed that participants within the four subgroups had significantly different attitudes toward termination of pregnancy for the different conditions in this study ($p < 0.001$).

Individual participants' attitudes to prenatal testing and termination for the 30 conditions

Participants could respond 'no', 'not sure', or 'yes' to the two questions for each of the 30 conditions. To summarise attitudes across conditions, these responses were scored as 0, 1 and 2, respectively, and added to calculate a total score for each participant for prenatal testing and for termination of pregnancy. A total score of 0 showed that participants did not want testing for any of the conditions and a total score of 60 showed that they would want testing for all the conditions. This total score shows considerable variation in parents' and relatives' attitudes for the 30 different conditions for both prenatal testing (Cronbach's alpha = 0.973) and termination of pregnancy (Cronbach's alpha = 0.966). For prenatal testing, 21% of the parents and 15% of the relatives had scores in the 55 to 60 range, indicating that they wanted testing for all or nearly all the conditions. Five percent of parents and 4% of the relatives did not want testing for any of the conditions (see Figure 3). For termination of pregnancy, 3% of the parents and none of the relatives had scores in the 55 to 60 range, while 20% of parents and 15% of relatives did not want termination for any of the conditions (see Figure 4).

Exploratory analysis, using nonparametric Mann–Whitney U tests, between the four subgroups (parents and relatives, subdivided by gender) showed that fathers had a significantly higher total score than male relatives, hence were more

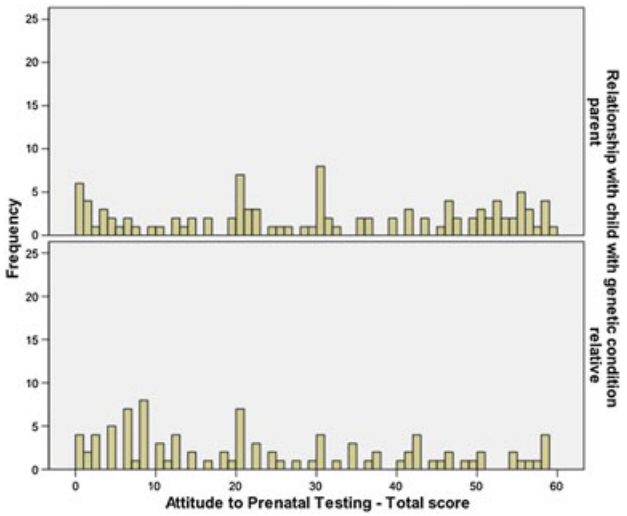

Figure 3 Parents' and relatives' attitudes to prenatal testing

accepting of both prenatal diagnosis ($z = -3.2$, $p < 0.001$) and termination of pregnancy ($z = -3.4$, $p < 0.001$). Other comparisons between males and females in the parents and relatives groups did not reach significance.

DISCUSSION

Similar to other studies,[21,25] we have shown that overall, parents of children with a recessive condition and their relatives were more accepting of PND than TOP for the majority of conditions. The variation in total scores for PND and TOP shows the participants varied from favourable views for the majority of

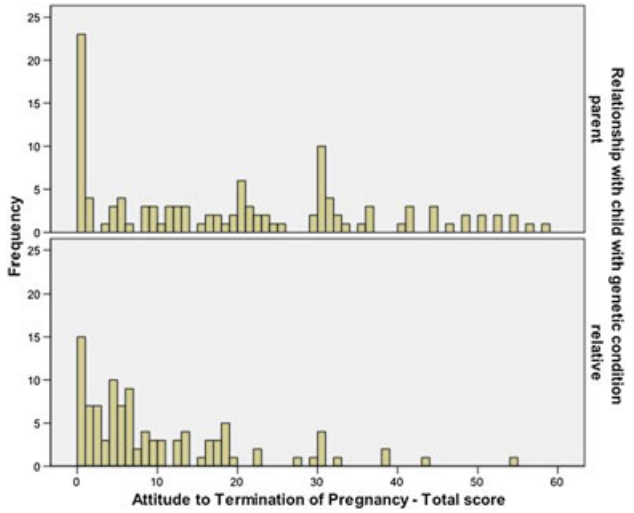

Figure 4 Parents' and relatives' attitudes to termination of pregnancy

conditions, through to little or no interest for any of the conditions, suggesting that participants distinguished between the conditions. There were no conditions for which participants did not want prenatal testing.

The higher level of interest in PND and TOP in the parents may be because they experience stigma and negative views about disability within their Pakistani community, where they and/or their affected children are often excluded from social (family and community) and religious gatherings.[30] The lower level of interest in PND and TOP in relatives may be due to lack of information disseminated by parents about their child's recessive inheritance and its implications for relatives, resulting in poor understanding of genetic risk in relatives.[31,32] There are a number of reasons why parents are unlikely to disseminate or disclose information about their child's recessive condition. First, parents may have poor comprehension of information provided during genetic counselling about recessive inheritance.[33] Hence, parents may feel ill equipped and lack confidence to disseminate this information to relatives,[31] particularly if it needs to be translated into another language. Second, the medical model of inheritance may not fit with lay beliefs, in particular, how a condition can run in the family when no-one else in the family has the condition.[34] Third, parents may feel 'that their extended families and communities also watch and judge them according to underlying negative beliefs about the causes of disability'.[30] Such social encounters may result in parents feeling responsible for their child's condition, resulting in social isolation for them and their child within their own family and community.[35]

There are a number of initiatives to raise awareness in British Pakistani populations about their risk of recessive genetic conditions, particularly in relation to cousin marriages. Whether such initiatives succeed in reducing consanguinity or increasing uptake of carrier testing remains to be seen. Providing proactive genetic counselling is probably the best approach to raise awareness of genetic risks, and to facilitate informed reproductive decision-making in at-risk relatives.[1] Currently, genetic counsellors in the UK encourage parents of children with an affected child to disseminate information to their relatives. However, our clinical experience with Pakistani families shows that at-risk relatives do not access genetic services, possibly because of poor information dissemination by parents.[35] Primary care services can also play an important part in educating at-risk relatives about the implications of their family condition,[36,37] where relatives may feel safe and supported.

Overall, the fathers in this study were more accepting of prenatal diagnosis and termination of pregnancy than the mothers. This may be because the fathers believed they had overall responsibility of care for their family, or because they experience financial and societal pressures. Nevertheless, there is a dearth of research on fathers' attitudes toward PND and TOP and a need for in-depth qualitative research to explore fathers' experiences and reasons for such attitudes.

Interestingly, antenatal screening guidelines focus on women as decision-makers.[38] However, the role of their partners should not be underestimated, given the evidence that women would prefer to make joint decisions about antenatal testing and termination of an affected pregnancy with them.[26,39] The fathers in our study had similar attitudes toward PND and TOP, suggesting that male partners could provide women with added support and reduce the burden of decision-making on them. Such joint decision-making may also help couples to adjust and cope after difficult reproductive decisions. Furthermore, male partners 'need to be informed, and supported both in their own right and to be supportive to their partners'.[40] Our clinical experience suggests that women often attend alone for prenatal counselling sessions. It is imperative that genetic counselling services proactively encourage both partners to attend their appointments so that counsellors can inform and facilitate joint decision-making. Furthermore, counsellors should acknowledge and meet the needs of fathers as individuals.[41]

The fathers in this study were also more accepting of both PND and TOP than their male relatives for most of the conditions, except for the more serious conditions, such as, quadriplegia and anencephaly. Given that the men in this study were all Muslims and had experience of a child with a genetic condition, either as a father, uncle or brother, differences in attitudes cannot be attributed to religion or understandings of the implications of the condition. There appears to be something specific about being the father of an affected child that results in them being more accepting of prenatal testing and termination of an affected pregnancy for a range of conditions. Similarly, research shows that individuals' views may change to become more accepting of termination of an affected pregnancy over time when they have a child with a genetic condition.[10,27] Exploratory qualitative research is needed to understand reasons for the difference between fathers' and their male relatives' attitudes to enhance our understandings of information dissemination within families and how to raise

awareness about genetic testing in Pakistani families considering consanguineous marriages.

The low level of interest in TOP in relatives may be due to socially desirable responses based on moral and religious objections for a hypothetical scenario. The actual behaviour of these relative may be different in a real situation. Furthermore, at the time of the study, participants were presented with a hypothetical scenario of noninvasive prenatal diagnosis using maternal serum. However, further developments in technology now provide the potential for population level noninvasive prenatal diagnosis (http://www.rapid.nhs.uk), making our hypothetical scenario a reality and our findings very timely.

WHAT'S ALREADY KNOWN ABOUT THIS TOPIC?

- There is a comparatively high incidence of autosomal recessive conditions among British Pakistanis in some areas of the UK.
- Advances in genetic analysis allow relatively cheap and rapid carrier testing for rare recessive conditions in consanguineous families with affected children.

WHAT DOES THIS STUDY ADD?

- Our findings present the attitudes, hence acceptability, of prenatal diagnosis and termination of pregnancy in British Pakistani parents and relatives of children with recessive genetic conditions.

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