GPs’ opinions of their role in prenatal genetic services: a cross-sectional survey

Nadeem Qureshi, Sarah Armstrong and Bernadette Modell


**Background.** In the UK about 4.5% of the population carry cystic fibrosis, whilst in the inner city areas an even higher proportion carry one of the haemoglobin disorders such as thalassaemia. Couples who both carry the same recessive disorder have a 1 in 4 risk of an affected child in every pregnancy.

**Objectives.** To assess GPs’ confidence in their ability to provide initial prenatal advice for couples carrying common autosomal recessive disorders (either the cystic fibrosis or thalassaemia gene), and their opinions of different approaches for referral to prenatal diagnostic services for such at-risk couples.

**Methods.** A cross-sectional postal survey of all 644 GPs in 388 general practices in Nottinghamshire. Practices were randomly allocated to receive either the cystic fibrosis or the thalassaemia scenario survey. The survey questions predominantly used six-point Likert scales to assess confidence and opinions of prenatal services.

**Results.** The questionnaire was returned by 62% (397) of GPs. Only 23% (91) were confident in providing prenatal advice to the at-risk carrier couples. GPs were more confident about advising cystic fibrosis carriers than thalassaemia carriers (*P* = 0.01). The least popular approach to prenatal service provision was direct referral to prenatal services after counselling with 52% (194) scoring this as useful, whilst 60.5% (233) of GPs scored referral to the obstetric services with the prenatal diagnosis organised by the obstetrician as useful.

**Conclusions.** GPs perceive that they lack the confidence to provide basic prenatal genetic advice to women at risk of the commonest recessive disorders, with particularly low confidence where the couple both carry thalassaemia. A significant knowledge gap was demonstrated by the poor awareness of the importance of rapid referral to prenatal diagnostic services.

**Keywords.** Family practice, medical genetics, prenatal diagnosis, survey, thalassaemia.

**Introduction**

Screening for risk of genetic disorders is becoming increasingly relevant in primary care. This includes identifying women at risk of giving birth to children with inherited disorders. Cystic fibrosis is the commonest severe autosomal recessive disorder among North Europeans, with a carrier rate of 4.5% in the British population. In the inner cities, the haemoglobin disorders (sickle cell disorders and thalassaemia) are even commoner, with carrier rates varying from 3 to 25% depending on ethnic group. Couples who both carry the same recessive disorder have a 1 in 4 risk of an affected child in every pregnancy.

In the UK screening for reproductive risk of Down’s syndrome and the haemoglobin disorders has long been standard practice in many areas, but there is a lack of consistency in the delivery of these services. The UK National Screening Committee is now developing national programmes to ensure equitable delivery of screening for these disorders. Further, antenatal screening for cystic fibrosis is supported in the USA. At present, in the UK these programmes are based on antenatal and/or neonatal screening within maternity
services, with minimal involvement of the general practice team. On the other hand, there is evidence that the participation of primary care is essential for the effective delivery of antenatal genetic screening. For example, most at-risk couples can only access first trimester prenatal diagnosis if their GP recognises their reproductive risk and refers them directly to specialist prenatal diagnostic and counselling services early in pregnancy.

This survey was designed to investigate GPs’ attitudes to involvement in initial prenatal genetic services. The results of a simultaneous investigation of how continuing medical education in genetics and familial cancer services should be provided are reported elsewhere. 

Methods

The surveyed area was the county of Nottinghamshire in central England served by a single tertiary genetic referral centre and with direct access to secondary care prenatal diagnosis and counselling services. Thus local GPs can access these facilities without the need to refer via Obstetric or Genetic services. The county has been providing cystic fibrosis neonatal screening for many years. Maternity services provide screening for haemoglobin disorders, and local haematology laboratories prompt GPs to consider screening for haemoglobin disorders if microcytosis is detected. Within the county, there is a heterogeneous group of general practices from single-handed inner city practices to six-partner rural practices.

The initial draft of the questionnaire was developed through a review of the genetic survey literature, as well as themes and models of genetic service provision that emerged in focus groups with British GPs and with professionals. To maximise response rate, the questionnaire was designed so that it could be quickly completed at one sitting without the need to refer to other resources, such as practice records or patient notes, and questions were presented through scenarios that may be seen in a normal GP’s surgery. The piloting stage included both face-to-face interviews with GP volunteers and a pilot postal survey.

The first section of the questionnaire addressed GPs’ involvement in prenatal genetic counselling services. The general practices were randomly allocated to receive questionnaires including either a cystic fibrosis or a thalassaemia scenario, as follows:

**Cystic fibrosis scenario**
A young couple joins your list. In their general practice records both are noted to be carriers of cystic fibrosis (based on a simple saliva test). The woman is at present 6 weeks pregnant. She has a 1 in 4 chance of a child with cystic fibrosis.

**Thalassaemia scenario**
A young Indian couple joins your list. In their general practice records both are noted to be carriers of thalassaemia (based on a simple blood test). The woman is at present 6 weeks pregnant. She has a 1 in 4 chance of a child with thalassaemia.

These scenarios were followed by a question asking the GP to indicate how confident he/she would feel in explaining the option of prenatal diagnosis to the couple. This was scored on a six-point Likert scale from ‘completely confident’ (scored 1) to ‘not at all confident’ (scored 6). The approach has been adopted in previous postal genetic surveys of primary care professionals. 

To underline the relevance of the topic to general practice, the scenarios were anchored to prenatal advice on Down’s syndrome risk, an area of prenatal diagnosis that GPs are more likely to come across. An associated question therefore enquired:

‘How confident would you be in explaining the option of prenatal diagnosis to this woman [thalaassaemia or cystic fibrosis carrier scenario] compared to explaining the option of prenatal diagnosis for Down’s syndrome to a 45-year-old pregnant woman?’ (More confident, as confident, less confident.)

These two questions were followed by a set of four questions about GPs’ opinions of different approaches for referral to prenatal diagnostic services, with levels of GP involvement ranging from minimal (refer at-risk couples directly to local obstetric services) to maximal (providing initial genetic counselling and obtaining telephone advice from the genetics services on appropriate referral) (Box 1). The GPs were asked to rate each model on a six-point Likert scale ranging from very useful approach to not useful at all.

**Box 1 Four different approaches for referral to prenatal diagnostic services**

<table>
<thead>
<tr>
<th>Number</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>a.</td>
<td>Refer to the obstetric services at the local hospital with the prenatal diagnosis organised by the obstetrician.</td>
</tr>
<tr>
<td>b.</td>
<td>Refer directly to prenatal services at the local hospital.</td>
</tr>
<tr>
<td>c.</td>
<td>Refer to prenatal services at the local hospital after counselling couple of their 1 in 4 genetic risk.</td>
</tr>
<tr>
<td>d.</td>
<td>Phone clinical genetics department for advice about follow up. Provide initial counselling and refer to appropriate department according to advice offered.</td>
</tr>
</tbody>
</table>

During the piloting phase GPs were reluctant to complete ranking questions which appeared to be testing knowledge. The GPs were therefore not asked to rank the different approaches, but to score each question independently.

As well as prenatal genetic services, the survey explored GPs’ opinions of familial cancer services and
their preferred approaches for genetic continuing medical education. The results are presented elsewhere.\textsuperscript{7,8} Finally the questionnaire collected data on the GP’s age, sex, country of graduation, perceived adequacy of genetic education, size and type of practice (urban or rural).

The questionnaire was sent with a covering letter from a local GP to all 644 GPs in 388 practices in the Nottinghamshire area. The county is split into 2 broad areas (predominately urban Greater Nottingham and predominately rural North Nottinghamshire). Based on 2001 Census data, the proportion of the population from ethnic minority groups at increased risk of haemoglobin disorders are 1.4\% in North Nottinghamshire and 9.1\% in Nottingham City and the surrounding Primary Care Trusts. The sampling frame was drawn from databases held by the respective Health Authority Departments of Public Health. The details held included GPs’ ages, genders and practice sizes. Reminder questionnaires were sent at 6 weeks and 10 weeks, and a post-card reminder at 12 weeks.

The data was analysed using SPSS. The asymmetrical ordinal Likert scales were analysed using the non-parametric Mann-Whitney U test. Chi-squared tests were used to assess the univariate association between categorical variables.

### Results

#### Response rate

The response rate was 62\% ($n = 397$). Responders were more likely to be under 50 ($\chi^2 = 10.173; P = 0.001$) and more likely to be female ($\chi^2 = 9.733; P = 0.002$) than non-responders. There was no difference between responders and non-responders in terms of practice size ($\chi^2 = 0.995; P = 0.608$). Just under 10\% (38) of responding GPs were overseas graduates, with 7.6\% (30) graduating outside Europe, predominately from the Indian subcontinent (26 of 30 GPs).

#### Review of all GP responders

Overall only 23\% (91) of GPs gave high confidence scores (scores 1 or 2 on the Likert scale) to giving prenatal advice for either the cystic fibrosis or thalassaemia scenario (Table 1). Further, 17\% (25) of GPs who gave a low confidence score (5 or 6) about giving prenatal advice for these conditions had as little or even less confidence in their ability to counsel a 45-year-old woman about prenatal diagnosis for Down’s syndrome.

Considering the usefulness scores GPs gave for the different service models, referring directly to obstetric services with the prenatal service organised by the obstetrician was given the highest scores (60.5\%; 233 scored 1 or 2). A smaller percentage gave a high score for direct referral to the prenatal services (56.7\%; 131). The lowest score was given to the service model requiring greatest GP involvement: 52\% (194) scored this as very useful (scores 1 or 2).

#### Comparison of cystic fibrosis and thalassaemia survey replies

GPs were significantly less confident in dealing with the thalassaemia scenario than the cystic fibrosis scenario (Mann-Whitney $P = 0.01$, difference in medians = 1, 

### Table 1

<table>
<thead>
<tr>
<th>Variable</th>
<th>Profile of ALL GPs completing survey n (%)</th>
<th>Profile of GPs completing thalassaemia survey n (%)</th>
<th>Profile of GPs completing cystic fibrosis survey n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>GPs’ confidence score</td>
<td>High (1 &amp; 2) 91 (23.2)</td>
<td>41 (18.6)</td>
<td>50 (29.1)</td>
</tr>
<tr>
<td></td>
<td>Medium (3 &amp; 4) 160 (40.5)</td>
<td>85 (38.5)</td>
<td>74 (43.0)</td>
</tr>
<tr>
<td></td>
<td>Low (5 &amp; 6) 143 (36.4)</td>
<td>95 (43.0)</td>
<td>48 (27.9)</td>
</tr>
</tbody>
</table>

### Table 2

<table>
<thead>
<tr>
<th></th>
<th>Thalassaemia survey</th>
<th>Cystic fibrosis survey</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Median</strong></td>
<td><strong>IQ Range</strong></td>
<td><strong>Median</strong></td>
</tr>
<tr>
<td><strong>Confidence in explaining prenatal diagnosis to couples</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Refer directly to hospital obstetric service (MINIMAL GP involvement)</td>
<td>2</td>
<td>(2, 3)</td>
</tr>
<tr>
<td>Refer directly to hospital prenatal service</td>
<td>2</td>
<td>(2, 3)</td>
</tr>
<tr>
<td>Refer to hospital prenatal service at hospital AFTER explain risk to couple</td>
<td>2</td>
<td>(2, 3)</td>
</tr>
<tr>
<td>Phone Clinical Genetics department and advise appropriately (MAXIMAL GP involvement)</td>
<td>2</td>
<td>(1, 4)</td>
</tr>
</tbody>
</table>

IQ Range = interquartile range, giving the scores between the 25th and 75th centile.
TABLE 3  Confidence with offering prenatal diagnosis advice for carrier couples compared to advising an ‘older’ woman about prenatal diagnosis for Down’s syndrome

<table>
<thead>
<tr>
<th>Confidence in providing prenatal advice to at-risk carrier woman</th>
<th>Type of survey</th>
<th>Profile of GPs completing thalassaemic survey (%)</th>
<th>Profile of GPs completing cystic fibrosis survey (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>More confident than Down’s syndrome advice</td>
<td></td>
<td>20 (9.0)</td>
<td>15 (8.7)</td>
</tr>
<tr>
<td>As confident as Down’s syndrome advice</td>
<td></td>
<td>59 (26.7)</td>
<td>61 (35.3)</td>
</tr>
<tr>
<td>Less confident than Down’s syndrome advice</td>
<td></td>
<td>142 (64.3)</td>
<td>97 (56.1)</td>
</tr>
</tbody>
</table>

95% CI = 0 to 1) (Table 2). Similarly, by comparison with advising on Down’s syndrome prenatal diagnosis, GPs were less confident in dealing with the thalassaemia scenario than the cystic fibrosis scenario (Table 3). This however did not reach statistical significance ($\chi^2 = 2.724$, $P = 0.10$, OR = 0.86, 95% CI = 0.97–1.51).

GP’s country of graduation, type and size of practice and sex made no difference to their confidence scores. However, perceived adequacy of genetic education was related to confidence. GPs who reported adequate genetic education at medical school were more confident to offer prenatal advice to the couple at-risk for cystic fibrosis (Mann-Whitney $P = 0.002$, difference in medians = 1, 95% CI = 0 to 1). By contrast, there was no such relationship with the thalassaemia scenario.

There was very little difference for both recessive disorders in GPs’ scores on the usefulness of different approaches for referral to prenatal services. Further there was no significant difference between the two recessive disorders for each approach (Table 2).

Discussion

Principal findings

There was a lack of confidence in providing basic prenatal genetic advice for all three genetic disorders included in the survey. GPs were less confident dealing with the thalassaemia scenario than the cystic fibrosis scenario. But also, to a lesser extent, the survey indirectly demonstrated a lack of confidence in providing prenatal advice about the risk of Down’s syndrome. In reality, Down’s syndrome counselling is more complex because it depends on age-related empirical risk assessment, whilst risk of the two autosomal recessive disorders depends on simple Mendelian calculations.

GPs scored referral to obstetric departments as slightly more useful than direct referral to prenatal services. Although this may indicate a lack of awareness of direct access to local prenatal diagnostic services, it does indicate that GPs were unaware that at-risk couples face unnecessary delay in prenatal diagnosis when referral is organised via obstetric services. In fact couples of African Caribbean and Pakistani origin are less likely to accept prenatal diagnosis unless it is available in the first trimester. The importance of early referral to prenatal services needs to be brought to the attention of GPs.

The perceived inadequacy of undergraduate and postgraduate genetic education has previously been highlighted. However, this survey also indicates that those GPs who reported adequate genetic education at medical school had greater confidence in dealing with cystic fibrosis but the same did not apply for the thalassaemia scenario. This may suggest a lack of undergraduate exposure to genetic screening for haemoglobin disorders.

Strength and weakness

We attempted to improve the content validity of the survey by constructing the questionnaire using themes derived from qualitative work with GPs on their attitude to genetics. Further, this survey was administered in an English county with a wide spectrum of practices of difference sizes and from various localities. There was also a good response rate for a survey of GPs. These factors enhance the generalisability of the study. However the age and sex response bias needs to be taken into account in interpreting these results.

Implications of the study

The survey indicates that GPs lack the confidence to provide a basic initial prenatal genetic service. However, in the near future they will be exposed to increasing numbers of antenatal genetic screening tests and, based on these tests, have to provide basic advice to pregnant women and their partners. The UK White Paper on genetic health policy identifies a role for GPs with a special interest in genetics. These specialist GPs may have a remit to support other GPs (and primary health care professionals) in developing the skills and knowledge to fulfil this antenatal screening responsibility. Ideally, genetic education and information should take place at the GP’s place of work. Educational outreach has been shown to be effective in improving the process of care. With the advent of the NHS net and the assurance that all British GPs will be given broadband access by the end of 2004, this educational outreach could be supplemented by the provision of internet resources in the GP’s consulting room. The Accessible Publishing of Genetic Information (APoGI) project (www.chime.ucl.ac.uk/APoGI/) offers such an opportunity. The APoGI materials are designed to help health professionals to provide accurate initial
counselling and advice on haemoglobin disorders, together with written information for their patients.

**Future research**

Educational outreach visits to GPs about screening for haemoglobin disorders improved the number of practices offering screening tests. A further pragmatic controlled trial needs to be undertaken to include evaluation of internet information resources, incentives to improve general practice involvement, competence of the general practice team and hard clinical outcomes. The latter outcomes include the ability of at-risk couples to make informed reproductive choices and access first trimester prenatal diagnosis.

**Acknowledgements**

The paper is based on work submitted for a Masters in Health Service Research at the Medical Research Care Unit, University of Sheffield. We would like to thank all of the course tutors.

**Declaration**

Funding: this work received a proportion of its funding from the NHS Research and Development levy; the views expressed in this publication are those of the authors and not necessarily those of the NHS executive. Ethical approval: Nottingham and North Nottinghamshire Local Medical Committees approved the study.

Conflicts of interest: None.

**References**