The views of general practitioners on community carrier screening for cystic fibrosis

MARY BOULTON

CAROLE CUMMINGS

ROBERT WILLIAMSON

SUMMARY

Background. *Recent developments in molecular genetics have made it possible to identify carriers of the cystic fibrosis (CF) mutation, regardless of family history, before they have an affected child. Using these techniques, population or 'community' carrier screening can offer informed reproductive choice to individuals and couples who would not otherwise know of their risk of having a CF child.*

Aim. *This study set out to assess the views of general practitioners (GPs) on community carrier screening for CF and to consider the factors that influence their willingness to offer it themselves.*

Method. *A self-administered questionnaires was sent to all 616 GPs in four areas of North Thames (West) Region.*

Results. *Two-thirds of respondents indicated that identifying carrier couples to offer genetic counselling before conception was a very important benefit of community carrier screening. Two-thirds felt that general practice was the most appropriate place in which to offer it, and similar proportions that the most appropriate times to do so were when a close relative was diagnosed and when seeking family planning. About half wanted to offer community carrier screening themselves; this was related to experience with CF patients and CF carrier testing, and estimates of the numbers of CF carriers in the practice.*

Conclusions. *There is considerable support among GPs for community carrier screening for CF in general practice, particularly in the context of family planning services. Knowledge and experience of CF increase GPs' willingness to offer it themselves.*

Keywords: cystic fibrosis; screening, genetics.

Introduction

CYSTIC fibrosis (CF) is the most common of the severe autosomal recessive disorders in Caucasian populations. Affected individuals suffer chronic lung disease which requires lifelong treatment and considerably shortens life expectancy. In the UK, the incidence of individuals affected by CF is one in 2500 births in the Caucasian population, while the incidence of CF carriers is 100 times higher at one in 25. Carriers are at a one in four risk of having an affected child if their partner is also a carrier. As only about 10% of carriers have a family history of CF, few know they are at risk. Most children affected by CF are born to parents who were not aware that they could have a CF child.

Following the identification of the CFTR gene,[1] this need no longer be the case. It is now possible to identify about 85% of CF carriers and 70% of carrier couples so they can be informed of their risk before they have an affected child and counselled on the range of reproductive options available to them. These include assisted conception, prenatal diagnosis (with or without termination of affected pregnancies) or taking the risk of having a CF child. Evidence suggests that the majority of those identified as carriers wish to avoid the birth of a CF child,[2] although this may change if developments in clinical management offer effective treatments. For the foreseeable future, however, advances such as gene therapy are unlikely to provide a 'cure' and may prove too expensive for general use.[3]

Whatever the developments in the treatment of CF, carrier screening will remain central to informed reproductive choice. Because over 90% of carriers have no family history of CF, there is limited value in screening programmes directed at relatives of CF patients and it is generally accepted that a population approach, screening Caucasians regardless of family history, is required.[2,8] A number of studies have shown that this is acceptable to the public, administratively feasible and relatively inexpensive.[4-7] What continues to be debated is where and when such population or 'community' carrier screening should be offered.[2,8-11] Possible target times include at birth,[12] during secondary school,[7] when seeking family planning,[6] at marriage[2] and when pregnant.[13-15] Possible places include hospitals,[13,14] schools,[7] community health clinics[6] and general practice.[6,15,16]

General practice is the context for community carrier screening favoured by the lay public[5] and increasingly by other health care professionals as well.[17-19] What general practitioners' (GPs) opinions are, however, remain unclear. This paper presents the results of a survey of GPs regarding their views on community carrier screening for CF and their willingness to provide it themselves.

Method

Self-administered questionnaires were sent to all 616 GPs in four areas of the North Thames (West) Region. Fourteen were returned marked 'retired' or 'moved'; 388 were returned adequately completed—a response rate of 64% of eligible GPs. The questionnaire used mainly fixed choice answers or Likert scales. Data were analysed using SPSS for Windows; relationships between variables were assessed using Spearman's rank order correlation (r_s) and the chi-square test (χ^2).

Results

Sample characteristics

About two-thirds of the respondents were men (234, 61%). One-quarter had been a GP principal for less than 5 years (93, 24%) and half for more than 10 years (191, 50%). Forty-six (12%) were in single-handed practices; the others were evenly divided between practices of two to four partners (174, 45%) and practices of five or more partners (163, 43%). Similar proportions of respondents were in practices with a list size under 6000 patients

M Boulton, PhD, senior lecturer, Academic Department of Public Health, St Mary's Hospital Medical School, London. C Cummings, RGN, research assistant, Department of Biochemistry and Molecular Genetics, St Mary's Hospital Medical School, London. R Williamson, PhD (formerly, professor, Department of Biochemistry and Molecular Genetics, St Mary's Hospital Medical School), professor and director, Murdoch Institute, Melbourne, Australia.

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(136, 36%), 6000–9999 patients (121, 32%) and 10 000 or more patients (128, 33%).

Compared with non-responders, the sample of responders contained a higher proportion of female GPs ($\chi^2 = 5.98$, 1 d.f., $P = 0.01$), and a higher proportion of GPs from medium-sized practices (2–4 partners) and a lower proportion from single-handed practices ($\chi^2 = 27.52$, 2 d.f., $P < 0.001$).

Experience of CF patients and carrier screening

Only half the respondents (199, 51%) currently had a CF patient registered with their practice and only a few more (215, 56%) had ever cared for a patient with CF during their time as a GP. Less than one in five (70, 18%) had ever suggested that a patient be tested for CF carrier status.

Prevalence of CF carriers

General practitioners were asked for their estimates of the number of CF carriers in their own practice population. Only about one-third (124, 38%) gave an estimate that was within the range that would be expected on the basis of epidemiological evidence, i.e. between 3 and 5% of their practice list size; over half (197, 60%) gave a low estimate of less than 3%; and only seven (2%) gave a high estimate of over 5%.

Benefits and problems of carrier screening

General practitioners were asked how important they thought were four benefits and three problems associated with community carrier screening. Two-thirds (248, 67%) indicated that identifying carrier couples to offer genetic counselling before conception was a very important benefit (Table 1).

Table 1. General practitioners views on the importance of selected benefits and problems of community carrier screening for those with no family history of CF.

	Not at all important	Minimally important	Moderately important	Very important
Benefits				
To inform individuals about themselves	46 (13%)	133 (36%)	129 (35%)	59 (16%)
To identify carrier couples to offer genetic counselling before conception	5 (1%)	27 (7%)	91 (25%)	248 (67%)
To identify pregnant women at risk, to offer prenatal diagnostic tests	10 (3%)	46 (13%)	130 (35%)	181 (49%)
To identify affected fetuses, to offer early termination	23 (6%)	53 (15%)	143 (40%)	141 (39%)
Problems				
Increased anxiety in those identified as carriers	7 (2%)	59 (16%)	174 (47%)	131 (35%)
Labelling those identified as carriers as 'abnormal' or 'ill'	22 (6%)	94 (25%)	163 (44%)	91 (25%)
Inappropriate reassurance to 'false negatives'	7 (2%)	94 (26%)	172 (47%)	90 (25%)

Organization of carrier screening

General practitioners were given a list of possible times and places for conducting CF carrier screening, and for each, were asked to indicate up to three which they felt were most appropriate. Two-thirds (261, 67%) felt that general practice was an appropriate place in which to conduct CF carrier screening (Table 2). Similar proportions felt that appropriate times for screening were when a close relative had been diagnosed (252, 65%) and when seeking family planning (244, 63%) (Table 2).

Willingness to provide carrier screening

When asked whether they would like to include community screening for carriers of CF as one of the services they offered to patients in their own practices, almost half (172, 45%) said they would; 40% (153) were uncertain and 15% (58) did not want to provide screening.

Respondents who currently had a CF patient registered with their practice were more favourable towards screening (50, 34 and 16%; $\chi^2 = 8.27$, 2 d.f., $P < 0.05$), as were those who had already tested patients (57, 37 and 6%; $\chi^2 = 8.13$, 2 d.f., $P < 0.05$) and those who gave higher estimates of the number of carriers in their own practice ($r_s = 0.1890$, $P < 0.001$).

Discussion

This paper describes the results of a survey of GPs regarding their views on what may be the first population screening programme for carriers of a genetic condition offered to Caucasians. The response rate of 64% is good for a postal survey but, with an over-representation of female GPs and an under-representation of single-handed GPs, it is possible that those who returned the questionnaire had a greater interest in genetic screening than those who did not.

Overall, the respondents held essentially positive attitudes to community carrier screening for CF. They valued its benefits and saw general practice as an appropriate place in which to offer it. Almost half wanted to undertake it themselves, and while almost as many were uncertain, only a small minority were opposed to it. These findings are consistent with those of a previous study[20]

Table 2. General practitioners' views on the best times and places to offer CF carrier screening. (Totals are over 100% as GPs were asked to indicate up to three choices.)

Views	Number (%)
What do you consider the best time to offer CF carrier screening?	
At birth	99 (26%)
As a teenager	115 (30%)
When seeking family planning services	244 (63%)
During pregnancy	95 (25%)
When a close family relative is diagnosed with CF	252 (65%)
On request	172 (44%)
Which setting do you think that community carrier screening for CF is most appropriately conducted through?	
General practice	261 (67%)
Community health clinics (e.g. family planning clinics)	144 (37%)
School health services	112 (29%)
Hospital antenatal services	143 (37%)
Hospital paediatric services	72 (19%)
Regional genetic services	139 (36%)
Workplace screening programmes	64 (17%)

and suggest that there is widespread support for community carrier screening for CF among general practitioners.

Carrier screening was most highly valued for the opportunity it affords to identify carriers and carrier couples before they conceive, and was seen as most appropriately offered when individuals are seeking family planning advice and when they had a relative who was diagnosed with CF. It is perhaps not surprising that GPs support screening that is targeted at individuals who are known to be at high risk of having a CF child because of their family history. If testing is limited to such patients, however, it will be of little value in allowing informed reproductive choice for the vast majority of carriers and carrier couples.[2,8,11] Therefore, it is encouraging that GPs also wish to screen those with no family history of CF, particularly in the context of family planning services where a discussion of reproductive risks is clearly relevant. Previous studies have found similar support for community carrier screening in this context among both health professionals[20] and the lay public.[6] In contrast, few respondents saw pregnancy as an appropriate time or antenatal clinics as an appropriate place for screening, although currently community carrier screening for CF is most commonly conducted in this way.[13-15,22]

Although the great majority of respondents supported community carrier screening in principle, almost half were uncertain about whether they wanted to provide it themselves. This may be partly because CF is seen as a 'rare' condition. Cystic fibrosis may be the most common of the severe autosomal recessive disorders in the UK, but almost half the respondents had never cared for a patient with CF. As one said, 'In 30 years in practice with 10 000 patients, I have yet to see a single case.' Similarly, the majority of respondents estimated an improbably low number of carriers in their practice. Any plans to develop community carrier screening through general practice will clearly need to include appropriate training for GPs in the epidemiology and population genetics of CF.[23]

The identification of the gene defect which causes CF was a significant milestone for the 'new genetics'. With the rapid advances now being made, the genes associated with other disorders may soon be identified. Because carrier screening for CF is likely to provide a model for screening for other conditions, the way in which it is eventually organized will have major implications for the National Health Service. General practice is currently among the places most widely favoured for conducting community carrier screening.[2,17-19] The findings of this study suggest that, particularly in the context of family planning services, there is substantial support for it among GPs themselves

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Address for correspondence

Dr Mary Boulton, Academic Department of Public Health, St Mary's Hospital Medical School, Norfolk Place, London W2 1PG.