Attitudes to carrier screening for cystic fibrosis: a survey of health care professionals, relatives of sufferers and other members of the public

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SUMMARY. The gene which is mutated in cystic fibrosis has now been identified, thus permitting the detection of carriers in the general population. This paper reports pilot surveys in the North West Thames region of the health service to assess knowledge of people about cystic fibrosis and their attitudes towards screening. Three groups were surveyed: a group of relatives of those with cystic fibrosis (n = 268), a sample of the community (school pupils and family planning clinic attenders, n = 363), and a group of health care professionals (general practitioners and family planning clinic staff, n = 227).

The relatives of cystic fibrosis sufferers were unanimously in favour of the introduction of cystic fibrosis screening, and the results indicate that there is likely to be support from the relevant health professionals: approximately 75% of respondents in the group of health care professionals believe the introduction of screening would be worthwhile. Data from the community sample suggest that, although knowledge of cystic fibrosis within the general community is low (less than 50% of respondents realized that cystic fibrosis affects the lungs and that no cure is available), there is likely to be considerable demand for carrier testing from the general public. Approximately 75% of the community sample indicated that they would like to be tested. There was no clear consensus, either from the professionals or the public, as to the best time to offer screening.

Introduction

CYSTIC fibrosis is the most common of the severe autosomal recessive diseases of childhood affecting north Europeans. The birth incidence is approximately one in 2000. Treatment is improving, but the disease is still severe in most cases, and the average life expectancy is less than 25 years. The gene which is mutated in cystic fibrosis has recently been identified,[1-3] and in the UK 78% of cystic fibrosis chromosomes have the single, common three base pair deletion DF508.[4] Several other mutations have also been identified, some of which occur in the UK population at lower levels; at present, greater than 85% of the mutations causing cystic fibrosis in Britain can be identified, and screening could therefore detect 72% of carrier couples at a one in four risk of having a child with cystic fibrosis (0.85 x 0.85 = 0.72).

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Population screening for carriers of recessively inherited diseases in the UK at present exists only for ethnic minority groups: sickle cell anaemia for blacks, thalassaemia for the Cypriot and Asian community, and Tay-Sachs disease for those of Jewish ancestry. These programmes, while not without problems, have in general been successful.[5]

Carrier testing for cystic fibrosis meets the basic criteria set for screening programmes.[6] The disease is serious and treatment is not wholly effective. Testing can be performed rapidly and inexpensively using DNA extracted from a buccal swab, mouthwash or small blood sample using gene amplification by polymerase chain reaction.[7] For those who are identified as carriers, the results are unequivocal. The fact that only 85% of carriers can be identified at present, however, poses counselling problems, particularly for couples where one partner is positive and the other negative for the currently known mutations; for this reason, pilot studies rather than full community screening are currently recommended by a working party of the National Institutes of Health in the United States of America.[8]

There are several possible strategies for community screening, which are not necessarily mutually exclusive:

First, there is neonatal screening, which would be relatively easy to implement technically, but would be inefficient in reducing the incidence of cystic fibrosis. One in four carrier couples would already have an affected child before being identified as carriers, and a further 25% would not be identified because their child had inherited neither cystic fibrosis gene. There is also the problem of ensuring a carrier child receives full information regarding this result at an appropriate age.

Secondly, antenatal screening has the potential advantage of identifying all at risk couples. However, the disadvantages are considerable. Anxiety could be caused to all women identified as carriers, of whom only one in 20 would actually be at risk of having an affected child. Late booking in at the antenatal clinic restricts the use of chorion villus sampling, which in turn could lead to a large number of second trimester diagnoses. The short interval between testing and decision-making leaves little time for reflection, particularly as the majority of such couples will have no previous experience of cystic fibrosis.

Thirdly, preconception screening could involve testing either through schools or primary health care services (general practitioners and family planning clinics). Screening of individuals of reproductive age prior to pregnancy gives carrier couples a chance to consider the maximum range of reproductive options, including (among other options) prenatal diagnosis followed either by termination of affected pregnancies, or by early treatment of affected persons with cystic fibrosis from birth with associated improved prognosis.[9]

Educated professionals and a well informed community are a prerequisite for effective screening. The present study was an attempt to assess knowledge of cystic fibrosis and attitudes to screening both in the community and among primary health care professionals. We also wished to investigate opinions regarding the preferred time to make screening available.

Three questionnaire surveys were undertaken: one of health care professionals — general practitioners and family planning clinic staff — whom we view as key professionals for a com-

munity based screening programme; one among relatives of those with cystic fibrosis; and one in the general community (this last component extends a previous pilot study which attempted to assess attitudes of general practice patients and school children[10]).

We did not feel justified in alerting women in the antenatal or neonatal clinic to the risks of cystic fibrosis without offering a test and have therefore limited our community study to two specific groups: school pupils aged 16 years and over and women attending a family planning clinic. Both groups are easy to target and might be considered as suitable for initiating a population screening programme.

Method

Survey of health care professionals

Questionnaires were sent with reply envelopes to all general practitioners in two districts of North West Thames regional health authority, one in central London (North Parkside) and one suburban (North Bedfordshire) and to family planning clinic staff (general practitioners and nurses) in three districts (North and South Parkside and North Bedfordshire). The questionnaire asked about attitudes towards the introduction of population screening to detect carriers of cystic fibrosis. Responses were achieved from 227 people, a response rate of 52%. This level of response, although lower than, for example, a postal survey regarding the role of primary care in the management of human immunodeficiency virus (HIV) infection[11] is a typical response rate for a postal questionnaire distributed to general practitioners.[12,13]

Survey of relatives of cystic fibrosis sufferers

The Cystic Fibrosis Research Trust posted out a questionnaire to everyone on their mailing list in the North West London branch (242 individuals). The mailing list includes parents and relatives of cystic fibrosis sufferers and adults with cystic fibrosis. The questionnaire asked about attitudes towards population carrier screening for cystic fibrosis and how knowledge of carrier status may affect reproductive plans. We received 268 replies (from 92 parents, 32 siblings, 38 grandparents, 60 aunts/uncles, 21 cousins, two husbands, one affected individual and 12 family friends). The excess of replies can be accounted for by families copying the original questionnaire and distributing it to other relatives/close friends.

Survey of a community sample

Our community sample comprised two groups. First, a group of 177 school pupils (aged 16 years or over) attending one of eight different inner London schools. We wrote to the head of biology at several secondary schools within a 10 mile radius of St Mary's Hospital. In each case we were granted access to the school. Although we emphasized that we wished to see a range of pupils, in practice it was difficult to gain access to classes other than biology. We spoke to biology classes of different levels in seven schools and to the entire sixth form at one school. The sample is biased towards women and relatively high academic achievers.

Secondly, a sample of 186 consecutive attenders at a suburban family planning clinic (Stanmore) were surveyed over a three month period. This group was a representative sample of that subset of the population.

The questionnaire was in two parts; the first assessed knowledge of cystic fibrosis and the second addressed attitudes towards screening. Prior to answering the second part all school pupils were shown a short video on cystic fibrosis and were involved in a general class discussion. Care was taken to standardize this discussion which was led by the same individual in each case. The family planning clinic attenders were provided with an information sheet on cystic fibrosis. Participation in the survey was voluntary. No school pupils declined to participate, and only 2% of the family planning clinic attenders refused.

The characteristics of the community groups interviewed are shown in Table 1.

Table 1. Characteristics of the two samples from the community.

	School pupils ($n = 177$)		Attenders at family planning clinic ($n = 186$)	
Sex				
Male	47	*(27)*	0	*(0)*
Female	130	*(73)*	186	*(100)*
Ethnic origin				
Caucasian	126	*(71)*	108	*(58)*
Asian	23	*(13)*	16	*(9)*
Black	11	*(6)*	6	*(3)*
Unclassified	17	*(10)*	56	*(30)*
Age (years)				
Under 20	177	*(100)*	32	*(17)*
20–50	0		147	*(79)*
Unclassified	0		7	*(4)*
Education				
A level/GCSE	165	*(93)*	No data	
Private education	52	*(29)*	No data	
State education	125	*(71)*	No data	

Results

Health care professionals

Table 2 shows that approximately 75% of both general practitioners and family planning clinic staff thought that the introduction of cystic fibrosis carrier testing was appropriate, and less than 10% were opposed to it. Of the family planning clinic staff 76% were willing, in principle, to participate in pilot studies on this subject as were 50% of general practitioners. Seventy per cent of general practitioners and 57% of the family planning

Table 2. Attitudes towards cystic fibrosis carrier screening among health care professionals.

	Number (%) of respondents			
	General practitioners ($n = 178$)		Family planning clinic staff ($n = 49$)	
Support cystic fibrosis screening	129	*(72)*	38	*(78)*
Do not support cystic fibrosis screening	14	*(8)*	5	*(10)*
Believe best time to screen would be:				
At birth	58	*(33)*	7	*(14)*[a]
At school	45	*(25)*	17	*(35)*
Before marriage/planning family	69	*(39)*	30	*(61)*
During pregnancy	6	*(3)*	1	*(2)*
Would feel happy counselling cystic fibrosis families	124	*(70)*	28	*(57)*
Interested in participating in pilot study	89	*(50)*	37	*(76)*

[a] Six of the clinic staff ticked two options for the question regarding the best time to screen. n = total number of respondents.

clinic staff felt they would be happy counselling cystic fibrosis families.

There was no clear agreement among health care staff as to the best time to screen for carrier status, apart from a bias against screening during pregnancy.

Relatives of cystic fibrosis sufferers

The response of the parents and other relatives were in complete accord, and will be outlined together. Every respondent thought that carrier testing would be a good thing and 89% would wish to be tested themselves. Over 85% thought that testing should be made available on a voluntary basis. There was a division of opinion as to the best time to offer carrier testing, with equivalent numbers favouring testing at birth or before marriage/when planning a family (Table 3). Ninety per cent would request prenatal diagnosis of a high risk pregnancy, although approximately one in five stated that they would not consider termination and a further one in five were unsure. One in three would consider not having children (Table 4).

Table 3. Attitudes towards cystic fibrosis carrier testing among relatives of sufferers and other groups in the community.

	Number (%) of respondents		
	Relatives of cystic fibrosis sufferers (*n* = 268)	Family planning clinic attenders (*n* = 186)	School pupils (*n* = 177)
Believe carrier testing would be a good thing	268 *(100)*	167 *(90)*	145 *(82)*
Would want a test	238 *(89)*	140 *(75)*	136 *(77)*
Believe testing should be voluntary	231 *(86)*	152 *(82)*	147 *(83)*
Believe best time to screen would be:			
At birth	105 *(39)*	37 *(20)*	36 *(20)*
At school	39 *(15)*	35 *(19)*	67 *(38)*
Before marriage/ planning a family	116 *(43)*	97 *(52)*	74 *(42)*
No response	8 *(3)*	17 *(9)*	0 *(0)*

n = total number of respondents.

Table 4. Attitudes towards child bearing of those wishing to be tested for cystic fibrosis.

	Number (%) of respondents		
	Relatives of cystic fibrosis sufferers (*n* = 238)	Family planning clinic attenders (*n* = 140)	School pupils (*n* = 136)
Request prenatal diagnosis	214 *(90)*	135 *(96)*	124 *(91)*
Consider termination of pregnancy:			
Yes	138 *(58)*	87 *(62)*	50 *(37)*
No	42 *(18)*	14 *(10)*	31 *(23)*
Don't know	58 *(24)*	39 *(28)*	55 *(40)*
Consider not having children	89 *(37)*	50 *(36)*	20 *(15)*

n = number of respondents who would wish to be tested.

Members of the community

Table 5 shows that among the community sample 86% had heard of cystic fibrosis. However, fewer than half realized that cystic fibrosis affects the lungs and that the life span of sufferers is reduced. Sixty four per cent indicated that they knew cystic fibrosis was inherited, but only 9% answered all questions correctly.

Of the 177 school pupils and 186 family planning clinic attenders, 82% and 90% respectively thought that carrier testing would be a good thing, and 77% and 75% stated they wished to be tested themselves (Table 3). Of those wishing to be tested, over 90% in each group stated they would request prenatal diagnosis for a high risk pregnancy, although one in five school pupils and one in 10 of the family planning clinic attenders would not consider a termination. Over half of the family planning clinic group would consider a termination. There were no significant differences between responses from men and women.

When asked about the best time to screen, the largest proportion favoured offering tests before marriage or when planning a family.

The responses from men and women in these groups were similar.

Table 5. Knowledge of cystic fibrosis among school pupils and family planning clinic attenders.

	Number (%) of respondents		
	Family planning clinic attenders (*n* = 186)	School pupils (*n* = 177)	Total (*n* = 363)
Heard of cystic fibrosis	163 *(88)*	148 *(84)*	311 *(86)*
Of those who had heard of cystic fibrosis, those who knew:			
1. Incidence	53 *(33)*	30 *(20)*	83 *(27)*
2. Cystic fibrosis is inherited	88 *(54)*	109 *(74)*	197 *(63)*
3. Life span of sufferers is shortened	89 *(55)*	55 *(37)*	144 *(46)*
4. Lungs are affected	79 *(48)*	61 *(41)*	140 *(45)*
5. No cure available	80 *(49)*	65 *(44)*	145 *(47)*
Answered 1-5 correctly	18 *(11)*	10 *(7)*	28 *(9)*

n = total number of respondents.

Discussion

The relatives of those with cystic fibrosis have personal knowledge of the condition and (apart from cystic fibrosis sufferers) are those most directly affected by the disease. They responded enthusiastically to the questionnaire. Every respondent favoured carrier testing in the community; most stated that they would wish prenatal diagnosis, although almost one fifth were either opposed to or reluctant to consider termination of an affected pregnancy.

It was apparent that the health care professionals who responded were generally sympathetic to screening, and a large proportion felt competent to deliver counselling to those found to be carriers. This favourable response is vital to the success of any screening programme involving many millions of persons, which would have to be administered, at least in large measure, through primary health care services. However, it should be noted that there was a high proportion of general practitioners who did not respond to the questionnaire, and almost half of the family planning clinic staff felt unsure about giving counselling.

The great majority of those questioned in schools and when attending family planning clinics had heard of cystic fibrosis, but had at best an imperfect understanding of the nature and course of the disease. Two-thirds of those questioned knew that cystic fibrosis is inherited, but only one in 10 had a full range of accurate knowledge of the transmission and severity of the disease. These data clearly indicate that much education will be required before it can be assumed that those who may find they are carriers of cystic fibrosis have sufficient knowledge to make informed choices about child bearing. Any community-based screening programme should include provision of appropriate counselling facilities for those who are identified as carriers, in order to ensure that a correct range of options are put in a non-directive way to those at risk of having an affected child.

A large majority of those questioned supported the notion of population based carrier testing and stated a desire to be tested themselves. Caution, however, must be exercised in translating this desire into a likelihood that people will come for testing in practice; nonetheless, it does indicate a general sympathy with the concept of carrier testing in the community. Attitudes to or uptake rates of preventive screening for serious diseases affecting an individual (as with breast or cervical cancer screening), which may anyway be sub-optimal,[14,15] may not correspond to uptake rates for testing for cystic fibrosis, which has no immediate implications for the individual but poses choices about child bearing. Acceptance rates for cystic fibrosis carrier testing might be greater or less than for programmes which detect disease in those screened. The great majority of respondents agree with the profession and the World Health Organization that testing should be voluntary.

The data we have obtained indicates that knowledge of carrier status is likely to have an effect on the reproductive behaviour of individuals. The majority of those wishing to be tested would request prenatal diagnosis for a known high risk pregnancy. More than half of the relatives of cystic fibrosis sufferers would consider terminating an affected pregnancy. Among the community sample a higher proportion of the family planning clinic attenders would consider a termination following the diagnosis of cystic fibrosis in a pregnancy. By definition, the family planning clinic attenders are at risk of becoming pregnant and are a group who presumably have considered the issue of reproduction. This is not necessarily true of school pupils and it is not surprising that the largest proportion of the school sample were unsure how they would react to the diagnosis of an affected pregnancy. The results from both groups, however, should be considered in the context of the limited understanding of the nature of cystic fibrosis.

Approximately one third of the cystic fibrosis relatives and the clinic attenders felt that they might opt not to have children if they were in a high risk partnership. A lower proportion of the school sample considered this to be an option.

There was a division of opinion as to the optimal time to screen. In each group studied, approximately 50% favoured testing before pregnancy (either before marriage or before starting a family), but there were sizeable numbers supporting testing at birth and a significant proportion favouring screening at school.

We conclude that the attitudes of those with most experience of cystic fibrosis, and of those of reproductive age in the community, pose no major problem to the introduction of screening programmes which are community based. At least half of general practitioners and those who work in family planning centres would welcome the introduction of testing, and this would be critical to the success of any programme. However, in order to plan resource allocation, community education, and adequate staff and training for counselling, it would be advantageous to introduce pilot studies.

References

1. Rommens J, Ianuzzi M, Kerem BS, *et al.* Identification of the cystic fibrosis gene: chromosome walking and jumping. *Science* 1989; **245:** 1059-1065.
2. Riordan J, Rommens J, Kerem BS, *et al.* Identification of the cystic fibrosis gene: cloning and characterisation of cNDA. *Science* 1989; **245:** 1066-1072.
3. Kerem BS, Rommens JM, Buchanan JA *et al.* Identification of the cystic fibrosis gene: genetic analysis. *Science* 1989; **245:** 1074-1080.
4. Watson E, Williamson R, Brueton L, Winter R. Genetic counselling for cystic fibrosis based upon mutation/haplotype analysis. *Lancet* 1990; **336:** 191-192.
5. Modell B. Community control of hereditary anaemias: memorandum from a WHO meeting. *Bull WHO* 1983; **61:** 63-80.
6. Cuckle HS, Wald NJ. Principles of screening. In: Wald NJ (ed). *Antenatal and neonatal screening.* Oxford University Press, 1984.
7. Lench N, Stanier P, Williamson R. A simple, non-invasive method to obtain DNA for gene analysis. *Lancet* 1988; **1:** 1356-1358.
8. Statement from the National Institutes of Health workshop on population screening for the cystic fibrosis gene. *N Engl J Med* 1990; **323:** 70-71.
9. Wilken B, Chalmers G. Reduced morbidity in patients with cystic fibrosis detected by neonatal screening. *Lancet* 1985; **2:** 1319-1321.
10. Williamson R, Allison MED, Bentley TJ, *et al.* Community attitudes to cystic fibrosis carrier testing in England: a pilot study. *Prenatal Diagnosis* 1989; **9:** 727-734.
11. Roderick P, Victor CR, Beardow R. Developing care in the community: GPs and the HIV epidemic. *Aids Care* 1990; **2:** 127-132.
12. Owen P, Rutt G, Keir M, *et al.* Survey of general practitioners' opinions on the role of radiology in patients with low back pain. *Br J Gen Pract* 1990; **40:** 98-100.
13. Thomas K, Birch S, Milner P, *et al.* Estimates of a general practitioner workload: a review. *J R Coll Gen Pract* 1989; **39:** 509-513.
14. Calnan M, Rutter DR. Do health beliefs predict health behaviour? An analysis of breast self examination. *Soc Sci Med* 1986; **22:** 673-678.
15. Johnson IS, Milner PC, Todd JN. An assessment of the effectiveness of cervical cytology screening in Sheffield. *Community Med* 1987; **9:** 160-170.

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