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What do young people think about screening for cystic fibrosis?

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Abstract

We investigated the knowledge of cystic fibrosis and the views about neonatal and carrier screening in 216 school students aged 14 to 16 years. This work was completed before the published identification of the cystic fibrosis gene in September 1989. Although initial knowledge of cystic fibrosis was low (only 17% of the students knew that the disease affected the lungs), there was good recall of basic information about cystic fibrosis and of recessive inheritance after a brief lecture. A total of 86% considered that carrier detection should be offered routinely and 88% felt that an offer of prenatal diagnosis for cystic fibrosis should be made if both prospective parents were known to be carriers. We believe that pilot studies of cystic fibrosis carrier screening in schools should be undertaken.

The identification of the cystic fibrosis (CF) gene and of its major mutations[1 2] has made it feasible to screen large populations for the carrier state. In the UK around 4·6% of Caucasians are carriers and of these about 77% could now be detected by molecular genetic techniques.[3] Screening of couples before pregnancy, or in the first trimester, would therefore provide an opportunity for genetic counselling of those couples in which both partners were CF heterozygotes. This would give such high risk couples options which were previously available only to those who had an affected subject in their family.

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Although the technology is now available to perform such screening programmes, little is known of the attitudes of those in the community who might be offered such screening. Williamson *et al*[4] reported the results of a community survey of 166 persons sampled from two general practices, a family planning clinic, and two schools in or around London. They found that over 80% would be interested in knowing their CF carrier status. The ages ranged widely and 24% of those questioned were above child bearing age. Their data suggested that the school children knew less about CF and were slightly less likely than older people to wish for carrier screening. Ten Kate and Tijmstra[5] reported that of 100 second year medical students 96% considered that carrier screening for CF should be offered while 80% of 127 mothers who had just had their first baby were in favour of such screening.

We now report a study of 14 to 16 year old school children in Edinburgh which was carried out some months before the isolation of the CF gene by Tsui and his colleagues.

Materials and methods

In Edinburgh there are 35 secondary schools (14 fee paying and 21 state funded). Fifteen head teachers were asked by letter if they would permit their school to take part in this project and 11 responded positively. From these, six schools were chosen so that our sample of students included those at three fee paying schools (one all male, one all female, and one mixed) and at three state schools (all mixed sex). We were not allowed to include a Roman Catholic school, because the group we wished to study had not yet been taught about the issue of abortion and therefore discussion of prenatal diagnosis was considered by the head teachers to be unsuitable.

The schools chosen to take part in the study were asked if a random selection of pupils in the third year could be offered questionnaires investigating their knowledge, experience, and understanding of CF as a genetic disease and also of their opinions regarding carrier screening, prenatal diagnosis, and neonatal screening. The initial questionnaire, testing basic knowledge, was administered before a short lecture by one of us (EC) covering genetic principles and also

information about CF and neonatal screening. The recall and opinions of the group on neonatal screening were checked with a second questionnaire and then a further talk was given about the present and future possibilities for carrier screening and prenatal diagnosis. A third questionnaire examining attitudes to these possibilities then followed.

Results

In total, 216 third year pupils attending the six schools filled out the three questionnaires: the ages ranged from 14 to 16 years. Students were eliminated from the two individual questionnaires asking about opinions on (1) neonatal screening and (2) carrier screening if they showed that they had not grasped the salient points regarding CF and methods involved in screening. Of the original 216 questionnaires, 166 were included for analysis of opinions regarding neonatal screening and 154 were left in the group concerning carrier detection and prenatal diagnosis.

Table 1 summarises the data from the 216 study pupils and their initial knowledge, particularly about CF. A low percentage (17%) had any real knowledge of CF before the lecture, but 75% had heard of the condition. Table 2 shows the change observed immediately after the short lectures. After the short talks on cystic fibrosis, 88% knew that to be affected a subject required two recessive genes, one from the mother and one from the father, 98% knew that carriers had no disease, and 99% knew that the average lifespan was considerably reduced.

Table 3 shows the views about neonatal screening. A very large majority considered that neonatal testing was a good idea and that it should be routine. Of the pupils who understood enough about CF, 95% were in favour of neonatal screening.

Table 4 shows that of all the pupils questioned, 86% thought that routine carrier screening should be offered and 87% thought that prenatal diagnosis should be offered if both parents were found to be carriers. Of the 71% who understood the genetic principles behind carrier detection and the risks of amniocentesis, 86% were for routine carrier screening and 88% were for offering prenatal diagnosis to carrier parents.

Table 2 Genetic/CF knowledge immediately after lecture.

	No	%
Understood what recessive genes were	189	88
Knew that carriers were not affected	212	98
Knew that lifespan is much reduced	213	99

Table 3 Views on neonatal testing for CF.

	No	%
Approved of neonatal screening	207	96
Thought neonatal screening should be routine	204	94
Understood about CF	174	94
Pupils who understood CF and thought that neonatal screening should be routine	166	95

Table 4 Views on carrier detection and prenatal diagnosis.

	No	%
All pupils		
Thought routine carrier screening should be offered	186	86
Thought prenatal diagnosis should be offered if both parents are carriers	187	87
Understood sufficient about carrier screening and prenatal diagnosis	154	71
Pupils with adequate understanding		
Carrier detection should be routine	133	86
Prenatal diagnosis should be offered if both parents carriers	135	88

Tables 2, 3, and 4 do not show the results from individual schools since none of them differed significantly from any other (χ^2 $p>0{\cdot}1$).

Discussion

Our study shows that knowledge about CF of third year school students is minimal. Only in those few who knew of an affected person was there any understanding of the condition, and this was slight. Haydon *et al*[6] found that even CF sufferers and their sibs were poorly informed about the condition. Thus there is an essential need for community education to cover the problems of CF before any population screening is started.[5 7–9]

Table 1 CF knowledge of third year pupils in six Edinburgh schools.

School	No	Females (%)	Heard of CF	Knew CF affected lungs	Knew someone with inherited disease
A Fee paying	24	0 (0)	18 (75)	0 (0)	8 (33)
B State funded	9	5 (56)	6 (67)	1 (11)	3 (33)
C State funded	22	6 (27)	13 (59)	2 (9)	5 (23)
D Fee paying	89	89 (100)	71 (80)	14 (16)	36 (40)
E State funded	29	11 (38)	15 (52)	2 (7)	6 (21)
F Fee paying	43	22 (51)	38 (88)	18 (42)	13 (30)
Total (%)	216	133 (62)	161 (75)	37 (17)	71 (33)

Those who were in the third year classes at the time of our study will now be approaching the stage when carrier screening might be offered, either in a pre-conception situation at health centres and family planning clinics, or during the early stages of a pregnancy. Our main findings suggest that these subjects would be in favour of both neonatal screening for CF and also of screening for carriers. We feel that, in the hypothetical situation which we presented, the influence of family pressures and attitudes was reduced. Also, in our study, the attitudes were not influenced by the actual availability of a carrier test or by the imminent need to decide about prenatal diagnosis, as would occur in young pregnant women. There would seem to be a very positive attitude to carrier screening and to the offer of prenatal tests of those couples shown both to be carriers.

Before setting up carrier screening programmes for carriers of Tay–Sachs disease in Montreal, pilot programmes were established to determine the most effective approaches.[10] Community leaders helped with such studies to determine the expectations of this type of preventative medicine, taking into account the previous education of the community and the need for follow up counselling. A study of haemoglobinopathies by Scriver *et al*[11] showed that the highest response to carrier screening was by school pupils, and Williamson *et al*[4] reported that 77% of school pupils (aged 16 to 18) favour carrier testing. Therefore, we think that the later school years would be the best time to offer carrier detection for cystic fibrosis, rather than at antenatal clinics, as suggested for the thalassaemias by Mouzouras *et al*.[12] However, since most pupils would be under the age of 18 when offered testing, their parents and also the wider community would have to be involved.

Taken as a whole, our results, and those of Williamson *et al*[4] and of Ten Kate and Tijmstra,[5] are comparable to those obtained in the Montreal schools when views about screening for Tay–Sachs disease were investigated.[10] They also accord with the views of many geneticists who have tested parental attitudes to carriers tests. Our experience in Edinburgh schools has drawn our attention to another important factor, the positive role of the school teacher in facilitating this type of community education. Not only were the schools who took part in this survey helpful in arranging for us to visit and to offer students the opportunity to take part, but several schools suggested that we spoke to other pupils who were more specifically interested in biological topics. Although these could not be included in the study, the potential for community education in the schools before the development of screening programmes is encouraging.

The most common mutation of the CF gene can be screened for using a non-radioactive technique.[13] However, only 70% of CF carriers would be identified by this test and perhaps a further 10% by tests for less frequent mutations. A further element must therefore be introduced into community teaching, that is, that not all carriers are identified by current screening methods. This fact can be contrasted with the present situation in the absence of carrier screening. Although 1 in 20 of the population are carriers, only one couple out of 1600, or less, will know of this after the neonatal testing of their first child. Using the screening tests available now, and assuming an 80% take up of these tests and a 70% identification of carriers, from 1600 couples offered testing, 45 carriers would be identified and also two or three couples with a 1 in 4 risk of having a child with CF. Clearly counselling support would be required for the couples in which one or other was a carrier and the greatest support must be directed at those couples where both are carriers.

We believe that the community will accept such carrier testing and, provided that the counselling support available is adequate and non-directive, we recommend that pilot screening should be undertaken.

It is a pleasure to record the help we have had from the schools who took part and from many individual teachers and pupils.

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