LETTERS TO THE EDITOR

Cystic fibrosis screening and community genetics

In the excellent review by Modell (*J Med Genet* 1990;27:475-9), she suggests that an immediate question is "How acceptable is prenatal diagnosis for CF to the families at risk?". Stimulated by her article we mailed out a questionnaire to the membership of the NSW Cystic Fibrosis Foundation to try to answer this question. Of the 471 questionnaires mailed out, 227 (48.2%) were returned. Four were from persons with CF (all were in favour of prenatal diagnosis) and the rest were replies from parents from 223 different families. In NSW a neonatal screening programme has been in operation since 1981, prenatal diagnosis by amniotic fluid enzyme analysis since 1985, and by RFLPs since 1988. A summary of the questionnaire follows.

Question I	Would members of your family be interested in carrier detection?	Yes 99.5% No 0.5%
Question II	It is generally agreed that children should not be tested until they themselves can consent to testing and understand the information they receive. At what age do you think this is?	See figure
Question III	asked for a list of their children and the age of diagnosis of CF	
Question IV	listed a choice of reproductive options and asked for an opinion of which they chose or might have chosen	See table

Reproductive choices.

Had no further children	121
Would like or would have liked prenatal diagnosis	78 (64.5%)
Would not like prenatal diagnosis	13 (10.7%)
Did not give an opinion	30 (24.8%)
Had further children after birth of proband	102
Monitored by prenatal diagnosis	23 (22.6%)
Would have liked prenatal diagnosis	31 (30.4%)
Would not have liked prenatal diagnosis	16 (15.7%)
Had conceived before diagnosis of proband	16 (15.7%)
Had an unaffected child by other means: adoption, artificial insemination, another partner	13 (12.7%)
Did not give an opinion	3 (2.9%)

Age at which respondents thought children could be tested.

The results show that in those who expressed an opinion, 83% wished for prenatal diagnosis whereas 17% did not. No question was asked about termination deliberately, but two respondents wrote that they would wish for prenatal diagnosis but would not want a termination.

We have no information about the selection bias introduced by being a member of the CF Foundation or by returning or not returning questionnaires, but at least the membership is a well informed group on the newer developments through the CF Newsletter.

This study provided some answers to the questions posed by Dr Modell and may be of help to those genetic departments seeking funding for family and community screening programmes.

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False positive results with immunoreactive trypsinogen screening for cystic fibrosis owing to trisomy 13

In Northern Ireland cystic fibrosis, the commonest autosomal recessive disorder, has a prevalence rate of 1 in 1850 livebirths.¹ Since August 1983, it has been routine to screen all infants using immunoreactive trypsinogen (IRT) activity on dried blood spots taken between 5 and 8 days of age.² Since that time, nine trisomy 13 infants have been born, of whom three survived long enough to have IRT screening for cystic fibrosis. Two had positive IRT results and one had a high normal value.

The first, a male born in 1987, had IRT values of 81 µg/l and 118 µg/l (upper limit of normal range 45 µg/l). Sweat testing gave normal results. At the age of 20 months, he is still alive with marked neurological difficulties but no clinical evidence of cystic fibrosis. The second, a male born in 1990, had IRT activities of 62 µg/l and 65 µg/l (upper limit of normal range 45 µg/l). He died aged 10 weeks before sweat