Testing carrier status in siblings of patients with cystic fibrosis

Ian Balfour-Lynn, Su Madge, Robert Dinwiddie

Abstract
Altogether 114 parents of patients attending a cystic fibrosis clinic and 27 regional genetics units were surveyed for their views on whether healthy siblings should be tested for carrier status during childhood. Most parents wanted to know their child’s carrier status and felt it was their right; almost all would tell the children if they were carriers. However, 37% of the units never tested siblings and 40% said the parents had no right to this knowledge. Furthermore, 60% would withhold the information from parents.

Keywords: cystic fibrosis, screening, carrier state.

Unaffected siblings of children with cystic fibrosis have a two in three chance of being carriers of the recessive gene. Depending on the genotype of the index case, it is often possible to determine the carrier status of these siblings. Although there is agreement that carrier testing should be offered to families of patients with cystic fibrosis, opinions vary as to whether this should be performed on children too young to be involved in the decision. The recent report from the Clinical Genetics Society (UK) was not in favour of testing children for recessive disorders that was of purely reproductive significance to that child in the future. This study was designed to elicit the views of parents of children with cystic fibrosis over this issue.

Methods
Parents of patients with cystic fibrosis attending the cystic fibrosis clinic at Great Ormond Street Hospital for Children were approached directly and asked to fill in a questionnaire. A separate questionnaire was sent to the heads of department of the 30 regional genetics services listed in the Cystic Fibrosis Trust booklet entitled Genetics, carrier tests and tests during pregnancy.

Results
Parents of 115 children were approached and 114 agreed to fill in the questionnaire (113 parents, one grandparent). The one refusal was from a father who was thought to have difficulty reading. Ninety eight per cent understood the term ‘cystic fibrosis carrier’ and 86% had thought about the issue of carrier testing before. Of those who had other children (n=102), 33% already knew the siblings’ carrier status. Results from parents are shown in tables 1 and 2. Parents were also asked who had the most rights over knowledge of a child’s carrier status: 11% thought the parents, 14% the child, 67% that parents and child had equal rights, 2% thought doctors, 3% were unsure, and 5% didn’t answer.

Out of 30 genetics units contacted, replies were received from 27 (90% response rate); 10 units (37%) never tested siblings and 17 units (63%) did. Most units only tested after formal counselling and the impression given was that they would try to dissuade the parents or postpone testing until the child was older. The majority could not answer the questions without many additional comments; a representation of their views is given within the discussion section.

Discussion
It would appear from these findings that the parents of children with cystic fibrosis have a different viewpoint over testing the carrier status of their unaffected children from that put forward by the Clinical Genetics Society1 and most clinical geneticists we surveyed.

WHY PARENTS WANT TO KNOW
The vast majority of parents wanted to know the carrier status of their other children, and a third had already found out. This study did not ascertain their reasons, but in the case of the younger children, it is likely that it was simply for the sake of knowing. Knowledge of carrier status offers no advantage to the children as there is no risk to health from being a cystic fibrosis carrier. Information on carrier status is primarily important for informed reproductive decision making, which is not relevant until mid to late teens. Perhaps many parents feel that as they will eventually know the answer,
they might as well find out sooner. Seventy nine per cent of parents (in whom this question was applicable) thought the children themselves wanted to know their carrier status; this is not surprising considering the children belong to a family where the consequences of having a child with cystic fibrosis are only too evident.

## Conclusion

In conclusion, we have reported this study in order to stimulate further debate, particularly among clinicians looking after children with cystic fibrosis and their families. We have not said whether one side is right or wrong. It is unlikely that a consensus can be reached but each family must be taken on its own merit. As with many difficult ethical issues in clinical paediatrics, the problem still remains as to who should be making these sorts of decisions. Clearly it is important to involve all parties in the discussion and informed pretest counselling is obligatory. Although it may be prudent to wait until the child is old enough to make the decision, problems arise when parents of younger children are insistent. Determined parents will only go elsewhere to have the test done and it is likely that commercial pressures will eventually make genetic testing available both in private laboratories and from the local chemist (see a report in the Daily Express (8/8/94) entitled ‘£25 DIY test to check for deadly genes’). Further discussion of these issues is now vital.

We wish to thank all the parents and geneticists who participated in this study.