Cystic fibrosis newborn screening and detection of carriers

M Super

Detection of carriers can be managed

A decision to introduce nationwide cystic fibrosis (CF) newborn screening in the United Kingdom has now been taken, although the exact procedure is still being determined, except in Scotland where screening began in February 2003. In Scotland, as well as in a number of regions in England and Wales where ad hoc newborn screening was started some time ago, an immune reactive trypsin (IRT)/DNA system of newborn screening is used. DNA analysis is being undertaken on those above a cut off point of IRT. The multiplex DNA arm covers about 86% of the most common mutations in the native UK population. There is evidence that the more severe mutations of the cystic fibrosis transmembrane regulator (CFTR) gene are the ones most likely to cause an increase in IRT.

There are a number of advantages of IRT/DNA programmes: earlier full diagnosis within the first few weeks of life becomes possible for those in whom both mutations are found; there is a reduction in sweat testing and in the uncertainty that may follow inadequate sweat collection; also many fewer couples need to be faced with the worry that their baby might have CF than when IRT is used alone. In those from ethnic minorities where the mutation spectrum is largely unknown, one has no choice other than to proceed to sweat test in those with a persistently raised IRT.

The disadvantage of the IRT/DNA method is that the genetic analysis step results in the discovery of some healthy carrier infants, with the spectre of CF. The one was refusal by the genetic counsellor (department) to check their other children to see if they were carriers. Parents saw an inconsistency in this. One of the authors (Angus Clarke) chaired a working party for the Clinical Genetics Society on genetic testing in children. The tone of this report was generally against such testing, suggesting that children should make their own decision on whether to be tested when of an age to do so. It is refreshing to note that the authors are prepared to question their current practice in response to strongly expressed wishes of parents. The Genetic Interest Group (GIG) have long campaigned on behalf of parents in this respect, adopting the attitude expressed by many of their members, “Parents generally make decisions for the benefit of their families and children.”

The other negative aspect that parents found most difficult was the suggestion that they contact their relatives to inform them of their potential carrier status. The authors suggest that their reluctance to do so signifies dysfunctional family relationships. I disagree with them on this and find the parents’ behaviour understandable. They are in a state of grace after the birth of their healthy baby and are understandably

Abbreviations: CF, cystic fibrosis; IRT, immune reactive trypsin; CFTR, cystic fibrosis transmembrane regulator

www.archdischild.com
reluctant to cast a shadow over this. Their situation is entirely different from that of the parents of a child affected by the disease CF. Although active cascade testing of relatives may be an excellent idea in many circumstances, such practice applied to discovered carrier newborns is inappropriate. Simply mentioning that carrier testing for the extended family is available and giving the parents the pamphlet on cascade testing of relatives prepared by the Cystic Fibrosis Trust (available at http://www.cftrust.org.uk) should suffice, with no pressure on parents to contact relatives. The pamphlet contains all necessary information for those who wish to avail themselves of tests. In discussing the carrier state, while mentioning the very occasional disadvantage of some symptoms in common with CF in carriers, it also mentions the probable health advantage that the vast majority enjoy. This is likely to help people to see CF carrier status in a more positive light.

In a larger study than that reported by Parsons et al, Wheeler and colleagues discuss their experiences with the parents of 102 newborns discovered to have one CF mutation in an IRT/DNA screening programme in Massachusetts. The parents were counselled in the genetics department on the same day as the sweat test, in the hour before the result became known. As there was a low chance that the child would have CF, discussion of the disease was brief and more for the purposes of explaining why carrier testing was being offered to the parents. In their study, most parents gave the chance that they might both be carriers as their reason for opting to have themselves tested. Although it would require some organisation, this is an approach in which clinical genetics and IRT/DNA screening units could fruitfully cooperate in the United Kingdom. Again it would be useful to have written national protocols to follow.

The advantages of including DNA analysis in newborn CF screening outweigh the disadvantages, but special care must be taken in managing the parents of neonates discovered to be carriers.

Arch Dis Child Fetal Neonatal Ed 2003;88:448–449

Author’s affiliation
M Super, Royal Manchester Children’s Hospital, Manchester M27 4HA, UK

Correspondence to: Dr Super; maurice.super@man.ac.uk

REFERENCES


Cystic fibrosis newborn screening and detection of carriers

M Super

Arch Dis Child Fetal Neonatal Ed 2003 88: F448-F449
doi: 10.1136/fn.88.6.F448

Updated information and services can be found at:
http://fn.bmj.com/content/88/6/F448.1

These include:

References
This article cites 8 articles, 2 of which you can access for free at:
http://fn.bmj.com/content/88/6/F448.1#BIBL

Email alerting service
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Topic Collections
Articles on similar topics can be found in the following collections

- Screening (epidemiology) (234)
- Screening (public health) (234)
- Cystic fibrosis (15)
- Pancreas and biliary tract (54)

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/