Ethics watch

CARRIER TESTING IN MINORS: CONFLICTING VIEWS

Carrier tests are used to determine whether an individual carries a mutated gene or a balanced chromosomal rearrangement. Contradictions are emerging in the already active debate about whether parents should be allowed to test for the carrier status of their children once a genetic disorder has been diagnosed in their family.

Genetic and medical associations have argued against parents being entitled to request a carrier test for their children[1]. The grounds for this objection are that information about carrier status does not provide immediate medical benefit to the individual who is tested, and that testing violates the rights of privacy, confidentiality and autonomous decision-making of the child once they reach adulthood. Moreover, it has been maintained that testing at a young age might harm a child's self-esteem or affect his or her self-image. It has also been argued that knowledge of carrier status might distort the perception of the child by family members, possibly leading to psychological maladjustment or stigmatization. Finally, a significant level of misunderstanding among the public and the consequently erroneous interpretation of the carrier status have been advanced as reasons to defer carrier testing in minors.

The growing use of DNA testing in newborn screening programmes, however, challenges this professional recommendation, and also raises ethical considerations that were not present when inherited disorders were screened solely by biochemical means. As well as identifying affected infants, DNA-mutation analysis can incidentally identify genetic carriers. More than a decade ago, professional guidelines from the American Medical Association and the German Society of Human Genetics recommended that such incidentally discovered carrier status should not be conveyed to the parents or to third parties. Rather, they suggested that carrier status should be discussed with the child when he or she reaches reproductive age. The guidelines from the American Medical Association provided instructions for maintaining the confidentiality of this genetic information, stating that this privileged information should be kept in a separate portion of a patient's medical record to prevent accidental disclosure. However, no clear instructions were offered about the precise time of disclosure, or about who should provide this information.

Various newly introduced screening programmes[2,3] challenge this policy further. Professionals are now more and more instructed to tell parents, before the test, about the possibility of detecting carrier infants and to inform them of the result, together with providing adequate counselling by a health-care professional.

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A Practice Guideline[4] from the American College of Medical Genetics that aims to standardize practices in the field of newborn screening in the United States acknowledged that practices in the field vary, but recommended that "…clear policies should be in place about communicating such information…" to parents.

This protocol is in line with recommendations offered by the British Medical Association[5] and the American Academy of Pediatrics[6], which had in the past defended the view that results of tests for carrier status that are obtained incidentally should be conveyed to the parents of the newborn. The argument in favour of disclosing the carrier status of an infant is that this information is immediately useful to his or her parents in planning future pregnancies.

Because many more carriers than affected children are identified in newborn screening programmes, informing and counselling the families of carriers is a major challenge. Many questions about the implications of this disclosure still have to be answered. Should information be provided before and/or after screening? Who should provide this information? Should parents and other family members be invited to undergo carrier testing? Who should ensure that the minor is informed of his or her carrier status when they reach reproductive age?

As a consequence of the opposing policies in the context of familial genetic counselling and newborn screening programmes, some couples might come into a situation in which they have received information about the identification of carrier status in their infant after neonatal screening, but are counselled to delay carrier testing for another child until this child has reached reproductive age. The expected expansion of newborn screening programmes to other diseases will further increase pressure to alter the professional recommendation to defer carrier testing in a family context until minors are mature enough to understand the implications of having such a test. The existence of two contradicting recommendations concerning carrier status in minors clearly needs further analysis, discussion and harmonization.

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