Psychological aspects of von Recklinghausen neurofibromatosis (NF1)

In the December 1995 issue of your journal, Mouridsen & Sorensen reviewed the psychological aspects of NF1. They provided an excellent insight into the range of significant pairwise IQ, child reading profile, and attention span deficits. In children with NF1, the exact nature of this association and its relationship to the number, volume, and location of lesions remains to be elucidated. The available evidence suggests that these T2 signal deficits represent areas of dysplastic gliosis and aberrant myelination in the developing brain. If the relationship between MRI lesions and cognitive deficits in NF1 is validated then this association may provide important insight into the pathogenesis of cognitive deficits in patients with NF1.

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The male excess in Down’s syndrome

Mutton et al confirmed earlier reports of a male excess in cases of Down’s syndrome (DS). The cause of this excess is not immediately obvious. It may be associated with a more selective spontaneous abortion in their data, these cases numbered 63 males and 51 females. I should like to suggest a cause of this excess. It is that in cases of DS, the timing of insemination in relation to ovulation is not optimal. It is usually believed (at least among non-geneticists) that the best time for fertilization of the ovum in the human menstrual cycle is associated with offspring sex ratio, male zygotes being preferentially formed when the fruitful insemination is either early or late. In a meta-analysis of the data, Gray estimated that fruitful inseminations around ovulation have a relative risk of only 90% of yielding males as contrasted with early or late inseminations. A similar phenomenon has been reported in other species, such as the domesticated deer,1 Barbary macaque, golden hamster, and Norway rat.2 If the present hypothesis were true, one might expect an excess of DS in cases of rhythm failure. The evidence on this point is equivocal, but suspicion is raised by the reportedly high maternal age specific rates in children born to Catholic women.3

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Predictive genetic testing in children

The paper by Michie et al (J Med Genet 1996;33:313-18) describes a situation which is likely to rise with increasing frequency as more dominantly inherited disorders become reliably detectable by molecular methods. The discussion focuses on the views of the parents and of the professionals but there is no evidence that the children at the ages of 4 and 2 years they are too young to give their opinion, but perhaps a proxy should have done this for them.

For a few disorders (for example, retinoblastoma) surveillance starts in infancy but usually predictive testing for risk of malignant disease is done with a view to prevention of disease by regular surveillance into adult life. We need to know how balanced the procedure is most likely to lead to a responsible attitude to the irksome and unpleasant screening regimens. Parents have their children’s best interests at heart but may find it difficult to remember that the child’s still young. Older children may develop into rebellious teenagers or into 20 year olds who know they are invincible. The poor compliance of diabetics at this stage of life is well known.

Instinct tells me that compliance is likely to be higher when the child has been actively involved in the decision on the timing of the test. Discussion of the need for a test, the time line and importance of the test may increase the child’s cooperation. When the whole family is aware of the disease and can take part in the test together, the result may be greater cooperation.
resent a parent testing him or her "when you were too young to remember". Controlled surveys are unrealistic but we could discover the views of 20 to 20 year olds. Has anyone asked them?

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This letter was shown to Drs Mickie and Marteau, who reply as follows.

Dr Berry raises several important questions concerning predictive genetic testing in children: the age at which a child's opinion about his/her own testing should be sought, whether a proxy should give an opinion in the case of a young child, and the extent to which a child should have a say in the decision about whether and when to be tested.

In the absence of data to inform these questions directly, research concerning children's informed consent in other areas can shed some light.1 The general trend over the last decade has been one of discovering genetic differences in young children before they are clinically apparent2 and of giving children responsibility for decisions about, for example, their medical treatment and participation in research.3 Views about whether parents should be allowed to make such decisions for their children vary and, again, we lack the evidence of impact on children and family life of parents either being allowed to make this decision or of not being allowed to make this decision. Our recently reported single case study suggested that the latter can lead to anger both within the family and with health professionals.4 Dr Berry argues that teenagers may resent having been tested as a child. In the case study found resentment of not having been tested as a child. As Dr Berry states, we need more information about what children and teenagers think about these issues.

We are currently conducting a multicentre trial to investigate the psychological impact of predictive test results on children and their parents. Much more research is needed if we are to have an informed debate on these important issues.

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Predictive genetic testing in children.

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