LETTERS TO THE EDITOR

Psychological aspects of von Recklinghausen neurofibromatosis (NF1)

In the December 1995 issue of your journal, Mouridsen and Sorensen reviewed the psychological aspects of NF1. They provided an excellent insight into many issues, such as the frequency of poor self-image and psychiatric disturbance. However, there have been a number of recent publications, which were not included in the review, and which provide a number of areas of consensus concerning the cognitive phenotype of patients with NF1.


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 yet established and may not be sex 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 widely believed (at least among non-geneticists) that the timing of fruitful coitus within 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 studies, 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 recently observed in other species of captiveReality: in a long-finned pilot whale, 3 Barbary macaque, 4 golden hamster, and 5 Norway rat. 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.

WILLIAM H JAMES

The Galton Laboratory, University College London, Woburn House, 4 Stephenson Way, London NW1 2HE, UK


5 Pratt NC, Hack UW, Link RD. Offspring sex ratio in hamsters is correlated with vaginal pH at certain times of mating. Behav Neural Biol 1987;48:310-16.

6 Hedricks C, McClintock MK. Timing of insemination is correlated with the secondary sex ratio of Norway rats. Physiol Behav 1990;48:5-11.


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 arise 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 little to give the 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 adulthood. We need to know that this procedure is most likely to lead to a responsiable 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 children's years olds 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 and the reasons early age, but let parent and child together await the result. A teenager may well
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 the very young to 20 year olds. Has anyone asked them?

A CAROLINE BERRY
Division of Medical & Molecular Genetics, Guy's Hospital, 7th & 8th Floors Guy's Tower, St Thomas Street, London SE1 9RT UK

This letter was shown to Drs Michie 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 susceptibilities in young children than previously attributed2 and of giving them more responsibility for decisions about, for example, their medical treatment and participation in research.

Views about whether parents should be allowed to make such decisions for their children vary and, again, we lack the evidence of the 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.3 Dr Berry argues that teenagers may resent having been tested as a child, 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.

SUSAN MICHIE
THeresa Marteau
Psychology & Genetics Research Group, UMDS, Ground Floor, Old Medical School Building, Guy's Campus, London SE1 9RT UK

2 Karmilof-Smith A. Beyond modularity: a develop-
3 British Paediatric Association. Guidelines for the ethical conduct of medical research involving chil-

If you wish to order or require further information regarding the titles reviewed here, please write to: Books, The BMJ, Bookshop, PO Box 295, London WC1H 9JR. Tel 0171 383 6244. Fax 0171 383 6662. Books are supplied post free in the UK and for BFPO addresses. Overseas customers should add 15% for carriage and packing. Payment can be made by cheque in sterling drawn on a UK bank or by credit card (Mastercard, Visa, or American Express) stating card number, expiry date, and full name. (The price and availability are occasionally subject to revision by the Publishers.)


As possession of the Y chromosome provides the clearest evidence for a genotype with a predisposition to antisocial behaviour, a possi-

Copyright © 2017 BMJ Publishing Group Ltd. All rights reserved.

Downloaded from http://jmg.bmj.com/ on November 6, 2017 - Published by group.bmj.com.
Predictive genetic testing in children.

A C Berry

*J Med Genet* 1996 33: 806-807
doi: 10.1136/jmg.33.9.806-b

Updated information and services can be found at:
http://jmg.bmj.com/content/33/9/806.3.citation

**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.

**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/