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CORRESPONDENCE

Genetic counselling and genetic registers

Sir,—Dr A J Caro and his colleagues (14 August, p 420) have recently emphasised the need for detecting individuals at risk of having children who might develop Huntington's chorea so that they may be given genetic counselling. While I entirely agree with these sentiments, the problem is in fact much wider than this, for it exists with many other dominant disorders with late onset (such as polyposis coli, polycystic kidney disease, and myotonic dystrophy) and several X-linked disorders (such as haemophilia and Duchenne muscular dystrophy) where there are often many family members at risk of having affected children who may be unaware of this.

In a study of the families of individuals referred to this department for genetic counselling over a four-year period we estimated that there were 950 relatives at high risk (greater than 1 in 10) of having a child with a serious genetic disorder, yet of these only 138 (15%) had had any genetic counselling. Further, 81 affected children were born to parents who a priori were at high risk of having affected children and at least a proportion of these might have been prevented by genetic counselling.

From these findings it would seem reasonable to assume that the extent of the problem is much greater than is reflected by the number of individuals who attend a genetic counselling clinic. There is at present no defined procedure for ascertaining and follow-up of such individuals. Herein lies the value of a genetic register system which, for this reason, has been advocated by a WHO scientific group. ¹ Such a system was first established in Edinburgh in 1970 under the acronym RAPID (Register for the Ascertainment and Prevention of Inherited Disease) ² and has been fully operational for at least a year. For ease of storage, updating, and retrieval of family data the system has been computerised, and strict safeguards for confidentiality have been incorporated into the system. Individuals are included in the register system only with their full approval and written consent (or that of their parents). No relatives are contacted without the permission of the index case and also the relatives' family doctor. The latter is considered especially important as there may be factors unknown to the medical geneticist or index case which would make it imprudent or unnecessary to contact certain family members. Particularly important in such a system is the maintenance of data on individuals who are at high risk of having affected children or of developing a genetic disorder but as yet are too young for counselling.

In the past few years a number of similar genetic register systems have been established in various countries, including Canada, Norway, the United States, and Belgium. ³ The latter is particularly interesting because it has been organised on a national basis. This is important if a genetic register system is to be really effective in helping prevent serious genetic disease within the community.

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National Health Service Consultants' Association

S P BAXTER, MRCPGTCH

Consultants' contract

J M CUNDY, FRAMC

Industrial action

P L HAYS, MB; D C Hogg, MB; Margaret C Watkinson, MB; P D Griffiths, MD; D M Bowes, LRCPED

Consultants' increments

A P J ROSS, FRCS; M J BROWN, MRCPATH

Madness of the profession

H CAIRNS, MRCP

Juniors' contract dispute

R F Massey, MB; K J J TAYLOR, FRCS, and others

United profession

E R C Walker, FFCPED

Private practice and the NHS

K T H MOORE, FRCS

Medical manpower and hospital staffing

I M LIBRACH, MD

Professional unity

R A Keable-Elliot, FRCPED

Genetic counselling and genetic registers

Sir,—A recent paper (14 August, p 420) by Dr A J Caro and his colleagues emphasises the need for detecting individuals at risk of having children who might develop Huntington's chorea so that they may be given genetic counselling. I have been concerned about the wider implications of this, however, and have been invited by the Department of Health and Social Security to comment on the establishment of a national genetic register. It is a common experience of geneticists that the public are aware of genetic diseases such as the haemophilias, muscular dystrophy, and Huntington disease because of high profile cases, but are less aware of the much greater problem of genetic diseases which tend to affect small groups of individuals and are not widely publicised in the media. It is important for the geneticist to publicise the disease when a child with the condition is seen, but it is perhaps more important to be aware of the problem when there are no clinical symptoms.

There is a need for a centralised register of genetic disease in the United Kingdom, and a national register would be an important educational tool. It is planned that those who consult at the Royal Hospital for Sick Children, Edinburgh, will be included in this register, which will then form the basis of a database from which one can draw conclusions about the prevalence of genetic disease in the UK.

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