Clinical genetics services in 1990 and beyond

Purchasers' guidelines to genetic services in the NHS

A SUMMARY OF TWO REPORTS OF WORKING GROUPS OF THE CLINICAL GENETICS COMMITTEE OF THE ROYAL COLLEGE OF PHYSICIANS

The most important principles of medical genetics are those that insist that people must have fully informed personal choice to accept or reject genetic screening. However, clinical genetics has the potential for the prevention of disease, since couples at high risk of having a child with a serious long-term handicap choose either to avoid pregnancy or to have an abortion. Recent advances of molecular genetics, cytogenetics and obstetric techniques have led to great improvements in the services offered to high risk families. These comprise the identification of carriers, with the opportunity for intervention, and the provision of early prenatal diagnostic tests. Such facilities have meant that the majority of high risk relatives can have their anxiety relieved and be reassured that they are not carriers, while medical resources are directed at the known carriers. Such resources include the presymptomatic treatment of metabolic disease, screening for cancer in high risk genetic conditions (such as retinoblastoma and familial polyposis coli) and the offer of early prenatal diagnosis so that termination of pregnancy, if desired, only occurs in affected pregnancies.

These dramatic advances in the management and prevention of genetic diseases have implications for organisation, manpower and training. Immediate discussions are important because the future of the genetic services depends upon the current NHS changes [1], and so two new reports from the Royal College of Physicians are timely. The first concern, which is reiterated in both reports, is that genetic services should continue to be organised at regional level. There is a worry that they may be fragmented to districts in spite of the Joint Statement from the medical Royal Colleges, endorsed by the Department of Health, that regional organisation be preserved. It would be damaging to the welfare of patients and families at risk for genetic disease, who frequently live in more than one health district, if regional genetic services were splintered by devolution to districts; in addition, the quality of service could not readily be controlled or monitored.

The report: *Clinical genetics services in 1990 and beyond* [2] was generated initially by the College's need for accurate manpower data in negotiations with JPAC, but an urgent general review of genetics facilities became essential because of the upheaval in the NHS just when advances in clinical genetics were accelerating. The report surveys the current clinical manpower establishment, genetic family registers and the availability of molecular genetics services (laboratory manpower is reviewed elsewhere). Comparison with what is needed to maintain adequate levels of clinical management and disease prevention revealed a significant disparity. For example, the number of trained clinical geneticists and co-workers (the equivalent of 165) appeared to be insufficient to cope with the number of patients and their relatives at high risk of developing single gene disorders who make up 2–3 per cent of the population (and a greater proportion if part-genetic conditions are included). The report inevitably therefore makes recommendations for increasing the number of clinical geneticists, trainees and co-workers, and this has already proved to be valuable. Even before publication, the data helped obtain a doubling of senior registrar numbers and the establishment for the first time of a separate quota of registrars in clinical genetics. It remains to be seen whether the reformed NHS is capable of funding new consultant and co-worker posts and the facilities they require.

However, genetic diseases are collectively common, and susceptibility to most common diseases is genetically programmed. The numbers and diversity of diseases make it inevitable that all clinicians will require some genetic training. While the College's Genetics in Medicine Education Task Group grapples with undergraduate and postgraduate teaching, a major initiative, recommended by the College report [2] and funded by the department of Health, has been developed by the College. It has set up a Confidential Enquiry Into Counselling for Genetic Disorders and its aims are to educate clinicians in other specialties about screening, diagnosis and management of genetic disorders.

The second report, *Purchasers' guidelines to genetic Services in the NHS* [3], was prepared by the Clinical

Genetics Committee to help overcome some of the problems arising from the new purchaser-provider arrangements and to give general managers information necessary to plan genetic services according to the needs of their residential populations. In the pragmatic world of cost-effectiveness and contracts there is a danger that 'minor' specialties dealing with rare disorders will be overlooked. A manager may not be overimpressed by the needs of, for example, the 10–20 patients with Huntington's disease in his district compared with the thousands with diabetes, osteoarthritic hips, etc. It is therefore important to give districts reliable data on the commoness of genetic disorders, the numbers of high risk and presymptomatic individuals, and their susceptibility to helpful interventions in order to justify funds being allocated for the management and prevention of genetic diseases. Thus the Guidelines for purchasers of genetic services include epidemiological data on genetic diseases and what manpower and standard of facilities are required to cope with these disorders in the population of the average health district.

References

1 Harris R. Genetic services in Britain; a strategy for success after the National Health Service and Community Care Act 1990. *J Med Genet* 1990;**27**:711–14.

2 Royal College of Physicians. *Clinical genetic services in 1990 and beyond.* A report of the Clinical Genetics Committee. Royal College of Physicians, London 1991.

3 Royal College of Physicians. *Purchasers' guidelines to genetic services in the NHS.* An aid to assessing the genetic services required by the resident population of an average health district. Report of a working group of the Clinical Genetics Committee. Royal College of Physicians, London, 1991.

The reports summarised above are available from the Royal College of Physicians, price £4.00 each, plus 50p postage and packing.

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