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Medical genetics

Rodney Harris

Molecular genetics has been responsible for some of the most striking recent developments in medicine. Enormous new opportunities result from the human genome project, with virtually all single gene diseases already mapped to their chromosomal locations. The application of the information gained in this project to clinical practice should be consistent with the aims of medical genetics, which are:

- To ensure the maximum range of options for those at risk of genetic disease by providing accurate diagnosis and screening, empathic genetic counselling, and support
- To prevent genetic disease and unnecessary anxiety by facilitating personal informed choices from among these options
- To aid appropriate clinical management of genetic disease and to identify preventable complications by early and accurate diagnosis.

Below I discuss why medical genetics should be recognised as a key area in the consultations initiated by *The Health of the Nation*.¹

The case for genetics

Genetic diseases and congenital malformations fulfil all three of the government's criteria for a key area. They are an important cause of premature death and avoidable ill health, there are interventions offering scope for improvement in health, and it is possible to set targets and then to monitor them.

Five per cent of the population will have a genetic disorder by the age of 25. This figure rises to 65% in a lifetime if common diseases with a strong genetic predisposition are included. In addition, 2-3% of couples are at high risk of having handicapped offspring.^{2,3} There is also the fear and anxiety generated by recurring genetic disease among relatives at risk of such diseases.

SPECIFIC VULNERABLE GROUPS

The green paper calls for "specific initiatives to address the health needs of particularly vulnerable

groups." In medical genetics these groups are pregnant women, neonates, people from ethnic minorities, and those who are found to be positive for genetic defects in population screening programmes.

People with a family history of serious genetic disease are a special group because many common genetic disorders within families can now be avoided. Consistent with the aims of medical genetics, genetic family registers of those at highest risk are the most effective way of assisting relatives of patients with severe genetic disease including hereditary cancer, Duchenne muscular dystrophy, Huntington's chorea, and adult polycystic kidney disease. In the future additional vulnerable groups may be recognised as knowledge grows about the genetic predisposition to common disease (cancer, coronary artery disease, diabetes mellitus, psychosis, etc).

INTERVENTIONS OFFERING IMPROVEMENTS IN HEALTH

There are well established programmes for preventing genetic and congenital disorders, notably Down's syndrome, neural tube defect, thalassaemia, Tay-Sachs disease, and phenylketonuria. The cost-effectiveness of these programmes has been studied. Screening with ultrasound and of maternal serum α fetoprotein concentrations has contributed to the dramatic reduction in the number of babies born with neural tube defect, while screening based on maternal age and biochemical markers is capable of identifying at least 60% of pregnancies with fetuses affected with Down's syndrome.⁴

New techniques in genetics may facilitate and simplify existing procedures and are often effective in more than one laboratory discipline. Thus the polymerase chain reaction has revolutionised molecular genetics and has applications throughout laboratory medicine. Non-isotopic in situ hybridisation offers greater safety than conventional isotopic methods and has wide applicability in cytogenetics, molecular genetics, and biochemical genetics.

Medical genetics is a dynamic subject in which change occurs very rapidly. Screening for the following

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conditions will become a public health issue in the near future.

Cancer in families—All cancers are the result of genetic mutations or a failure of gene regulation, often aided and abetted by environmental carcinogens. However, some cancers are inherited in a mendelian fashion. For example, colorectal cancer kills 21 000 people a year—10 times the figure for cervical cancer—but while bowel screening is technically possible it is not currently practical for the whole adult population at risk. Some colorectal cancers, however, occur in families, including familial adenomatous polyposis and non-adenomatous familial bowel cancer. Such cancers should be preventable by family counselling, regular bowel screening, and appropriate early surgery. These interventions reduce the 10 year mortality from over 80% to 40%.⁵ Advances in molecular genetics have made screening more effective and, as a bonus, most relatives could soon be reassured that they are unlikely to carry the cancer gene. This not only gives great relief to the relatives but saves the NHS the expense of regular screening for them and their descendants. There are strong hints that common, sporadic bowel cancer may share similar genetic mutations, and this pattern recurs for many genetic cancers and other disorders. In breast cancer molecular techniques also promise advances in screening in younger women, those for whom mammography is least useful.

Marfan syndrome—The gene for this disorder has now been identified⁶ and the basic biochemical defect elucidated. This will provide a new certainty in diagnosis, which is particularly important as the clinical expression of the disease is so variable. The first beneficiaries will be older children and young adults whose close relatives have died of dissecting aneurysm of the aorta. Those who are shown to have the gene will require regular cardiac ultrasonography, prophylactic drugs, and elective heart surgery—thus greatly improving their prognosis. Most relatives will be shown not to be carriers and will require no further screening, which will thus save NHS resources. Relatives will also enjoy considerable relief from anxiety about their own and their children's health.

Fragile X syndrome—Recent molecular genetic advances in the fragile X syndrome^{7,8} (carried by perhaps one in 1000 British women) indicate that screening programmes may soon be feasible for carriers of this the most common form of severe inherited mental retardation.

Premature coronary artery disease—One in every 500 of the white population is a carrier of familial hypercholesterolaemia, and all carriers are liable to premature death due to coronary occlusion. Screening and prevention are now possible, but relatives at high risk need to be identified at an early age if prevention is to be effective. An important limiting factor is the need for input from clinical geneticists; at present there are too few to deal with such large numbers of families.

INTERSECTOR ADVANTAGES

Medical genetics depends on multidisciplinary cooperation in the NHS and the community. It has strong advantages in many health sectors because prevention of genetic disease by health service initiatives reduces chronic disability and results in savings which are at their maximum in the community and social services. However, such savings are not always easy to quantify in accountancy terms and tend to be overlooked.

Identifying and monitoring targets

Targets must be identified which are consistent with the fundamental principles of medical genetics, which were outlined at the beginning of the article. The

Department of Health anticipated the need for evaluation and funded a four year special medical development to assess the value of molecular genetics in clinical genetics. This showed that genetic services were both cheap and effective, even with the limited procedures then available.⁹ Now, specifically to evaluate the management and prevention of genetic disorders by many specialties, the department has commissioned the Royal College of Physicians of London to set up a confidential enquiry into the quality of counselling for genetic disorders. The inquiry will review clinical records of marker disorders (cystic fibrosis, Down's syndrome, familial polyposis, haemophilia, multiple endocrine neoplasia type 2, neural tube defect, and thalassaemia). The targets in each case will be 100% of clinical records having documented evidence of appropriate counselling, of relevant genetic services being offered, and of follow up, especially where potentially preventable late onset disorders are concerned.

The case against

Genetic disorders have, in the past, been regarded as rare and untreatable. There are also reservations about termination of pregnancy, which has erroneously been perceived as the only preventive measure available. Now new genetic screening procedures for familial cancers and other adult onset genetic disease offer different approaches. Reservations about prenatal diagnosis have also been mitigated by rapid technical advances bringing much earlier and more precise diagnosis.¹⁰ Prenatal diagnosis in most cases gives normal results providing enormous relief from anxiety. Indeed, in these situations women are encouraged to begin and to continue pregnancies that might otherwise have been terminated. Medical genetics emphasises the importance of fully informed free choice in reproduction and of the confidentiality of genetic information. This is not only consistent with the ethical importance of recognising the autonomy of the individual but experience shows that it in no way impedes effectiveness because most of those at risk of having offspring with serious handicapping disorders will choose prenatal diagnosis if it is available and if they are given all the relevant information.

There is now overwhelming evidence of general public support for the use of medical genetic advances, as was evident during the proceedings of the Embryo Research Bill. Nevertheless, the unique value of human life must continue to be emphasised, with thoughtfulness and restraint being exercised in all such research. Support for medical genetics is particularly consistent and articulate from lay organisations such as the Genetic Interest Group, which represents more than 60 medical genetic charities.

Need for better education

The generally poor teaching of genetics to medical students and the sometimes doubtful clinical relevance of what they have been taught¹¹ is a potential barrier at present to the effective involvement of doctors in genetic services. The Royal College of Physicians has also surveyed postgraduate education in genetics (A W Johnston, personal communication). Apart from specialist training for clinical geneticists, postgraduate education is even poorer and constitutes a major barrier to the uptake of genetic services. To improve the teaching of genetics to medical students the college has established a medical genetic education task force, while the confidential enquiry into the quality of counselling should improve postgraduate medical understanding of genetic disorders.

Functions of the regional genetic services

Strategies for reaching targets

Several general strategies are required. National feasibility studies for screening programmes for the presymptomatic diagnosis of late onset cancers and other genetic diseases should be a high priority. It is hoped that the new central research and development committee¹² will promote genetic programmes of this type. There is also a need to ensure that all appropriate patients have equal access to screening for Down's syndrome and to other well evaluated screening programmes, including those for thalassaemia and Tay-Sachs disease. Additional screening should be introduced for carriers of cystic fibrosis and the fragile X syndrome as soon as these tests have been evaluated. In parallel with these screening programmes there must be study of the social, community, educational, ethical, and logistic issues that may be expected. It will also be important to ensure by medical audit that the highest standards of clinical and laboratory services are achieved and maintained.

The Health of the Nation identifies the "role of the centre to develop new key areas." This is particularly

important in medical genetics, which is a new and vulnerable specialty. Regional genetic services must be preserved (figure). This need is recognised by the Royal College of Physicians of London¹³ and by other royal colleges and has been endorsed by the Department of Health. It will be extremely damaging to the welfare of patients with genetic disorders and their families, who often live in different health districts, if regional genetic services are splintered by hasty devolution to districts.

In addition to central initiatives, regional genetic committees with multispecialty representation should be charged with strategic planning. They should also provide impartial professional advice to the regional health authority, recommend priorities for developments, and advise on monitoring regionally purchased genetic contracts so as to achieve and maintain an efficient quality service with equal access for patients in all districts.

Advances in medical genetics have implications for other medical specialties. All clinicians should pay increasing attention to this aspect of patient care.

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The New NHS: Six Months On

West Suffolk: changes in the offing

Jane Smith

Nobody in West Suffolk thinks that patients will have noticed much difference in the health service—either for better or for worse. But six months into the reformed NHS there undoubtedly have been changes, and more are in the offing. The plan to redevelop Newmarket Hospital as a community hospital is before the Secretary of State for Health, as is a proposal to merge East and West Suffolk districts from October 1992. Both the acute and the community units have applied to become third wave trusts. On the other hand, some of the same old problems are there: as far as the general practitioners are concerned patients are still on waiting lists, and West Suffolk Hospital suffered

ward closures throughout August in an attempt to save money.

Extracontractual referrals

One headline hitting problem that does not seem to have affected West Suffolk's patients is extracontractual referrals. The district set aside 1.5% of its budget for these, and all requests have so far been honoured, with the district still within its budget. Dr Roger West, the director of public health, wishes, however, that extracontractual referrals were not costed at average specialty costs: one patient having a

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