Genetic counselling and testing in Europe

ABSTRACT — The Genetic Enquiry Centre in Manchester has designed a three-pronged health services research programme to address current issues in genetics. The issues are: whether doctors who are not trained in genetics can manage the genetic problems they meet in their practice; whether there are enough resources in specialist centres to cope with current and imminent referrals; and whether providers of primary care recognise genetic problems and refer patients appropriately. The three studies providing the basis of the programme – the National Confidential Enquiry into Counselling for Genetic Disorders, the Concerted Action on Genetics Services in Europe and the Primary Care for Genetics Services study – are discussed. The first two provide unique views of genetic counselling in the UK and of the access to and quality of health services for patients with or at risk of genetic disorders throughout Europe, and make recommendations based on their findings. The third is a continuing study that aims to determine the effects of patterns of referral and care in the different healthcare systems in Europe. Although it is unlikely that there will ever be enough medical geneticists to cope with the consequences of genetic advances on health services that are largely unprepared, specialist genetic centres are the natural core resource for future multi-specialty genetic services. This will give clinical geneticists an extended role complementary to that of diagnosing rare syndromes.

Advances in genetic screening, diagnosis and prevention of disease can be safely exploited only if there are enough knowledgeable healthcare professionals working in effective healthcare systems. Otherwise, patients will be denied the benefits of advances and will be deprived of access to informed medical advocates who would help them avoid discrimination. Medical genetics, with its wealth of clinical, laboratory and ethical experience, is a paradigm for other clinical activities and is therefore an invaluable resource for many clinical specialities. However, in the UK opportunities to use this resource may be restricted by the serious understaffing of specialist genetic centres reported by the Royal College of Physicians of London[1,2]. Surveys by the Royal College also found that the teaching of genetics to medical students in the UK was 'often of doubtful clinical relevance'[3]. Postgraduate education, outside clinical genetics itself, seemed to be no better as 'there is no overall plan to educate doctors (in genetics) and in most centres there appears to be nothing but occasional unplanned lectures'[4]. These reports suggest that most doctors have received little education in genetics and that there is unlikely to be enough trained clinical geneticists to cope with the burgeoning need. They indicate that health services in Europe must tackle three crucial questions in relation to genetic counselling, screening and diagnosis:

- do doctors currently have enough knowledge to manage genetic problems encountered in their practice?
- to what extent are available resources in specialist genetic centres able to cope with current and imminent referrals?
- do primary care providers recognise genetic problems and refer them appropriately to specialist centres?

The Genetic Enquiry Centre in Manchester has designed a three-pronged health services research programme to address these three questions. First, we have audited genetic counselling by non-geneticists in the UK for five genetic disorders (the National Confidential Enquiry into Counselling for Genetic Disorders, CEGEN)[5]. Second, we have documented the current situation of specialist genetic centres in 31 countries in Europe (the Concerted Action on Genetics Services in Europe, CAGSE)[6]. Third, we are investigating the interface between specialist genetic centres and the non-geneticist doctors who refer patients to them in seven European countries. This is the Primary Care for Genetics Patients programme, PrimGen.

UK National Confidential Enquiry into Counselling for Genetic Disorders (CEGEN)

Genetic counselling does not aim to prevent couples from having children with genetic diseases. Preventing genetic disorders, although important, is secondary to good clinical practice, which identifies couples at risk and by empathic counselling allows them to make their own informed choice about prenatal diagnosis, termination of pregnancy or other aspects of management. Informed choice without external coercion should distinguish medical genetics from eugenics, which has the contrary aim of improving the communal gene pool.

CEGEN is a multi-disciplinary investigation that asks how well paediatricians, obstetricians, general practitioners and other specialists provide services for patients and their families at risk of genetic disorders. CEGEN has audited 'memorable events' – procedures indicated on the grounds of maternal age, family history or available screening tests – and has studied individual case records for unambiguous evidence of informed choices made by patients. Five reports have been published[7–11]. Four concern prenatal screening, diagnosis and reproductive choices in the following:

- Down's syndrome pregnancies during 1990–91 in women aged 38 years or more"
neural tube defect births in 1990–91,
births, in 1991–95, of children with cystic fibrosis into families where a sibling had previously been diagnosed with the disease,
pregnancies during 1990–94 in which the fetus was affected and beta thalassaemia or which resulted in an infant with beta thalassaemia (report in preparation).

A fifth report described counselling and screening for a form of inherited cancer, multiple endocrine neoplasia type 2, diagnosed between 1980 and 1991.

These disorders were selected because the ‘memorable events’ could be reliably ascertained for study and they allowed the audit of clinical practice by a range of non-geneticists, including obstetricians, midwives, endocrinologists, surgeons, paediatricians and haematologists. If all cases had been managed correctly, it would have been possible to offer counselling and screening in good time to avoid the relevant disorder.

Audit concentrated on documented evidence of appropriate counselling, the offer of relevant genetic services and of follow up. Clinicians involved their clinical teams as part of the teaching process and reviewed their own records. When the inquiry team examined a sample of the reviewed case notes, they found that questionnaires had been completed with great accuracy and that co-operation and response rates were excellent. The proportions of questionnaires completed for appropriate cases ranged from 74% to 98%, and there was no evidence of systematic bias in compliance.

Inadequate case notes and delays

Poor recording of genetic events in hospital case notes made it difficult to assess the provision and content of counselling. This is particularly hazardous with events critical to a couple’s reproductive plans and has potential medico-legal implications. Doctors responsible for individual case management could not always confirm from hospital records that offers of prenatal diagnosis had been made or that termination of pregnancy had been preceded and followed by counselling. In hereditary cancer, a pedigree was not consistently recorded in the case notes and close relatives were not always offered screening and prophylactic interventions. Delayed referral by general practitioners led to missed opportunities for counselling and prenatal diagnosis, while late diagnosis of cystic fibrosis sometimes prevented counselling in a subsequent pregnancy and contributed to the birth of a second child affected by the disorder. We do not have directly comparable data from other European countries.

Recommendations

Recommendations made to overcome these shortcomings and to improve education and training for all health professionals stress that records of genetic counselling are at least as important as the virtually obligatory notes of surgical operations, consent and prescribing. A standard of care that documents information, counselling and decisions, together with written consent before genetic procedures, including genetic testing for late onset disorders, is needed. Without good records of patients’ informed choices, how can we be sure that medical genetics is not neo-eugenics, albeit with technical improvements?

Concerted Action on Genetic Services in Europe (CAGSE)

CAGSE recruited a network of senior medical geneticists in 31 European countries to report on the current state of services provided by specialist genetic centres and, eventually, to assist dissemination of the findings. Initially fairly disparate collaborators became a cohesive group, thanks to a series of workshops funded by the European Union that provided opportunities to link on an individual basis and to share common experience in relation to genetic services in each country. They achieved international comparability and accuracy by using a standardised format of seven dimensions and independent evaluators to confirm the data in each country. The report gives detailed information about the quality of genetic services in each of the 31 countries and relates this to their particular economic situation, geography, demography, health services settings, primary care arrangements, history and politics. The CAGSE report documents comparative availability data on staff numbers, screening of newborn babies and for carriers, access, genetic registers, education, and official recognition of the specialty of medical genetics. CAGSE provides a snapshot of genetics services in Europe in the mid-1990s and has updated partial studies in several countries.

Multidisciplinary collaborations

Clinical genetics is evolving rapidly. Many doctors (and lay workers) other than medical geneticists are becoming involved as a result of genetic screening, diagnosis and understanding of the pathogenesis of diseases that are not classically part of the medical geneticist’s remit. These disorders include Alzheimer’s disease, cancer, coronary heart disease, diabetes and psychoses. Recognising the importance of multi-disciplinary collaborations, CAGSE participants agreed three principles on which medical genetics services should be based. In doing this, they deliberately avoided detailed ex cathedra recommendations that might be difficult to apply in the widely varying conditions found in the 31 countries.

Principles for specialist genetic services

The three CAGSE principles for specialist genetic services are:

- official recognition of the specialty of medical genetics at a national level, and national strategic planning

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by medical genetics organisations linked to other specialties, to patient organisations and to government

- development of regional medical genetic centres as an important point of delivery of specialist genetic care in collaboration with other specialties, community and other medical services

- joint education and training programmes to promote the teaching of genetics to medical and other students, and training programmes with assessment for specialist geneticists and for other health care workers.

Based on the wide experience of the CAGSE collaborators, these principles provide a framework for the development of future services.

**Spreading the message**

The report has been widely disseminated and authors in all 31 countries are making presentations based on the three principles to national genetic societies, medical organisations, health administrators and governments. Publication of the report already seems to have encouraged the development of services appropriate to the special needs of each country. For example, the Bulgarian minister of health, Professor P Boyadjiev, wrote: 'Many ideas from the (Bulgarian) chapter will be taken into consideration in the process of updating of the National Program for Genetics ... the right ideas will be accomplished step by step in the future'. Other official representatives of collaborating countries have also emphasised the value of the report.

**Primary care for patients with genetic problems (PrimGen)**

PrimGen is a current research project that aims to determine the extent to which access to and delivery of genetic care depends on the different healthcare systems operating in European countries. Primary care is of particular interest because patients do not usually approach clinical geneticists directly – referrals come from other specialists. In the UK, and some other countries, primary care is provided by specially trained general practitioners who act as 'gatekeepers' to specialist care. In these countries geneticists receive referrals directly from general practitioners or indirectly through other specialists. In countries where general practice is not well developed, patients are usually free to go directly to whichever specialist they wish, within financial limits. Primary care is then provided by paediatricians, internists, obstetricians etc. Providers of primary care who are well informed about genetics should have a highly beneficial clinical and economic role in patient care. However, those who are poorly informed could have an extremely limiting and even detrimental effect on the welfare of patients and their families, as well as being more costly. The danger that genetic centres will be submerged beneath a flood of inappropriate referrals also exists. PrimGen concentrates on two inter-related issues: referral, that is the route and quality of referrals to genetic centres; and continuing care and co-ordination after the specialist genetic consultation is complete.

**Referral**

PrimGen is assessing how referrers’ awareness (or lack of awareness) of the particular implications and characteristics of genetic problems influences successful clinical outcomes, including patient satisfaction and confidence in medical services. Do referrers prepare patients adequately for what to expect at the genetic centre? What is the impact of the referrers on the volume, type and appropriateness of referrals to specialist genetic centres? Do letters of referral or other means of communication describe adequately the genetic problem, identify the patient’s needs, and provide a basic family history? PrimGen studies the routes of referral, the number of contacts with other professionals and the referral rates as a proportion of the population prevalence for different genetic disorders.

**Continuing care**

The second major issue is the quality, availability and provision of continuing care and co-ordination. Continuity of care is important for genetic disorders that have serious implications for other family members. This is especially true for increasing numbers of people who are currently healthy but are found to be at risk, including those who have positive genetic predictive tests. A whole new set of continuity problems is likely to arise as we identify genes that convey clinically important susceptibility – but not certainty – of common disease occurring sometime in the future. Continuity requires effective means of identifying and following up family members who are at risk, and in some countries genetic family registers assist this process. Follow up requires stringent precautions to ensure informed consent, confidentiality and protection against potential discrimination. Genetic disorders frequently involve more than one body system and require multiple investigations in a variety of different specialities, so co-ordination of clinic visits is essential to avoid unnecessary duplication and major disruption of the patient’s life.

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