Introduction

Although genetic disorders are individually rare, there are so many thousands of them that collectively they are recognized as an important cause of poor health. They also create social and financial burdens for affected individuals, their families and society. Nevertheless the diagnosis and management of genetic disorders have traditionally been regarded as arcane, the preserve of clinical geneticists and other specialists working in tertiary centres. This perception is now outmoded because of the great leap forward in molecular genetic technology. This allows for precise diagnosis based on DNA analysis, so that diseases formerly regarded as indistinguishable can now be split into specific groups, while mutation analyses lead to new knowledge about the protean structure and function of disease genes.

Remarkably these advances are also uncovering inborn genetic susceptibilities to such common diseases as breast, ovary and bowel cancer, as well as to Alzheimer's disease, coronary artery disease, diabetes mellitus and schizophrenia. This progress will enable radically new approaches to treatment and prevention specifically related to the individual's genetic profile of disease susceptibility. There is also new optimism about gene therapy, although there are only modest expectations of immediate clinical success (Verma and Somia, 1997).

These topics are subject to great exposure in the media, which portray them perversely either as medical panaceas or as extreme threats to civilization as we know it. Just as it is not possible to stand aside from the influence of computer technology, the impact of genetics on medicine and medical care is all pervasive and will impinge on most medical specialties and on primary care, as well as raising thorny ethical problems.

Medical advances are already transcending the ability of hospital specialists to respond to them and to accommodate patient needs and anxieties. Fortunately the primary care team is in a particularly favourable position to recognize genetic problems and to utilize these new developments to the benefit of patients and their families. General practitioners, like clinical geneticists, are commonly concerned with the family as a unit. Their existing involvement with community-based preventive measures, such as immunizations and cervical cancer screening, makes the primary care setting a logical one for genetic screening programmes, such as for carriers of the cystic fibrosis mutation.

The need for appropriate training in genetic issues for those in primary care and for links between genetics specialists and primary practitioners is increasingly recognized (Harris and Harris, 1995), and represents a major challenge. The relevance of primary care to the long-term problems experienced by patients and families with genetic disorders is emphasized in the recent RCGP report (RCGP, 1996):

> *"The characteristics that distinguish effective and efficient primary care are its accessible front line position, its longitudinal nature, its focus on the individual, its responsibility for dealing with most common problems in the population, its co-ordinating functions for integrated patient care, and its orientation to prevention in addition to traditional clinical functions. (A)...balance must be struck between molecular medicine, person-centred medicine, and population medicine in undergraduate and postgraduate programmes if the application of scientific advances is to be optimum in the future."*

Background

1.1 General practitioners have been involved in detection and management of genetic diseases for many years. All neonates are screened routinely for phenylketonuria and hypothyroidism. Many practices are also involved in screening selected populations for Down's syndrome or Tay-Sachs disease. Many will have a family registered with the practice in whom there is an inherited condition, such as cystic fibrosis or neurofibromatosis. Even more will be consulted by women in whom there is a strong family history of breast or ovarian cancer.

1.2 Knowledge in relation to genetically inherited disorders is increasing and technology in relation to their treatment is advancing. Yet genetics remains to many general practitioners a distant specialty whose relevance to everyday practice is not yet clear.

1.3 Recent advances in genetics have been reported in four reports (House of Commons Science and Technology Committee on Human Genetics, 1995; Department of Health, 1995 (two reports); Harris H et al). The increasing involvement of primary care was the subject of a recent editorial in the *British Medical Journal* (Harris and Harris, 1995).

1.4 The most logical way for general practitioners to begin to address the advances in genetics is to develop a partnership between primary care and specialist centres in medical genetics. This could provide the basis for the development of solutions to the problems and opportunities generated by the advances of the Human Genome Project (an international collaboration to map and sequence all human genes). Genetic disorders are particularly appropriate for shared care between specialist and generalist because they are frequently chronic conditions, and may involve relatives in successive generations.

1.5 General practice in the United Kingdom is particularly well-suited for this partnership. Registration of patients encourages continuity of care and allows effective co-ordination of other caring and support services together with cost effective access to secondary care. Proper use of computers can provide rapidly accessible disease registers for the general practice population. General practitioners and their teams are skilled in counselling, screening and health promotion, but most importantly they have a special understanding of health and disease as it affects patients and their families. They are particularly skilled in interpreting the meaning of a diagnosis to particular patients according to their individual need and their social context. The primary care team is in a privileged position to offer continuing support in the light of that diagnosis.

1.6 Genetic counselling by specialists is very detailed but may comprise only a single consultation. In primary care there is opportunity for 'staged' counselling – the giving and reinforcing of information in small amounts over a number of consultations. This is an advantage to many patients who may have difficulty in absorbing all the implications of a genetic diagnosis on a single occasion. Medical genetics and other hospital services provide specialist diagnostic and therapeutic services which are complementary to that provided by the primary care team.

The issues facing primary care

Medical genetics in general practice

2.1 Generally, genetic counselling is not a one-off event, and therefore cannot be effectively completed during a single visit. There needs to be review and reinforcement on more than one occasion. For example patients at risk of Huntington's disease may well reject genetic tests (or even counselling) when they are 20 years old but feel quite differently when 40 and approaching the age when they might get the disease. Some genetic centres have registers to keep in touch with patients at long term risk, but most other specialists will see such patients only once or at most occasionally.

2.2 In primary care, the patients' knowledge of their carrier status identified in a population screening programme (for example, for cystic fibrosis or thalassaemia) needs to be documented and used appropriately if pregnancy occurs. Provision for continuing care for individuals and families is essential, and this is the core role of general practice. But there is much overlap in the skills needed by primary care and genetic teams and both are faced with the same torrent of new information from the Human Genome Project.

Opportunities for family history taking

2.3 General practitioners look after families but are not always aware of family relationships within their own practice. Sometimes this becomes more clearly defined during pregnancy and at times of

family crisis, for example, during terminal illness or sudden bereavement. New genetic information and awareness will be the catalyst for change in the recording of family history information, as this knowledge becomes both important and relevant to patient care. The taking of a clinical family history in primary care should be a complement to pedigree drawing by specialists. The latter consists of the construction of a detailed family tree indicating both those members affected and those currently unaffected by a relevant hereditary illness or carrier state.

2.4 There are a number of opportunities in general practice when family history taking can be formalized and integrated into the consultation or health check:

- New registration checks could be preceded by a questionnaire completed by the patient who could check a list of named conditions which may have a genetic component. Further details can be recorded by the nurse or doctor who completes the new registration medical.
- The first antenatal consultation or the preconception clinic is a particularly important opportunity to identify those conditions which can be screened during the antenatal period, for example, Down's syndrome, cystic fibrosis, spina bifida and haemoglobinopathies.
- Well person screening checks and patients attending for contraceptive advice and hormone replacement therapy are further opportunities for taking a structured family history relating to, for example, insulin dependent diabetes mellitus, cancers of the breast, ovary, colon and prostate.

Team working

2.5 Family history taking is not a single event. It is constantly changing and needs updating in the light of new births, new cases of genetic disorders, new family information and new genetic applications. The patients' own knowledge of their family history can be prompted by a questionnaire. This should be simple and easy to complete, with particular reference to family members affected and age of diagnosis.

2.6 The nurse or doctor who is carrying out health checks is often best placed to include a family history protocol. Other members of the primary health care team, particularly the midwife and health visitor can contribute to this process. Some practices are introducing a non-medically trained member of the ancillary staff to collect family data from patients and input them to computer records.

2.7 General practitioners' personal involvement in family history taking may be only occasional and opportunistic but it is important that they perceive the relevance of the information collected by other members of the team and are able to use the data when appropriate.

2.8 Occasionally doctors have expressed doubts as to whether patients will be willing to divulge what may be sensitive family histories to non-medical staff. In fact there is no evidence that this is true and certainly, in other areas of primary care, patients may sometimes prefer to discuss problems with nurses rather than with doctors. It needs to be emphasized to patients that the same code of confidentiality which applies to doctors is equally applicable to other members of the primary care team.

New relationships

2.9 A new and intimate partnership will need to be forged between general practices and genetic centres using much speedier and more comprehensive communications. There is an opportunity for locally derived and regularly up-dated clinical guidelines. Studies carried out by general practitioners show that a partnership with genetics centres does work (Harris et al, 1993). For example, screening in general practice for carriers of cystic fibrosis early in pregnancy is popular with patients and it can be successfully integrated into early antenatal care by the primary care team. Only about 10 per cent of the pregnant women who should be offered tests are missed, usually due to lack of time. However, attention may need to be given to relationships with some obstetric and paediatric colleagues, who may doubt the ability of general practice to cope with the implications of genetic disease.

Referral for specialist services

3.1 The four most common reasons for referral to genetics centres are:

- the diagnosis of a Mendelian genetic disease
- family history of a disorder with a genetic component

- interpretation of the results of genetic screening
- the birth of a child with dysmorphic features (abnormal anatomy) and/or mental and physical handicap.

Diagnosis of a Mendelian genetic disorder in a family

3.2 When someone in a family has been diagnosed as having a Mendelian genetic disorder, such as Marfan syndrome, neurofibromatosis, Duchenne muscular dystrophy, adult polycystic kidney disease, or another disorder, this has implications for other family members. Indeed some of them may already be affected themselves without realizing it. There may be both affected and unaffected individuals in a family who may want to have more information about the condition, how it might affect them, what the risks are to themselves and to their children. Depending on the mode of inheritance of these conditions they may also want to consider the possibility of having genetic tests, whether this is a pre-symptomatic predictive test, a carrier test or a pre-natal diagnosis.

Family history of a disorder with genetic component

3.3 A positive family history involving several relatives with an early onset of a disorder which has a genetic component (but does not usually follow Mendelian inheritance), may suggest increased risks for family members. Examples are breast, ovary and bowel cancer, coronary heart disease and learning disabilities. In other families recurrent obstetric problems, miscarriages, still births or neo-natal deaths suggest an underlying genetic factor.

Results of genetic screening

3.4 Couples are referred when an abnormality has been unexpectedly identified during pregnancy by biochemical, ultrasound or amniocentesis particularly when the significance of the finding is not clear. Risk estimation related to pregnancy outcome needs timely, detailed and sensitive explanation. Without this, screening programmes like the triple test for Down's syndrome may fall into disrepute. There is a clear need for well written understandable literature for patients and general practitioners. The latter should be immediately informed about the outcome of screening tests.

Birth of child with dysmorphic features

3.5 Children who have congenital abnormalities, dysmorphic features (unusual appearance) or learning disabilities in any combination are frequently referred to a genetic centre initially for diagnosis and genetic counselling.

The role of the specialist geneticist

4.1 The construction of detailed pedigree information will normally remain the province of the specialist clinical geneticist or co-worker. However the accurate recording and transmission of the relevant data from primary care is important. This can simplify and speed up the risk assessment for an individual patient. For example, a general practitioner's recording of an analysis for a carrier of cystic fibrosis will enable precise risk estimation for a close relative's pregnancy.

Making a clinical diagnosis

4.2 It is always desirable, but not always possible, to make a precise clinical diagnosis before genetic counselling. In the absence of a family history this will depend on the clinical geneticist's knowledge and experience and the judicious use of diagnostic tests. When a child has an unusual combination of physical abnormalities which defy local diagnosis, it may be necessary to seek diagnostic assistance at one of the regular national meetings of dysmorphologists.

Genetic consultation and counselling

4.3 The term *genetic consultation* is preferred by many clinicians as *genetic counselling* does not adequately convey the need for formal diagnostic evaluation as well as careful and empathic explanation. Some people do benefit greatly from counselling but increasingly, some patients are anxious to confirm the diagnosis and may regard counselling as patronizing, or even directive. Ideally the general practitioner or a specialist genetic nurse will have identified what the family wants in advance of a genetic consultation.

Genetic centres as sources of information

4.4 Genetic centres have information on all genetic disorders and the latest diagnostic methods, and

act as a resource for information and education. For detailed medical information, genetics departments use a variety of databases and these not only help with differential diagnoses but they provide prints of information on individual conditions, the features of the conditions, and a list of references.

Specialist trained genetic nurses and family support groups

4.5 One of the functions of the genetics department is to provide support to families whilst they come to terms with the diagnosis of a genetic disease and its implications within the family. Genetic centres have specialist trained genetic nurses who are experienced in supportive counselling, and who provide part of this service to these families. Forging links between academic departments and primary care is a natural extension of their present role. This will enable training of the primary health care team and may enable genetic screening and research projects to be established. Voluntary family support groups exist for virtually every genetic disorder. These groups organize meetings, put families in contact with each other and provide basic information, brochures and newsletters. This is valuable both for the family and for the professionals helping to care for them.

Specialist investigations

Laboratory tests

4.6 Cytogenetic analysis is still one of the most important investigations for those who present with congenital abnormalities or learning disabilities. The molecular genetics laboratory also forms an integral part of the genetics unit and molecular scientists are now able to do DNA analysis for an increasing number of single gene disorders.

Interpretation of screening tests

4.7 Prenatal tests are mainly used to detect fetuses with potentially serious disorders that the parents may choose to have aborted. Neo-natal tests are most commonly used to detect infants with preventable biochemical disorders including phenylketonuria and congenital hypothyroidism, or with genetic disorders who may benefit from early diagnosis, for example cystic fibrosis. Finally there is an increasing number of tests which detect genes causing common diseases in later life, such as cancer and coronary artery disease, for which there may be effective prevention. For a family afflicted by genetic disease, the effect of a negative test both in terms of relieving anxiety and in removing the need for further investigation, should not be underestimated. However the possible unreliability of such a result must be explained.

Ethics

5.1 The subject of ethics has been reviewed elsewhere (Nuffield Council on Bioethics, 1993; Harper and Clarke, 1997). Ethics is presently being considered by a BMA Genetic Steering Group under the chairmanship of the Most Reverend Michael Holloway, Bishop of Edinburgh. In most respects the issues concerning professional conduct while practising genetic medicine are the same as those which apply in other areas of clinical practice.

Family involvement

5.2 There is at least one important difference from normal practice, however. Receiving genetic information about one individual frequently has relevance for other members of the family. This can create great difficulty. For example, in a family with Huntington's disease where a grandparent is affected, a prenatal diagnosis will reveal the status of the parent and have implications for the siblings. There is immediately a conflict of interest between the maintenance of confidentiality for the patient and the right of other potentially affected family members to know. If possible, with sensitive advice, the patient might be persuaded to transmit this information directly, or give permission for this to be given. This assumes that the relative wishes to know, which in some cases may not be true.

5.3 This highlights another issue for the family. The patient will always be counselled first so that informed consent can be given for a test to be undertaken. The possible effect on other family members must be part of the information offered. Clearly no other family member can give this informed consent. On the other hand, even if they were identifiable in advance, they could not justifiably interfere with the patient's right to request testing.

5.4 The affected family member might be the patient's spouse or partner. Tests on the couple relating to a pregnancy may cast doubt on the

paternity of the fetus. Should this possible outcome be discussed before the tests are undertaken?

Termination of pregnancy

5.5 Genetic screening tests, which may lead to consideration of the termination of pregnancy of an affected fetus, present ethical challenges for the patient and the doctor. It is important that patients understand that the screening path onto which they step may have termination as a possible outcome and that if they hold strong religious or moral views, they may wish to decline the tests. There are particular difficulties in giving a balanced picture of a disease and what this will mean to a family when a screening test has detected a risk to the pregnancy for a condition of which the family has had no previous experience and little awareness. (Ninety per cent of cystic fibrosis births occur in families with no previous family history and the majority of these are now theoretically preventable by carrier screening and detection of carrier couples.)

Testing of children

5.6 The genetic testing of children has been considered by a working party of the Clinical Genetics Society (UK) (Clinical Genetics Society, 1994). Legally, young people under the age of 16 can consent to testing provided they are of sufficient maturity and competence to understand its implications. The working party concludes that predictive genetic testing of children is appropriate where the onset of a disorder regularly occurs in childhood and medical intervention can be offered, for example, through diet, medication, ongoing surveillance. Predictive testing for the onset of the disorder in adulthood should generally not be undertaken if the child is healthy and if no useful medical interventions can be offered in the event of a positive result. Testing children for carrier status for recessive disorders and balanced familial chromosomal rearrangements is more difficult but is best deferred until the result is of current relevance (that is, before pregnancy). If testing is not carried out in childhood, then a certain obligation rests upon the health care system and the family to ensure that testing is offered when the child is older. The importance of comprehensive general practitioner records (usually on computer) to include genetic data cannot be overstated if this recommendation is to be realized.

Employment

5.7 Many employers insist on a pre-employment medical examination. Participation in screening tests should be voluntary, and the result should not be used to exclude individuals from employment but to allow them to make their own decision. There will be occasions when the presence of a genetic disorder will put the individual or colleagues at particular risk in the proposed employment environment. It may well be in this person's interest to be aware of the problem in advance.

Insurance

5.8 The House of Commons *Science and Technology Report on Human Genetics* recommended in July 1995 that the insurance industry be allowed one year in which to propose a solution acceptable to Parliament. It should be possible to regulate the use of genetic information in life insurance, which would enable as many people as possible to obtain insurance and protect the insurance companies themselves. As the balance between state and private funding of pensions and health care alters, this area will increase in importance.

5.9 This whole area remains unresolved and unsatisfactory. If the consequence of genetic testing by general practitioners is that this information may be transferred subsequently to insurance companies, patients may well decline to have the test, or opt to be tested elsewhere (even 'over the counter') and request that the results are not passed to their general practitioner. This would be a highly undesirable outcome. The issue has been discussed extensively elsewhere (Harper and Clarke, 1997).

The general practitioner's role

5.10 As in many other fields, the general practitioner will increasingly act as the patient's advocate. Many of these difficult ethical questions may present as a snap shot for the obstetrician, paediatrician, oncologist or clinical geneticist who is involved with the patient's care at that point. For general practitioners who have knowledge of the family dynamics, the level of appropriate counselling and ongoing support needs sensitive and skilled handling, and may affect families over a long period of time.

Impact on primary care workload

6.1 Coping with new knowledge and skills will impact on general practice workload. However, this increase could be reduced through effective teamwork. One practice is already evaluating the effect of using the practice team to take detailed family histories. It is likely that as patients become better informed about inherited conditions, they will consult more frequently. General practitioners will need to be abreast of recent developments and will require rapid access to information about family histories, cross referenced to other family members. Current electronic recording systems will need to be extended to cope with this.

6.2 As clinical generalists it is essential that general practitioners prepare to take on this additional work. There may be a need for additional resources, for example, trained practice nurses and more sophisticated electronic records. Links with genetic nurses working in genetic centres will prevent fragmentation of services and ensure that referrals to secondary or tertiary care are appropriate. There will be cost implications but it is important that decisions on genetic issues are taken in partnership with the patients and this must involve both primary and secondary health care teams.

Education and training in genetics

7.1 If primary care is to provide front line services for patients who are concerned about inherited diseases or who have a strong family history, all members of the health care team should have the appropriate knowledge and skills to identify the presenting problem and formulate a management plan. *The Nature of General Medical Practice* (RCGP, 1996) has highlighted the skills of the clinical generalist in making a diagnosis using knowledge of the disease together with an understanding of the patient and the social context. These skills form an essential backdrop to working with genetic diseases.

Knowledge of disease

7.2 Genetics is a fast moving field. All members of the clinical health care team will need sufficient knowledge of genetic disease to understand the different inheritance patterns, laboratory tests, and unusual appearances associated with genetic disorders. It may not be possible to maintain that knowledge in the forefront of the clinical repertoire but the primary care clinician should be sufficiently knowledgeable to be suspicious and should use local or regional expertise. Equally a primary care clinician should have sufficient understanding of disease and test results to be able to put into perspective the risk of genetic illness and advise concerned patients or parents appropriately.

7.3 Some patients will need referral for a consultation with a secondary or tertiary centre. The primary care clinician should be familiar with the services available and the existing resource limitations in local secondary and tertiary care providers. Equally many patients with genetic disease will require the general practitioner to be able to mobilize social services and to be familiar with local provision for continuing and terminal care for serious genetic disorders. Care plans should be developed by a partnership between the specialist, who will have experience of rare genetic disease and its progress, and the primary health care team who will deliver support and terminal care and have more detailed knowledge of family dynamics.

Clinical skills

Family history

7.4 Managing genetic disease in primary care requires specific skills. As has been discussed (see para. 2.3), the ability to take an accurate family history is crucial. All practitioners should be able to take a family history of clinically apparent inherited disorders, but should also be able to take a fuller family history if inherited disease is known or suspected.

Genetic counselling

7.5 Having identified a positive family history the primary care clinician should have sufficient counselling skills to be able to place the likelihood of inherited disease into a meaningful comparison for a particular patient. This will require a knowledge of the prevalence of disease and an ability to interpret that disease occurrence within a familiar context. They should be able to explain the meaning of positive and negative results of diagnostic tests and use their consultation and communication skills to respond to the patient's concerns and fears. Primary care clinicians should be sufficiently familiar with

secondary care to be able to alert patients to the tests which they are likely to encounter at the hospital.

Genetic screening

7.6 Primary care clinicians should be familiar with the essentials of screening. This includes the pros and cons, ethical implications and possible future developments for pregnancy, family and population screening. Some practices may want to consider offering screening and would need to understand concepts of false positive and negative, sensitivity and specificity and the variations relating to ethnic, gender and age differences.

Team working skills

7.7 One important feature of primary care has been the development of the practice team to deliver health care. In genetics as in many other spheres the ability to work as a member of a team is paramount. This will require the ability to negotiate and delegate, to be able to work to others' strengths and support them through difficult situations.

Skills in clinical decision-making and risk management

7.8 There is always a danger that clinicians working in a rapidly changing field will be swayed by media coverage and populist pressure. An effective primary care clinician should have skills in critical evaluation and possess the ability to minimize and manage risk. Although these skills are not specific to work in inherited diseases, they are fundamental if patients are to be managed sympathetically and secondary care is to be used efficiently.

Professional values

7.9 Many of the professional values associated with clinical genetics are common to those of a competent clinician. They will include issues such as the importance of informed consent (autonomous decision-making) before genetic testing, confidentiality of genetic information and the general practitioner acting as the patient's advocate as well as seeking equity in access to care and avoiding harm.

7.10 They will also include the other professional values we would expect of any clinician, such as keeping abreast of new developments, being aware of one's own clinical limitations and knowing when to seek expert advice. These are skills that are particularly important in a fast-moving field such as clinical genetics. Information published today, especially in text books, is likely to be rapidly out of date. Additional skills are needed such as being able to access electronic information, either through a CD-ROM or the Internet.

Educational programmes

7.11 Educational programmes for primary care may need to ensure a basic knowledge of clinical genetics, although much of this could be covered through pre-course reading material. Important aspects of courses are the opportunities to practise skills such as breaking bad news, taking an accurate family history and counselling. These skills are best learned in a multi-professional setting and there should be opportunity for general discussion of ethical issues and professional values.

The undergraduate curriculum

7.12 Genetics is an overarching theme in undergraduate learning, encompassing basic science and clinical practice at the cutting edge of scientific discovery. Furthermore it involves patient counselling, ethical dilemmas, the application of hard medical science to patients and their families in specialized, secondary and primary care settings. It spans all age groups and potentially connects different specialties.

7.13 The use of genetics as a learning aid in basic medical sciences is then easily picked up as a link into its application into clinical practice. For example whilst the link between clearly inherited disorders and illness in early or later life can be explored, so can the implications of inherited susceptibilities and risks and multifactorial interactions with the environment. This triggers exploration of, for example, ethical themes, prevention and screening.

7.14 Used wisely it should enhance the learning of links between basic science and clinical problems, their impact on doctor-patient and inter-professional communication, and ethics.

Vocational training

7.15 It is vital that the knowledge and skills that are needed by a competent general practitioner in the sphere of genetics should be covered during the three year training programme. Although it may

be important for the purposes of summative examination to be able to quote incidence of disease it is much more important that a practitioner completing vocational training is aware of the major facts in relation to genetic disease, such as the familial pattern of certain cancers and is confident in taking a family history. Many of the skills which are relevant to clinical genetics will be covered in other areas, such as teaching and learning the consultation. Finally the professional values of keeping up-to-date and knowing the limits of one's ability will be more crucial at this stage of a doctor's career.

Continuing professional development

7.16 There is a growing trend for general practitioners to focus their education on perceived gaps in their knowledge and skills. With the passage of time, not only will practitioners' knowledge decay, but unless they strive to keep up-to-date they will not be informed about recent developments. One of the most potent ways of learning is through significant event analysis. Many, but by no means all, practices have introduced clinical meetings. These are usually open to all members of the practice team. Through case presentation and discussion the practice team learns together. Developments in genetics may be brought to such events, so raising awareness of lack of knowledge or skills. It may be easy to discuss an incident in a superficial way, which is less likely to identify learning needs. However, a confident team, or a skilled facilitator should enable deeper discussion to allow sharing of professional values and identification of knowledge or skill gaps. The individuals concerned may choose a specific course or programme to rectify their deficits, or if they are more skilled they may be able to identify their deficiencies more precisely and thereby arrange training to meet specific needs. Evaluation of effectiveness of any learning could be checked through audit.

(For those requiring more formal training there are MSc courses in several centres including London, Manchester, and Glasgow.)

Training the trainers

7.17 Reading this paper may have increased awareness of inherited disease, but at the same time it may have raised concerns in relation to competence and confidence. General practice has three main groups of doctors who are responsible for postgraduate and continuing education – trainers, course organizers and general practice tutors. Each of these groups will have a slightly different function, but they all need to be aware of developments in genetics. They all need to understand basic clinical skills such as the ability to take a family history or how to break bad news, and they must be able to help people learn and refine these skills in a protected setting. Most importantly, their own professional values as medical educators should motivate them to offer educational programmes which will provide practitioners with the competence to deal adequately with genetic issues. Insufficient knowledge may well serve only to raise anxieties in those they are training.

The public perception

8.1 The potential for public misunderstanding of genetics is great since the field is highly complex and difficult for even the expert clinician to grasp fully. However, the impact of knowledge about the new genetics – disseminated through the media – is enormous. As one author has observed:

"Though it is only one conceptual mode, 'genetics' is increasingly identified as the way to reveal health and disease, normality and abnormality" (Lipman, 1994).

8.2 The radical growth of clinical genetics can offer powerful explanations for a range of health problems ranging from relatively trivial conditions to lethal syndromes. These stories compete effectively in everyday descriptions of illness with ideas about infection and lifestyle that have a much longer history (Chapple et al, 1995[a]). Even so, levels of knowledge in the general population, about the means by which such disorders are transmitted remain low (Decruyenaere et al, 1992). In this context, it is vital that health professionals talking about genetic disorders with patients and their relatives have some understanding of their patients' beliefs and knowledge about inheritance. Failure to take account of these beliefs may substantially diminish the effectiveness and utility of any information provided (Richards, 1993).

8.3 The presence of a genetic disorder means that individuals and families have to respond to a highly complex body of medico-scientific knowledge. Many people find it hard to cope with such information when it first becomes relevant to them. Beyond the issue of complexity – which

can be resolved by sensitive explanation of the causal mechanisms involved in the transmission of genetic disorders, and of their implications – lies the uncertain nature of much of the knowledge about clinical genetics itself. There are many thousands of known genetic disorders, and while some are relatively well understood, many are poorly described (Gilbert, 1993).

8.4 An important aspect of public understanding of genetic disorders is the extent to which they are believed to be unavoidable and that it is impossible to protect the individual since they are by definition present from the moment of conception. However this ignores widely applied and successful neo-natal screening for preventable conditions (for example phenylketonuria and congenital hypothyroidism), prenatal diagnosis for a wide range of genetic disorders and, most importantly for the future, the slow but certain progress towards understanding, for example, the genetics of cancer, coronary heart disease and diabetes mellitus.

8.5 It is important to recognize how individuals perceive their own role in transmission, and the part played by other family members. In one British study, feelings of anxiety and culpability have been shown to result from even very sensitive and well conducted expert counselling (Chapple et al, 1995[b]).

8.6 Such feelings may be easily translated into hostility and blame-seeking between family members, since parents and grandparents are, to all intents and purposes, the identifiable sources of the condition (Chapple and May, 1996) and this may amplify the perceived stigma. However much the genetic science is explained, certain groups are likely to continue to believe genetic problems are due to 'God's will', 'the Evil Eye', or key events, for example, during pregnancy.

8.7 Passing on the necessary highly complex information to individuals and their families, therefore, often creates anxiety and apprehension. Limited understanding is associated in a number of studies with difficulties of reconciling a sense of blame – both personally and collectively (Chapple et al, 1995[b]).

8.8 There are wide ranging implications for informing and educating patients, relatives and the general public. Educational interventions of any kind must be tailored to the experience, educational levels and emotional states of the individuals concerned. Information needs to be organized to alleviate anxieties which inhibit understanding, and add to feelings of guilt.

Information technology

9.1 As developments in genetics progress, there will be an increasing need for sophisticated electronic recording systems which will search and organize information in an appropriate way. These systems, based in the practice, will need to be quickly accessible during the consultation if they are to be available when decisions are taken with the patient.

9.2 One way of achieving this rapid access is to hold a genetic information database on the practice server's hard disk. This can be updated automatically at predetermined intervals by downloading over the Internet from a frequently revised central database. This technique is now regularly made available by commercial software providers to their customers. The construction and maintenance of the database would appropriately be a responsibility of the Royal College of General Practitioners.

Evidence-based medicine

9.3 The practice of clinical genetics will need to be based very firmly on evidence. There is no background of experience or 'case law' on which to rely and desirable outcomes are difficult to define (Royal College of Physicians, 1997). As the evidence changes rapidly general practice will need access to the most up-to-date information.

9.4 This imperative will encourage clinicians to look to electronic systems which incorporate genetic protocols or guidelines. However this may not be straightforward as nationally generated guidelines are sometimes perceived to be inappropriate for local issues. As we create more and more guidelines, one looming dilemma is that these may become centralized, complex and interacting. Changing one guideline would then have a ripple-like effect through a whole host of other guidelines. Computer knowledge based systems should solve this problem but will not avoid the desire to develop local guidelines which local clinicians feel they can 'own'.

Information super highway: the Internet

9.5 Increasingly the answer seems to lie in the Internet, an enormous collection of computers in a

huge network across the world. Already many general practitioners can reach the Internet through their practice computers. For them there is immediate access to medical information sources including the *British Medical Journal*[a], genetic information in a massive catalogue of Mendelian disease written by Victor McKusick[b], and medical biosciences[c]. The Cochrane collaboration[d] will be widely used on the network, and this is designed specifically as a guide to treatments which have been assessed by controlled trials.

9.6 The number, range and relevance of Internet sources is expanding very rapidly and groups will be able to publish locally on the Internet to answer the most commonly asked questions. For example a local group of practices working with medical geneticists could set up and regularly update its own guidelines on the Internet. This is not possible with paper-based communication.

9.7 However this will create problems as well as solutions. For example, patients also have access to a plethora of information. Currently there is no control on the quality of information on the Internet. This is likely to be a huge issue over the next few years and will be difficult to manage unless most practices are on line and are aware of what their patients have seen (which, of course, may not always be accurate).

Recording family history

9.8 Few, if any, currently available general practice computer systems have the facility for recording family trees of genetic information. A computer program which incorporates family history taking for cancers of the breast, ovary and colon is in the development stage. The output, based on the input family history, advises the clinician either to refer the patient for specialist advice or, in the majority of cases, permits immediate reassurance without the need for referral.

9.9 There are many software applications which are designed for the construction of flow diagrams, of which a family tree is a simple example. It should not be difficult for suppliers of general practice systems to incorporate these programs. We believe they should do so..

9.10 When considering the relevance of a family history of a disease, the age at diagnosis in each affected family member is a crucial factor. Programs designed to record family history should incorporate a prompt that ensures that this information, if available, is recorded.

Conclusions

Because of many technical advances and patients' anxieties, primary care will increasingly face new demands from patients with real or perceived genetic problems, and demands for genetic screening programmes. These new commitments will be unavoidable because of genuine clinical and preventive opportunities fuelled by the frenetic publicity to which patients are exposed. Medical genetics is in fact the quintessential arena for shared care. The technology and the concepts of probability and risk are sufficiently complex to demand intermittent specialist involvement. Yet only primary care can cater for the life-time needs of patients and families with chronic genetic disabilities and with threat of future disease in family members.

Recommendations

A To consider the commissioning and publication, in the RCGP's clinical series, of an educational package in genetics for the primary care team.

B To consider establishing a task force charged with making detailed recommendations for the effective development of genetic medicine in primary care, perhaps through collaboration with existing groups.

These general principles are expanded below:

1. Consider engaging partners in the planning and delivery of seamless genetics services, training and continuing medical education:

    - The British Society for Human Genetics includes four arms of medical genetics: Clinical Genetics Society, Association of Clinical Cytogeneticists, Clinical Molecular Genetics Society and the Association of Genetics Nurses and Social Workers.
    - The Cancer Family Group is a related and extremely effective professional organization.
    - The Genetic Interest Group (GIG) includes all genetic patient support groups and has an active record of reviewing genetics services and of training courses including those for general practitioners.

    Links with other specialists are also appropriate, for example, with the Royal Colleges of Obstetricians and Gynaecologists and of Pathologists, and with midwives in prenatal and postnatal diagnosis.
2. Consider encouraging the linking of primary and secondary genetics care perhaps by using genetics associates and specialist genetic nurses.

3. Define the implications for manpower and resources.
4. Define new priorities in primary care resulting from a progressive shift of emphasis to genetic screening and prediction, prevention and counselling.
5. Identify and commission genetics training packages for general practitioners, general practitioner registrars, practice nurses and other attached staff.
6. Work with the Directors of Postgraduate General Practice Education (Regional Advisers in General Practice) to raise awareness of genetics in training and continuing medical education programmes.
7. Initiate appropriate continuing medical education courses.
8. Review current research, development, implementation and evaluation in primary care genetics.
9. Encourage the development of general practice computer systems which incorporate facilities for the recording of family trees of those with genetic disorders.
10. Incorporate prompts in family history taking software to ensure that the date of diagnosis and/or death in affected members is always recorded.

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[a] http://www.bmj.com/bmj/
[b] http://www3.ncbi.nlm.nih.gov/Omim/
[c] http://www.ohsu.edu/cliniweb/wwwvl/
[d] http://www.jr2.ox.ac.uk/Bandolier/band16/b16-7.html
Alternatively use http://cebm.jr2.ox.ac.uk/