The general practitioner and the "new genetics"

Linda Mann

ABSTRACT

- GPs are involved in long term care of patients and families with complex conditions. They juggle the need for medical expertise, the relationships between family members, the cost of expertise, limitations of access, and the medicolegal environment. With this background, the GP is ideally placed to play an active role in the "new genetics".
- GP consultations involving the new genetics will include diagnostic testing for patients with clinical problems, preconception and prenatal testing for couples in relation to pregnancy, predictive testing for families with some genetic conditions, and community genetic screening in some circumstances.
- GPs will need to understand the language of the new genetics, undergo continuing education, and receive ongoing support to enable them to communicate effectively with patients and their families.
- Different models of care incorporating GPs, specialists and allied health professionals can be developed to provide maximum delivery of relevant genetic data for both genetic and common multifactorial disorders.

MJA 2003; 179: 109–111

GENERAL PRACTICE should provide comprehensive, coordinated and continuing medical care, drawing on biomedical, psychological, social and environmental understanding of health.[1] The recent advances in knowledge of genetics and the human genome will inevitably require GPs to incorporate the "new genetics" into aspects of daily consultation.

Patients consult their GP for many reasons.[2] At present, most genetic consultations are for single gene disorders such as thalassaemia, cystic fibrosis or haemochromatosis. This is changing fast, as genes are identified that predispose to multifactorial illnesses such as deep venous thrombosis, Alzheimer disease and some forms of cancer. The GP will have a particularly important role in interpreting these tests for patients, and then ensuring that relevant family members are offered appropriate tests in turn.

The new genetics and GP consultations

Diagnostic DNA testing

DNA testing can help diagnose the causes of many of the problems for which patients present to GPs. A common presenting problem is tiredness. In the appropriate circumstances, the GP will order tests, usually including a full blood count and iron studies. The results may lead to further investigations, including DNA tests. Haemochromatosis, a common autosomal recessive condition, is more likely to be diagnosed in the general practice setting by the early symptom of tiredness than by the textbook description of diabetes and skin pigmentation. Similarly, the finding of anaemia or microcytosis will indicate iron deficiency and/or thalassaemia. In this situation, DNA studies may be appropriate to identify an underlying thalassaemia, particularly α-thalassaemia. Factor V Leiden deficiency, or other genetic causes of thrombophilia, may be found with DNA testing after deep vein thrombosis, a condition increasingly treatable in general practice.

Except where limited by the Health Insurance Commission or health department directives, there are no restrictions on GPs ordering DNA tests. Indeed, there are important reasons why GPs should have access to DNA testing options. These include:

- patients have easier access to a GP than to specialist or public hospital services;
- the interval from the taking of blood to the patient receiving the result is often faster;
- the overall costs are likely to be cheaper; and
- a GP who understands the full implications of a DNA test is well placed to communicate this result to the patient (and family), particularly if he or she has established a relationship with the patient.[3]

The testing laboratory should provide information to ensure that the significance of the test (and its limitations) are recognised. Further specialist investigation and intervention can be undertaken by referral.

It is also reasonable to expect that some GPs will refer DNA testing to specialists, because of concern about their knowledge of the new genetics,[4] and the medicolegal responsibility for the consequences of the test. The GP will also carry some responsibility for ensuring that tests are offered to relatives of the index case.

Preconception counselling and prenatal DNA testing

More than 50% of women plan their pregnancy.[5] A consultation before conception (eg, when a woman presents for advice on contraception) is an opportunity to explore genetic conditions that may be suitable for prenatal or preimplantation genetic diagnosis. The family history, which the GP will often be aware of, is highly relevant in determining whether there are particular risks to the fetus, especially if the mother is older.

93-95 Balmain Road, Leichhardt, NSW.
Linda Mann, DipRACOG, FRACGP, General Practitioner.
Reprints will not be available from the author. Correspondence: Dr L Mann, 93–95 Balmain Road, Leichhardt, NSW 2040.
lmann@med.usyd.edu.au

Prenatal testing needs to be completed by a certain gestational age, and the hospital clinic or obstetrician will not always see at-risk patients early enough for prenatal diagnosis to be undertaken in a calm and considered manner. GPs supervise an increasing number of pregnant women in shared-care programs.[6] As the person of first contact, the GP is ideally placed to facilitate DNA prenatal testing by arranging for early investigation or referral to determine if prenatal testing is feasible. Various useful resources are available to guide GPs in these scenarios.[7,8]

Patients expect their GP to inform them in a timely manner of the existence of various prenatal testing options, from carrier testing of the couple for specific conditions, such as cystic fibrosis,[9] to nuchal translucency[10] in the fetus to test for Down syndrome. The new genetics increases the test options the GP needs to be aware of. For example, material collected after chorionic villus sampling in the first trimester of pregnancy can be used for a range of tests, such as fluorescent in situ hybridisation to identify specific chromosome abnormalities,[11] or as a source of DNA to look for mutations in genes (eg, dystrophin to identify the risk of developing Duchenne muscular dystrophy).

Predictive or presymptomatic DNA testing

The presence of a genetic disorder in a family now allows other family members who are at risk to undertake predictive or presymptomatic DNA testing to quantify their risk or exclude the risk before clinical features of the disease become apparent.[12] There are three situations GPs will face:

- A known genetic condition exists in a family member (eg, disorders such as haemochromatosis, familial adenomatous polyposis, or Huntington disease).
- Patients may observe family patterns of disease (eg, cancer), and request DNA testing to predict their own risk.
- Patients may ask their GP about DNA testing options that have been raised in the media.

Conditions like familial adenomatous polyposis or haemochromatosis can be treated. Perhaps more importantly, preventive measures are possible in both. In contrast, Huntington disease is a rare and ultimately fatal neuropsychiatric disorder without any established treatment or prevention option. Therefore, the result from a predictive test in Huntington disease carries with it considerable psychological and societal implications. For optimal patient care, patients who need DNA testing for disorders requiring specialist treatment such as surgery (familial adenomatous polyposis), or rare and serious disorders requiring intensive workup and ongoing support (eg, Huntington disease), should be referred to a clinical geneticist initially. On the other hand, GPs may elect to undertake DNA tests in patients and family members at risk of common or easily treatable conditions such as haemochromatosis. These individuals can be referred for specialist treatment when needed.

Patients may exaggerate their risk of heritable cancer. The information disseminated to GPs by cancer councils[13,14] is useful for providing patients with realistic risk assessments. Specific cancer genes such as *BRCA1* and *BRCA2* are a rare cause of breast or ovarian cancer that runs in families, accounting for 1%–2% of these cancers in the overall population. Risk assessment by pedigree allows GPs to put a patient's concern into perspective. Most patients will end up needing the same level of surveillance as the general population (eg, screening mammography). Patients with appropriate family histories benefit most from referral to the specialist cancer genetics clinics.

Community DNA screening

GPs see unreferred patients, and see them repeatedly over years. Therefore, they are well placed to support and promote community screening programs, which may be based on ethnicity (eg, Tay Sachs in Ashkenazi Jews), newborn screening for genetic disorders (eg, cystic fibrosis and phenylketonuria), or opportunistically (eg, thalassaemia trait in asymptomatic patients with microcytosis).

Challenges in integrating the new genetics into general practice

Specialist medical activity based at teaching hospitals is funded by state departments of health, and supported by university affiliations. Such activity spreads out to general practice for economic as well as medical reasons, as GP consultations are a Commonwealth cost. GPs need to participate in the new genetics, to be part of a team and to have access to suitable referral sources. They should be consulted before changes in their practice leave them undertaking work for which they may feel unskilled.

Who pays for upskilling general practitioners?

Possible funding sources are the Commonwealth, through Divisions of General Practice, or state health departments, through academic and hospital-based departments. It is the state health departments that will benefit most from developing a cohort of GPs knowledgeable in the new genetics and so enhancing community-based care, and ensuring better targeted referrals.

How should genetics training for GPs be delivered?

Computer tools that improve the accuracy of risk assessment, and so enhance referral patterns,[15] are being developed for general practice. Suitable web-based resources are being tested in Victoria and are available to members of the Royal Australian College of General Practitioners (RACGP).[2] Many GPs prefer more traditional methods of learning, which can be expensive and teacher-intensive. Watson et al compared an information pack to inhouse education for upskilling GPs on the genetic aspects of breast cancer. Both systems improved referral decisions significantly, while inhouse sessions increased doctor confidence.[16]

In the United Kingdom, there are initiatives such as a genetics nurse,[17] who could spend three hours per month in each of 20 practices to provide GP education as well as GP and patient support. Another suggestion is a genetic coun-

sellor,[18] who could liaise with the local genetics clinic and the practice, providing genetic counselling to patients. However, the current Australian financial environment has no means to provide such initiatives with ongoing funding.

Models of care for GPs and the new genetics

In the Australian context, GPs are the entry to further medical care in all but some emergency situations. They are gatekeepers of expensive medical care, and they look after the chronic health needs of people with complex conditions.

GPs balance their own knowledge and ability to supervise a patient's medical needs, and the needs of the family, with the availability of specialists, on a background of limited resources for public care, and limited patient means for private care. To decrease the isolation of GPs undertaking the primary care of patients with complex conditions, and to accommodate the shift in care of these patients from state-based institutions to Commonwealth-funded areas of care, new models are being developed. Some are summarised below to illustrate possible approaches that would involve the GP in the new genetics.

Traditional care

This is characterised by GP referral of a patient to a specialist unit that will continue to look after that patient, with variable reference to the GP. Inter-specialist referral ensures high quality scientific care, but communication with the GP may be lacking. Patients (and their families) may suffer uncoordinated care as a result.

Shared care

This can occur informally. In more formalised programs, an institution shares the care of a patient with a professional who has undergone some form of training, and who will then follow an agreed protocol and some form of continuing education. Communication is enhanced, often with a patient-held record.[19] Standard care is expected, but there is often little audit of this.[6]

HIV and HCV model

This model of care has been developed to acknowledge the significant increase in knowledge needed for care in this area, the continuing and changing knowledge, and the limited numbers of GPs who wish to undertake care at this level.[20] If this model were applied to the new genetics, GPs would attend a training program and satisfy the relevant assessment. Training could occur in two stages: an introductory course designed for all GPs with an interest in the new genetics, and an advanced component specifically tailored to GPs who wish to expand their role and, for example, counsel in genetics clinics or in groups. The GPs would be required to designate genetics specialists with whom they would collaborate in patient management and model evaluation. To keep their status, they would be required to maintain a suitable level of continuing medical education to demonstrate their understanding of current trends. The GPs would be able to undertake community-based research in the aspects of genetics that affect their patients.

Different models of care incorporating GPs, specialists and allied health professionals can be developed to provide maximum delivery of relevant genetic data for both genetic disorders and common multifactorial disorders.

References

1. RACGP. Definition of general practice and general practitioner. Available at: http://www.racgp.org.au/document.asp?id=6234 (accessed Apr 2003).
2. Britt H, Bhasale A, Miles DA, et al. The sex of the general practitioner: a comparison of characteristics, patients, and medical conditions managed. *Med Care* 1996; 34: 403-415.
3. Fairhurst K, May C. Knowing patients and knowledge about patients: evidence of modes of reasoning in the consultation? *Fam Pract* 2001; 18: 501-505.
4. Metcalfe S, Hurworth R, Newstead J, Robins R. Needs assessment study of genetics education for general practitioners in Australia. *Genet Med* 2002; 4: 71-77.
5. Marsack CR, Alsop CL, Kurinczuk JJ, Bower C. Pre-pregnancy counselling for the primary prevention of birth defects: rubella vaccination and folate intake [published erratum appears in *Med J Aust* 1996; 165: 130]. *Med J Aust* 1995; 162: 403-406.
6. Gunn J, Lumley J, Young D. Involvement of Victorian general practitioners in obstetric and postnatal care. *Aust Fam Physician* 1998; 27: S78-S83.
7. NSW Genetics Education Program. Prenatal diagnosis: special tests for your baby during pregnancy [booklet]. Sydney: NSW Genetics Education Program, 2001.
8. The Centre for Genetics Education [website]. Available at: http://www.genetics.com.au/ (accessed May 2003).
9. Super M, Barnes R, Greig D. Failure to refer for testing for cystic fibrosis. Doctors must ensure that those with a family history are advised appropriately [editorial]. *BMJ* 2001; 322: 310-311.
10. Snijders RJ, Noble P, Sebire N, et al. UK multicentre project on assessment of risk of trisomy 21 by maternal age and fetal nuchal-translucency thickness at 10–14 weeks of gestation. Fetal Medicine Foundation First Trimester Screening Group. *Lancet* 1998; 352: 343-346.
11. FISH: (Fluorescent *In Situ* Hybridization) [website]. Available at: http://www.slh.wisc.edu/cytogenetics/procedures/FISH/FISH.html (accessed May 2003).
12. Goizet C, Lesca G, Durr A. Presymptomatic testing in Huntington's disease and autosomal dominant cerebellar ataxias. *Neurology* 2002; 59: 1330-1336.
13. The Cancer Council of New South Wales. Screening for cancer: a guide for general practitioners [pamphlet]. Sydney: Cancer Council of New South Wales, 2001.
14. The Cancer Council New South Wales [website]. Available at: http://www.cancercouncil.com.au/ (accessed May 2003).
15. Glasspool DW, Fox J, Coulson AS, Emery J. Risk assessment in genetics: a semi-quantitative approach. *Medinfo* 2001; 10: 459-463.
16. Watson E, Clements A, Yudkin P, et al. Evaluation of the impact of two educational interventions on GP management of familial breast/ovarian cancer cases: a cluster randomised controlled trial. *Br J Gen Pract* 2001; 51: 817-821.
17. Shickle D, Hapgood R, Qureshi N. The genetics liaison nurse role as a means of education and supporting primary care professionals. *Fam Pract* 2002; 19: 193-196.
18. Donnai D, Kerzin-Storrar L, Craufurd D, et al. Tensions in implementing the new genetics. Genetic counsellors could be based in genetic centres but be formally linked to general practice. *BMJ* 2000; 321: 241.
19. Having a baby in Victoria. Models of care explained [website]. Available at: http://maternity.health.vic.gov.au/models/ (accessed May 2003).
20. Australasian Society for HIV Medicine Inc [website]. Available at: http://www.ashm.org.au/ (accessed May 2003).

(Received 27 Sep 2002, accepted 8 Apr 2003) ❑