
The Genetics File

A Resource for General Practitioners

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This resource folder has been produced in collaboration with the following organisations, as part of a Genetics Education Strategy for General Practitioners.

The Victorian Department of Human Services
The Murdoch Childrens Research Institute
Genetic Health Services Victoria
Biotechnology Australia
Royal College of General Practitioners, Victoria
The Cooperative Research Centre for the Discovery of Genes for Common Human Diseases
The University of Melbourne

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Collaborating Organisations



Department of
Human Services

Department of Human Services, State Government of Victoria.

The mission statement, "To enhance and protect the health and well-being of all Victorians, emphasising vulnerable groups and those most in need" encompasses the Department's broad responsibilities and reflects its commitment to maintaining and improving human services in Victoria.



**Murdoch Childrens
Research Institute**

Murdoch Childrens Research Institute

MCRI is the largest research centre in Australia specialising in infant, child and adolescent health. It is based in the Royal Children's Hospital, and has world class research programs in areas as diverse as childhood obesity, diabetes, infectious disease, heart disease, cerebral palsy, cystic fibrosis, leukaemia, asthma, epilepsy, ataxia and prematurity.



Genetic Health Services Victoria

Genetic Health Services Victoria is a partnership between the Department of Human Services (DHS), the Murdoch Childrens Research Institute (MCRI), other health service providers and the community. Genetic Health has the mandate to provide genetic diagnosis and counselling to the people of Victoria and Tasmania.



Biotechnology Australia

Biotechnology Australia is a collaboration of five Commonwealth Government departments. It was created to assist in coordinating the Government's approach to biotechnology, and reports to the Commonwealth Biotechnology Ministerial Council on its progress and achievements.

Biotechnology Australia aims to increase the public's general awareness of biotechnology and its uses, through the provision of balanced and factual information explaining the technology, its applications, and regulations to safeguard people and the environment.

We established the Gene Technology Information Service to meet the growing community need for balanced information on gene technology. Contact the service by the Australia-wide freecall number 1800 631 276.

Email us at ba@biotechnology.gov.au or visit our website at <http://www.biotechnology.gov.au> for further information.



The Royal Australian College
of General Practitioners

Royal Australian College of General Practitioners

The Royal Australian College of General Practitioners (RACGP) is the national leader in setting and maintaining the standards for quality practice, education and research in Australian general practice. It is a strong, viable, professional organisation, which works to advance key concerns for general practitioners, and to improve the standards of health care for all Australians. This includes attention to the role of general practitioners working with Australians living in rural and remote areas, Aboriginal people, people of low socio-economic status, people of culturally and linguistically diverse backgrounds and people with chronic health care problems.



The Cooperative Research Centre for Discovery of Genes of Common Human Diseases

The Cooperative Research Centre for Discovery of Genes for Common Human Diseases (Gene CRC) was established in July 1997 under the Australian Government's Cooperative Research Centre scheme. The mission of the Gene CRC is to enhance the capacity of Australian academia, industry and society to apply the wealth and diversity of genetic information that will accrue over the coming decade and beyond.



The University of Melbourne

The vision is of a University of Melbourne international in character and focus, and world class in the staff and students it attracts, the academic standards to which it adheres; a university adding immense intellectual, cultural and professional energy to the City of Melbourne, and serving Victoria and Australia by performing and being acknowledged as one of the finest universities in the world.

Introduction

The recent explosion of genetic information resulting from the Human Genome Project and other developments in genetics and genetic technology is expected to have major implications for general practitioners. The "GP Genetic Education Strategy Steering Committee" was formed at RACGP Victoria to look at the ways to assist general practitioners to incorporate new genetic knowledge and technology into their day to day practice.

As part of the Project, the Centre for Program Evaluation, University of Melbourne was commissioned to gather information from GPs about their experiences in dealing with genetics in their practices and their needs in regards to genetic education. Their findings are summarised in "Genetics Education for GPs in Victoria - A Needs Assessment" (see appendix for executive summary).

This resource has been developed in response to some of the suggestions made by GPs who participated in the needs assessment and covers those topics identified by GPs as of most relevance and interest.

Terminology

Best practice in genetic counselling recommends using terms such as 'altered genes', 'genes with alterations' and 'variations in genes', rather than the term 'mutation', especially when counselling individuals and families. Consequently, 'altered genes' or equivalent is the term generally used in the content of sections.

HOW TO USE THIS RESOURCE

Each section represents a discrete topic, however the Talking with Families section covers aspects of genetic counselling that are relevant to all sections.

Each section generally follows the format:

- Contents/Acknowledgements
- Key Points
- Background Information
- Appendices
- Patient and Further Information (last page of section)
- Where to refer

The information in "Key Points" of each section provides a 'snap shot' of the topic.

GPs wanting a brief summary of the condition may wish to refer only to the "Key Points". More detailed content is given in the "Background Information" and some of the material from the "Key Points" is repeated as needed.

The content is designed to provide information to assist decision making and is based on the best evidence available at the time of printing. Every effort has been made to ensure that the material in this resource is accurate, however the authors and producers of the resource are not responsible for any inaccuracies or liability whatsoever that may arise from the use of the material provided.

Appendix: Executive Summary from Needs Assessment¹

The explosion of genetic information resulting from the Human Genome Project, and other developments in genetics and genetic technology, is expected to have major implications for GPs in general practice. Some of the new roles and knowledge that GPs will be expected to incorporate include:

- Identifying individuals with, or who may be at risk of, a genetic condition, such that they would benefit from a genetic consultation
- Being familiar with the clinical features of common genetic conditions
- Being able to provide basic non-directive genetic counselling skills thus ensuring that patients can make informed decisions about genetic choices
- Possessing a sound knowledge of the availability of clinical genetics services and being able to refer patients appropriately
- Having the ability to work closely, as part of a health professional team, in the provision of ongoing care for patients with genetic conditions

However, since genetics has never been a major focus in the training of general practitioners, a number of studies from the USA and the UK have clearly identified a lack of relevant knowledge of genetics and appropriate referral practices in currently practising, primary care providers. Furthermore, there are several barriers to the increased uptake of genetics in primary care.

Meanwhile, in Australia there is negligible published information regarding knowledge and attitudes to genetics, genetic technologies or genetics education, among general practitioners. Such information is essential for developing and implementing appropriate continuing medical education strategies, and for evaluating their effectiveness.

As an initial attempt at filling this knowledge gap, the study documented in the second part of this report, was carried out in order to determine the genetics knowledge and education needs of GPs in Victoria. Between late 1999 and early 2000, data about Victorian GPs' experiences in dealing with genetics in their practices and their needs in regards to genetics education, were obtained from five focus group of GPs, one focus group of genetics experts from the Victorian Clinical Genetics Services (VCGS), interviews with two GP educators, a statewide survey of GPs, a survey of Victorian Divisions of General Practice and a group consultation with the Genetics Support Network of Victoria (GSNV). The aim of such data collection was to acquire information that would assist with both the development of appropriate education programs and the creation of useful resources.

Patient Management

All focus groups reported that they had had some recent, if sporadic, experience in managing patients with genetic conditions. The most common conditions faced were pregnancy-related, haemochromatosis, Huntington disease and cancers. In many instances management is patient-driven and often patients know more about the condition than the GP.

Particular problems that GPs faced in management of patients were associated with:

- difficulties explaining genetics in lay terms
- not being able to provide definitive answers
- dealing with patient fear and anxiety.
- counselling effectively, and
- dealing with ethical issues that arise

¹ Hurworth R, Robins R and Metcalfe SA. Genetic Education for GPs in Victoria: A Needs Assessment. A report to the DHS, June 2000.

In relation to ordering genetic tests GPs were often confused about what constituted such a test. Furthermore, very few GPs had much experience in ordering such tests (even though most would have ordered a test some time during their careers). Rather, they would refer to specialists, although again experience in doing so was limited. GPs also said they had little or no experience in referring patients to support groups and were most likely to leave the searching for such groups to the patients themselves. However, there was a belief that those who belong to such groups find them to be beneficial.

In summarising GPs' ability to manage patients with genetic conditions, the tone of response was fairly negative.

Explanations for this include:

- Exposure to need for such management is not an everyday occurrence in general practice (and therefore a deep knowledge about genetics was not seen as particularly relevant)
- Knowledge about genetics is changing so rapidly that GPs find it hard to keep up-to-date
- The management of patients requires sensitive counselling and such activity was perceived as difficult and often not well carried out.

GPs' Current Knowledge and Education Levels

GPs generally admitted that their knowledge concerning genetics was poor.

They felt that in most cases their training in genetics had been severely limited because the area was:

- undertaught
- taught poorly
- presented in a dry and complicated way
- not presented in a systematic way
- not taught again after undergraduate training
- only one of many areas which GPs have to know about and when they had to be selective, genetics was given a low priority

Areas That Need Strengthening and Particular Topics Requested

Areas of knowledge that were said to need strengthening included:

- the genetic bases of some common diseases such as cancers
- risk factors and incidence of genetic diseases
- the latest information and development of genetics
- tests that are available for particular genetic disorders
- antenatal tests and screening
- how to deal with ethical issues

There was also a request for much more on genetic counselling skills, specifically in relation to:

- counselling patients about risk
- pregnancy counselling
- allaying patient fears

A large number of conditions were listed as potential topics.

However, by far the commonest cited in both focus groups and survey were:

- cancers (and in particular bowel and breast cancers)
- haemochromatosis
- antenatal testing
- cystic fibrosis
- thalassaemia
- fragile X syndrome
- Huntington disease
- dementia/Alzheimer disease

However, there was a common request that whatever is taught needs to have a practical application and be able to make a difference. Knowledge for its own sake without being able to change management was not thought to be desirable.

Potential Organisation of Genetics Programs

There was general consensus that any program developed should be short, and be no longer than 10-15 hours. Suggested modes of delivery varied according to learning styles and prior experience.

Possibilities included:

- By distance/on-line mode
- Working in collaboration with the GPDV and local Divisions of General Practice to launch local programs, with appropriate input from genetics services, such as the Victorian Clinical Genetics Services
- A program based on a case-based approach
- An integrated approach where genetics is taught alongside, or as part of, other topics
- Seminars

Other points raised were that programs needed to be:

- relevant to a GP's day-to-day practice
- simple
- interesting and lively
- able to assist GPs to work more closely with genetics experts and consumers

Resources and Materials

GPs asked for resources to be provided in a variety of formats. Both electronic (such as CD-ROM and Internet) and printed material (such as resource folders, workbooks, reference guides) were thought to be necessary to accommodate the various working-styles of individuals. However, rural GPs were especially interested in electronic forms.

GPs also emphasised that such material should contain concise and simple information.

Specific resources suggested included:

- a reference folder
- a resource index
- a telephone contact list for specialists
- a telephone advisory service
- a hot line for up-to-date information
- a guide detailing where to refer patients
- concise points about what to tell patients with certain conditions
- information sheets for patients.

The responses to the survey specifically indicated that GPs would benefit most from access to a reference folder, a telephone advisory service or seminars. This was at odds with the literature which suggested that interventionist approaches have proved to be more effective in fostering change. Therefore, GP 'needs' may be at variance with GP 'wants'.

Much of what was raised by GPs in relation to genetics education and resources was repeated during the Genetics Support Network of Victoria (GSNV) group consultation (such as GPs needing to learn good communication skills and to know where to find appropriate resources). However, participants added that GPs need to learn about how to work with, and appreciate the knowledge of, families in which there are individuals with genetic conditions.

Suggested strategies for developing appropriate genetics education programs and resources are outlined in the full report.^{1,2}

¹ Hurworth R, Robins R and Metcalfe SA. Genetic Education for GPs in Victoria: A Needs Assessment. A report to the DHS, June 2000.

² Metcalfe SA, Hurworth R, Newstead J and Robins R. A Needs Assessment Study of Genetics Education for General Practitioners in Australia. *Genetics in Medicine*, 4: 71-77, 2002.

