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Working in partnership with support services in the era of the "new genetics"

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ABSTRACT

- Patient care in the "new genetics" era encompasses not only the diagnosis of a genetic condition or risk, but also managing the psychosocial, familial and ethical sequelae.
- Partnerships between the medical professional and expert clinical genetics services, support groups, registries and genetics education services provide a framework for this management.
- More than 750 Australian support groups assist individuals and families with genetic conditions through contact with peers, information and education resources for patients and professionals, practical advice about coping and advocacy.

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THE DIAGNOSIS OF A GENETIC DISORDER or an inherited susceptibility in a family member has implications for all relatives. Doctors not only have to address the impact of such a diagnosis on their patients and their families, but also facilitate informed decision-making in regard to the options available and provide support in the aftermath of such decisions.

A range of specialist services is available to general practitioners and other specialists to assist in this role (Box 1). It is essential that such services are not provided in isolation, as each contributes to patient care and provides opportunities to meet the diverse and changing needs of patients and their families.

Support groups

Families affected by a genetic diagnosis relate that the issues often not fully understood by the medical profession include:[1]

- the impact and shock of diagnosis;
- a feeling of social isolation and stigma;
- an overwhelming need for practical and up-to-date information;
- difficulty in dealing with family reactions and questions;
- the need for emotional support systems; and
- development of coping skills through contact with another person with the same genetic condition.

In addressing these issues, support groups are increasingly important resources. More than 750 groups that provide peer support and information for familial conditions are listed in the 2002–2003 Australasian *Genetics Resource Book*.[2] Collaboration between healthcare practitioners and support groups can result in a "union of skills" that addresses these issues.[3] GPs who work with self-help organisations state that this improves the relationship with their patients and the quality of healthcare provided.[3]

Yet, even among GPs who have considerable involvement with and understanding of support groups, linking patients with the groups by direct referral, or even informing their patients of the group, is not routine.[3] This was also observed in the results of a survey of the needs and experiences of 643 genetic support groups and their members conducted by the Centre for Genetics Education, Sydney, in 2000 (unpublished data). Although 93% of respondents ($n = 703$) reported that they would have liked to be informed of a support group by their medical practitioner, this had occurred in only 22% of cases. Instead, respondents obtained information about the support groups from the media, talking to other patients in clinics, friends and family, community centres and through the Internet. Written information at the time of the diagnosis on the available options and services, as well as the impact of the diagnosis, was considered essential in the often difficult task of educating other family members and other involved healthcare professionals. Importantly, respondents' information needs changed with time, as seen by the areas of interest to patients at least one year after diagnosis of a genetic condition in themselves, a child or another family member (Box 2).

In addition to the support groups for people with specific conditions, there are a number of "umbrella" groups throughout Australia (Box 3). Although there is some variation in their approaches, their aims include:

- providing peer support and information for family members affected by a genetic condition;
- facilitating contact with other affected individuals;
- facilitating networking and enhancing communication between professionals and patients and family members;
- improving community and professional knowledge of genetic conditions, their impact and available services;
- advocating for people with genetic conditions; and
- participating in planning of services and policy.

These groups may be, in effect, a centralised "referral service" to help GPs to identify relevant groups to meet the needs of their patients and their own information needs.[3]

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Other support and clinical services

Clinical genetics services

A specialist clinical genetics consultation optimally involves a multidisciplinary team approach with clinical geneticists, genetic counsellors, laboratory specialists and social workers, and involves more than just providing information and diagnostic procedures. The clinical genetics team can guide the patient in making use of the information; enable discussion of the options presented in the context of the patient's beliefs and values, and, by considering the perception and understanding of the risks and burden of the disorder, may assist in facilitating decision-making. This process requires expertise and is demanding and time-consuming. Maximal benefit for the patients and their families will only be achieved by appropriately using the skills and resources provided by the clinical genetics teams together with the GP or specialist.

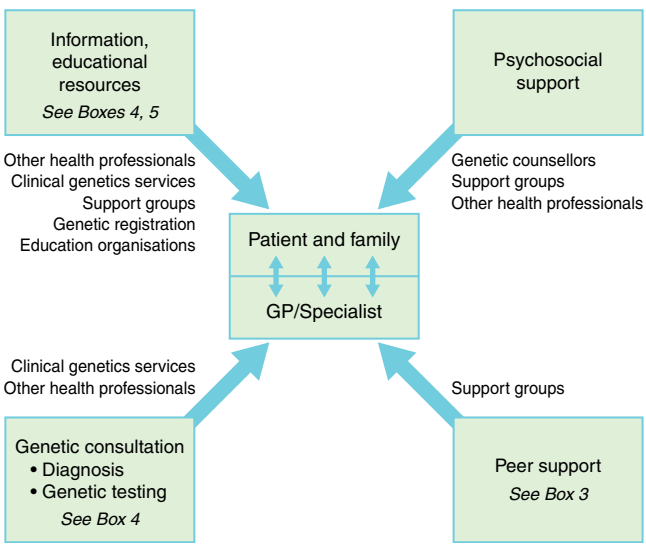
Other healthcare professionals

The GP, specialists or clinical genetics team may refer patients to various other healthcare professionals for medical and psychosocial support. These may include social workers and counsellors (as distinct from genetic counsellors), who provide practical support and long term counselling where the impact of the diagnosis, testing process or decision-making has taken its toll. They are an important part of the multidisciplinary team that works together in a clinical genetics setting.

Genetics registers

Genetics registers have been established for a number of genetic conditions, such as hereditary bowel cancer (familial adenomatous polyposis, and hereditary non-polyposis colon cancer). Register staff can assist in improving risk assessment, provide information about the condition, prevent duplication of genetic testing, help validate genetic test results and facilitate research.[4] Although they are all State-based, the registers liaise to meet the needs of families across geographic borders worldwide.

1: Support services for assisting GPs and patients



Various sources of genetic information, education and support can work in partnership with the treating doctor to meet the family's information and support needs.

Working in partnership in the individual journeys

Diagnostic genetic testing

If symptoms are indicative of a rare clinical condition, or a diagnosis is unclear, a clinical genetics consultation may facilitate a diagnosis. For many, having a name or label to explain their symptoms, and therefore knowing what the future may hold, brings enormous relief.[5] However, it can be isolating to be affected by a very rare condition. This isolation can be ameliorated by contact with other families experiencing the same difficulties and uncertainties.

The importance of support groups after diagnosis of a genetic condition is illustrated by the comments made at an initial support group meeting held by the Association of Genetic Support of Australasia (AGSA) for XXY (Klinefelter) syndrome (incidence of about 1 in 800).[6] The mean age of affected individuals who contacted AGSA was 21 years. Most families had minimal understanding of the condition: fathers thought they were responsible; others wanted to know if the condition would result in their son being homosexual. Parents either had conflicting information about hormone treatment at puberty, or they knew nothing at all. Most families expressed a sense of bewilder-

2: The five most important types of information for different groups at least one year after diagnosis of a genetic condition*

Person with condition	
1	Research update
2	Any changes in information previously provided
3	Practical information on condition management
4	Social service/disability options
5	Contact with other people with the same condition
Parent of person with condition	
1	What will happen in the future
2	Research update
3	Any changes in information previously provided
4	Educational services
5	Social service/disability options
Child or partner of person with condition	
1	Any changes in information previously provided
2	What will happen in the future
3	Research update
4	Practical information on condition management
5	Impact of the diagnosis

* Reported by respondents to a survey of members of support groups (Barlow-Stewart, unpublished data).

ment, anger and frustration. Rather than a perception of being there to "pick up the pieces",[6] as in this situation, support groups should be part of the team working with healthcare providers when a genetic condition is diagnosed (Box 1). Progress in this area is seen by the fact that healthcare professionals comprise more than 50% of the AGSA membership.[6]

Prenatal genetics testing

With prenatal genetic diagnosis, some parents may wish to meet with other parents with a similarly affected child before deciding about continuing the pregnancy. O'Reilly (initiator of the Support After Fetal Diagnosis of Abnormality [SAFDA] support group) acknowledges the sense of isolation that such a diagnosis imposes and the importance of peer support in reducing it.[7] However, she suggests that supporting this course of action may need careful assessment, as support groups may view providing unbiased information and non-directive counselling to families considering terminating the pregnancy as being in conflict with their mission to be pro-active on behalf of those affected by genetic conditions. Some support groups recognise this dilemma and provide distinct services and resources for these families. GPs believe the credibility of a self-help group is influenced by the ability of its members to represent the group, rather than just themselves or their own interests.[3] To balance the need for information and support, referral to a genetics or fetal medicine service for counselling to assist with informed decision-making at this time is essential.

It is in the aftermath of the decision that support groups are an important part of the multidisciplinary team. Where the choice was to terminate the pregnancy, support and information is available from a number of sources (Box 3). O'Reilly reports that the shared-experience meetings held by the SAFDA network (Box 3) enabled opportunities for parents to re-experience, and thus validate, their loss and grief.[7] It was also a chance to address the feelings of numbness, stupor and being distant from reality that can occur at the time of the diagnosis and to be able to speak openly without fear of negative judgement.

> I felt able to express my sadness to other parents and knew they understood. We didn't feel alienated if we wished to talk about our daughter.[8]

If a decision to continue the pregnancy is made, support and information from healthcare professionals and support groups can assist in managing the weeks ahead. The decision may not have been universally supported by doctors or family members, and it can be a very difficult time, but the burden can be ameliorated by enabling contact with other families affected by the same or similar conditions.[8] If the condition is rare, contact with other affected families may be possible by approaching international groups through their websites. During the pregnancy and after the birth, the GP will be central to coordinating the healthcare professionals required for the care of the baby and the parents (Box 1).

3: Support groups in Australia

Support groups for genetic disorders

Australia and New South Wales
Association of Genetic Support of Australasia (AGSA)
66 Albion Street
Surry Hills NSW 2010 — Phone (02) 9211 1462
Email: agsa@ozemail.com.au — Fax (02) 9211 8077
Website: www.agsa-geneticsupport.org.au

Queensland
Self Help Qld Inc
PO Box 353
Sunnybank QLD 4109 — Phone (07) 3344 6919
Email: qnosho@gil.com.au — Fax (07) 3344 6919
Website: www.selfhelpqld.org.au

Victoria
Genetic Support Network of Victoria
10th Floor, Royal Children's Hospital
Flemington Road — Phone (03) 8341 6315
Parkville VIC 3052 — Fax (03) 8341 6390
Email: info@gsnv.org.au
Website: www.gsnv.org.au

Western Australia
Genetic Support Council WA
Level 1, Oasis Lotteries House
37 Hampden Road — Phone (08) 9389 6722
Nedlands WA 6009
Email: info@geneticsupportcouncil.org.au
Website: www.geneticsupportcouncil.org.au

Support groups for abnormality before birth

New South Wales
Support After Fetal Diagnosis of Abnormality (SAFDA)
Royal Hospital for Women
Barker Street — Phone (02) 9382 6670
Randwick NSW 2031 — Fax (02) 9382 6513

Queensland
Queensland Clinical Genetics Service
Royal Children's Hospital — Phone (07) 3636 1686
Herston QLD 4029 — Fax (07) 3636 1987
Email: qcgs@health.qld.gov.au

South Australia
C/- Social Work Department
Women's and Children's Hospital — Phone (08) 8161 7375
North Adelaide SA 5006 — Fax (08) 8161 6088

Victoria
Support After Fetal Diagnosis of Abnormality (SAFDA)
Genetic Services, CHSD Royal Women's Hospital
132 Grattan Street — Phone (03) 9344 2121
Carlton VIC 3053 — Fax (03) 9344 2156

Western Australia
C/- Genetics Services of Western Australia
King Edward Memorial Hospital for Women
374 Bagot Road — Phone (08) 9340 1525
Subiaco WA 6008 — Fax (08) 9340 1678

4: Useful web sites and databases

The Human Genetics Society of Australasia www.hgsa.com.au
Links to current policies and guidelines, including aspects of genetic practice, including genetic testing and counselling.

The Cooperative Research Centre for the Discovery of Genes for Common Human Diseases www.genecrc.org
Includes a learning centre with information about genes and genetics.

National Human Genome Research Institute www.nhgri.nih.gov
Provides information on the Human Genome Project, recent scientific advances, frequently asked questions in genetics and some genetic conditions.

GeneClinics: Medical Genetics Knowledge Base www.geneclinics.org
A medical knowledge base relating genetic testing to the diagnosis, management, and genetic counselling of individuals and families with specific inherited disorders. It is funded by the NIH and developed at the University of Washington, Seattle.

Online Mendelian Inheritance in Man www3.ncbi.nlm.nih.gov/Omim
A catalogue of human genes and genetic disorders, authored and edited by Dr VA McKusick and colleagues at Johns Hopkins and elsewhere, and developed for the Web by the National Center for Biotechnology Information. The database contains textual information, pictures, and reference information.

Betterhealth Channel www.betterhealth.vic.gov.au
Information on genetics and genetic conditions, as well as general health information.

National Breast Cancer Centre www.nbcc.org.au
This site contains a wide range of information about breast cancer and a list of resources.

The Genetics File www.racgp.org.au/folder.asp?id=606
Information for general practitioners about selected genetic conditions.

The Dolan DNA Learning Centre at Cold Spring Harbor Laboratory www.dnalc.org
Clinical, research and educational resources.

Information for genetic professionals, University of Kansas Medical Center www.kumc.edu/gec/geneinfo.html
Provides links to clinical, research and educational resources for genetic counsellors, clinical geneticists and medical geneticists, as well as information on training programs in genetics.

March of Dimes www.modimes.org
Offers information on specific birth defects or infant health problems, pre-pregnancy, pregnancy, teen pregnancy, newborn care, effects of exposure to drugs and environmental hazards during pregnancy, support groups and genetics.

The Centre for Genetics Education www.genetics.com.au
Contains information about genetics services and their contact details, resources available from the program (including fact sheets), a section for students, links to other websites (such as genetic support groups) and an online ethics survey. There are links to other important sites from this website.

NORD Home Page www.rarediseases.org
Provides education, advocacy, research and service for voluntary health organisations serving people with rare disorders and disabilities.

In touch www.intouchlive.com
A site explaining various aspects of cancer, including an educational resource for cancer genetics.

For all of these couples, each future pregnancy is often a "tentative pregnancy",[9] and professional support and information is essential, but it should not be assumed that couples will take the same course of action if a fetal abnormality is found again.

> There have been three occasions on which I have had to consider terminating a pregnancy, which may or may not have been abnormal. Each time, my past experiences, my circumstances, my support systems, my faith and my attitudes have been different; therefore my decision each time has been different also.[7]

Screening and cascade testing

When a "positive" result is found on a screening test, for example for Tay–Sachs disease conducted in the Jewish community[10] or as a result of newborn screening, the condition becomes a "family affair". Education of the family or community is an important component of screening programs, aiming to improve understanding, minimise stigmatisation, and provide options. This may involve a combined approach by clinical genetics services and medical practitioners. Support groups are often contacted directly by the family for information. Usually they can provide information about carrier genetic testing in the case of disorders such as cystic fibrosis. However, the doctor may be able to facilitate better understanding by using information provided by the support groups and material from genetics education providers (Box 4) and discussing it with their patients.

Predictive/presymptomatic genetic testing

Support groups such as the Huntington Disease Association[11] have been at the forefront in working with professional groups to develop predictive testing protocols that are viewed as the paradigm for testing for dominantly inherited, adult-onset genetic conditions (eg, neurological and cancer syndromes).

These protocols address counselling and, in some instances, medical management, highlighting the need for partnership to provide optimal care. Adherence to surveillance recommendations can be affected by emotional and psychosocial reactions to the test result.[12] Both positive and negative emotional and psychosocial responses can occur as a result of these genetic tests.[13] Many may consider knowledge of their genetic status beneficial, but have not considered that a person who does not inherit the disease-causing copy of the gene may no longer share the family bond of being at risk,[5] while those receiving a positive genetic test result can express emotions such as grief, guilt, shame, denial, anger and depression. Genetic counselling prepares individuals to receive and cope with test results, considering the unique experiences, family relationships and coping strategies of the individual. In addition to the ongoing psychosocial support provided by professional counsellors and peers, medical management may need to be revised following genetic testing. Information sources can assist the GP or specialist in providing optimal care for these families.[5]

Public and professional education providers

Clinical genetics services have a major role in providing genetics education for both the community and healthcare professionals in each State and Territory.[14] In some States, dedicated genetics education programs have been established. These providers produce a wide range of pamphlets, booklets and fact sheets designed for patients and their families, as well as material produced for professionals. Various websites in Australia and internationally (Box 4) also provide high quality, updated genetic information.

Medical professionals are increasingly expected to be responsive to the changing needs of their patients and families affected by genetic conditions. Consequently, there is a pressing need for enhanced education and training in the area of genetics.[1,15] Draft recommendations by the Australian Law Reform Commission and the Australian Health Ethics Committee produced as part of the Inquiry into Genetic Privacy[15] are for this to be conducted at the undergraduate, postgraduate and professional levels in areas such as clinical genetics, genetic counselling and related ethical issues. Currently, such education and training are being offered by the staff of clinical genetics services and genetics education programs in consultation with Divisions of General Practice.[15]

Conclusion

Developments in genetics are rapid and are having an increasing impact on the practice of medical professionals. Families seek up-to-date information and psychosocial support from a range of sources. GPs and specialists are able to enhance patient care by availing themselves of the educational activities and resources provided by clinical genetics services and forming interactive relationships with clinical genetics services and support groups. In taking a pro-active approach, GPs and specialists remain central to the patient's care and ensure their varying and evolving needs are met.

Competing interests

None identified.

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