



Genetics

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Definition

General practice genetics deals with the general practice management of hereditary issues, problems and conditions, including those involving the mechanisms of hereditary transmission.

Genetic counselling is a procedure by which patients and their families are given support and advice about the nature and consequences of inherited disorders, the possibility of being affected or having affected children, and the various options available for prevention, diagnosis and management of such conditions.¹

Curriculum in practice

Typical presentations that illustrate how the genetics curriculum applies to general practice include:

- Gary, aged 60 years, has been feeling tired and run down. He says he has been ‘putting on a bit of weight’ and feels uncomfortable in his upper abdomen, but is more troubled by recent joint swelling and tenderness. He has been a construction worker most of his life and believes this is all part of the aging process. He says: “Even the old fella won’t work as well as he used to.” Examination identifies hepatomegaly, but you also notice his skin is a grey-bronze colour. What family history might you specifically ask for and what genetic testing would be appropriate?
- Anna, aged 23 years, is planning her first pregnancy. During her pre-conception counselling you discover her younger brother died when he was 16 years of age from complications of cystic fibrosis. Her husband was originally from the Middle East. Does this increase or decrease the risk that their child might be affected? What tests should she be offered prior to and following conception?
- Stephanie, aged 47 years, has a younger sister who has just been diagnosed with breast cancer. Her older brother commenced treatment for prostate cancer 2 years ago. She is now worried about her own risk of developing cancer and is keen to be tested for everything. Assuming she is currently well and her examination is normal, what advice is appropriate for managing her genetic risk?

Rationale and general practice context

The last decade has witnessed significant advances in genetic medicine, such as the mapping of the human genome and the understanding of genetic causes of disease.

These advances have increasing relevance to clinical care in general practice including the management of people with a family history of cancer and heart disease, carrier testing for common autosomal recessive conditions, and the diagnosis of inherited diseases such as haemochromatosis and thrombophilia.

The role of the GP in genetics

Australian²⁻⁴ and international⁵⁻⁸ studies have highlighted the need for general practitioners to develop genetic literacy and to understand the important role primary care plays in the management of genetic conditions.⁹⁻¹¹ This includes:

- taking and using the family history to determine the risks of common diseases such as cancer and heart disease
- identifying patients with specific genetic conditions who may benefit from referral to genetic counselling or specialist management services
- pre-pregnancy counselling from a genetic perspective, including discussion of prenatal screening and diagnostic tests for genetic conditions
- identifying, assessing and, when appropriate, referring children and adults with developmental delay, developmental disability or dysmorphic features for diagnosis and specialist services
- using genetic tests appropriately, including those listed on the Medicare Benefits Schedule
- being aware of the growing field of genomics and the use of genetic markers to determine therapeutics
- discussing newborn screening programs with parents and managing children who test positive
- supporting families with genetic conditions and co-ordinating their care between clinical genetics services and other clinical specialties.

Genetics can affect many areas of general practice care, and genetic issues may also occur in other curriculum statements.

Training outcomes of the five domains of general practice

1. Communication skills and the patient-doctor relationship

- GENT1.1 Apply communication strategies (appropriate to those receiving the information), when discussing the implications of a genetic diagnosis or genetic test result, including the implications for family members.
- GENT1.2 Address the potential personal impact of a diagnosis of a genetic condition for the patient and their family.
- GENT1.3 Understand the inherent variation in risk perception and use a range of strategies for communication to support informed decision making.
- GENT1.4 Discuss strategies that the patient may use for communication of genetic risk with other family members.
- GENT1.5 Respect the different belief systems that may have an impact on perceptions of health, disability, kinship and understanding of genetic risk.
- GENT1.6 Communicate sensitively when exploring family relationships, including issues of adoption, paternity and consanguinity.
- GENT1.7 Communicate the implications and limitations of genetic tests and their potential to lead to uncertainty.
- GENT1.8 Recognise and develop strategies to support families in the face of uncertainty or when there is lack of clinical diagnosis.

2. Applied professional knowledge and skills

- GENT2.1 Use a three-generation family history to recognise patterns of inherited disease or disability.
- GENT2.2 Use family history information to identify patients who are at increased risk of common, preventable multifactorial conditions.
- GENT2.3 Be aware of the wide range of conditions that may have a genetic factor in their aetiology and the role of disease predisposition genes.
- GENT2.4 Understand the importance of ethnicity in determining risk of certain common inherited conditions.
- GENT2.5 Understand the implications of genetic conditions for other family members who may benefit from genetic counselling.
- GENT2.6 Know the clinical indications for ordering common genetic tests including those on the Medicare Benefits Schedule.
- GENT2.7 Understand the role of genetic tests in the assessment of people with developmental delay, developmental disability and/or dysmorphic features.
- GENT2.8 Know the diagnosis and management of general practice genetic conditions including those described in the NHMRC publication, *Genetics in family medicine: the Australian handbook for general practitioners*.

3. Population health and the context of general practice

- GENT3.1 Understand the process of newborn screening, which conditions are included, and be aware of issues relating to retention and access to the newborn screening cards.
- GENT3.2 Discuss the value of pre-pregnancy counselling from a genetic perspective, including discussing prenatal screening and diagnostic tests for genetic conditions and the protective role of folate.
- GENT3.3 Discuss prenatal screening tests that are available in both public and/or private sectors to support informed reproductive choices.
- GENT3.4 Adhere to screening guidelines for genetic conditions as summarised in the RACGP *Guidelines for preventive activities in general practice* (the 'red book').
- GENT3.5 Recognise that genetic conditions are often lifelong, reflective of many issues related to chronic conditions and disability.
- GENT3.6 Be familiar with, and encourage the appropriate use of, community services such as genetic support groups.

4. Professional and ethical role

- GENT4.1 Recognise the impact of rapid scientific developments on the ability to provide current information and diagnosis, and the benefits of specialist referral in this context.
- GENT4.2 Be aware of your own values and belief systems and how these may have an impact on patient care when dealing with the implications of a genetic diagnosis, or the result of a genetic test (eg. the decision whether to continue or terminate a pregnancy). Also recognise the need, where necessary, for timely referral to another medical practitioner.
- GENT4.3 Understand the family context of genetic conditions and the ethical issues, including the right of access to and need for consent in the disclosure of genetic risk or genetic test results to blood relatives.
- GENT4.4 Discuss the personal and family implications of third party interest, such as employers and insurers, for a genetic diagnosis in a family member or a predictive genetic test result.
- GENT4.5 Recognise the psychosocial impact of a genetic diagnosis or genetic risk and provide patients with appropriate support or referral.

5. Organisational and legal dimensions

- GENT5.1 Discuss the ethical, legal and social implications of common genetic tests.
- GENT5.2 Maintain confidential medical records to include information about genetic conditions and genetic risks.
- GENT5.3 Maintain confidential medical records that adhere to privacy legislation when recording or disclosing information to, or about, other family members.
- GENT5.4 Understand how privacy laws can have an impact on communication about genetic conditions within families.
- GENT5.5 Understand the role of clinical genetics services and how to access them.

Learning objectives across the GP professional life

Medical student

1. Communication skills and the patient-doctor relationship

- GENLM1.1 Demonstrate sensitivity to the personal beliefs of patients and their family, and the impact this has on a genetic diagnosis and the actions that follow this diagnosis.
- GENLM1.2 Describe how common genetic conditions arise and what their impact might be on the individual and their family.

2. Applied professional knowledge and skills

- GENLM2.1 Be able to notate a three-generation family tree and recognise modes of inheritance.
- GENLM2.2 Describe how DNA technology is applied in diagnostic investigations.
- GENLM2.3 Demonstrate a functional understanding of the molecular basis of inheritance and the DNA processes involved in different modes of inheritance.

3. Population health and the context of general practice

- GENLM3.1 Describe the importance of gene environment interactions in predisposition to disease and/or disability.

4. Professional and ethical role

- GENLM4.1 Describe the ethical and personal issues and privacy implications for the patient, their family and the doctor in genetic diagnosis.

5. Organisational and legal dimensions

- GENLM5.1 Describe the role of genetic counselling.

Learning objectives across the GP professional life

Prevocational doctor

Assumed level of knowledge – medical student

1. Communication skills and the patient-doctor relationship

GENLP1.1 Describe how to undertake pre-pregnancy counselling and advise on available prenatal testing and discuss patient options.

2. Applied professional knowledge and skills

GENLP2.1 Demonstrate knowledge of common genetic conditions and the GP's role in the multidisciplinary team that cares for patients with these conditions.

3. Population health and the context of general practice

GENLP3.1 Understand the genetic implications in multifactorial common medical conditions.

4. Professional and ethical role

GENLP4.1 Demonstrate an awareness of the ethical and personal issues and privacy implications for the patient, their family and the doctor in genetic diagnosis.

5. Organisational and legal dimensions

GENLP5.1 Understand the appropriate use of genetic testing and referral for assessment and care by clinical genetic services in the prevocational setting.

Learning objectives across the GP professional life

Vocational registrar

Assumed level of knowledge – prevocational doctor

1. Communication skills and the patient-doctor relationship

GENLV1.1 Demonstrate the ability to undertake prenatal counselling, recognise complexity and refer accordingly, and support the parents over the consequences of testing.

2. Applied professional knowledge and skills

GENLV2.1 Demonstrate how to recognise and manage the general practice aspect of the care of patients with genetic conditions over time, including considerations of the patient within their family and community.

GENLV2.2 Describe the implications and consequences of predictive, predisposition testing for later onset disorders.

GENLV2.3 Outline the diagnosis and management of general practice genetic conditions including those described in the NHMRC publication, *Genetics in family medicine: the Australian handbook for general practitioners*.

3. Population health and the context of general practice

GENLV3.1 Develop and apply practice systems that support routine screening for genetic conditions according to the RACGP 'red book'.

4. Professional and ethical role

GENLV4.1 Manage tensions between the patient with a genetic condition and their right to privacy, the implications for the patient's family, third party interest in the condition, and the doctor's own values and social beliefs.

5. Organisational and legal dimensions

GENLV5.1 Understand the appropriate use of genetic testing and referral for assessment and care by clinical genetic services in the prevocational setting.

Learning objectives across the GP professional life

Continuing professional development

Assumed level of knowledge – vocational registrar

1. Communication skills and the patient-doctor relationship

GENLC1.1 Identify gaps in communication skills and attitudes, including genetic counselling in relation to genetic conditions.

2. Applied professional knowledge and skills

GENLC2.1 Identify gaps in knowledge, skills and attitudes in relation to genetic conditions including screening and its consequences.

3. Population health and the context of general practice

GENLC3.1 Identify gaps in knowledge in relation to population based issues of genetic conditions including screening and its consequences.

4. Professional and ethical role

GENLC4.1 Identify and access professional development and resources in the area of genetic conditions and genetic counselling to maintain functional knowledge of this rapidly developing domain.

5. Organisational and legal dimensions

GENLC5.1 Maintain and update knowledge of community resources to support patients with genetic conditions, including specialist centres and community support groups.

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