

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

Rationale

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 general practitioner in genetics

Australian²⁻⁴ and international⁵⁻⁷ studies have highlighted the need for general practitioners (GPs) to develop genetic literacy and understand the important role played by primary care in the management of genetic conditions. 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 for genetic counselling or specialist management
- 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
- be 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 coordinating 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.

The five domains of general practice – genetics

Communication skills and the patient – doctor relationship

General practitioners need to be able to:

- apply communication strategies, appropriate for those receiving the information, in discussing the implications of a genetic diagnosis or genetic test result including the implications for family members
- address the potential personal impact of a diagnosis of a genetic condition in themselves or their family
- understand the inherent variation in risk perception and use a range of strategies for its communication to support informed decision making
- discuss strategies that the patient may use for communication of genetic risk with other family members
- respect the different belief systems that may impact on perceptions of health, disability, kinship and understanding of genetic risk
- communicate sensitively when exploring family relationships, including issues of adoption, paternity and consanguinity
- communicate the implications and limitations of genetic tests and their potential to lead to uncertainty, and
- recognise and develop strategies to support families in the face of uncertainty or lack of a clinical diagnosis.

Applied professional knowledge and skills

General practitioners need to:

- use a three generation family history to recognise patterns of inherited disease or disability
- use family history information to identify patients who are at increased risk of common, preventable multifactorial conditions
- be aware of the wide range of conditions that may have a genetic factor in their aetiology and the role of disease predisposition genes
- understand the importance of ethnicity in determining risk of certain common inherited conditions
- understand the implications of genetic conditions for other family members who may benefit from genetic counselling
- know the clinical indications for ordering common genetic tests including those on the Medicare Benefits Schedule, and
- understand the role of genetic tests in the assessment of people with developmental delay, developmental disability and/or dysmorphic features.

Population health and the context of general practice

General practitioners need to be able to:

- 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
- discuss the value of prepregnancy counselling from a genetic perspective, including discussing prenatal screening and diagnostic tests for genetic conditions and the protective role of folate
- discuss prenatal screening tests that are available in both public and/or private sectors to support informed reproductive choices
- adhere to screening guidelines for genetic conditions as summarised in the RACGP *Guidelines for preventive activities in general practice* (the 'red book')⁸
- recognise that genetic conditions are often lifelong, reflecting many issues related to chronic conditions and disability, and
- be familiar with, and encourage the appropriate use of, community services such as genetic support groups.

Professional and ethical role

General practitioners need to be able to:

- 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
- be aware of own values and belief systems and how these may 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) and the need where necessary, for extremely timely referral to an alternative medical practitioner
- understand the family context of genetic conditions and the ethical issues, including right of access to, and need for, consent in the disclosure of genetic risk or genetic test results to blood relatives
- discuss the personal and family implications of third party interest (eg. employers and insurers) in a genetic diagnosis in a family member or a predictive genetic test result, and
- recognise the psychosocial impact of a genetic diagnosis or genetic risk and provide patients with appropriate support or referral.

Organisational and legal dimensions

General practitioners need to:

- discuss the ethical, legal and social implications of common genetic tests
- maintain confidential medical records to include information about genetic conditions and genetic risks
- maintain medical records that adhere to privacy legislation when recording or disclosing information to, or about, other family members
- understand how privacy laws impact on communication about genetic conditions within families, and
- understand the role of clinical genetics services and how to access them.

Learning objectives across the GP professional life

Medical student

Communication skills and the patient-doctor relationship

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

Applied professional knowledge and skills

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

Population health and the context of general practice

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

Professional and ethical role

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

Organisational and legal dimensions

- Describe the role of genetic counselling.

Learning objectives across the GP professional life

Prevocational doctor

Assumed level of knowledge – medical student

Communication skills and the patient-doctor relationship

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

Applied professional knowledge and skills

- Demonstrate knowledge of common genetic conditions and the GP's place in the team that cares for patients with these conditions.

Population health and the context of general practice

- Understand the genetic implications in multifactorial, common medical conditions.

Professional and ethical role

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

Organisational and legal dimensions

- 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

Communication skills and the patient-doctor relationship

- Demonstrate the ability to undertake prenatal counselling, recognise complexity and refer accordingly, and support the parents with the consequences of testing.

Applied professional knowledge and skills

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

Population health and the context of general practice

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

Professional and ethical role

- 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.

Organisational and legal dimensions

- Understand the appropriate use of genetic testing and referral for assessment and care by clinical genetic services in the community setting.

Learning objectives across the GP professional life

Continuing professional development

Assumed level of knowledge – vocational registrar

Communication skills and the patient-doctor relationship

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

Applied professional knowledge and skills

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

Population health and the context of general practice

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

Professional and ethical role

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

Organisational and legal dimensions

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

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