COMMON PRESENTATIONS AND CONDITIONS

Basic Trainees will require a sufficient depth of knowledge of these presentations and conditions.

- Cardiac syndromes
  » Brugada syndrome
  » long QT syndrome
- Cystic fibrosis
- Dementias
- Familial cancer syndromes
- Haemochromatosis
- Huntington disease
- Inherited dementia syndromes
- Klinefelter syndrome
- Marfan syndrome
- Multiple endocrine neoplasia
- Myotonic dystrophy
- Neurofibromatosis (NF-1)
- Trisomy 21
- Turner syndrome

For all common presentations, Basic Trainees will need to know how to:

Synthesise
  » incorporate epidemiology, pathophysiology and clinical science
  » recognise the clinical presentation
  » take a relevant clinical history
  » conduct an appropriate examination
  » establish a differential diagnosis
  » plan and arrange appropriate investigations

Manage
  » provide initial, evidence-based management
  » discuss the principles of ongoing management
  » apply quality use of medicines
  » recognise potential complications of the disease and its management, and initiate preventative strategies
  » refer appropriately

Consider other factors
  » identify broader considerations and their impact on diagnosis and management
LESS COMMON OR MORE COMPLEX PRESENTATIONS AND CONDITIONS

Basic Trainees will need to have an awareness of, and an understanding of appropriate resources that should be used to help manage patients with these presentations and conditions.

- Other genetic neurocutaneous syndromes such as Sturge-Weber syndrome
- Urea cycle disorders

For all less common and more complex presentations, Basic Trainees will need to know how to:

**Synthesise**
- incorporate epidemiology, pathophysiology and clinical science
- recognise the clinical presentation
- take a relevant clinical history
- conduct an appropriate examination
- establish a provisional diagnosis
- plan and arrange appropriate initial investigations

**Manage**
- initiate therapy in consultation
- discuss broad therapeutic options
- recognise potential complications
- refer appropriately

**Consider other factors**
- identify broader considerations and their impact on diagnosis and management

EPIDEMIOLOGY, PATHOPHYSIOLOGY AND CLINICAL SCIENCE

Basic Trainees will be able to describe the principles of the foundational sciences.

- Basic principles of pharmacogenetics and individualised medicine
- Definitions of polymorphism, mutation, sex-linked, multifactorial inheritance and polygenic inheritance
- Genetic testing techniques, such as polymerase chain reaction (PCR), gene sequencing, chromosomal microarray, and exome and genome sequencing
- Principles of major cancer genetics
- Inheritance:
  - Mendelian
  - mitochondrial
  - parental disomy
  - polygenic
  - repeating triplet sequences
  - sex-linked
- Process of defining pathogenicity of mutations
- Structure and function of human cells, genes, DNA, RNA, and proteins
INVESTIGATIONS AND PROCEDURES

Basic Trainees will know how to select and interpret the results of these investigations and procedures.

- Know how to select and interpret the results of:
  » chromosome microarray (CMA)
  » conventional karyotype
  » single gene testing (e.g. cystic fibrosis (CF), myotonic dystrophy, spinocerebellar ataxia)
- Discuss the results of other commonly performed genetic testing:
  » attenuated phenotype
  » cystic fibrosis mutation testing
  » variants of uncertain significance (VOUS) or reduced penetrance
- Indications for appropriate referral to Clinical Genetics services, and the process for this, including referral for prenatal testing, carrier testing, and pre-implantation genetic diagnosis

IMPORTANT SPECIFIC ISSUES

Basic Trainees will be able to identify important specialty-specific issues and their impact on diagnosis and management.

- Constructing and interpreting genograms, particularly in relation to determining mode of inheritance
- Goals and potential benefits of the Human Genome Project
- Legal and ethical principles of genetic testing including:
  » need for written consent
  » predictive testing processes
  » ethical barriers to testing minors for adult onset conditions
  » implications to a patient and family of a genetic diagnosis
- Patient and family counselling regarding findings of variants of uncertain significance (VOUS) and incidental findings in genetic testing, including the absence of prognostic information, need for family studies, and possibility of functional studies

LEARNING METHODS

Suggested opportunities, activities, and resources to assist with learning.

- Clinical experience with genetics patients in a range of settings
- Centre For Genetics Education www.genetics.edu.au
- Gene Reviews Series www.genereviews.org
- Genetic databases such as:
  » Pictures of Standard Syndromes and Undiagnosed Malformations (POSSUM) www.possum.net.au
  » Online Mendelian Inheritance in Man (OMIM) www.omim.org