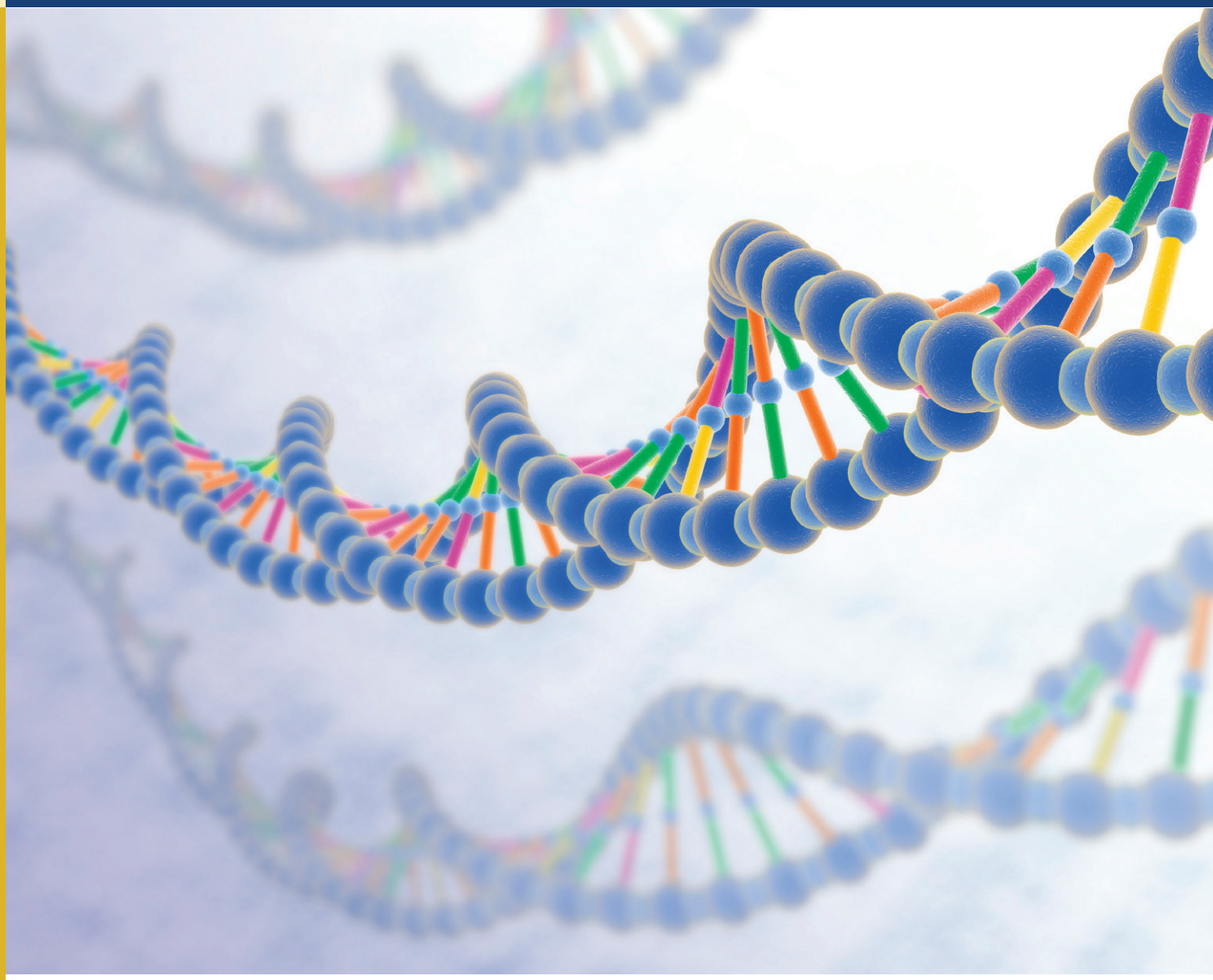
The Royal Australasian College of Physicians

Clinical Genetics

Advanced Training Curriculum

Adult Medicine Division

Paediatrics & Child Health Division



Human Genetics Society of Australasia

The Royal Australasian College of Physicians

Physician Readiness for Expert Practice (PREP) Training Program

Clinical Genetics Advanced Training Curriculum

TO BE USED IN CONJUNCTION WITH:

Basic Training Curriculum - Adult Internal Medicine

Basic Training Curriculum - Paediatrics & Child Health

Professional Qualities Curriculum

ACKNOWLEDGEMENTS

Fellows, trainees and RACP staff have contributed to the development of this curriculum document.

The College specifically thanks those Fellows and trainees who have generously contributed to the development of these curriculum documents, through critical comments drawn from their knowledge and experience and the donation of their time and professional expertise.

The following Fellows and Genetic Counsellors deserve specific mention for their contribution:

- Prof Agnes Bankier, FRACP
- Dr Joanne Dixon, FRACP
- A/Prof Michael Gabbett, FRACP
- Dr Edwin Kirk, FRACP
- Ms Mona Saleh
- Prof Ravi Savarirayan, FRACP
- Dr Graeme Suthers, FRACP
- Dr Anne Turner, FRACP
- Dr Callum Wilson, FRACP
- A/Prof Stephen Withers, FRACP
- Dr Sharron Worthington

We also thank the members of the independent review panel who critically evaluated the penultimate draft.

- Dr Christopher Barnett, FRACP
- Dr Gareth Baynam, FRACP
- Prof John Christodoulou, FRACP
- Prof Martin Delatycki, FRACP
- Dr Ken Maclean, FRACP
- A/Prof Julie McGaughran, FRACP
- Dr Kate Neas, FRACP
- Dr Nicola Poplawski, FRACP
- Dr Rani Sachdev
- Dr Sarah Sandaradura

Development of the Clinical Genetics Advanced Training Curriculum content was overseen by the Specialist Advisory Committee in Clinical Genetics.

The process was managed by the Curriculum Development Unit within the College's Education Deanery, who designed the document, drafted content material, organised and facilitated writing workshops, developed resource materials, and formatted the final document.

The RACP gratefully acknowledges the contribution of the Human Genetics Society of Australasia to the development of this curriculum.

CONTACT DETAILS

THE ROYAL AUSTRALASIAN COLLEGE OF PHYSICIANS

AUSTRALIA
145 Macquarie Street
Sydney
NSW 2000
Australia

Tel: (+61) (2) 9256 5444
Fax: (+61) (2) 9252 3310

Email: racp@racp.edu.au
Website: www.racp.edu.au

NEW ZEALAND
5th Floor
99 The Terrace
Wellington
New Zealand

Tel: (+64) (4) 472 6713
Fax: (+64) (4) 472 6718

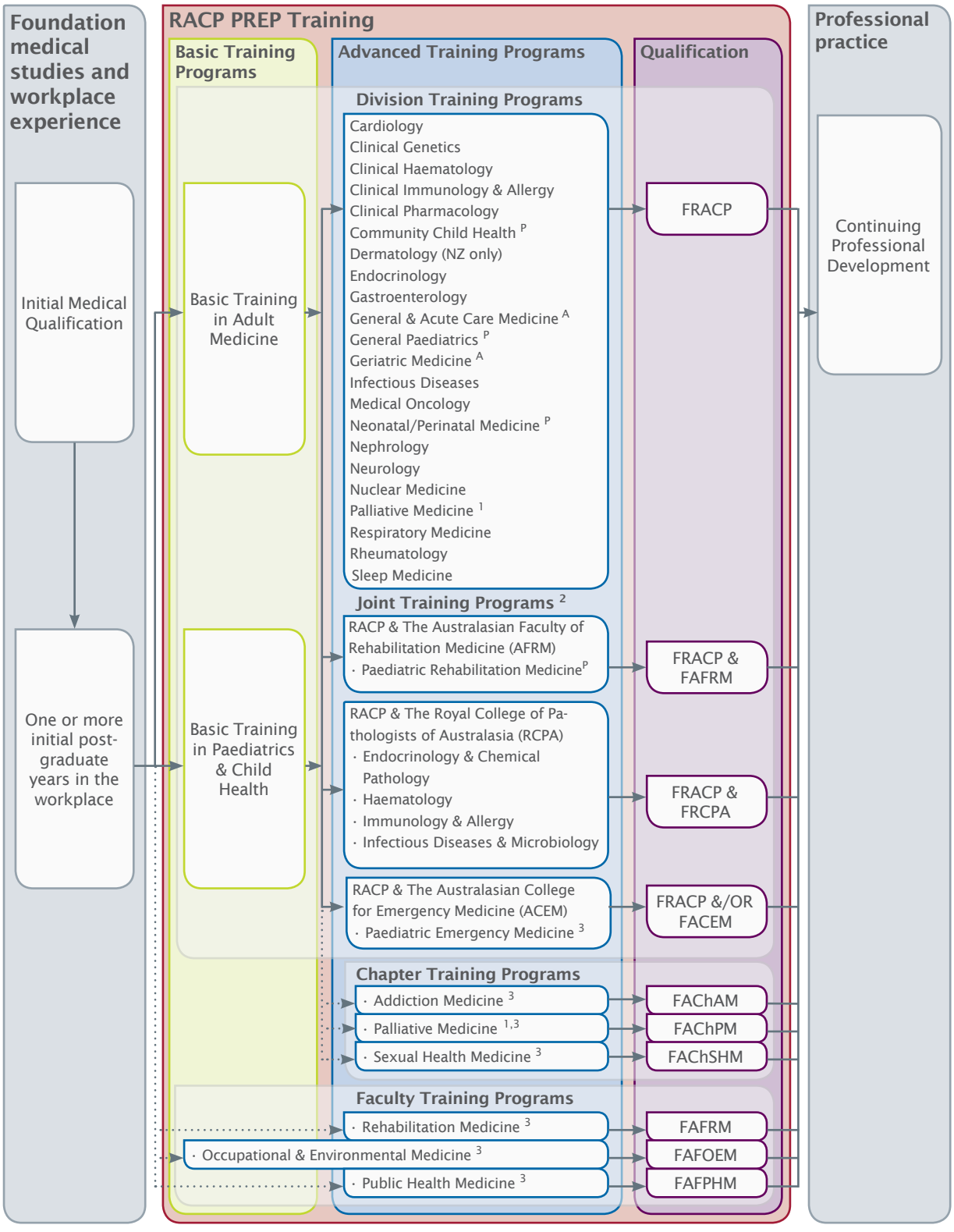
Email: racp@racp.org.nz
Website: www.racp.edu.au

COPYRIGHT

1st edition 2010 (revised 2013).

Please note: No Domains, Themes or Learning Objectives have been updated for this edition; design changes ONLY.

RACP FELLOWSHIP TRAINING PATHWAYS AND THE CONTINUUM OF LEARNING



P Trainees must complete Basic Training in Paediatrics & Child Health to enter this program.
A Trainees must complete Basic Training in Adult Medicine to enter this program.
1 Trainees who have entered Advanced Training in Palliative Medicine via a RACP Basic Training Program will be awarded FRACP upon completion and may subsequently be awarded FAChPM. Trainees who have NOT entered Advanced Training in Palliative Medicine via a RACP Basic Training Program will only be awarded FAChPM upon completion.
2 The Child & Adolescent Psychiatry Joint Training Program with the Royal Australian and New Zealand College of Psychiatrists (RANZCP) is currently under review by the RACP and RANZCP and closed to new entrants at present.
3 Alternative entry requirements exist for these training programs; please see the corresponding PREP Program Requirements Handbook for further information.

NB1: This diagram only depicts training programs that lead to Fellowship. Please see the RACP website for additional RACP training programs.
NB2: For further information on any of the above listed training programs, please see the corresponding PREP Program Requirements Handbook.

OVERVIEW OF THE SPECIALTY

Clinical genetics is a subspecialty which is taking on greater importance in the management of patients throughout life. We are continuing to identify genetic mutations which lead to disease processes. As a result of this, our ability to provide timely and useful advice to patients and families is steadily improving. Clinical genetics requires trainees to become well schooled in both the science of genetics and the art of genetic counselling. In addition to developing skills in syndrome identification, it is also necessary to understand the complexities and limitations of different tests that are currently available. With the rapid progress in technology, it is necessary for clinical geneticists to be in touch with the latest advances in the field so that they are able to provide the best available advice during consultations. Trainees must become skilled in critical assessment of relevant literature.

CURRICULUM OVERVIEW

Clinical Genetics – Advanced Training Curriculum

This curriculum outlines the broad concepts, related learning objectives and the associated theoretical knowledge, clinical skills, attitudes and behaviours required and commonly utilised by clinical genetic physicians within Australia and New Zealand. The curriculum includes programs in genetic metabolic medicine and cancer genetics.

The purpose of Advanced Training is for trainees to build on the clinical skills acquired during Basic Training to acquire the clinical skills of a subspecialist in the chosen field. At the completion of the Clinical Genetics Advanced Training Program, trainees should be competent to provide, at consultant level, unsupervised comprehensive medical care in clinical genetics.

Attaining competency in all aspects of this curriculum is expected to take three years of training. It is expected that all teaching, learning and assessment associated with the Clinical Genetic Advanced Training Curriculum will be undertaken within the context of the physician's everyday clinical practice and will accommodate discipline-specific contexts and practices as required. As such it will need to be implemented within the reality of current workplace and workforce issues and the needs of health service provision.

There may be learning objectives that overlap with or could easily relate to other domains; however, to avoid repetition, these have been assigned to only one area. In practice it is anticipated that within the teaching/learning environment, the progression of each objective would be explored.

Note: The curricula should always be read in conjunction with the relevant College Training Handbook available on the College website.

Professional Qualities Curriculum

The Professional Qualities Curriculum (PQC) outlines the range of concepts and specific learning objectives required by, and utilised by, all physicians, regardless of their specialty or area of expertise. It spans both the Basic and Advanced Training Programs and is also utilised as a key component of the Continuing Professional Development (CPD) program.

Together with the various Basic and Advanced Training Curricula, the PQC integrates and fully encompasses the diagnostic, clinical, and educative-based aspects of the physician's/paediatrician's daily practice.

Each of the concepts and objectives within the PQC will be taught, learnt and assessed within the context of everyday clinical practice. It is important, therefore, that they be aligned with, and fully integrated into, the learning objectives within this curriculum.

EXPECTED OUTCOMES AT THE COMPLETION OF TRAINING

Graduates from this training program will be equipped to function effectively within the current and emerging professional, medical and societal contexts. At the completion of the Clinical Genetics Advanced Training Program, as defined by this curriculum, it is expected that a new Fellow will have developed the clinical skills and have acquired the theoretical knowledge for competent clinical genetic practice. It is expected that a new Fellow will be able to:

- draw and interpret family history data
- obtain medical history and carry out a clinical examination as it relates to genetic diseases
- diagnose genetic disease using clinical evaluation and genetic testing
- choose appropriate investigations and interpret results
- provide accurate information and effective genetic counselling to individuals and families
- write clear summaries of genetic clinic consultations in post-clinic letters to colleagues and patients
- formulate management plans for genetic/hereditary disorders
- perform risk calculation, including the use of Bayes theorem
- carry out phlebotomy, skin biopsy, hair root extraction, cheek swab collection, and clinical photography
- conduct literature searches and use medical genetics databases
- store and retrieve genetic data in genetic registers
- work effectively in a team with other colleagues providing genetic services
- liaise appropriately with colleagues from other specialties
- work with lay organisations to support patients and families with genetic diseases
- communicate and explain genetic issues to colleagues and the lay public
- work effectively with colleagues in other disciplines
- conduct research within the discipline
- participate in teaching
- understand ethical, legal, social and cultural issues in the context of clinical genetics and in the context of clinical, epidemiological and laboratory research.

CURRICULUM THEMES AND LEARNING OBJECTIVES

Each of the curriculum documents has been developed using a common format, thereby ensuring a degree of consistency and approach across the spectrum of training.

Domains

The domains are the broad fields which group common or related areas of learning.

Themes

The themes identify and link more specific aspects of learning into logical or related groups.

Learning Objectives

The learning objectives outline the specific requirements of learning. They provide a focus for identifying and detailing the required knowledge, skills and attitudes. They also provide a context for specifying assessment standards and criteria as well as providing a context for identifying a range of teaching and learning strategies.

LEARNING OBJECTIVES TABLES

DOMAIN 1	**SCIENTIFIC BASIS OF CLINICAL GENETICS**
Theme 1.1	Fundamentals of Inheritance and Genetics Theory
Learning Objectives	
1.1.1	Define cellular and molecular mechanisms that underpin inheritance in human beings
1.1.2	Define patterns of inheritance and undertake risk assessment
1.1.3	Describe the evolution of genetic knowledge
DOMAIN 2	**DIAGNOSTIC METHODS, INVESTIGATIONS AND MANAGEMENT**
Theme 2.1	Clinical Skills
Learning Objectives	
2.1.1	Investigate, diagnose and manage genetic conditions
2.1.2	Use available resources to inform diagnostic techniques
Theme 2.2	Genetic Testing
Learning Objectives	
2.2.1	Apply appropriate diagnostic procedures and interpret results of genetics tests
2.2.2	Interpret genetic laboratory results

2.2.3	Describe emerging genetics technologies and their application
Theme 2.3	Genetic Screening Programs and Registers
Learning Objectives	
2.3.1	Explain the processes to establish and operate genetic screening programs
Theme 2.4	Genetic Counselling
Learning Objectives	
2.4.1	Provide genetic counselling as part of a multidisciplinary team
DOMAIN 3	**GENETIC DISORDERS AND DISEASES**
Theme 3.1	Prenatal Assessment
Learning Objectives	
3.1.1	Conduct prenatal genetic assessments and counselling
Theme 3.2	Neurogenetics
Learning Objectives	
3.2.1	Assess, diagnose and treat patients with neurogenetic disorders
Theme 3.3	Skeletal Dysplasias
Learning Objectives	
3.3.1	Assess, diagnose and treat patients with skeletal dysplasias
Theme 3.4	Dysmorphic Syndromes
Learning Objectives	
3.4.1	Assess, diagnose and treat patients with dysmorphic syndromes
Theme 3.5	Cancer Genetics
Learning Objectives	
3.5.1	Assess, diagnose and contribute to the multidisciplinary management of patients with genetic cancer syndromes
Theme 3.6	Other Genetic Disorders and Diseases
Learning Objectives	
3.6.1	Assess, diagnose and treat patients with other genetic disorders and diseases

DOMAIN 4	PROFESSIONAL QUALITIES OF A CLINICAL GENETICIST
Theme 4.1	Ethics
Learning Objectives	
4.1.1	Identify ethical and legal issues related to clinical genetics practice
Theme 4.2	Health Education
Learning Objectives	
4.2.1	Provide education on risk factors and management of genetic disease
Theme 4.3	Research
Learning Objectives	
4.3.1	Plan and execute a clinical or basic genetics research project
Theme 4.4	Dealing with Medical Uncertainty
Learning Objectives	
4.4.1	Identify and discuss factors contributing to uncertainty in clinical genetics practice
DOMAIN 5	**METABOLIC MEDICINE**
Theme 5.1	Scientific Basis of Metabolic Medicine
Learning Objectives	
5.1.1	Interpret information on the scientific basis of metabolic conditions
Theme 5.2	Diagnostic Methods, Investigations and Management in Metabolic Medicine
Learning Objectives	
5.2.1	Elicit a comprehensive history from a patient
5.2.2	Conduct an examination
5.2.3	Select, perform and interpret appropriate investigations
5.2.4	Synthesise findings to formulate a diagnosis
5.2.5	Manage patients with metabolic disorders

DOMAIN 1	SCIENTIFIC BASIS OF CLINICAL GENETICS
Theme 1.1	Fundamentals of Inheritance and Genetics Theory
Learning Objective 1.1.1	Define cellular and molecular mechanisms that underpin inheritance in human beings

Knowledge	Skills
• define the chromosomal basis of heredity (mitosis and meiosis) • describe the mechanisms of origin of numerical and structural chromosome abnormalities • describe the behaviour of structural chromosome abnormalities at meiosis • identify the chemical structure of DNA and replication • describe the central dogma of cell biology and explain the transcription and translation process • describe the principles of genetic variation.	• identify and critically evaluate information to inform diagnoses • recognise different inheritance patterns in pedigrees • conduct pedigree-based calculations of segregation ratios for structural chromosome abnormalities.

DOMAIN 1	SCIENTIFIC BASIS OF CLINICAL GENETICS
Theme 1.1	Fundamentals of Inheritance and Genetics Theory
Learning Objective 1.1.2	Define patterns of inheritance and undertake risk assessment

Knowledge	Skills

• describe modes of inheritance, Mendelian and non-Mendelian, including mitochondrial inheritance
• explain how empiric risks are derived and used
• conduct risk calculations, including combinatorial probability and Bayes theorem
• describe the mechanism of imprinting and triplet repeat mutations
• describe the regulation of gene expression
• describe the molecular basis of somatic mutations and the cause and consequences of somatic chromosomal variation.
	• evaluate primary sources of data to assess risk
	• conduct empiric risk calculations, occurrence and recurrence risks
	• perform Bayesian risk calculations including:
	  • linkage-based risk calculations
	  • analyse simple genetic linkage by logarithm of odds (LOD) score methods
	  • calculate gene frequencies
	  • calculate Hardy-Weinberg equilibrium
	  • conduct chi-square tests of departure
	• identify and critically evaluate information to inform diagnoses.

DOMAIN 1	SCIENTIFIC BASIS OF CLINICAL GENETICS
Theme 1.1	Fundamentals of Inheritance and Genetics Theory
Learning Objective 1.1.3	Describe the evolution of genetic knowledge

Knowledge	Skills
• identify advances in research relating to gene therapy, stem cells and therapeutic cloning • describe the rapid shifts in understanding of major genetic principles over the past century • explain the history of use and abuse of genetic information • identify community attitudes to genetic information and genetic technology.	• evaluate new advances in genetics theory and their application to clinical practice.

DOMAIN 2	DIAGNOSTIC METHODS, INVESTIGATIONS AND MANAGEMENT
Theme 2.1	Clinical Skills
Learning Objective 2.1.1	Investigate, diagnose and manage genetic conditions

Knowledge	Skills
• define the pathophysiological basis of signs and symptoms of genetic conditions • recognise the signs and symptoms of genetic disease • define the clinical signs observed in genetic diseases, including appropriate clinical measurements • plan appropriate questions to elicit genetic history • identify information sources, including databases and literature searches • recognise evolving knowledge about the genetic basis of common and complex disorders • discuss the importance of obtaining informed consent prior to investigation • discuss any financial implications of testing for the patient/family • discuss the potential impact of investigations, their results and the diagnoses of genetic conditions on individuals and their families • define the indications for investigation	• record and analyse a clinical history • elicit family history information • analyse relevant patient and family information • perform an examination to elicit relevant signs of genetic disease, recognising the potential need for a chaperone and involving relatives as appropriate • recognise and refer when additional specialist examination is required • calculate genetic risk by various means, including Mendelian principles etc • use genetic databases and registers for information retrieval • elicit and record complex pedigrees, including consanguinity loops • perform investigations and interpret the results • formulate differential diagnoses for genetic disorders • present genetic information to a patient in a sensitive and understanding manner

DOMAIN 2	DIAGNOSTIC METHODS, INVESTIGATIONS AND MANAGEMENT
Theme 2.1	Clinical Skills
Learning Objective 2.1.1	Investigate, diagnose and manage genetic conditions

Knowledge	Skills

• explain the purpose, extent and limitations of investigation results
• explain the risks and benefits of investigations
• describe the causes, frequency and implications of erroneous test results
• identify the features of potential diagnoses and disorders
• explain concepts such as:
  • variable expressivity
  • reduced penetrance
  • somatic mosaicism
  • gonadal mosaicism
• describe the implications of ethnic difference in the incidence of genetic disease
• explain the natural history of genetic diseases
• explain the therapeutic use of medical intervention in genetic disease
• describe management issues relevant to genetic syndromes
• discuss the significance and importance of family history and consanguinity in rare genetic conditions
• distinguish between common general paediatric presentations and signs that are indicative of genetic conditions.
	• present undiagnosed cases to colleagues, including Dysmorphology Club meetings
	• conduct clinics which require specialist diagnoses, assessment and genetic counselling
	• discuss management options and/or surveillance with individuals, families and the professionals involved in their care
	• devise management strategies as part of a multidisciplinary team
	• provide genetic advice in multidisciplinary clinics, such as child development, vision, hearing, endocrine, skeletal dysplasia, neurological, craniofacial, cancer genetics and prenatal diagnosis clinics
	• overcome difficulties of language and physical and intellectual impairment
	• perform examinations appropriately in situations involving cultural sensitivity.

DOMAIN 2	DIAGNOSTIC METHODS, INVESTIGATIONS AND MANAGEMENT
Theme 2.1	Clinical Skills
Learning Objective 2.1.2	Use available resources to inform diagnostic techniques

Knowledge	Skills

• identify relevant teaching resources and medical journals
• identify key websites such as:
  • Online Mendelian Inheritance in Man (OMIM)
  • POSSUM-web
	• apply the use of available resources to enhance patient care and professional development
	• access and utilise genetic websites, specialist databases and statistics programs

DOMAIN 2	DIAGNOSTIC METHODS, INVESTIGATIONS AND MANAGEMENT
Theme 2.1	Clinical Skills
Learning Objective 2.1.2	Use available resources to inform diagnostic techniques

- London Dysmorphology Database (LDDB)
- Metab-l
- Online Metabolic and Molecular Basis of Inherited Disease
- Genetests
- PubMed

- identify relevant conferences such as:
  - Human Genetics Society of Australasia
  - Society for the Study of Inborn Errors of Metabolism
  - Australasian Society for Inborn Errors of Metabolism
- retrieve and use data recorded in clinical systems
- recognise main local and national projects and initiatives in information technology related to genetics
- describe the range of possible uses for clinical data and information
- identify the risks and benefits of aggregating clinical data
- identify issues relating to confidentiality applied to information technology in clinical practice.
- use and appraise available software packages for genetics data, including CYRILLIC and PROGENY
- undertake literature searches
- explain how to access and use available resources and support groups
- recognise personal gaps in knowledge, and identify and use appropriate resources to resolve these gaps
- identify and use appropriate references, including textbooks and the internet, to further understanding of laboratory results and clinical findings.

DOMAIN 2	DIAGNOSTIC METHODS, INVESTIGATIONS AND MANAGEMENT
Theme 2.2	Genetic Testing
Learning Objective 2.2.1	Apply appropriate diagnostic procedures and interpret results of genetic tests
Genetic tests include:	

- phlebotomy
- hair root extraction
- cheek swab collection
- Wood's light
- skin biopsy
- clinical measurements
- clinical photography.

DOMAIN 2	DIAGNOSTIC METHODS, INVESTIGATIONS AND MANAGEMENT
Theme 2.2	Genetic Testing
Learning Objective 2.2.1	Apply appropriate diagnostic procedures and interpret results of genetics tests

Knowledge	Skills
• describe the indications for performing procedures • describe the associated risks, complications, and management of complications • define and explain techniques of the procedure • discuss the risks and benefits of the procedure, and obtain informed consent • explain the indications for clinical photography and obtain informed consent for the uses of the photographs, e.g. clinical application, teaching, publication etc • recognise the terminology related to clinical measurement in genetic diseases • describe the importance and confidentiality of use and storage of photographs • recognise appropriate photographic views required for a particular clinical context.	• perform procedures related to clinical genetics • apply standard precautions in dealing with blood tissue or bodily fluids • perform, record, plot and interpret clinical measurements • take photographs for clinical use.

DOMAIN 2	DIAGNOSTIC METHODS, INVESTIGATIONS AND MANAGEMENT
Theme 2.2	Genetic Testing
Learning Objective 2.2.2	Interpret genetic laboratory results

- describe techniques for chromosome analysis in different tissues
- describe laboratory techniques for diagnosing chromosome breakage syndromes
- describe molecular genetic techniques in common usage, e.g. DNA extraction, Southern blotting, polymerase chain reaction (PCR), DNA sequencing
- describe the principles of DNA-based zygosity testing
- describe the potential application of new DNA technologies, e.g. micro arrays, multiplex ligation-dependent probe amplification (MLPA)
- use common methodologies to form a biochemical diagnosis of inborn errors of metabolism, e.g. organic acid tests
- distinguish between screening and diagnostic tests
- describe the special considerations in predictive genetic testing and such testing in children
- obtain informed and specific consent for genetic testing
- obtain consent in relation to storage of DNA samples and cell lines.

- use sensitivity and specificity to interpret laboratory tests
- use the International System for Human Cytogenetic Nomenclature (ISCN) and standard mutation nomenclature
- apply DNA based testing for gene mapping, linkage and mutation detection
- use DNA and molecular cytogenetic methods in pre-implantation diagnosis
- interpret clinical consequences of abnormal karyotypes and enzyme deficiencies
- interpret clinical results of pharmacogenetics testing
- analyse test results in a clinical diagnostic context, liaising with molecular and cytogenetic scientists
- provide advice to the laboratory on the wording of reports to referring clinicians
- conduct Bayesian calculations to calculate genetic risk, using information from different sources, e.g. pedigree, creatine kinase (CK), DNA linkage data
- explain the uncertainties and limitations of genetic testing for both screening and diagnostic tests
- liaise with colleagues to interpret laboratory results.

DOMAIN 2	DIAGNOSTIC METHODS, INVESTIGATIONS AND MANAGEMENT
Theme 2.2	Genetic Testing
Learning Objective 2.2.3	Describe emerging genetics technologies and their application

Knowledge	Skills
• describe advances in new diagnostic tests, e.g. MLPA, array comparative genomic hybridisation (CGH), next generation sequencing • describe the applications of pharmacogenetic testing • describe potential benefits and harms of genetic technologies • identify public concerns about the application of genetic technologies • identify community attitudes to genetic information and genetic technology • recognise the ethical, legal, social and cultural implications of genetic technologies • describe the issues concerning prioritisation of application and equity of access in the environment of limited resources • describe the role of patents and their applicability in the public sector.	• evaluate the efficacy and timely application of new technologies.

DOMAIN 2	DIAGNOSTIC METHODS, INVESTIGATIONS AND MANAGEMENT
Theme 2.3	Genetic Screening Programs and Registers
Learning Objective 2.3.1	Explain the processes to establish and operate genetic screening programs

Knowledge	Skills

• identify World Health Organisation (WHO) recommendations for requirements to implement a screening program
• describe the limitations of these recommendations
• describe potential applications of screening for conditions which do not meet the criteria
• describe the genetic characteristics in different populations, including gene mutation frequencies and disease prevalence
• describe contributing factors for the establishment of population screening programs for genetic diseases
• identify current screening programs, including newborn screening
• describe the operation of local and national antenatal and newborn genetic disease screening programs
• evaluate the costs and benefits of screening programs
• discuss the counselling support needed for screening programs
• discuss the public health outcomes of screening programs
• identify population-based genetic registers
• identify practical issues arising from the operation of genetic registers
• describe the methods of data collection and their limitations
• describe the principles of first and second degree prevention and screening
• discuss the ethical, legal, social and cultural issues associated with genetic and newborn screening programs, including:
  • the central role of public education
  • the benefits of central coordination for maintenance of standards
  • civil rights concerns
  • cultural and social sensitivities
	• assess an individual patient's risk factors
	• contribute to multidisciplinary teams to:
	  • conduct 'cascade screening'
	  • provide genetic services for extended families with common single gene disorders
	• interpret the sensitivity, specificity, and predictive values of screening tests
	• explain the benefits of the newborn screening program
	• contribute to the operation of screening programs for common carrier states, e.g. cystic fibrosis (CF), thalassaemia
	• explain the benefits and consequences of screening programs
	• encourage participation in appropriate disease prevention or screening programs
	• educate patients effectively about epidemiological screening.

DOMAIN 2	DIAGNOSTIC METHODS, INVESTIGATIONS AND MANAGEMENT
Theme 2.3	Genetic Screening Programs and Registers
Learning Objective 2.3.1	Explain the processes to establish and operate genetic screening programs

• public concern about unauthorised secondary use of screening samples • concerns about unauthorised creation of DNA banks from screening samples.	

DOMAIN 2	DIAGNOSTIC METHODS, INVESTIGATIONS AND MANAGEMENT
Theme 2.4	Genetic Counselling
Learning Objective 2.4.1	Provide genetic counselling as part of a multidisciplinary team

Knowledge	Skills

• describe the role of a genetic counsellor
• identify normal reactions to grief and loss, recognising pathological grief
• discuss the effects of grief and loss on decision making regarding genetic risks
• recognise and discuss factors that will impact on a genetic counselling interview, including:
  • patient and family anxieties
  • the influence of personal experiences
  • cultural beliefs and ethnic background
  • cross-cultural issues such as consanguinity and arranged marriages
  • religious and philosophical beliefs and attitudes to prenatal diagnosis and assisted reproduction techniques
• prepare interview questions to identify the patient's:
  • concerns
  • problem list and priorities
  • agenda
  • expectations
  • understanding
  • acceptance
• describe communication strategies to:
  • assist patients to consider their available choices
  • empower patients to make decisions
  • identify the familial implications of genetic diagnosis
	• identify 'at-risk' patients and make appropriate referrals
	• plan a genetic counselling interview in an appropriate setting
	• discuss the features, natural history, genetic basis, and risks of patients developing or passing on genetic disorders
	• explain genetic information and risks to patients and family members in manner they can understand. This may involve:
	  • tailoring communication, both verbal and written, according to the patient's age/ educational level/cultural background
	  • providing additional information in lay language, including fact sheets, useful websites and support group materials
	  • communicating both verbally and in writing to patients whose first language may not be English
	  • using interpreters where appropriate
	• provide clear information and feedback to patients and share information with relatives when appropriate
	• discuss the patient's and family members' understanding of the information received
	• discuss strategies to assist patients and/or family members to adjust to the disorder

DOMAIN 2	DIAGNOSTIC METHODS, INVESTIGATIONS AND MANAGEMENT
Theme 2.4	Genetic Counselling
Learning Objective 2.4.1	Provide genetic counselling as part of a multidisciplinary team

- facilitate the discussion with respect, empathy, honesty and sensitivity
- ask difficult and awkward questions in a sensitive and thoughtful manner
- engage in realistic discussions regarding sensitive subjects
- communicate bad news in a caring and sensitive manner

- identify skills required for 'non-directive' counselling, such as:
  - active listening
  - use of open questions followed by appropriate closed questions
  - avoid jargon and use familiar language
  - encourage questions
  - reassure 'worried well' patients
- describe actions required following the interview, such as:
  - advise patient of available community supports
  - plan for short- and long-term support as indicated
- discuss the role of counselling supervision in:
  - debriefing
  - gaining insight into one's own responses to issues
  - learning new strategies and skills.

- discuss strategies to minimise the risk of recurrence of the disorder
- identify and support patients in distress
- reflect on own genetic counselling style and effectiveness, and identify strategies for improvement
- discuss the encounter with colleagues after counselling sessions.

DOMAIN 3	GENETIC DISORDERS AND DISEASES
Theme 3.1	Prenatal Assessment
Learning Objective 3.1.1	Conduct prenatal genetic assessments and counselling

Common prenatally diagnosed disorders include:

- autosomal and sex chromosome aneuploidy syndromes
- Duchenne muscular dystrophy
- Huntington disease
- spinal muscular atrophy
- haemophilia
- fetal anomaly diagnosed on antenatal imaging.

Knowledge	Skills

• describe fetal pathology
• describe the natural history, prognosis, and risks of inheritance of prenatally diagnosed conditions
• discuss the process and limitations of clinical and laboratory diagnostic procedures in the areas of:
  • preimplantation genetic diagnosis (PGD)
  • imaging
  • antenatal diagnosis
• identify screening programs in pregnancy and pre-pregnancy
• identify laws pertaining to termination of pregnancy for fetal abnormality
• discuss management options available for fetal abnormality
• discuss the psychosocial aspects of infertility.
	• interpret family history data and trace medical records
	• provide genetic advice for women who may consider prenatal diagnosis
	• assess the risk of fetal abnormality through the evaluation of chromosome, DNA and fetal imaging studies
	• perform risk assessments when pregnancies are exposed to hazards such as congenital infections, teratogenic medications, alcohol and ionising irradiation
	• use syndrome databases to inform diagnosis
	• formulate differential diagnoses and assess prognosis in collaboration with the fetal medicine team
	• provide counselling regarding genetic risks, abnormal test results or diagnoses in the antenatal period
	• perform post-mortem clinical analysis of the fetus, including:
	  • examination
	  • measurements
	  • photography
	  • radiology
	  • tissue sampling
	  • storage for diagnostic studies
	• contribute to ultrasound sessions and maternal-fetal medicine meetings.

DOMAIN 3	GENETIC DISORDERS AND DISEASES
Theme 3.2	Neurogenetics
Learning Objective 3.2.1	Assess, diagnose and treat patients with neurogenetic disorders

Common neurogenetic disorders include:

- myotonic dystrophy
- Huntington disease
- spinocerebellar ataxias (SCA)
- inherited neuropathies (demyelinating vs. neuronal pathology).

Knowledge	Skills

• describe the molecular basis of neurogenetic disorders, including mitochondrial cytopathies
• describe molecular issues in triplet repeat disorders, including mutable normal alleles, anticipation, and intermediate 'grey zone' alleles
• describe the basic neuropathology of early onset inherited dementias
• describe the natural history of childhood and adult onset genetic neuromuscular conditions
• recognise common clinical presentations of neurogenetic conditions, such as:
  • hypotonic neonate/infant
  • motor developmental delay
  • progressive weakness and/or ataxia
  • movement disorders
  • dementia/cognitive decline
• identify appropriate investigations for suspected neurogenetic disorders, such as:
  • indications for serum biochemistry (CK, lactate)
  • nerve conduction studies
  • electromyogram
  • intracranial imaging
  • muscle biopsy
  • molecular studies
• describe the management options available for childhood and adult onset genetic neuromuscular conditions
• participate in the provision of predictive testing for autosomal dominant adult onset neurogenetic conditions such as Huntington's disease.
	• recognise the medical presentations and family histories that indicate a risk of familial neurological disease
	• conduct a clinical examination of the neurological system
	• recognise the different presentations of neuromuscular diseases in diverse age groups
	• formulate a differential diagnosis and institute appropriate genetic testing
	• apply protocols for presymptomatic predictive testing of late-onset neurodegenerative disorders
	• discuss the impact caused by the risk or eventuality of neurodegeneration on an individual
	• refer appropriately to other specialists.

DOMAIN 3	GENETIC DISORDERS AND DISEASES
Theme 3.3	Skeletal Dysplasias
Learning Objective 3.3.1	Assess, diagnose and treat patients with skeletal dysplasias

Common skeletal dysplasias include:

- thanatophoric dysplasia
- achondroplasia
- osteogenesis imperfecta
- achondrogenesis.

Knowledge	Skills
• describe the natural history, prognosis, and risks of inheritance of skeletal dysplasias • recognise common clinical presentations of skeletal dysplasias • identify basic radiographic signs of common skeletal dysplasias, e.g. achondroplasia • identify specialist centres and/or individuals for potential referral • describe the management options for common skeletal dysplasias.	• interpret family history data and trace medical records • identify key radiographic features • formulate a diagnosis and institute appropriate genetic testing • discuss management issues with patients, family members, and colleagues • refer appropriately to other specialists.

DOMAIN 3	GENETIC DISORDERS AND DISEASES
Theme 3.4	Dysmorphic Syndromes
Learning Objective 3.4.1	Assess, diagnose and treat patients with dysmorphic syndromes

Common dysmorphic syndromes include:

- Noonan syndrome
- Sotos syndrome
- velocardiofacial syndrome
- Alagille syndrome
- Beckwith-Wiedemann syndrome.

DOMAIN 3	GENETIC DISORDERS AND DISEASES
Theme 3.4	Dysmorphic Syndromes
Learning Objective 3.4.1	Assess, diagnose and treat patients with dysmorphic syndromes

Knowledge	Skills

• differentiate between teratogenesis and normal embryonic development
• evaluate the teratogenesis of drugs and alcohol
• outline the impact of perinatal factors on birth defects and development
• explain morphogenesis in terms of:
  • deformation
  • malformation
  • disruption
  • dysplasia
• identify dysmorphic syndromes from standard clinical photographs
• identify commonly presented dysmorphic syndromes and the tests available to confirm genetic diagnosis
• identify specialist centres and/or individuals to assist in the diagnosis of rare dysmorphic syndromes
• discuss the importance of clinical judgement, timing, and tact when diagnosing and informing parents of an infant with serious malformation or handicap
• discuss the emotional reactions of parents following early diagnosis of a syndrome or recognition of developmental delay
• discuss the importance of accurate diagnosis in providing reproductive choice
• describe indications for investigation of children with delayed development and/or dysmorphic features
• discuss the adverse reaction families may experience following retraction of a previous diagnosis.
	• take a relevant history and perform an appropriate examination identifying dysmorphic features
	• record cases of dysmorphic syndromes using clinical photography
	• evaluate database information and case reports to identify uncertainty and subjectivity in syndrome diagnosis
	• diagnose delayed development based on normal development milestones
	• actively advocate and be involved in public education regarding preventable birth defects, e.g. folate and alcohol
	• present cases at dysmorphology meetings
	• discuss the nature of the clinical findings and differential diagnosis of cases with colleagues.

DOMAIN 3	GENETIC DISORDERS AND DISEASES
Theme 3.5	Cancer Genetics
Learning Objective 3.5.1	Assess, diagnose and contribute to the multidisciplinary management of patients with genetic cancer syndromes

Genetic cancer syndromes include:

- breast and ovarian cancer – arising from BRCA1 and BRCA2 mutations
- colorectal cancer - arising from familial adenomatous polyposis (FAP) or hereditary non-polyposis colorectal cancer (HNPCC)
- multiple endocrine neoplasia type 1 and 2 (MEN1 and MEN2)
- melanoma – arising from CDKN2A or CDK4 mutations
- neurofibromatosis types 1 and 2.

Knowledge	Skills
• identify genetic and environmental factors that affect risk of cancer • describe current recommendations concerning tumour surveillance in cancer prone families • define clinical features of genetic cancer syndromes • describe medical and family history features of inherited cancers • describe genetic mechanisms in cancer, including Knudson's two-hit hypothesis • discuss the impact of inherited cancer on the individual and their at-risk family • discuss current recommendations concerning tumour surveillance and genetic testing in families at risk of cancer • discuss surveillance recommendations for those at low to moderate risk of cancer • discuss the impact of cancer risk on individuals and families • discuss the impact of mutation positive diagnosis on individuals and their families • describe the roles primary care physicians and genetic counsellors play in assessing families where relatives are at risk of developing cancer.	• recognise when a monogenic familial predisposition to cancer is likely to be present in a family • verify a reported cancer history • evaluate and prioritise gene testing for cancer diagnosis • use genetic and disease registers to support follow-up of affected and at-risk patients, e.g. von Hippel-Lindau disease • assess screening protocols for at-risk relatives • recommend appropriate interventions for individuals who are identified as being at increased risk of cancer • identify at-risk patients and relatives who are eligible to participate in trials of cancer prevention strategies • educate patients about lifestyle factors that affect cancer risk, emphasising risk factor avoidance and promoting behaviours that reduce the risk of developing disease • support general practitioners with the long-term management of selected patients with familial cancer syndromes • liaise with other specialists regarding cancer screening and treatment options • attend multidisciplinary cancer genetic meetings.

DOMAIN 3	GENETIC DISORDERS AND DISEASES
Theme 3.6	Other Genetic Disorders and Diseases
Learning Objective 3.6.1	Assess, diagnose and treat patients with other genetic disorders and diseases

Common genetic disorders and diseases include:

- CF
- Duchenne muscular dystrophy
- fragile X syndrome.

Knowledge	Skills

• describe the clinical, biochemical, metabolic and genetic features of:
  • inherited renal disorders, e.g. polycystic kidney disease (PKD)
  • inherited vascular disorders and hypercholesterolemias
  • mitochondrial diseases
  • psychiatric disorders
• discuss the diagnosis and management of childhood-onset disorders in adults, e.g. CF, haemophilia
• discuss the diagnosis and management of adult onset sensory disorders, including deafness and ophthalmological disorders
• discuss the diagnosis and management of inherited cardiac disorders, including:
  • syndromal and nonsyndromal forms of congenital heart disease
  • disorders of cardiac rhythm, including long QT syndrome, Brugada syndrome, inherited forms of atrial fibrillation and supraventricular tachycardia, catecholaminergic polymorphic ventricular tachycardia
  • cardiomyopathies, including dilated cardiomyopathy, hypertrophic cardiomyopathy, left ventricular noncompaction
  • Marfan and Loeys-Dietz syndromes.
	• identify family history data that suggest inherited disease
	• verify the diagnoses of genetic disorders and diseases from hospital records
	• confirm clinical signs of genetic disorders and diseases in affected individuals
	• interpret the results of genetic disorder investigations, including CT and MRI Scans
	• refer patients appropriately to other specialists, such as neurologists, psychologists, psychiatrists and speech therapists.

DOMAIN 4	PROFESSIONAL QUALITIES OF A CLINICAL GENETICIST
Theme 4.1	Ethics
Learning Objective 4.1.1	Identify ethical and legal issues related to clinical genetics practice

Knowledge	Skills

• discuss privacy, consent and access issues in relation to the creation and use of genetic registers
• discuss relevant strategies to ensure confidentiality
• identify situations where confidentiality might be broken
• discuss different cultural attitudes to genetics and genetic testing
• discuss the impact of genetic disorders on individuals, families and cultural groups
• discuss the process for gaining informed consent
• discuss the importance of genetic registers in genetic condition research and management
• differentiate between the legal and Privacy Act requirements of the patient's medical record and that of the genetic register
• discuss issues relevant to consent for post mortem examinations, including differing cultural beliefs about death and treatment of the body after death
• discuss the ethical basis of needing approval and consent prior to commencing a research project
• identify the key sources of advice on the responsibilities of medical practitioners in serious criminal matters
• discuss the legal issues related to criminal matters that may arise during the management of patients with genetic disorders, e.g. discovery of an incestuous relationship between parents of a patient
• outline the professional guidelines published by the National Health and Medical Research Council (NHMRC) relating to human research
• outline the issues involved in the genetic testing of minors
• discuss the issues of 'duty to warn' and 'obligation to recontact'
• discuss the need for equity of services
	• communicate the value of genetic registers to colleagues, patients and their families
	• provide patient details to the appropriate genetic registers
	• record linkage
	• establish or maintain congenital malformation registers, e.g. European Surveillance of Congenital Abnormalities (EUROCAT)
	• contribute to the maintenance of departmental genetic register systems
	• consult with patients before disclosing information
	• seek advice regarding cultural aspects of care
	• adapt counselling considering specific cultural beliefs and attitudes
	• appropriately use and share genetic information
	• disseminate appropriate information in lay terms and gain informed consent from patients
	• obtain suitable evidence when criminal matters arise and consult with appropriate bodies when necessary
	• provide 'non-directive' genetic advice to patients and their families
	• discuss with patients, colleagues and the public the ethical issues concerning:
	  • assisted reproduction
	  • confidentiality
	  • informed consent
	  • genetic testing of children
	  • late termination of pregnancy
	  • population screening for genetic disease
	  • potential impact of testing an individual on other family members, employment/life insurance
	  • predictive genetic testing
	  • prenatal/pre-implantation diagnosis
	• consult with experienced colleagues on difficult cases

DOMAIN 4	PROFESSIONAL QUALITIES OF A CLINICAL GENETICIST
Theme 4.1	Ethics
Learning Objective 4.1.1	Identify ethical and legal issues related to clinical genetics practice

Knowledge	Skills
• consider resource allocation issues, including limitations on budget for genetic testing • summarise the diversity of public opinion on ethical and moral aspects of the practice of clinical genetics.	• refer cases when conflict exists between personal values and those of the patient.

DOMAIN 4	PROFESSIONAL QUALITIES OF A CLINICAL GENETICIST
Theme 4.2	Health Education
Learning Objective 4.2.1	Provide education on risk factors and management of genetic disease

Knowledge	Skills

• describe the course and manifestations of genetic disease
• describe genetic disease investigation procedures and possible alternatives, including the possible risks and costs
• recognise management strategies for genetic disease
• identify risk factors that may influence certain genetic diseases, including:
  • lifestyle
  • occupation
  • smoking
  • alcohol
  • medication
  • maternal/paternal age
  • consanguinity
• discuss issues of addiction and other complexities that may underpin lifestyle changes.
	• encourage patient's questions
	• discuss genetic disease management plans and follow-up arrangements with patient
	• involve patients in developing mutually acceptable investigation plans
	• coordinate patient care with other practitioners
	• encourage patients to access further information and patient support groups
	• advise patients on lifestyle changes for the management of genetic conditions
	• advise patients on the teratogenic potential of medication.

DOMAIN 4	PROFESSIONAL QUALITIES OF A CLINICAL GENETICIST
Theme 4.3	Research
Learning Objective 4.3.1	Plan and execute a clinical or basic genetics research project

Knowledge	Skills
• identify a research question based on lack of current knowledge in an area • discuss the statistical methods utilised in genetic research and describe their use • outline the principles of ethical research and the role of research ethics committees • discuss the importance of ethical approval and patient consent for clinical research • identify potential sources of genetic research funding.	• set up and test a hypothesis • design a genetic research study in consultation with individuals experienced in conducting research • obtain ethics committee approval for genetic research as required • write scientific papers.

DOMAIN 4	PROFESSIONAL QUALITIES OF A CLINICAL GENETICIST
Theme 4.4	Dealing with Medical Uncertainty
Learning Objective 4.4.1	Identify and discuss factors contributing to uncertainty in clinical genetics practice

Knowledge	Skills

• identify the role of uncertainty as an integral part of all medical practice, including clinical genetics
• identify that uncertainty is a limitation of current knowledge and does not indicate clinician failure
• develop a solid foundation of accurate and current knowledge; undertake a reasonable literature search and consult colleagues if necessary
• recognise that uncertainty can provoke anxiety in the patient and the clinician
• discuss the many types of uncertainty in clinical genetics, including:
  • unexpected antenatal detection of abnormality: the consequences for the fetus are uncertain and the issue of whether to terminate or continue the pregnancy often arises
  • no known syndrome diagnosis in person with intellectual disability, +/- malformations, +/- dysmorphism, leading to uncertainty about cause, prognosis and recurrence risks
	• disclose and openly acknowledge areas of uncertainty to patients
	• encourage open discussion of the uncertainties of clinical genetics
	• communicate to a patient that there may be no right or wrong decision in settings of uncertainty, e.g. termination of pregnancy
	• assist in the patient's decision making process to reach a comfortable conclusion
	• reframe uncertainty by breaking it down into manageable parts, e.g. encourage consideration of best and worse case scenarios
	• elicit how a patient has previously dealt with uncertainty in their life and use this when formulating decisions
	• reduce time pressure on a patient in settings of uncertainty, recognising that there is time, even in acute settings, to reach a mutually comfortable decision.

DOMAIN 4	PROFESSIONAL QUALITIES OF A CLINICAL GENETICIST
Theme 4.4	Dealing with Medical Uncertainty
Learning Objective 4.4.1	Identify and discuss factors contributing to uncertainty in clinical genetics practice

• known diagnosis but uncertain prognosis, e.g. neurofibromatosis type 1 (NF1) diagnosed in young child • significant recurrence risk exists, but no prenatal test available, or prenatal test does not distinguish severity (e.g. fragile X syndrome in female) - uncertain outcome for current or future pregnancy • discuss the value of support from a trusted friend, relative, religious advisor and/or community group for the patient • discuss the need for ongoing support in acute and long term settings.	

DOMAIN 5	METABOLIC MEDICINE
Theme 5.1	Scientific Basis of Metabolic Medicine
Learning Objective 5.1.1	Interpret information on the scientific basis of metabolic conditions

Knowledge	Skills
• recognise normal physiology • discuss the principles of biochemistry, including particular common metabolic pathways pertaining to metabolic disease, such as the Krebs cycle and urea cycle • describe the role of the lysosome, peroxisome and mitochondria in cell biochemistry • outline the principles of nutrition, including normal requirements, micronutrients and special requirements during times of physiological stress • identify common general paediatric presenting complaints and potential metabolic diagnoses.	• apply the fundamentals of biochemistry and metabolism to the everyday management of metabolic disease.

DOMAIN 5	METABOLIC MEDICINE
Theme 5.2	Diagnostic Methods, Investigations and Management in Metabolic Medicine
Learning Objective 5.2.1	Elicit a comprehensive history from a patient

Knowledge	Skills
• identify potential diagnoses and disorders through revision of previously recorded patient notes and investigations • discuss the natural history of metabolic disease • plan appropriate questions to elicit metabolic history • recognise the significance and the importance of previous family history and consanguinity in rare diseases • show attention to detail and accuracy in collecting and checking family history and medical data • appreciate the confidentiality, cultural aspects and ethical issues arising from family history gathering • recognise the importance of psychosocial and cultural factors of patients and relatives.	• record and analyse a clinical history • elicit family history information • analyse relevant patient and family information • evaluate a patient's history of metabolic investigations • elicit and record complex pedigrees, including consanguinity loops.

DOMAIN 5	METABOLIC MEDICINE
Theme 5.2	Diagnostic Methods, Investigations and Management in Metabolic Medicine
Learning Objective 5.2.2	Conduct an examination

Knowledge	Skills
• define the pathophysiological basis of the physical signs of metabolic conditions • define the clinical signs of metabolic diseases and the corresponding clinical measurements • identify when additional specialist examination is required.	• perform a reliable and appropriate examination to elicit relevant signs of metabolic disease • perform a neurological examination • perform examinations appropriately in situations involving cultural sensitivity.

DOMAIN 5	METABOLIC MEDICINE
Theme 5.2	Diagnostic Methods, Investigations and Management in Metabolic Medicine
Learning Objective 5.2.3	Select, perform and interpret appropriate investigations

Knowledge	Skills

• define the pathophysiological basis of specific metabolic investigations
• define the indications for metabolic investigation
• describe the methodology of metabolic investigations, their uses and their limitations
• identify the methodology, risks, benefits and limitations of metabolic investigation, including the assessment of:
  • amino acids
  • organic acids
  • fatty acids
  • lysosomal, peroxisomal and mitochondrial disease
  • glycogen storage diseases
• discuss the cost effectiveness of individual investigation
• describe the methodology, uses and limitations of the following laboratory metabolic investigations:
  • gas chromatography-mass spectrometry (GCMS)
  • chromatography
  • tandem mass spectrometry
  • enzymology
• discuss the need for further testing if initial investigations are inconclusive.
	• prioritise metabolic investigations appropriately through consultation with colleagues
	• explain to patients the rationale for investigations, detailing possible unwanted effects or findings
	• observe tests performed in a metabolic laboratory
	• identify, and refer appropriately to, laboratories that specialise in specific areas of metabolism.

DOMAIN 5	METABOLIC MEDICINE
Theme 5.2	Diagnostic Methods, Investigations and Management in Metabolic Medicine
Learning Objective 5.2.4	Synthesise findings to formulate a diagnosis

Knowledge	Skills

• identify key metabolites and their implications for the diagnosis of specific metabolic diseases
• describe the common laboratory findings that may indicate underlying metabolic disease
• discuss the need to confirm a suspected diagnosis with further biochemical testing, enzymology or molecular analysis
• define the symptoms of metabolic disease, particularly in regard to:
  • episodes of metabolic decompensation
  • reactions to certain diets
  • progression of symptoms
  • symptoms during periods of stress, infection and exercise
• discuss the need to evaluate all organ systems when diagnosing metabolic syndromes
• describe the symptoms of metabolic neurological, cardiac, hepatic, renal, pulmonary and gastrointestinal disease.
	• formulate differential diagnoses for metabolic disorders
	• present metabolic information to patients and their family members in a sensitive and understanding manner
	• evaluate findings from history, examination and investigations to form a diagnosis.

DOMAIN 5	METABOLIC MEDICINE
Theme 5.2	Diagnostic Methods, Investigations and Management in Metabolic Medicine
Learning Objective 5.2.5	Manage patients with metabolic disorders

Knowledge	Skills

• identify the issues relevant to managing metabolic syndromes
• outline the principles of metabolic disease management, including:
  • specialised diets
  • treatment with 'simple' fluids
  • medications
  • haemofiltration
  • bone marrow transplantation
  • chaperone therapy
  • enzyme replacement therapy
  • organ replacement
• discuss the therapeutic use of medical intervention in metabolic disease
• outline the principles of palliative care and epilepsy treatment
• outline the neurological symptoms of metabolic syndromes
• discuss the importance of multidisciplinary teams when managing metabolic diseases
• discuss the need to consult other professionals, including international metabolic experts
• discuss the need to develop treatment protocols and specific treatment plans for individual patients with metabolic conditions.
• outline the neurological symptoms of metabolic syndromes
• discuss the importance of multidisciplinary teams when managing metabolic diseases
• discuss the need to consult other professionals, including international metabolic experts
• discuss the need to develop treatment protocols and specific treatment plans for individual patients with metabolic conditions.
	• develop clear action plans regarding certain medical situations for patients and treating clinicians
	• consult appropriately with other professionals, including international metabolic experts, in the diagnosis and management of metabolic conditions
	• refer appropriately to other specialists.

ACRONYMS AND INITIALISMS

CF	cystic fibrosis
CGH	comparative genomic hybridisation
CK	creatine kinase
CT	computed tomography
EUROCAT	European Surveillance of Congenital Abnormalities
FAP	familial adenomatous polyposis
GCMS	gas chromatography-mass spectrometry
HNPCC	hereditary non-polyposis colorectal cancer
ISCN	International System for Human Cytogenic Nomenclature
LDDB	London Dysmorphology Database
LOD	logarithm of odds
MEN	multiple endocrine neoplasia
MLPA	multiplex ligation-dependent probe amplification
MRI	magnetic resonance imaging
NF1	neurofibromatosis type 1
NHMRC	National Health and Medical Research Council
OMIM	Online Mendelian Inheritance in Man
PCR	polymerase chain reaction
PGD	preimplantation genetic diagnosis
PKD	polycystic kidney disease
SCA	spinocerebellar ataxias
WHO	World Health Organisation