NEW CURRICULA

Advanced Training in Clinical Genetics

Curriculum standards

RACP
Specialists. Together

About this document

This document outlines the draft curriculum standards for Advanced Training in Clinical Genetics for trainees and supervisors.

The curriculum standards should be used in conjunction with the Advanced Training in Clinical Genetics learning, teaching, and assessment programs.

For more information or to provide feedback contact curriculum@racp.edu.au.

Contents

Program overview

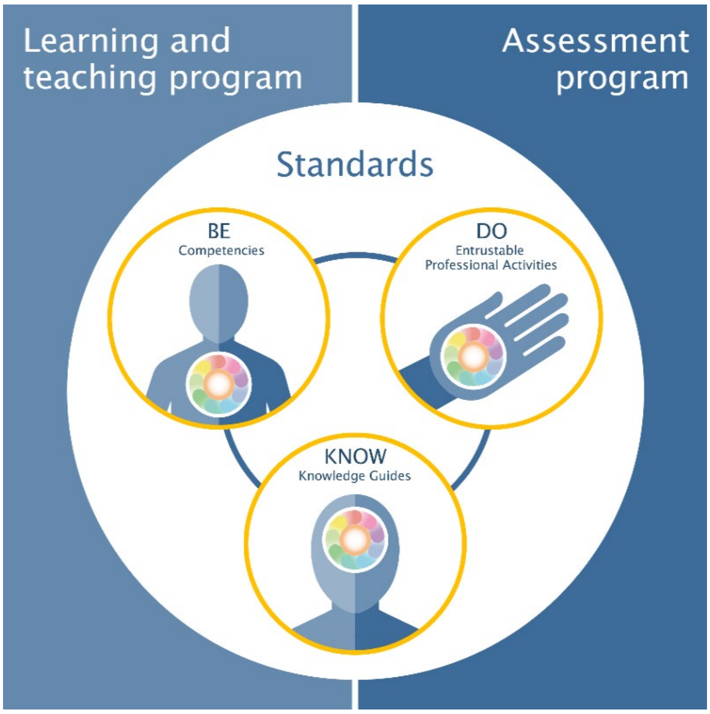
Purpose of Advanced Training

The RACP offers Advanced Training in 33 diverse medical specialties as part of Division, Chapter, or Faculty training programs.

The purpose of Advanced Training is to develop a workforce of physicians who:

- have received breadth and depth of focused specialist training, and experience with a wide variety of health problems and contexts
- are prepared for and committed to independent expert practice, lifelong learning, and continuous improvement
- provide safe, quality health care that meets the needs of the communities of Australia and Aotearoa New Zealand.

RACP curriculum model



The **RACP curriculum model** is made up of curricula standards supported by learning, teaching, and assessment programs.

Learning and teaching programs outline the strategies and methods to learn and teach curricula standards, including required and recommended learning activities.

Assessment programs outline the planned use of assessment methods to provide an overall picture of the trainee’s competence over time.

The **curricula standards** outline the educational objectives of the training program and the standard against which trainees’ abilities are measured.

- **Competencies** outline the expected professional behaviours, values, and practices of trainees in 10 domains of professional practice.

- **Entrustable Professional Activities** (EPAs) outline the essential work tasks trainees need to be able to perform in the workplace.

- **Knowledge guides** outline the expected baseline knowledge of trainees.

Professional Practice Framework

The Professional Practice Framework describes 10 domains of practice for all physicians.

Learning, teaching, and assessment (LTA) structure

The learning, teaching and assessment structure defines the framework for delivery and trainee achievement of the curriculum standards in the Advanced Training program.

Advanced Training is structured in three phases. These phases will establish clear checkpoints for trainee progression and completion.

1	**Specialty foundation**	• Orient trainees and confirm their readiness to progress in the Advanced Training program
2	**Specialty consolidation**	• Continue trainees' professional development in the specialty and support progress towards the learning goals
3	**Transition to Fellowship**	• Confirm trainees' achievement of the curriculum standards, completion of Advanced Training, and admission to Fellowship • Support trainees' transition to unsupervised practice

Figure 1: Advanced Training learning, teaching, and assessment structure

- An **entry decision** is made before entry into the program.
- A **progress decision**, based on competence, is made at the end of each phase of training.
- A **completion decision**, based on competence, is made at the end of the training program, resulting in eligibility for admission to Fellowship.

Advanced Training is a **hybrid time- and competency-based training program**. There is a minimum time requirement of full-time equivalent experience, and progression and completion decisions are based on evidence of trainees' competence.

Clinical genetics specialty overview

Clinical genetics involves the clinical assessment, diagnosis, genetic counselling, and management of individuals and families with genetic disorders and disease predispositions.

Clinical geneticists integrate genomic science with clinical medicine to diagnose, manage, and counsel individuals and families with genetic conditions. Training as a clinical geneticist focuses on developing expertise across clinical assessment, genomic interpretation, counselling and communication, patient and subspecialty education, multidisciplinary care, ethics, and ongoing adaptation to advances in genomic medicine.

Clinical geneticists:

- **conduct comprehensive assessments and investigations.** Clinical geneticists examine patients and use a variety of investigations to diagnose genetic conditions affecting each body system, making a broad clinical knowledge essential.
- **diagnose and manage patients and families with genetic conditions.** This includes preparing and interpreting family history data, conducting clinical examinations, and diagnosing genetic conditions using clinical evaluation and genetic testing. Following diagnosis, clinical geneticists formulate management plans to support patients and their families.
- **provide accurate information and effective genetic counselling** to individuals and families. This may include providing information on genetic risks, reproductive counselling, prevention methods, and liaising with support groups and other organisations.
- **facilitate genomic education,** upskilling, and uptake of genomics in liaison with genomic laboratories, adjacent medical subspecialties, and primary care.
- **keep up with the latest advances** in precision medicine, including genomic diagnostics, research into novel genes and rare diseases, and gene therapies and clinical trials, working with the relevant specialities to do this while providing genetic input.
- **subspecialise** further in areas such as cardiac, dermatology, endocrine, fertility and reproduction, immunology, neurology, ophthalmology, and renal and skeletal dysplasia genetics. Clinical geneticists can focus their training and practice in adult, paediatric, reproductive, metabolic and/or cancer genetics. There are also options for joint training in genetic pathology, and many opportunities for genomic research in this rapidly expanding field.

Clinical geneticists are at the forefront of genomic advances, research, and precision medicine, and have a vital role in managing the counselling, psychosocial, and ethical aspects of new genetic diagnostics and technologies for patient care. As they provide patient- and family-centred care for genetic conditions, excellent verbal and written communication are core skills.

Other essential skills include the ability to:

- **apply a scholarly approach.** Clinical geneticists conduct literature searches and use medical genetics databases, conduct research within the discipline, participate in teaching, and interpret genomic data for diagnostics and research.

- **work effectively as part of a multidisciplinary team** with other colleagues, providing genetic services in subspecialty medicine and genomic diagnostic laboratories. They liaise and work collaboratively with genetic counsellors and colleagues from other specialties, and work with lay organisations and consumer groups to support patients and families with genetic conditions.
- **understand ethical, legal, social, and cultural issues** in the context of clinical genetics and clinical, epidemiological, and laboratory research.

Clinical Genetics learning goals

The curriculum standards are summarised as 17 learning goals. The learning goals articulate what trainees need to be, do, and know, and are assessed throughout training.

BE Competencies	1. Professional behaviours
DO EPAs	2. Team leadership 3. Supervision and teaching 4. Quality improvement 5. Clinical assessment and management 6. Management of transitions in care 7. Longitudinal care 8. Communication with patients 9. Investigations 10. Clinic management
KNOW Knowledge guides	11. Clinical sciences 12. Laboratory-based clinical genomics 13. Cancer genetics 14. Common adult and paediatric genetic conditions 15. Subspecialty genetics 16. Genetic counselling 17. Metabolic genetics

Curriculum standards

Competencies

Competencies outline the expected professional behaviours, values, and practices that trainees need to achieve by the end of training.

Competencies are grouped by the 10 domains of the professional practice framework.

Competencies will be common across training programs.

Learning goal 1: Professional behaviours

Medical expertise **Professional standard:** Physicians apply knowledge and skills informed by best available current evidence in the delivery of high-quality, safe practice to facilitate agreed health outcomes for individual patients and populations.
Knowledge: Apply knowledge of the scientific basis of health and disease to the diagnosis and management of patients.
Synthesis: Gather relevant data via age- and context-appropriate means to develop reasonable differential diagnoses, recognising and considering interactions and impacts of comorbidities.
Diagnosis and management: Develop diagnostic and management plans that integrate an understanding of individual patient circumstances, including psychosocial factors and specific vulnerabilities, epidemiology, and population health factors in partnership with patients, families, whānau, or carers[1], and in collaboration with the healthcare team.

---

[1] References to patients in the remainder of this document may include their families, whānau, and/or carers.

Communication

Professional standard: Physicians collate information, and share this information clearly, accurately, respectfully, responsibly, empathetically, and in a manner that is understandable.

Physicians share information responsibly with patients, families, carers, colleagues, community groups, the public, and other stakeholders to facilitate optimal health outcomes.

Effective communication: Use a range of effective and appropriate verbal, nonverbal, written and other communication techniques, including active listening.

Communication with patients, families, and carers: Use collaborative, effective, and empathetic communication with patients, families, and carers.

Communication with professionals and professional bodies: Use collaborative, respectful, and empathetic clinical communication with colleagues, other health professionals, professional bodies, and agencies.

Written communication: Document and share information about patients to optimise patient care and safety.

Privacy and confidentiality: Maintain appropriate privacy and confidentiality, and share information responsibly.

Quality and safety

Professional standard: Physicians practice in a safe, high-quality manner within the limits of their expertise.

Physicians regularly review and evaluate their own practice alongside peers and best practice standards, and conduct continuous improvement activities.

Patient safety: Demonstrate a safety focus and continuous improvement approach to own practice and health systems.

Harm prevention and management: Identify and report risks, adverse events, and errors to improve healthcare systems.

Quality improvement: Participate in quality improvement activities to improve quality of care and safety of the work environment.

Patient engagement: Enable patients to contribute to the safety of their care.

Teaching and learning

Professional standard: Physicians demonstrate a lifelong commitment to excellence in practice through continuous learning and evaluating evidence.

Physicians foster the learning of others in their profession through a commitment to mentoring, supervising, and teaching.[2]

Lifelong learning: Undertake effective self-education and continuing professional development.

Self-evaluation: Evaluate and reflect on gaps in own knowledge and skills to inform self-directed learning.

Supervision: Provide supervision for junior colleagues and/or team members.

Teaching: Apply appropriate educational techniques to facilitate the learning of colleagues and other health professionals.

Patient education: Apply appropriate educational techniques to promote understanding of health and disease among patients and populations.

Research

Professional standard: Physicians support creation, dissemination and translation of knowledge and practices applicable to health.[3]

They do this by engaging with and critically appraising research, and applying it in policy and practice to improve the health outcomes of patients and populations.

Evidence-based practice: Critically analyse relevant literature and refer to evidence-based clinical guidelines and apply these in daily practice.

Research: Apply research methodology to add to the body of medical knowledge and improve practice and health outcomes.

---

[2] Adapted from Richardson D, Oswald A, Chan M-K, Lang ES, Harvey BJ. Scholar. In: Frank JR, Snell L, Sherbino J, editors. The Draft CanMEDS 2015 Physician Competency Framework – Series IV. Ottawa: The Royal College of Physicians and Surgeons of Canada; 2015 March.

[3] Adapted from Richardson D, Oswald A, Chan M-K, Lang ES, Harvey BJ. Scholar. In: Frank JR, Snell L, Sherbino J, editors. The Draft CanMEDS 2015 Physician Competency Framework – Series IV. Ottawa: The Royal College of Physicians and Surgeons of Canada; 2015 March.

Cultural safety

Professional standard. Physicians engage in iterative and critical self-reflection of their own cultural identity, power, biases, prejudices, and practising behaviours. Together with the requirement of understanding the cultural rights of the community they serve, this brings awareness and accountability for the impact of the physician's own culture on decision making and health care delivery. It also allows for an adaptive practice where power is shared between patients, family, whānau, and/or community and the physician, to improve health outcomes.

Physicians recognise the patient and population's rights for culturally safe care, including being an ally for patient, family, whānau, and/or community autonomy and agency over their decision making. This shift in the physician's perspective fosters collaborative and engaged therapeutic relationships, allows for strength-based (or mana-enhanced) decisions, and sharing of power with the recipient of the care, optimising health care outcomes.

Physicians critically analyse their environment to understand how colonialism, systemic racism, social determinants of health, and other sources of inequity have and continue to underpin the healthcare context. Consequently, physicians then can recognise their interfacing with, and contribution to, the environment in which they work to advocate for safe, more equitable and decolonised services, and create an inclusive and safe workplace for all colleagues and team members of all cultural backgrounds.[4]

Critical reflection. Engage in iterative and critical self-reflection and demonstrate cultural safety in the context of their own cultural identity, power, biases, prejudices and practising behaviours.

Allyship. Recognise the patient and population's rights to culturally safe care, including being an ally for patient, family, whānau and/or community autonomy and agency over their decision-making.

Inclusive communication. Apply culturally safe communication, acknowledging the sharing of power, and cultural and human rights to enable patients, families and whānau to engage in appropriate patient care decisions.

Culturally safe environment. Contribute to a culturally safe learning and practice environment for patients and team members. Respect patients may feel unsafe in the healthcare environment.

---

[4] The RACP has adopted the Medical Council of New Zealand's definition of cultural safety (below):
Cultural safety can be defined as:

- the need for doctors to examine themselves and the potential impact of their own culture on clinical interactions and healthcare service delivery
- the commitment by individual doctors to acknowledge and address any of their own biases, attitudes, assumptions, stereotypes, prejudices, structures, and characteristics that may affect the quality of care provided
- the awareness that cultural safety encompasses a critical consciousness where health professionals and health care organisations engage in ongoing self-reflection and self-awareness, and hold themselves accountable for providing culturally safe care, as defined by the patient and their communities.

Curtis et al. "Why cultural safety rather than cultural competency is required to achieve health equity". International Journal for Equity in Health (2019) 18:174

Ethics and professional behaviour

Professional standard: Physicians' practice is founded upon ethics, and physicians always treat patients, their families, communities, and populations in a caring and respectful manner. Physicians demonstrate their commitment and accountability to the health and well-being of individual patients, communities, populations, and society through ethical practice.

Physicians demonstrate their commitment and accountability to the health and wellbeing of individual patients, communities, populations, and society through ethical practice.

Physicians demonstrate high standards of personal behaviour.

Beliefs and attitudes: Reflect critically on personal beliefs and attitudes, including how these may impact on patient care.

Honesty and openness: Act honestly, including reporting accurately, and acknowledging their own errors.

Patient welfare: Prioritise patients' welfare and community benefit above self-interest.

Accountability: Be personally and socially accountable.

Personal limits: Practise within their own limits and according to ethical principles and professional guidelines.

Self-care: Implement strategies to maintain personal health and wellbeing.

Respect for peers: Recognise and respect the personal and professional integrity, roles, and contribution of peers.

Interaction with professionals: Interact equitably, collaboratively, and respectfully with other health professionals.

Respect and sensitivity: Respect patients, maintain appropriate relationships, and behave equitably.

Privacy and confidentiality: Protect and uphold patients' rights to privacy and confidentiality.

Compassion and empathy: Demonstrate a caring attitude towards patients and endeavour to understand patients' values and beliefs.

Health needs: Understand and address patients', families', carers', and colleagues' physical and emotional health needs.

Medical and health ethics and law: Practise according to current community and professional ethical standards and legal requirements.

Judgement and decision making

Professional standard: Physicians collect and interpret information, and evaluate and synthesise evidence, to make the best possible decisions in their practice.

Physicians negotiate, implement, and review their decisions and recommendations with patients, their families and carers, and other health professionals.

Diagnostic reasoning: Apply sound diagnostic reasoning to clinical problems to make logical and safe clinical decisions.

Resource allocation: Apply judicious and cost-effective use of health resources to their practice.

Task delegation: Apply good judgement and decision making to the delegation of tasks.

Limits of practice: Recognise their own scope of practice and consult others when required.

Shared decision making: Contribute effectively to team-based decision-making processes.

Leadership, management, and teamwork

Professional standard: Physicians recognise, respect, and aim to develop the skills of others, and engage collaboratively to achieve optimal outcomes for patients and populations.

Physicians contribute to and make decisions about policy, protocols, and resource allocation at personal, professional, organisational, and societal levels.

Physicians work effectively in diverse multidisciplinary teams and promote a safe, productive, and respectful work environment that is free from discrimination, bullying, and harassment.

Managing others: Lead teams, including setting directions, resolving conflicts, and managing individuals.

Wellbeing: Consider and work to ensure the health and safety of colleagues and other health professionals.

Leadership: Act as a role model and leader in professional practice.

Teamwork: Negotiate responsibilities within the healthcare team and function as an effective team member.

Health policy, systems, and advocacy

Professional standard: Physicians apply their knowledge of the nature and attributes of local, national, and global health systems to their own practices. They identify, evaluate, and influence health determinants through local, national, and international policy.

Physicians deliver and advocate for the best health outcomes for all patients and populations.

Health needs: Respond to the health needs of the local community and the broader health needs of the people of Australia and Aotearoa New Zealand.

Prevention and promotion: Incorporate disease prevention, health promotion, and health surveillance into interactions with individual patients and their social support networks.

Equity and access: Work with patients and social support networks to address determinants of health that affect them and their access to needed health services or resources.

Stakeholder engagement: Involve communities and patient groups in decisions that affect them to identify priority problems and solutions.

Advocacy: Advocate for prevention, promotion, equity, and access to support patient and population health needs within and outside the clinical environment.

Resource allocation: Understand the factors influencing resource allocation, promote efficiencies, and advocate to reduce inequities.

Sustainability: Manage the use of healthcare resources responsibly in everyday practice.

DO
Entrustable
Professional Activities

Entrustable Professional Activities

Entrustable Professional Activities (EPAs) outline the essential work tasks trainees need to be able to perform in the workplace.

#	Theme	Title
2	**Team leadership**	Lead a team of health professionals
3	**Supervision and teaching**	Supervise and teach professional colleagues
4	**Quality improvement**	Identify and address failures in health care delivery
5	**Clinical assessment and management**	Clinically assess and manage the ongoing care of patients
6	**Management of transitions in care**	Manage the transition of patient care between health professionals, providers, and contexts
7	**Longitudinal care**	Manage and coordinate the longitudinal care of patients / families with genetic conditions
8	**Communication with patients**	Discuss diagnoses and management plans with patients
9	**Investigations**	Select, organise, and interpret investigations
10	**Clinic management**	Manage an outpatient clinic

Learning goal 2: Team leadership

Theme	**Team leadership**
Title	**Lead a team of health professionals**
Description	This activity requires the ability to: • prioritise workload • manage multiple concurrent tasks • articulate individual responsibilities, expertise, and accountability of team members • understand the range of team members' skills, expertise, and roles • acquire and apply leadership techniques in daily practice • collaborate with and motivate team members • encourage and adopt insights from team members • act as a role model.

Behaviours

Professional practice framework domain	**Ready to perform without supervision** Expected behaviours of a trainee who can routinely perform this activity without needing supervision The trainee will:	**Requires some supervision** Possible behaviours of a trainee who needs some supervision to perform this activity The trainee may:
Medical expertise	• synthesise information with other disciplines to develop optimal, goal-centred plans for patients[5] • use evidence-based care to meet the needs of patients or populations • assess and effectively manage clinical risk in various scenarios • demonstrate clinical competence and skills by effectively supporting team members	• demonstrate adequate knowledge of healthcare issues by interpreting complex information • assess the spectrum of problems to be addressed • apply medical knowledge to assess the impact and clinical outcomes of management decisions • provide coordinated and quality health care for populations or patients as a member of a multidisciplinary team
Communication	• provide support and motivate patients or populations and health professionals by effective communication • demonstrate a transparent, consultative style by engaging patients, families, carers, relevant professionals, and/or the public in shared decision making • work with patients, families, carers, and other health professionals to resolve conflict that may arise when planning and aligning goals	• communicate adequately with colleagues • communicate adequately with patients, families, carers, and/or the public • respect the roles of team members

---

[5] References to patients in the remainder of this document may include their families, whānau, and/or carers.

	• demonstrate rapport with people at all levels by tailoring messages to different stakeholders	
Quality and safety	• identify opportunities to improve care by participating in surveillance and monitoring of adverse events and 'near misses' • identify activities within systems to reduce errors, improve patient and population safety, and implement cost-effective change • place safety and quality of care first in all decision making	• participate in audits and other activities that affect the quality and safety of patients' care • participate in interdisciplinary collaboration to provide effective health services and operational change • use information resources and electronic medical record technology where available
Teaching and learning	• regularly self-evaluate personal professional practice, and implement changes based on the results • actively seek feedback from supervisors and colleagues on their own performance • identify personal gaps in skills and knowledge, and engage in self-directed learning • maintain current knowledge of new technologies, health care priorities, and changes of patients' expectations • teach competently by imparting professional knowledge • manage and monitor learners' progress, providing regular assessment and feedback	• accept feedback constructively, and change behaviour in response • recognise the limits of personal expertise, and involve other health professionals as needed • demonstrate basic skills in facilitating colleagues' learning
Cultural safety	• demonstrate culturally competent relationships with professional colleagues and patients • demonstrate respect for diversity and difference • take steps to minimise unconscious bias, including the impact of gender, religion, cultural beliefs, and socioeconomic background on decision making	• demonstrate awareness of cultural diversity and unconscious bias • work effectively and respectfully with people from different cultural backgrounds
Ethics and professional behaviour	• promote a team culture of shared accountability for decisions and outcomes • encourage open discussion of ethical and clinical concerns • respect differences of multidisciplinary team members • understand the ethics of resource allocation by aligning optimal patients and organisational care • effectively consult with stakeholders, achieving a balance of alternative views • acknowledge personal conflicts of interest and unconscious bias	• support ethical principles in clinical decision making • maintain standards of medical practice by recognising the health interests of patients or populations as primary responsibilities • respect the roles and expertise of other health professionals • work effectively as a member of a team • promote team values of honesty, discipline, and commitment to continuous improvement

	• act collaboratively to resolve behavioural incidents and conflicts such as harassment and bullying	• demonstrate understanding of the negative impact of workplace conflict
Judgement and decision making	• evaluate health services and clarify expectations to support systematic, transparent decision making • make decisions when faced with multiple and conflicting perspectives • ensure medical input to organisational decision making • adopt a systematic approach to analysing information from a variety of specialties to make decisions that benefit health care delivery • recognise their own limitations and seek help in an appropriate way when required	• monitor services and provide appropriate advice • review new health care interventions and resources • interpret appropriate data and evidence for decision making • inadequately consult with senior colleagues
Leadership, management, and teamwork	• combine team members' skills and expertise in delivering patient care and/or population advice • develop and lead effective multidisciplinary teams by developing and implementing strategies to motivate others • build effective relationships with multidisciplinary team members to achieve optimal outcomes • ensure all members of the team are accountable for their individual practice	• understand the range of personal and other team members' skills, expertise, and roles • acknowledge and respect the contribution of all health professionals involved in patients' care • participate effectively and appropriately in multidisciplinary teams • seek out and respect the perspectives of multidisciplinary team members when making decisions
Health policy, systems, and advocacy	• engage in appropriate consultation with stakeholders on the delivery of health care • advocate for the resources and support for healthcare teams to achieve organisational priorities • influence the development of organisational policies and procedures to optimise health outcomes • identify the determinants of health of the population, and mitigate barriers to access to care • remove self-interest from solutions to health advocacy issues	• communicate with stakeholders within the organisation about health care delivery • understand methods used to allocate resources to provide high-quality care • promote the development and use of organisational policies and procedures

Learning goal 3: Supervision and teaching

Theme	**Supervision and teaching**
Title	**Supervise and teach professional colleagues**
Description	This activity requires the ability to: • provide work-based teaching in a variety of settings • teach professional skills • create a safe and supportive learning environment • plan, deliver, and provide work-based assessments • encourage learners to be self-directed and identify learning experiences • supervise learners in day-to-day work, and provide feedback • support learners to prepare for assessments.
Behaviours	

Professional practice framework domain	**Ready to perform without supervision** Expected behaviours of a trainee who can routinely perform this activity without needing supervision The trainee will:	**Requires some supervision** Possible behaviours of a trainee who needs some supervision to perform this activity The trainee may:
Medical expertise	• combine high-quality care with high-quality teaching • explain the rationale underpinning a structured approach to decision making • consider the patient-centric view during consultations • consider the population health effect when giving advice • encourage learners to consider the rationale and appropriateness of investigation and management options	• teach learners using basic knowledge and skills
Communication	• establish rapport and demonstrate respect for junior colleagues, medical students, and other health professionals • communicate effectively when teaching, assessing, and appraising learners • actively encourage a collaborative and safe learning environment with learners and other health professionals • encourage learners to tailor communication as appropriate for different patients[6], such as younger or older people, and different populations	• demonstrate accessible, supportive, and compassionate behaviour

---

[6] References to patients in the remainder of this document may include their families, whānau, and/or carers.

	• support learners to deliver clear, concise, and relevant information in both verbal and written communication • listen and convey information clearly and considerately	
Quality and safety	• support learners to deliver quality care while maintaining their own wellbeing • apply lessons learnt about patient safety by identifying and discussing risks with learners • assess learners’ competence, and provide timely feedback to minimise risks to care • maintain the safety of patients and organisations involved with education, and appropriately identify and action concerns	• observe learners to reduce risks and improve health outcomes
Teaching and learning	• demonstrate knowledge of the principles, processes, and skills of supervision • provide direct guidance to learners in day-to-day work • work with learners to identify professional development and learning opportunities based on their individual learning needs • offer feedback and role modelling • participate in teaching and supervision of professional development activities • encourage self-directed learning and assessment • develop a consistent and fair approach to assessing learners • tailor feedback and assessments to learners’ goals • seek feedback, and reflect on own teaching by developing goals and strategies to improve • establish and maintain effective mentoring through open dialogue • support learners to identify and attend formal and informal learning opportunities • recognise the limits of personal expertise, and involve others appropriately	• demonstrate basic skills in the supervision of learners • apply a standardised approach to teaching, assessment, and feedback without considering individual learners’ needs • implement teaching and learning activities that are misaligned to learning goals • adopt a teaching style that discourages learner self-directedness
Research	• clarify junior colleagues’ research project goals and requirements, and provide feedback regarding the merits or challenges of proposed research	• guide learners with respect to the choice of research projects • ensure that the research projects planned are feasible and of suitable standards

	• monitor the progress of learners' research projects regularly, and may review research projects prior to submission • support learners to find forums to present research projects • encourage and guide learners to seek out relevant research to support practice	
Cultural safety	• role model a culturally appropriate approach to teaching • encourage learners to seek out opportunities to develop and improve their own cultural safety • encourage learners to consider culturally appropriate care of Aboriginal and Torres Strait Islander peoples and Māori into patients' management • consider cultural, ethical, and religious values and beliefs in teaching and learning	• function effectively and respectfully when working with and teaching with people from different cultural backgrounds
Ethics and professional behaviour	• apply principles of ethical practice to teaching scenarios • act as a role model to promote professional responsibility and ethics among learners • respond appropriately to learners seeking professional guidance	• demonstrate professional values, including commitment to high-quality clinical standards, compassion, empathy, and respect • provide learners with feedback to improve their experiences
Judgement and decision making	• prioritise workloads and manage learners with different levels of professional knowledge or experience • link theory and practice when explaining professional decisions • promote joint problem solving • support a learning environment that allows for independent decision making • use sound and evidence-based judgement during assessments and when giving feedback to learners • escalate concerns about learners appropriately • recognise their own limitations and seek help in an appropriate way when required	• provide general advice and support to learners • use health data logically and effectively to investigate difficult diagnostic problems • inadequately consult with senior colleagues
Leadership, management, and teamwork	• maintain personal and learners' effective performance and continuing professional development • maintain professional, clinical, research, and/or administrative responsibilities while teaching	• demonstrate the principles and practice of professionalism and leadership in health care • participate in mentor programs, career advice, and general counselling

	• create an inclusive environment whereby learners feel part of the team • help shape organisational culture to prioritise quality and work safety through openness, honesty, shared learning, and continued improvement	
Health policy, systems, and advocacy	• advocate for suitable resources to provide quality supervision and maintain training standards • explain the value of health data in the care of patients or populations • support innovation in teaching and training	• incompletely integrate public health principals into teaching and practice

Learning goal 4: Quality improvement

Theme	**Quality improvement**	
Title	**Identify and address failures in health care delivery**	
Description	This activity requires the ability to: • identify and report actual and potential ('near miss') errors • conduct and evaluate system improvement activities • adhere to best practice guidelines • audit clinical guidelines and outcomes • contribute to the development of policies and protocols designed to protect patients[7] and enhance health care • monitor one's own practice and develop individual improvement plans.	
Behaviours		
Professional practice framework domain	**Ready to perform without supervision** Expected behaviours of a trainee who can routinely perform this activity without needing supervision The trainee will:	**Requires some supervision** Possible behaviours of a trainee who needs some supervision to perform this activity The trainee may:
Medical expertise	• use population health outcomes to identify opportunities for improvement in delivering appropriate care • evaluate environmental and lifestyle health risks, and advocate for healthy lifestyle choices • monitor personal professional performance • analyse adverse incidents and sentinel events to identify system failures and contributing factors	• contribute to processes on identified opportunities for improvement • recognise the importance of prevention and early detection in clinical practice • use local guidelines to assist patient care decision making • raise appropriate issues for review at morbidity and mortality meetings
Communication	• support patients to have access to, and use, easy-to-understand, high-quality information about health care • provide written communication to relevant health professionals and patients where appropriate, outlining assessments, investigations, and management plans • provide a family letter where appropriate • support patients to share decision making about their own health care, to the extent they choose • assist patients' access to their health information, as well as complaint and feedback systems	• demonstrate awareness of the evidence for consumer engagement and its contribution to quality improvement in health care • apply knowledge of how health literacy might affect the way patients or populations gain access to, understand, and use health information • access and use genetic websites, specialist databases, and statistics programs

---

[7] References to patients in the remainder of this document may include their families, whānau, and/or carers.

	• discuss with patients any safety and quality concerns they have relating to their care • implement the organisation’s open disclosure policy • assist patients to access and assess clinical trial information	
Quality and safety	• demonstrate safety skills, including infection control, adverse event reporting, and effective clinical handover • participate in organisational quality and safety activities, including morbidity and mortality reviews, clinical incident reviews, root cause analyses, and corrective action preventative action plans • participate in systems for surveillance and monitoring of adverse events and ‘near misses’, including reporting such events • ensure that identified opportunities for improvement are raised and reported appropriately • use clinical audits and registries of data on patients’ experiences and outcomes, learnings from incidents, and complaints to improve care	• demonstrate awareness of a systematic approach to improving the quality and safety of health care • protect the confidentiality of patients and families with respect to genetic results
Teaching and learning	• translate quality improvement approaches and methods into practice • supervise and manage the performance of junior colleagues in the delivery of high-quality, safe care	• work within organisational quality and safety systems for the delivery of clinical care • use opportunities to learn about safety and quality theory and systems
Research	• ensure that any protocol for human research is approved by a human research ethics committee, in accordance with the national statement on ethical conduct in human research • write and present scientific papers to disseminate research findings	• recognise that patient participation in research is voluntary and based on an appropriate understanding about the ethics purpose, methods, demands, risks, and potential benefits of the research
Cultural safety	• undertake professional development opportunities that address the impact of cultural bias on health outcomes • seek advice regarding cultural aspects of care	• communicate effectively and safely with patients from culturally and linguistically diverse backgrounds
Ethics and professional behaviour	• align improvement goals with the priorities of the organisation • contribute to developing an organisational culture that enables and prioritises patients’ safety and quality care	• comply with professional regulatory requirements and codes of conduct

Judgement and decision making	• use decision-making support tools, such as guidelines, protocols, pathways, and reminders • analyse and evaluate current care processes to improve care • recognise their own limitations and seek help in an appropriate way when required	• access information and advice from other health practitioners to identify, evaluate, and improve patients' care management • inadequately consult with senior colleagues
Leadership, management, and teamwork	• formulate and implement quality improvement strategies as a collaborative effort, involving all key health professionals • support multidisciplinary team activities to lower patients' risk of harm, and promote interdisciplinary programs of education	• demonstrate attitudes of respect and cooperation among members of different professional teams • partner with clinicians and managers to ensure patients receive appropriate care and information on their care
Health policy, systems, and advocacy	• participate in all aspects of the development, implementation, evaluation, and monitoring of governance processes • participate regularly in multidisciplinary meetings where quality and safety issues are standing agenda items, and where innovative ideas and projects for improving care are actively encouraged • take part in the design and implementation of the organisational systems for: » clinical education and training » defining the scope of clinical practice » performance monitoring and management » safety and quality education and training • evaluate the efficacy and timely application of new genomic technologies, including their impact on service provision, equity of access, and resource allocation	• maintain a dialogue with service managers about issues that affect patient care • contribute to relevant organisational policies and procedures • help shape an organisational culture that prioritises safety and quality through openness, honesty, learning, and quality improvement

Learning goal 5: Clinical assessment and management

Theme	**Clinical assessment and management**
Title	**Clinically assess and manage the ongoing care of patients**
Description	This activity requires the ability to: • identify and access sources of relevant information about patients[8], including gathering clinical background through histories and examinations • examine patients • synthesise findings to develop provisional and differential diagnoses • discuss findings with patients, families, and/or carers • request appropriate investigations, relevant to differential diagnoses, in a timeline appropriate to the status of patients • choose appropriate treatments, such as targeted and precision therapies for genetic conditions, based on an understanding of disease pathophysiology and pharmacology • generate management plans • present findings to other health professionals.

Behaviours		
Professional practice framework domain	**Ready to perform without supervision** Expected behaviours of a trainee who can routinely perform this activity without needing supervision The trainee will:	**Requires some supervision** Possible behaviours of a trainee who needs some supervision to perform this activity The trainee may:
Medical expertise	• perform comprehensive physical examinations, including focused dysmorphology exams and investigation plans, to establish the nature and extent of health concerns, and to identify potential syndromic diagnoses • evaluate and counsel patients on the likelihood of having a pregnancy affected by a genetic condition • gather accurate, organised, and problem-focused medical histories, with emphasis on family history, past medical history, and relevant past assessments • assess prognosis in collaboration with other healthcare professionals, including fetal medicine teams • calculate empiric and recurrence risk using population data and family history when there is no specific genetic diagnosis	• take patient-centred histories, including reviewing family background and medical histories, and consider psychosocial factors • recognise medical presentations and family histories that indicate a risk of genetic disease • perform accurate physical examinations • recognise and correctly interpret abnormal findings • synthesise pertinent information to direct clinical encounters and diagnostic categories • develop appropriate investigation plans, including identifying potential complications of disease • establish techniques to identify and use accurate and current information in patients' care • identify and manage adverse events

---

[8] References to patients in the remainder of this document may include their families, whānau, and/or carers.

	• assess screening protocols for at-risk relatives • conduct appropriate investigations to confirm diagnoses, and assess extent of disease using diagnostic and surveillance guidelines where available, including before and during pregnancy • synthesise and interpret histories and examination findings to devise provisional diagnoses and reasonable differential diagnoses • develop management plans based on relevant information and multidisciplinary assessments, integrated with guidelines, and consider the balance of benefit and harm by taking patients’ personal sets of circumstances into account • recommend appropriate interventions for individuals who are identified as being at increased risk of cancer	• be aware of potential side effects and practical prescription points, such as medication compatibility and monitoring in response to therapies
Communication	• communicate openly, listen, and take patients’ concerns seriously, giving them adequate opportunity to ask questions • provide clear information and feedback to patients to enable them to make fully informed decisions from various diagnostic, therapeutic, and management options • communicate clearly, effectively, respectfully, and promptly with other health professionals involved in patients’ care • present genetic information to patients in a sensitive and understanding manner, and check for understanding • discuss the features, genetic basis, natural history, and risks of patients developing or passing on genetic conditions, and strategies for patients to adjust and minimise reoccurrence • advise patients of their rights to refuse medical therapy, including life-sustaining treatment • promote open discussion of the uncertainties of clinical genetics • plan genetic counselling interviews in appropriate settings • advise patients on the teratogenic potential of medication	• anticipate, read, and respond to verbal and nonverbal cues • demonstrate active listening skills • communicate patients’ situations to colleagues, including senior clinicians • communicate clearly with other team members in acute clinical scenarios, and coordinate efforts of multidisciplinary team members • develop realistic treatment goals, and determine and explain the expected prognoses and outcomes • employ communication strategies appropriate for younger patients or those with cognitive difficulties

	• anticipate queries to help enhance the likelihood of management plans being followed as advised • overcome difficulties of language that may occur with physical and intellectual impairment • incorporate appropriate LGBTQIA+ safe language, including gender affirming language	
Quality and safety	• demonstrate safety skills, including infection control, adverse event reporting, and effective clinical handover • recognise and effectively deal with aggressive and violent patient behaviours through appropriate training • obtain informed consent before undertaking any investigation or providing treatment (except in an emergency) • ensure patients are informed of the material risks associated with any part of proposed management plans • use appropriate guidelines and evidence-based medicine resources to maintain a working knowledge of current therapies for genetic disorders, including clinical trials and re-purposed medications • evaluate and explain the benefits and risks of clinical interventions and therapies based on individual patients' circumstances	• perform hand hygiene, and take infection control precautions at appropriate moments • apply standard precautions in dealing with blood tissue or bodily fluids • take precaution against assaults from confused or agitated patients, ensuring appropriate care of patients • document histories and physical examination findings, and synthesise with clarity and completeness • identify situations where confidentiality might be broken
Teaching and learning	• set defined objectives for clinical teaching encounters, and solicit feedback on mutually agreed goals • regularly reflect upon and self-evaluate professional development • obtain informed consent before involving patients in teaching activities • turn clinical activities into an opportunity to teach, appropriate to the setting • recognise personal gaps in knowledge, and identify and use appropriate resources to resolve these gaps	• set unclear goals and objectives for self-learning • self-reflect infrequently • deliver teaching considering learners' level of training • mentor and train others to enhance team effectiveness • seek guidance and feedback from healthcare teams to reflect on encounters and improve future patients' care
Research	• evaluate new advances in genetics theory, and their application to clinical practice • search for, find, compile, analyse, interpret, and evaluate information relevant to research participants	• recognise the limitations of evidence and the challenges of applying research in daily practice

	• integrate evidence related to questions of diagnosis, therapy, prognosis, risks, and cause into clinical decision making • evaluate database information and case reports to identify uncertainty and subjectivity in syndrome diagnosis • identify at-risk patients who are eligible to participate in trials • recognise the principles of human research ethics applications	• refer to guidelines and medical literature to assist in clinical assessments when required • refer to evidence-based clinical guidelines and protocols on patients with inherited conditions
Cultural safety	• use plain-language patient education materials, and demonstrate cultural and linguistical sensitivity • demonstrate effective and culturally safe communication and care for Aboriginal and Torres Strait Islander peoples and Māori, and members of other cultural groups • use professional interpreters, health advocates, or family or community members to assist in communication with patients, and understand the potential limitations of each • acknowledge patients’ beliefs and values, and how these might impact on health • explain the cultural aspects and confidentiality and ethical issues arising from family history gathering • recognise the importance of psychosocial and cultural factors of patients and relatives • negotiate health care decisions in a culturally appropriate way by considering variation in family structures, cultures, religion, or belief systems	• display respect for patients’ cultures, and attentiveness to social determinants of health • be cognisant of at least the most prevalent cultures in society, and display an appreciation of their sensitivities • appropriately access interpretive or culturally focused services • be cognisant of patients’ cultural and religious backgrounds, attitudes, and beliefs
Ethics and professional behaviour	• demonstrate professional values, including compassion, empathy, respect for diversity, integrity, honesty, and partnership to all patients • hold information about patients in confidence, unless the release of information is required by law or public interest • assess patients’ capacity for decision making, involving a proxy decision maker appropriately • use and share genetic information appropriately	• demonstrate professional conduct, honesty, and integrity • consider patients’ decision-making capacity • identify patients’ preferences regarding management and the role of families in decision making • not advance personal interest or professional agendas at the expense of patient or social welfare • follow regulatory and legal requirements and limitations regarding prescribing

	• provide patients' details to the appropriate genetic registers, with consent • refer cases when conflict exists between personal values and those of patients	• follow organisational policies regarding pharmaceutical representative visits and drug marketing
Judgement and decision making	• apply knowledge and experience to identify patients' problems, making logical decisions to achieve positive outcomes for patients in acute care, and when delivering end-of-life care • use a holistic approach to health, considering comorbidity, risk, and uncertainty • use the best available evidence for the most effective therapies and interventions to ensure quality care • identify 'at risk' patients, and make appropriate referrals • identify and critically evaluate information to inform diagnoses • reframe uncertainty to allow patients to break down the positive and negative implications of difficult decisions or ambiguous results • identify that uncertainty is a limitation of current knowledge and does not indicate clinician failure, but that it can provoke anxiety in patients and the clinician • evaluate new medicines in relation to their possible efficacy and safety profile for individual patients	• demonstrate clinical reasoning by gathering focused information relevant to patients' care • recognise personal limitations and seek help in an appropriate way when required • consult with experienced colleagues on difficult cases
Leadership, management, and teamwork	• work effectively as a member of multidisciplinary teams to achieve the best health outcomes for patients • demonstrate awareness of colleagues in difficulty, and work within the appropriate structural systems to support them while maintaining patients' safety • discuss the nature of clinical findings and differential diagnoses of cases with colleagues • analyse test results in a clinical diagnostic context • coordinate patients' care with other practitioners and professionals, such as cancer screening and treatment options • facilitate interactions within multidisciplinary teams, respecting values and encouraging involvement	• share relevant information with members of the healthcare team • attend multidisciplinary team meetings • consult appropriately with other professionals • refer appropriately to other specialists • participate in morbidity and mortality meetings

	• follow appropriate referral pathways to ensure ongoing health care, including surveillance • provide genetic advice in multidisciplinary clinics	
Health policy, systems, and advocacy	• participate in health promotion, disease prevention and control, screening, and reporting notifiable diseases • aim to achieve the optimal cost-effective patient care to allow maximum benefit from the available resources • encourage participation in appropriate disease prevention or screening programs • use care pathways effectively, including identifying reasons for variations in care • evaluate the literature regarding the management of genetic conditions, including environmental and lifestyle health risks, and advocate for healthy lifestyle choices • recognise the need for escalation of care, and escalate to appropriate staff or services	• identify and navigate components of the healthcare system relevant to patients' care • identify and access relevant community resources to support patients' care

Learning goal 6: Management of transitions in care

Theme	**Management of transitions in care**
Title	**Manage the transition of patient care between health professionals, providers, and contexts**
Description	This activity requires the ability to: • manage transitions of patients'[9] care to ensure the optimal continuation of care between providers • identify appropriate care providers and other stakeholders with whom to share patients' information • exchange pertinent, contextually appropriate, and relevant patient information • perform this activity in multiple settings, appropriate to clinical genetics, including ambulatory, critical care, and inpatient settings, and from paediatric to adult services.

Behaviours

Professional practice framework domain	**Ready to perform without supervision** Expected behaviours of a trainee who can routinely perform this activity without needing supervision The trainee will:	**Requires some supervision** Possible behaviours of a trainee who needs some supervision to perform this activity The trainee may:
Medical expertise	• facilitate an optimal transition of care for patients • identify and manage key risks for patients during transition • anticipate possible changes in patients' conditions, and provide recommendations on how to manage them • assess readiness for transfer of care from paediatric to adult services, and support and educate young adults and their families during this transition process	• interpret the details of patients' conditions, illness severity, and potential emerging issues, with appropriate actions • provide accurate summaries of patients' information with accurate identification of problems or issues
Communication	• write relevant and detailed medical record entries, including clinical assessments and management plans • write comprehensive and accurate summaries of care, including discharge summaries, clinic letters, and transfer documentation • initiate and maintain verbal communication with other health professionals, when required • communicate with patients about transitions of care, and engage and support them in decision making	• communicate clearly with clinicians and other caregivers • use standardised verbal and written templates to improve the reliability of information transfer and prevent errors and omissions • communicate accurately and in a timely manner to ensure effective transitions between settings and continuity and quality of care

---

[9] References to patients in the remainder of this document may include their families, whānau, and/or carers.

	• communicate with patients about determinants of health as they age, and factors that promote health and minimise harm • incorporate appropriate LGBTQIA+ safe language, including gender affirming language	
Quality and safety	• identify patients at risk of poor transitions of care, and mitigate this risk • use electronic tools (where available) to securely store and transfer patient information • use consent processes, including written consent if required, for the release and exchange of information • explain the medicolegal context of written communications • use genetic and disease registers to support follow-up of affected and at-risk patients • recognise and follow referral pathways to mitigate the risk of patients being lost to follow-up	• ensure that handover is complete, or work to mitigate risks if incomplete • ensure all outstanding results or procedures are followed up by receiving units and clinicians • keep patients' information secure, adhering to relevant legislation regarding personal information and privacy
Teaching and learning	• integrate clinical education / management protocols in handover sessions and other transition of care meetings • tailor clinical education to the level of the professional parties involved	• take opportunities to teach junior colleagues during handover, as necessary
Cultural safety	• communicate with careful consideration to health literacy, language barriers, and culture regarding patients' preferences, and whether they are realistic and possible, respecting patients' choices • recognise the timing, location, privacy, and appropriateness of sharing information with patients • recognise cultural, socioeconomic, and interpersonal familial issues that may be barriers to smooth transition to mitigate against patient / family disengagement	• include relevant information regarding patients' cultural or ethnic background in handovers, and whether an interpreter is required
Ethics and professional behaviour	• disclose and share only contextually appropriate medical and personal information • recognise the clinical, ethical, and legal rationale for information disclosure • share information about patients' care in a manner consistent with privacy law and professional guidelines on confidentiality	• maintain respect for patients and other health professionals, including respecting privacy and confidentiality

	• explain the medicolegal complexity related to genetic information, and seek appropriate advice about disclosure of such information • interact in a collegiate and collaborative way with professional colleagues during transitions of care	
Judgement and decision making	• ensure patients’ care is in the most appropriate facility, setting, or provider	• use a structured approach to consider and prioritise patients’ issues • recognise personal limitations and seek help in an appropriate way when required • inadequately consult with senior colleagues
Leadership, management, and teamwork	• share the workload of transitions of care appropriately, including delegation • recognise the medical governance of patient care, and the differing roles of team members • show respect for the roles and expertise of other health professionals, and work effectively as a member of professional teams • ensure that multidisciplinary teams provide the opportunity for patients’ engagement and participation when appropriate	• recognise factors that impact on the transfer of care, and help subsequent health professionals understand the issues to continue care • work to overcome the potential barriers to continuity of care, appreciating the role of handover in overcoming these barriers
Health policy, systems, and advocacy	• contribute to processes for managing risks, and identify strategies for improvement in transitions of care • engage in organisational processes to improve transitions of care, such as formal surveys or follow-up phone calls after hospital discharge • advocate for provision of skills and services within paediatric and adult care settings to facilitate the establishment of, or grow existing, transition services	• factor transport issues and costs to patients into arrangements for transferring patients to other settings

Learning goal 7: Longitudinal care

Theme	**Longitudinal care**
Title	**Manage and coordinate the longitudinal care of patients / families with genetic conditions**
Description	This activity requires the ability to: • develop management plans and goals in consultation with patients[10] • manage genetic conditions and their associated disabilities, comorbidities, and complications • collaborate with other care providers • ensure continuity of care • facilitate patients' self-management and self-monitoring • engage with the broader health policy context • develop a partnership with families, whānau, carers, and other stakeholders to optimise care.

Behaviours		
Professional practice framework domain	**Ready to perform without supervision** Expected behaviours of a trainee who can routinely perform this activity without needing supervision The trainee will:	**Requires some supervision** Possible behaviours of a trainee who needs some supervision to perform this activity The trainee may:
Medical expertise	• develop, assess, and review management plans for patients, including long-term surveillance of disease and treatment complications • use principles of dietary and pharmacological therapies, where appropriate • monitor therapy outcomes with appropriate tests, and accurately interpret results to make appropriate changes to management • provide supportive care where disease-specific therapy is not available, including referrals to appropriate specialists or allied health therapists • provide documentation on patients' presentation, management, and progress, including key points of diagnosis and decision making, to inform coordination of care • ensure patients contribute to their needs assessments and care planning	• assess patients' knowledge, beliefs, concerns, and daily behaviours related to their genetic condition and its management • contribute to medical record entries on histories, examinations, and management plans in a way that is accurate and sufficient as a member of multidisciplinary teams • support patients with undiagnosed conditions • support patients throughout the lifespan, including discussing reproductive options, genetic testing in deceased family members, stillbirth, and management of multiple family members across generations

---

[10] References to patients in the remainder of this document may include their families, whānau, and/or carers.

	• review the goals of care and treatment plans with patients if significant changes in patients' conditions or circumstances occur	
Communication	• estimate prognosis and communicate this appropriately, if requested, including the uncertainties around such estimates • encourage patients' self-management through education to take greater responsibility for their care, and support problem solving • encourage patients' access to self-monitoring devices and assistive technologies • communicate with multidisciplinary team members to coordinate care of patients • discuss clinical outcomes or monitoring test results with patients to make appropriate changes to management as needed • communicate with families going through difficulties, including breaking bad or uncertain news, and managing family dynamics • communicate effectively and in a timely manner with other health professionals involved in patients' care • incorporate appropriate LGBTQIA+ safe language, including gender affirming language	• promote optimal management of genetic condition/s, including clearly articulating the risks on health in lifestyle choices • work in partnership with patients, and motivate them to comply with agreed care plans
Quality and safety	• use innovative models of health care delivery, including telehealth and digitally integrated support services • integrate new research and processes to optimise workflow and patient care in line with best practices • review disease-specific therapies, and ensure patients understand safe medication administration to prevent poor clinical outcomes • support patients' self-management when independent care is deemed appropriate • participate in quality improvement processes impacting on patients' quality of life	• participate in continuous quality improvement processes and clinical audits on genetic disease management • identify management strategies that may improve patients' quality of life • identify gaps in current care and processes, and raise these with the team • evaluate different methods of delivering care and filling service gaps, including identifying what has been completed in other services

Teaching and learning	• contribute to the development of clinical pathways for genetic disease management, based on current clinical guidelines • create teaching resources and programs to improve genetic health literacy for other professionals • educate patients and their families on long-term management expectations, including recognition of acute deterioration	• use clinical practice guidelines for the management of genetic diseases • participate in the education of other health professionals on the long-term care of patients
Research	• search for and critically appraise evidence to resolve clinical areas of uncertainty • work with families who have an undiagnosed condition to assist in research and recruitment to find a genetic diagnosis	• search literature using problem / intervention / comparison / outcome (PICO) format • recognise appropriate use of review articles • identify available clinical trials, natural history studies, and disease registries pertaining to genetic disease management and development of new treatments
Cultural safety	• encourage patients from culturally and linguistically diverse backgrounds to join local networks to receive the support needed for long-term self-management • ensure the clinical environment is inclusive of the cultural needs of patients and their support networks and/or family, such as using interpreters, cultural support officers, and disability liaison officers, and using the correct pronunciation of names • recognise the influence of values, culture, and priorities of diverse populations in treatment decisions, including end-of-life care	• provide culturally safe long-term disease management • recognise inherent cultural bias, both implicit and explicit, and the potential impact on clinical care
Ethics and professional behaviour	• share information about patients' health care, consistent with privacy laws and professional guidelines on confidentiality • use consent processes for the release and exchange of health information • assess patients' decision-making capacity, and appropriately identify and use proxy decision makers • establish supportive relationships with patients based on understanding, trust, empathy, and confidentiality • ensure all team members discuss end-of-life care with patients, and act on expressed patients' preferences	• share information between relevant service providers • acknowledge and respect the contribution of health professionals involved in patients' care

	• recognise the complexity of ethical issues related to human life and death when considering the allocation of scarce resources • recognise feelings of moral distress and burnout in themselves and colleagues • effectively manage personal challenges of dealing with death and grief	
Judgement and decision making	• avoid unnecessary investigations or treatments, ensuring physical and psychosocial support • implement stepped care pathways in the management of genetic conditions • recognise patients' needs in terms of both internal resources and external support on long-term health care journeys • maximise patients' autonomy and their best interests when making treatment decisions	• recognise personal limitations and seek help in an appropriate way when required • inadequately consult with senior colleagues
Leadership, management, and teamwork	• coordinate whole-person care through involvement in all stages of the patients' care journey • use a multidisciplinary approach across services to manage patients with genetic disease • develop collaborative relationships with patients, families, carers, and a range of health professionals • support general practitioners and community-based specialists with the long-term management of patients	• participate in multidisciplinary care for patients with genetic conditions, including organisational and community care, on a continuing basis, appropriate to patients' context
Health policy, systems, and advocacy	• use health screening for early diagnosis, intervention, and management of genetic conditions, such as the newborn bloodspot screening program • assess alternative models of health care delivery to provide individualised care to patients with genetic conditions • participate in government initiatives for management of genetic conditions to improve disease outcomes and patients' quality of life • ensure equity of access for patients to access initiatives and services pertaining to individuals with genetic conditions	• demonstrate awareness of government initiatives and services available for patients with genetic conditions, and display knowledge of how to access them

Learning goal 8: Communication with patients

Theme	**Communication with patients**
Title	**Discuss diagnoses and management plans with patients**
Description	This activity requires the ability to: • select suitable contexts and include family and/or carers and other team members • adopt a patient[11]-centred perspective, including adjusting for cognition and disabilities • select and use appropriate modalities and communication strategies • structure conversations intentionally • negotiate mutually agreed management plans • verify patients' understanding of information conveyed • develop and implement plans to ensure actions occur • ensure conversations are documented.

Behaviours

Professional practice framework domain	**Ready to perform without supervision** Expected behaviours of a trainee who can routinely perform this activity without needing supervision The trainee will:	**Requires some supervision** Possible behaviours of a trainee who needs some supervision to perform this activity The trainee may:
Medical expertise	• seek to understand the concerns and goals of patients, and plan management in partnership with them • provide information, including advantages and disadvantages, to patients to enable them to make informed decisions about diagnostic, therapeutic, and management options • anticipate and be able to correct any misunderstandings patients may have about their conditions and/or risk factors • identify and support patients in distress • educate patients about lifestyle factors that affect disease risk • discuss disease management plans and follow-up arrangements with patients • involve patients in developing mutually acceptable investigation plans in the setting of informed consent	• demonstrate an awareness of clinical problems being discussed • formulate management plans in partnership with patients • provide advice for patients, such as prenatal diagnosis, predictive genetic testing, and dissemination of genetic information to the family • recognise that counselling required in the acute care setting is completed under highly stressful circumstances, with multiple stakeholders invested in the testing process and results

---

[11] References to patients in the remainder of this document may include their families, whānau, and/or carers.

	• advise or refer patients for lifestyle changes for the management conditions	
Communication	• use appropriate communication strategies and modalities for communication, such as emails, face-to-face, or phone calls • elicit patients' views, concerns, and preferences, promoting rapport • provide information to patients in plain language, avoiding jargon, acronyms, and complex medical terms • encourage questions from patients, and answer them thoroughly • ask patients to share their thoughts or explain their management plans in their own words, to verify understanding • convey information considerately and sensitively to patients, seeking clarification if unsure of how best to proceed • explain how to access and use available resources and support groups • treat children and young people respectfully, and listen to their views • recognise the role of family or carers, and, when appropriate, encourage patients to involve their family or carers in decisions about their care • provide impartial genetic counselling with empathy and expertise while empowering, facilitating, and promoting patient and family participation in informed decision making • incorporate appropriate LGBTQIA+ safe language, including gender affirming language	• select appropriate modes of communication • engage patients in discussions, avoiding the use of jargon and medical terminology • check patients' understanding of information • adapt communication style in response to patients' age, developmental level, and cognitive, physical, cultural, socioeconomic, and situational factors • collaborate with patient liaison officers as required
Quality and safety	• provide information to patients in a way they can understand before asking for their consent • consider young people's capacity for decision making and consent • recognise and take precautions where patients may be vulnerable, such as issues of child protection, self-harm, or elder abuse • participate in processes to manage patients' complaints	• inform patients of the material risks associated with proposed management plans • treat information about patients as confidential
Teaching and learning	• obtain informed consent or other valid authority before involving patients in teaching	• respond appropriately to information sourced by patients, and to patients' knowledge regarding their condition

	• discuss the aetiology of diseases, and explain the purpose, nature, and extent of the assessments to be conducted	
Research	• provide information to patients that is based on guidelines issued by the National Health and Medical Research Council and/or Health Research Council of New Zealand • provide information to patients in a way they can understand before asking for their consent to participate in research • obtain an informed consent or other valid authority before involving patients in research	• refer to evidence-based clinical guidelines • recognise the limitations of the evidence and the challenges of applying research in daily practice
Cultural safety	• demonstrate effective and culturally safe communication with Aboriginal and Torres Strait Islander peoples and Māori • communicate effectively with members of other cultural groups by meeting patients’ specific language, cultural, and communication needs • use qualified language interpreters or cultural interpreters to help meet patients’ communication needs • provide plain language and culturally appropriate written materials to patients when possible	• identify when to use interpreters, and how to best use them • allow enough time for communication across linguistic and cultural barriers
Ethics and professional behaviour	• encourage and support patients to be well informed about their health, and to use information wisely when they make decisions • identify which types of data may be shared with or without patient consent • encourage and support patients in caring for themselves and managing their health • demonstrate respectful professional relationships with patients • prioritise honesty, patients’ welfare, and community benefit above self-interest • consult with patients before disclosing information, and maintain confidentiality • develop a high standard of personal conduct, consistent with professional and community expectations • support patients’ rights to seek second opinions	• respect the preferences of patients • communicate appropriately, consistent with the context, and respect patients’ needs and preferences • maximise patients’ autonomy, and support their decision making • avoid sexual, intimate, and/or financial relationships with patients • respect patients, including protecting their rights to privacy and confidentiality • behave equitably towards all, irrespective of gender, age, culture, socioeconomic status, sexual preferences, beliefs, contribution to society, illness-related behaviours, or the illness itself • use social media ethically and according to legal obligations to protect patients’ confidentiality and privacy

Judgement and decision making	• communicate to patients that there may be no right or wrong decision in settings of uncertainty • assist in patients' decision-making processes • elicit how a patient has previously dealt with uncertainty in their life, and use this when formulating decisions • reduce time pressure on patients in settings of uncertainty, recognising that there is time, even in acute settings, to reach decisions • recognise their own limitations and seek help in an appropriate way when required	• disclose and openly acknowledge areas of uncertainty to patients • inadequately consult with senior colleagues
Leadership, management, and teamwork	• communicate effectively with team members involved in patients' care, and with patients • discuss medical assessments, treatment plans, and investigations with patients and primary care teams, working collaboratively with all • discuss patients' care needs with healthcare team members to align them with appropriate resources • facilitate an environment in which all team members feel they can contribute, and their opinion is valued • communicate accurately and succinctly, and motivate others on the healthcare team	• answer questions from team members • summarise, clarify, and communicate responsibilities of healthcare team members • keep team members focused on patient outcomes
Health policy, systems, and advocacy	• collaborate with other services, such as community health centres and consumer organisations, to help patients navigate the healthcare system • contribute to multidisciplinary teams to offer diagnostic or predictive testing • encourage patients to access further information and patient support groups	• communicate with and involve other health professionals as appropriate

Learning goal 9: Investigations

Theme	**Investigations**
Title	**Select, organise, and interpret investigations**
Description	This activity requires the ability to: • select, plan, and use evidence-based clinically appropriate investigations • prioritise patients receiving investigations (if there is a waiting list) • evaluate the anticipated value of investigations • work in partnership with patients[12] to facilitate choices that are right for them • provide aftercare for patients (if needed) • interpret the results and outcomes of investigations • communicate the outcome of investigations to patients.

Behaviours

Professional practice framework **Domain**	**Ready to perform without supervision** Expected behaviours of a trainee who can routinely perform this activity without needing supervision The trainee will:	**Requires some supervision** Possible behaviours of a trainee who needs some supervision to perform this activity The trainee may:
Medical expertise	• choose evidence-based investigations, and frame them as an adjunct to comprehensive clinical assessments • assess patients' concerns, and determine the need for specific tests that are likely to result in overall benefits • develop plans for investigations, identifying their roles and timing • recognise and correctly interpret abnormal findings and variants of uncertain significance, considering patients' specific circumstances, and act accordingly • interpret the sensitivity, specificity, and predictive values of screening tests • discuss the cost effectiveness of individual investigation • discuss the diagnostic yield of investigations offered, and the need for further testing if initial investigations are inconclusive	• provide rationale for investigations • recognise the significance of abnormal test results, and act on these • consider patient factors and comorbidities • consider age-specific reference ranges • observe tests performed in a laboratory • collaborate as part of a multidisciplinary team, where appropriate, to ensure the most appropriate investigations are performed • recognise the constraints of available investigations, and explore other potential methods • identify when rapid testing is required, and be able to outline steps required to facilitate this
Communication	• explain to patients the potential benefits, burdens, costs and risks of each option, including the option to have no investigations, including in predictive and prenatal scenarios	• discuss benefits, complications, indications, and risks of investigations with patients before ordering investigations • explain the results of investigations to patients

---

[12] References to patients in the remainder of this document may include their families, whānau, and/or carers.

	use clear and simple language, and check that patients understand the terms used and agree to proceed with proposed investigations identify patients' concerns and expectations, providing adequate explanations on the rationale for individual test ordering confirm whether patients understand the information they have been given, and whether they need more information before making a decision use written or visual material or other aids that are accurate and up to date to support discussions with patients explain findings or possible outcomes of investigations to patients provide information that patients may find distressing in a considerate way	arrange investigations, providing accurate and informative referrals, and liaise with other services where appropriate
Quality and safety	identify adverse outcomes that may result from proposed investigations, focusing on patients' individual situations	consider safety aspects of investigations when planning them seek help with the interpretation of test results for less common tests or indications or unexpected results
Teaching and learning	use appropriate guidelines, evidence sources, and decision support tools participate in clinical audits to improve test ordering strategies for diagnoses and screening apply the use of available resources to enhance patients' care and professional development	undertake professional development to maintain currency with investigation guidelines reflect on own genetic counselling style and effectiveness, and identify strategies for improvement
Research	provide patients with relevant information if a proposed investigation is part of a research program obtain written consent from patients if the investigation is part of a research program	refer to evidence-based clinical guidelines consult current research on investigations use genetic databases and registers for information retrieval
Cultural safety	be cognisant of patients' views and preferences about any proposed investigations and the adverse outcomes they are most concerned about	consider patients' cultural and religious backgrounds, attitudes, and beliefs, and how these might influence the acceptability of proposed investigations
Ethics and professional behaviour	remain within the scope of the authority given by patients (with the exception of emergencies) discuss patients' rights and clinicians' responsibilities across the testing timeline	identify appropriate proxy decision makers when required choose not to investigate in situations where it is not appropriate for ethical reasons

	• respect patients' decisions to refuse investigations, even if their decisions may not be evidence based • advise patients there may be additional costs or implications of testing, such as insurance policies • explain the expected benefits as well as the potential burdens and risks of any proposed investigations before obtaining informed consent or other valid authority • explain complex issues related to genetic information obtained from investigations, and subsequent disclosure of such information • contribute to the operation of carrier screening programs • obtain informed consent for genetic investigations, with reference to the possibility of additional findings, data protection, data sovereignty, and insurance implications	• practise within current ethical and professional frameworks • practise within own limits, and seek help when needed • involve patients in decision making regarding investigations, obtaining the appropriate informed consent, including financial consent, if necessary
Judgement and decision making	• evaluate the costs, benefits, and potential risks of each investigation in a clinical situation • adjust the investigative path depending on test results received • consider whether patients' conditions may get worse or better if no tests are selected	• choose the most appropriate investigation for clinical scenarios in discussion with patients • recognise personal limitations and seek help in an appropriate way when required • inadequately consult with senior colleagues
Leadership, management, and teamwork	• consider the role other members of the healthcare team might play, and what other sources of information and support are available • identify situations where a rapid test is required, and take initiative in leading this process • ensure results are checked in a timely manner, taking responsibility for following up results • liaise with colleagues to interpret laboratory results • facilitate the testing process, including counselling, obtaining consent, and communication between pathology labs to ensure timely testing and results	• demonstrate awareness of what parts of investigations are provided by different doctors or health professionals
Health policy, systems, and advocacy	• select and justify investigations regarding the pathological basis of disease, appropriateness, utility, safety, and cost effectiveness • consider resource use through peer review of testing behaviours	

Learning goal 10: Clinic management

Theme	**Clinic management**	
Title	**Manage an outpatient clinic**	
Description	This activity requires the ability to: • manage clinic services in the context of a multidisciplinary team • participate in quality improvement activities • communicate with patients[13] • liaise with other health professionals and team members • demonstrate problem-solving skills • responsibly use public resources • manage medical procedures and treatments.	
Behaviours		
Professional practice framework domain	**Ready to perform without supervision** Expected behaviours of a trainee who can routinely perform this activity without needing supervision The trainee will:	**Requires some supervision** Possible behaviours of a trainee who needs some supervision to perform this activity The trainee may:
Medical expertise	• effectively identify and address current clinical concerns, as well as longer-term clinical objectives, as appropriate to patients' context • create accurate and appropriately prioritised problem lists in the clinical notes or as part of ambulatory care reviews • update documentation in a timeframe appropriate to the clinical situation of patients within the priorities of the department • conduct clinics with the goal of diagnosing genetic conditions and providing genetic counselling	• recognise the importance of prevention, early detection, health maintenance, and chronic condition management
Communication	• help patients navigate the healthcare system to improve access to care by collaboration with other services, such as community health centres and consumer organisations • link patients to specific community-based health programs and group education programs • adjust communication style to meet the needs of patients and families, such as in the settings of intellectual disability and health literacy	• wherever practical, meet patients' specific language and communication needs • facilitate appropriate use of interpreter services and translated materials

---

[13] References to patients in the remainder of this document may include their families, whānau, and/or carers.

	• provide genetic counselling around genetic testing, diagnosis, and family / reproductive implications • raise issues and feedback with the clinical team • incorporate appropriate LGBTQIA+ safe language, including gender affirming language	
Quality and safety	• practice health care that maximises patient safety • adopt a systematic approach to the review and improvement of professional practice in the outpatient clinic setting • identify aspects of service provision that may be a risk to patients' safety • demonstrate reflective practice • identify service gaps and work with the team to integrate quality improvement activities into the workplace	• take reasonable steps to address issues if patients' safety may be compromised • follow a systematic approach to improving the quality and safety of health care • participate in organisational quality and safety activities, including clinical incident reviews
Teaching and learning	• evaluate own professional practice • demonstrate learning behaviour and skills in educating junior colleagues • contribute to the generation of knowledge • maintain professional continuing education standards	• recognise the limits of personal expertise, and involve other professionals as needed to contribute to patients' care • use information technology appropriately as a resource for modern medical practice
Research	• obtain informed consent or other valid authority before involving patients in research • inform patients about their rights, the purpose of the research, the procedures to be undergone, and the potential risks and benefits of participation before obtaining consent • review research in the context of clinic patients, and integrate this into the clinical setting • review current clinical trials for rare diseases, and provide the option for individuals to participate when relevant	• allow patients to make informed and voluntary decisions to participate in research
Cultural safety	• apply knowledge of the cultural needs of the community served, and how to shape service to those people • mitigate the influence of own culture and beliefs on interactions with patients and decision making • adapt practice to improve patient engagement and health outcomes	• acknowledge the social, economic, cultural, and behavioural factors influencing health, both at individual and population levels

Ethics and professional behaviour	• identify and respect the boundaries that define professional and therapeutic relationships • respect the roles and expertise of other health professionals • comply with the legal requirements of preparing and managing documentation • demonstrate awareness of financial and other conflicts of interest	• recognise the responsibility to protect and advance the health and wellbeing of individuals and communities • maintain the confidentiality of documentation, and store clinical notes appropriately • ensure that the use of social media is consistent with ethical and legal obligations
Judgement and decision making	• discuss clinics with team members in pre-clinic and clinic review meetings • integrate prevention, early detection, health maintenance, and chronic condition management, where relevant, into clinical practice • work to achieve optimal and cost-effective patient care that allows maximum benefit from the available resources • order relevant investigations • interpret investigations, and synthesise into the clinical picture and management • recognise their own limitations and seek help in an appropriate way when required	• consider the appropriate use of diagnostic interventions, health care facilities, human resources, and therapeutic modalities • inadequately consult with senior colleagues
Leadership, management, and teamwork	• prepare for and conduct clinical encounters in a well-organised and time-efficient manner • work effectively as a member of multidisciplinary teams or other professional groups • ensure that all important discussions with colleagues, multidisciplinary team members, and patients are appropriately documented • review discharge summaries, notes, and other communications written by junior colleagues • support colleagues who raise concerns about patients' safety	• attend relevant clinical meetings regularly
Health policy, systems, and advocacy	• demonstrate capacity to engage in the surveillance and monitoring of the health status of populations in the outpatient setting • maintain good relationships with health agencies and services • apply the principles of efficient and equitable allocation of resources to meet individual, community, and national health needs	• review common population health screening and prevention approaches • identify the determinants of health of the population, and mitigate barriers to access to care • consult with genetic support groups with respect to relevant and accurate information • advocate for vulnerable and at-risk groups for access to services

	- advocate and be involved in public education, such as regarding preventable birth defects	

Knowledge Guides

Knowledge guides (KGs) provide detailed guidance to trainees on the important topics and concepts trainees need to understand to become experts in their chosen specialty.

Trainees are not expected to be experts in all areas or have experience related to all items in these guides.

#	Title
11	Clinical sciences
12	Laboratory based clinical genomics
13	Cancer genetics
14	Common adult and paediatric genetic conditions
15	Subspecialty genetics
16	Genetic counselling
17	Metabolic genetics

EPIDEMIOLOGY, PATHOPHYSIOLOGY, AND CLINICAL SCIENCES

Advanced Trainees will have in-depth knowledge of the topics listed under each clinical sciences heading.

For the statistical and epidemiological concepts listed, trainees should be able to describe the underlying rationale, the indications for using one test or method over another, and the calculations required to generate descriptive statistics.

- Basic principles of genetics and genomics, such as:
  - DNA structure and function
  - factors influencing gene expression, pre- and post-translational factors, RNA processing, splicing, transcription, and translation
  - gene expression concepts, such as:
    - gene regulation
    - mosaicism
    - penetrance
    - variable expressivity
  - meiosis and mitosis
  - normal and abnormal:
    - cell function and division
    - chromosomal structure and function
  - relationship between molecular alternations and the underlying pathophysiological mechanisms of genetic disease
  - structure and function of the human genome
  - X-chromosome inactivation (Lyonisation)
- Heredity and principles of genetic variation, such as:
  - developmental biology as it applies to abnormalities in the embryo, fetus, and newborn
  - genetic variation in populations, including carrier states, founder variants, and ancestry / ethnicity
  - implications of ancestry, consanguinity, and family history on genetic testing
  - patterns of inheritance, such as:
    - Mendelian / monogenic
    - mitochondrial
    - epigenetics, such as imprinting disorders
    - mosaicism:
      - cytogenetic
      - molecular
    - multifactorial
    - oligogenic
    - polygenic
    - repeat disorders and anticipation
  - pedigree interpretation
  - Hardy–Weinberg principles
- Principles of somatic and mosaic variants
- Risk factors associated with birth defects and genetic disorders, such as:
  - consanguinity, including cultural awareness in addressing counselling challenges associated with these relationships
  - environment / lifestyle:
    - alcohol
    - intrauterine infection
    - medication
    - occupation

        - smoking
    - ethnicity
    - maternal / paternal age
- Genetic basis of common and complex conditions, including rapid shifts in the understanding of major genetic principles over the past century
- Major metabolic pathways, including their regulation, interdependencies, and clinical and biochemical phenotypes that arise from their disruption, such as:
    - carbohydrate metabolism
    - mitochondrial energy production
    - fatty acid oxidation
    - amino acid and nitrogen metabolism

INVESTIGATIONS, PROCEDURES, AND CLINICAL ASSESSMENT TOOLS

Advanced Trainees will know the scientific foundation of each investigation and procedure, including relevant anatomy and physiology. They will be able to interpret the reported results of each investigation or procedure.

Advanced Trainees will know how to explain the investigation or procedure to patients[14], families, and carers, and be able to explain procedural risk and obtain informed consent where applicable.

- Genetic test reports, such as American College of Medical Genetics (ACMG) criteria for copy number variants (CNV) and sequence variants (SV) classification
- Genome-wide association studies (GWAS) and polygenic risk scores
- Common biochemical tests & metabolic screening:
    - acylcarnitine profile
    - plasma amino acids
    - urine metabolic screen
- Newborn screening
- Purpose, extent, and limitations of investigation results, such as:
    - negative predictive values
    - positive predictive values
    - sensitivity
    - specificity
    - avenues of clarifying variants of uncertain significance
- Appropriate clinical measurements
- Risk calculations, such as:
    - Bayes theorem and combinatorial probability
    - empiric risks, and how they are derived and used
    - family history assessment of risk
    - online risk calculators
    - presence of consanguinity
- Research advances and emerging areas in genetics, such as:
    - gene therapy
    - stem cells
    - therapeutic cloning

IMPORTANT SPECIFIC ISSUES

Advanced Trainees will identify important specialty-specific issues and the impact of these on diagnosis, management and outcomes.

- The diversity of public opinion on ethical and moral aspects of the practice of clinical genetics
- The history of use and abuse of genetic information
- The need for equity of services
- Impact of genetic conditions on cultural groups, families, and individuals
- Importance of genetic registers in genetic condition research and management
- Legal and Privacy Act requirements of patients' medical records, and that of the genetic record

---

[14] References to patients in the remainder of this document may include their families, whānau, and/or carers.

- Local and national guidelines and legislation for management on issues such as:
    - child protection issues
    - complaints, confidentiality, and reporting
    - human research
    - unexpected / incidental findings
- Resource allocation issues, including limitations on budget for genetic testing

Genetic screening programs

- Cultural, ethical, legal, and social issues associated with genetic and newborn screening programs, including advantages and disadvantages
- Guidelines for the establishment of population screening programs for genetic conditions, including counselling support

EPIDEMIOLOGY, PATHOPHYSIOLOGY, AND CLINICAL SCIENCES

Advanced Trainees will have in-depth knowledge of the topics listed under each clinical sciences heading.

For the statistical and epidemiological concepts listed, trainees should be able to describe the underlying rationale, the indications for using one test or method over another, and the calculations required to generate descriptive statistics.

- Advantages and disadvantages of single gene, gene panels, exome, and genome analysis, and the indications for testing choice in different scenarios, such as:
  - acute
  - diagnostic
  - predictive
  - prenatal
  - screening
- Genomic variants, such as:
  - national and international variant classification guidelines, such as:
    - American College of Medical Genetics (ACMG)
    - Association for Molecular Pathology (AMP)
  - strategies to resolve variants of uncertain significance
  - functional significance, including the likely pathogenicity and impact at a gene / protein level, and on individuals
  - emerging complex genomic mechanisms leading to disease, such as:
    - intronic variants
    - noncoding RNA
    - regulatory regions
    - structural variants
    - differential splicing
    - repeat disorders
- Genomics in cancer genetics, clinical genetics, metabolic conditions, and precision medicine, including new emerging areas, such as:
  - advanced therapeutics
  - pharmacogenomics
  - polygenic analysis
  - population screening

INVESTIGATIONS, PROCEDURES, AND CLINICAL ASSESSMENT TOOLS

Advanced Trainees will know the scientific foundation of each investigation and procedure, including relevant anatomy and physiology. They will be able to interpret the reported results of each investigation or procedure.

- Cytogenetics, such as:
  - karyotypes (G-banded) for diagnosis of numeric and structural chromosomal aberrations
  - chromosomal microarrays in diagnostic, prenatal, and somatic testing, and interpretation of:
    - copy number variants, such as microdeletion and microduplication syndromes, and neurosusceptibility loci
  - adjunct cytogenetic tests, such as:
    - fluorescence in situ hybridisation (FISH)
    - quantitative fluorescent polymerase chain reaction (QF-PCR)
    - uniparental disomy (UPD) studies
- Molecular genetics and genomics, such as:
    - massively parallel (exome and genome) sequencing, such as:
      - variant curation and interpretation
      - bioinformatics
      - functional genomics
    - structural variant analysis

Advanced Trainees will know how to explain the investigation or procedure to patients[15], families, and carers, and be able to explain procedural risk and obtain informed consent where applicable.

  - » adjunct genetic tests, such as:
    - ○ epigenetic studies, such as:
      - ▪ episignatures
      - ▪ imprinting
      - ▪ methylation
    - ○ multiplex ligation-dependent probe amplification (MLPA)
    - ○ repeat sequence testing, such as:
      - ▪ Huntington disease
      - ▪ myotonic dystrophy
  - » functional genomic testing, such as:
    - ○ in vivo and in vitro assays used in research
    - ○ protein expression studies, such as western blot
    - ○ reverse transcription polymerase chain reaction (RT-PCR)
    - ○ RNA sequencing and splicing studies
  - » gene panel testing, and curation of genes used in panels
  - » screening genomic tests, such as:
    - ○ non-invasive prenatal screening (NIPS)
    - ○ preconception carrier screening
  - » single gene (Sanger) sequencing
- Testing germline and somatic samples
- Carrier testing and screening

Applications of diagnostic testing

- Molecular and genomic testing, such as:
  - » diagnostic germline genomic testing (prenatal, paediatric, adult, post-mortem)
  - » diagnostic somatic genomic testing in lesional tissue and cancer
- Prenatal testing paradigms, such as NIPS and prenatal genomic testing, and preimplantation genetic diagnosis
- Role of functional genomics and novel gene discovery in characterising new genes and novel variants

Applications of presymptomatic and predictive testing

- Presymptomatic cancer genetic testing
- Predictive testing in the prenatal and preimplantation genetic setting
- Predictive testing of minors for adult-onset conditions

IMPORTANT SPECIFIC ISSUES

Advanced Trainees will identify important specialty-specific issues and the impact of these on diagnosis, management and outcomes.

- Community attitudes and concerns regarding genetic information and genetic technology
- Cultural, ethical, legal, and social implications of genetic technologies

Emerging applications of genomics

- Gene therapies and precision medicine
- Pharmacogenomics
- Polygenic risk scores

---

[15] References to patients in the remainder of this document may include their families, whānau, and/or carers.

Ethical issues

- Consent in relation to storage of DNA samples and cell lines
- Informed and specific consent for genetic testing, including in children
- Reanalysis of genomic data for diagnosis, research, and screening with consent
- The importance of ethical approval, patient consent, and research governance for clinical research

Legal and regulatory issues

- Difference between research and diagnostic genomic testing, and their roles in patient diagnosis
- Issues concerning prioritisation of application of genetic testing technologies and equity of access in the environment of limited resources
- Potential benefits and harms of genetic technologies
- Special considerations in prenatal genetic testing, predictive testing, and population genetic screening
- The regulation of biobanking and data management, including genomic data sovereignty and data usage agreements and confidentiality

Learning goal 13 – Cancer genetics

Advanced Training in Clinical Genetics

KEY PRESENTATIONS AND CONDITIONS

Advanced Trainees will have a comprehensive depth of knowledge of these presentations and conditions.

Presentations

- Cancer predisposition syndromes, including:
  - diagnostic genomic testing
  - predictive / pre-symptomatic genetic testing
  - cancer risk assessment and risk management strategies
- Family history of:
  - cancer predisposition syndromes and/or other relevant non-cancer features
  - common cancers, such as:
    - breast and/or ovarian
    - colorectal
    - prostate
    - childhood onset-cancer
  - rare cancers
- Incidental finding of germline pathogenic variants in cancer predisposition genes
- Somatically identified genetic variants, including the role and indications for germline testing
- Common somatic screening test results, including:
  - immunohistochemistry
  - microsatellite instability testing
- Personal history of:
  - cancer predisposition syndromes
  - congenital anomalies
  - neoplasm, including both malignant and non-malignant tumours with abnormal somatic or germline test results
  - rare cancers and/or multiple cancers
  - environmental and lifestyle related cancer risk factors

Conditions (including but not limited to)

- Hereditary breast and ovarian cancer
- Lynch syndrome
- Li–Fraumeni syndrome

For each presentation and condition, Advanced Trainees will **know how to:**

Synthesise

- » recognise the clinical presentation
- » identify relevant epidemiology, prevalence, pathophysiology, and clinical science
- » take a comprehensive clinical history
- » conduct an appropriate examination
- » establish a differential diagnosis
- » plan and arrange appropriate investigations
- » consider the impact of illness and disease on patients[16] and their quality of life when developing a management plan

Manage

- » provide evidence-based management
- » recognise potential complications of disease and its management, and initiate preventative strategies
- » involve multidisciplinary teams
- » identify and link patients with appropriate clinical trials, registries and other research options.

Consider other factors

- » identify individual and social factors and the impact of these on diagnosis and management

---

[16] References to patients in the remainder of this document may include their families, whānau, and/or carers.

	• Polyposis syndromes including adenomatous, hamartomatous, juvenile and serrated/hyperplastic polyps • Phosphatase and tensin homolog (PTEN) hamartoma tumour syndrome (PHTS) • Neurofibromatosis (NF) type 1 • Schwannomatosis • Hereditary diffuse gastric cancer	
LESS COMMON OR MORE COMPLEX PRESENTATIONS AND CONDITIONS Advanced Trainees will understand these presentations and conditions. Advanced Trainees will understand the resources that should be used to help manage patients with these presentations and conditions.	**Presentations** • Treatment-focused genetic testing in cancer, including the role of precision medicine • Moderate risk cancer predisposition genes • Polygenic risk scores for cancer • Circulating tumour DNA testing • Polygenic risk scores for cancer • Clonal haematopoesis • Non-invasive prenatal testing results suspicious for maternal malignancy • Tumour multi-omics testing, such as: » methylation » RNA **Conditions** • Cancers potentially associated with cancer predisposition syndromes, including the genetic testing approach to: » breast and ovarian » colorectal » kidney » retinoblastoma » sarcoma » thyroid » haematological » melanoma (cutaneous and uveal) » medulloblastoma » mesothelioma » neuroblastoma » nephroblastoma, including Wilms tumourparaganglioma and phaeochromocytoma • Von Hippel–Lindau syndrome • Tuberous sclerosis complex • Peutz-Jeghers syndrome • Multiple endocrine neoplasia (MEN) syndromes	

	• Paraganglioma-phaeochromocytoma syndromes • Beckwith-Wiedemann syndrome • Birt-Hogg-Dube syndrome • Hereditary leiomyomatosis and renal cell cancer syndrome • Constitutional mismatch repair deficiency (CMMRD) • DICER1 cancer predisposition syndrome • Nevoid basal cell carcinoma (Gorlin) syndrome • Rhabdoid tumour predisposition syndromes	
EPIDEMIOLOGY, PATHOPHYSIOLOGY, AND CLINICAL SCIENCES Advanced Trainees will have a comprehensive depth of knowledge of the principles of the foundational sciences.	• Basic approach to common neoplasms and haematological malignancies, and the role of genetic testing • Clinical application of polygenic cancer risk scores • Clinicopathological features of cancers in cancer predisposition syndromes, including tumour screening programs and tests • Current recommendations concerning tumour surveillance in families with cancer predisposition syndromes • Estimation of probability of an underlying pathogenic variant in a cancer predisposition gene based on personal and family history • Genetic and environmental factors that affect risk of developing cancer • Genetic mechanisms in cancer, such as: » proto-oncogenes » tumour suppressor genes • Pathophysiology of cancer development • Medical and family history features of inherited cancers syndromes, such as monogenic high penetrance familial predisposition to cancer	
INVESTIGATIONS, PROCEDURES, AND CLINICAL ASSESSMENT TOOLS Advanced Trainees will know the scientific foundation of each investigation and procedure, including relevant anatomy and physiology. They will be able to interpret the reported results of each investigation or procedure. Advanced Trainees will know how to explain the investigation or procedure to patients, families, and carers, and be able to explain procedural risk and obtain informed consent where applicable.	• Germline single gene and gene panel based genetic tests • Somatic screening test and somatic genetic testing • Online tools and guidelines, such as: » CanRisk » European Society for Medical Oncology (ESMO) Guidelines » eviQ	

IMPORTANT SPECIFIC ISSUES

Advanced Trainees will identify important specialty-specific issues and the impact of these on diagnosis and management and integrate these into care.

- Approach to cascade testing in families:
  - consent for genomic investigations, including data governance and sovereignty, and discussion of additional findings
  - consent and ethical challenges related to testing children for pathogenic variants usually associated with adult-onset cancer, in both the predictive and diagnostic settings
  - impact of cancer risk and genetic diagnosis on individuals and families, including access to risk management and insurance implications
  - return of results related to cancer risk detected through incidental findings in genetic testing
  - the roles primary care physicians and genetic counsellors play in assessing families where relatives are at risk of developing cancer

Learning goal 14 – Common adult and paediatric genetic conditions

Advanced Training in Clinical Genetics

KEY PRESENTATIONS AND CONDITIONS

Advanced Trainees will have a comprehensive depth of knowledge of these presentations and conditions.

- Common Mendelian (single gene) disorders:
  - autosomal dominant
  - autosomal recessive
  - X-linked
  - mitochondrial
  - imprinting disorders
- Chromosome disorders, such as:
  - aneuploidy
  - major chromosomal structural variants (duplications, deletions, translocations, inversions)
  - chromosomal copy number variants (CNVs), such as microdeletions and microduplications
- Intellectual disability and autism
- Developmental regression
- Neurological symptoms, including:
  - myopathy
  - neuropathy
  - seizures
- Teratogenic disorders
- Congenital anomalies and abnormal growth or development, including:
  - ambiguous genitalia
  - arthrogryposis
  - cleft lip / palate
  - craniofacial malformation
  - craniosynostosis
  - eye anomaly
  - facial dysmorphism
  - growth retardation
  - hemihypertrophy
  - limb anomaly
  - obesity
  - overgrowth
  - skeletal dysplasia
  - vascular anomaly

For each presentation and condition, Advanced Trainees will **know how to:**

Synthesise

- recognise the clinical presentation
- identify relevant epidemiology, prevalence, pathophysiology, and clinical science
- take a comprehensive clinical history
- conduct an appropriate examination
- establish a differential diagnosis
- plan and arrange appropriate investigations
- consider the impact of illness and disease on patients[17] and their quality of life when developing a management plan

Manage

- provide evidence-based management
- recognise potential complications of disease and its management, and initiate preventative strategies
- involve multidisciplinary teams
- identify and link patients with appropriate clinical trials, registries and other research options

Consider other factors

- identify individual and social factors and the impact of these on diagnosis and management

---

[17] References to patients in the remainder of this document may include their families, whānau, and/or carers.

	Examples of common syndromal genetic conditions • Achondroplasia • Beckwith–Wiedemann syndrome • Coffin–Siris syndrome • Recurrent chromosomal copy number variant disorders • Fragile X syndrome • Kabuki syndrome • KBG syndrome • Marfan syndrome • Mitochondrial conditions • Neurofibromatosis type 1 • Noonan syndrome • Osteogenesis imperfecta • 22q11.2 microdeletion • Russell–Silver syndrome • Sotos syndrome • Tuberous sclerosis • Williams syndrome	
LESS COMMON OR MORE COMPLEX PRESENTATIONS AND CONDITIONS Advanced Trainees will understand these presentations and conditions. Advanced Trainees will understand the resources that should be used to help manage patients with these presentations and conditions.	• Numerous rare and newly identified genetic syndromes and developmental sequences may be encountered over the course of training, including adult presentations	
EPIDEMIOLOGY, PATHOPHYSIOLOGY, AND CLINICAL SCIENCES Advanced Trainees will have a comprehensive depth of knowledge of the principles of the foundational sciences.	• Approach to conditions with a range of aetiologies, such as: » cerebral palsy » connective tissue disorders » muscular dystrophy » myopathy and neuromuscular disorders » neurodevelopmental disorders • Normal embryologic development • Presentation mechanisms at different stages of life, including: » prenatal » neonatal » childhood » adulthood » postmortem • Teratogenesis	

INVESTIGATIONS, PROCEDURES, AND CLINICAL ASSESSMENT TOOLS

Advanced Trainees will know the scientific foundation of each investigation and procedure, including relevant anatomy and physiology. They will be able to interpret the reported results of each investigation or procedure.

Advanced Trainees will know how to explain the investigation or procedure to patients, families, and carers, and be able to explain procedural risk and obtain informed consent where applicable.

- Newborn screening
- Basic radiographic signs
- Genomic-first diagnosis through reverse phenotyping
- Standard clinical photographs to review with colleagues for possible syndromes
- Use of technology to inform diagnosis, such as artificial intelligence (AI) and face-matching

IMPORTANT SPECIFIC ISSUES

Advanced Trainees will identify important specialty-specific issues and the impact of these on diagnosis and management and integrate these into care.

- Consent for genomic investigations, including data governance and sovereignty and discussion of additional findings
- Emotional reactions of families, including following early diagnosis of a syndrome or recognition of developmental delay, and retraction of a previous diagnosis
- Importance of accuracy, clinical judgement, tact, and timing when diagnosing and informing parents of infants with a serious condition impacting development, or when providing reproductive choice
- Specialist centres and/or individuals for potential referral and to assist in the diagnosis of rare dysmorphic syndromes or other presentations, including multidisciplinary teams, such as:
    - neuromuscular meetings
    - undiagnosed disease programs

Learning goal 15 – Subspecialty genetics

Advanced Training in Clinical Genetics

KEY PRESENTATIONS AND CONDITIONS

Advanced Trainees will have a comprehensive depth of knowledge of these presentations and conditions.

- Prenatal genetics:
  - abnormal antenatal:
    - genetic screening result
    - ultrasound scan findings
  - abnormal prenatal diagnostic result
  - family history of known genetic condition
  - preimplantation genetic diagnosis
  - fetal postmortem genetic testing
  - fetal exposure to teratogens
- Cardiac genetic conditions, such as:
  - arrhythmia
  - cardiomyopathy
  - sudden cardiac death
- Neurogenetic presentations, such as:
  - epilepsy
  - structural brain malformations
  - neuromuscular conditions
  - neurodegenerative conditions
- Connective tissue disorders
- Dermatological conditions
- Endocrinological conditions
- Gastrointestinal conditions
- Haematological conditions
- Immunological conditions
- Ophthalmological conditions, such as:
  - inherited retinal dystrophy
- Renal conditions, such as:
  - cystic kidney disease
  - Alport syndrome
- Respiratory conditions, such as:
  - cystic fibrosis
  - interstitial lung disease and pulmonary fibrosis

For each presentation and condition, Advanced Trainees will **know how to:**

Synthesise

- recognise the clinical presentation
- identify relevant epidemiology, prevalence, pathophysiology, and clinical science
- take a comprehensive clinical history
- conduct an appropriate examination
- establish a differential diagnosis
- plan and arrange appropriate investigations and appropriate referrals
- consider the impact of illness and disease on patients[18] and their quality of life when developing a management plan

Manage

- provide evidence-based management plans
- recognise potential complications of disease and its management, and initiate preventative strategies
- identify and link patients with appropriate clinical trials, registries and other research options.

Consider other factors

- identify individual and social factors including employment and lifestyle and the impact of these on diagnosis and management

---

[18] References to patients in the remainder of this document may include their families, whānau, and/or carers.

	• Skeletal disorders and dysplasias, such as: » primary skeletal dysplasias » osteogenesis imperfecta and genetic osteoporosis » disorders of bone mineralisation • Vascular genetic disorders	
LESS COMMON OR MORE COMPLEX PRESENTATIONS AND CONDITIONS Advanced Trainees will understand these presentations and conditions. Advanced Trainees will understand the resources that should be used to help manage patients with these presentations and conditions.	• Numerous and newly identified genetic subspecialties may be encountered over the course of training	
EPIDEMIOLOGY, PATHOPHYSIOLOGY, AND CLINICAL SCIENCES Advanced Trainees will have knowledge of the principles of the foundational sciences.	• Normal embryologic development • Presentation mechanisms at different stages of life, including: » prenatal » neonatal » childhood » adulthood » postmortem	
INVESTIGATIONS, PROCEDURES, AND CLINICAL ASSESSMENT TOOLS Advanced Trainees will know the scientific foundation of each investigation and procedure, including relevant anatomy and physiology. They will be able to interpret the reported results of each investigation or procedure. Advanced Trainees will know how to explain the investigation or procedure to patients, families, and carers, and be able to explain procedural risk	• Genomic investigations for subspecialties, such as: » genomic testing » adjunct tests, including: ○ methylation signatures ○ other investigations ○ repeat disorder studies ○ functional genomics • Other relevant investigations, such as medical imaging and tissue biopsy, as appropriate	

and obtain informed consent where applicable.	
IMPORTANT SPECIFIC ISSUES Advanced Trainees will identify important specialty-specific issues and the impact of these on diagnosis and management and integrate these into care.	• Trainees should develop an overall approach to new areas of genomics and need to have an awareness that new developments driven by genomic research are changing the landscape of subspecialty genetics at a rapid pace. This means a comprehensive list of specific subspecialty knowledge is quickly outdated and may not fully reflect future practice

CLINICAL SCIENCES

Advanced Trainees will describe the principles of the foundational sciences.

- Role of the genetic counsellor, and how to co-consult collaboratively in supporting the patient as they integrate genomic information and make decisions about testing and family planning
- Patient-centred care
- Uncertainty regarding testing limitations and potential diagnosis
- Normal and pathological reactions to grief and loss, and its effects on decision making regarding genetic risks
- Counselling, and communication theories that underpin genetic counselling
- Formulation of questions to identify patients' acceptance, agenda, concerns, expectations, priorities, and understanding
- Advantages and disadvantages of both diagnostic and predictive genetic testing
- Mendelian and non-Mendelian modes of inheritance
- Non-disclosure genetic testing in preimplantation genetic diagnosis (PGD) or prenatal testing

ELIGIBILITY CONSIDERATIONS

Advanced Trainees will assess the patient's[19] current condition and plan the next steps.

- Counselling as a patient-centered framework that considers:
  - Aboriginal and Torres Strait Islander peoples and Māori health perspectives
  - complex family structures
  - cultural beliefs and sensitivities, and ancestry background
  - health literacy
  - lived experiences of patients and clinicians
  - patients' and family members' anxieties
  - religious and philosophical beliefs and attitudes
  - the influence of personal identity, including gender diversity
- Cultural and community attitudes to genetics and genetic testing
- Empiric and recurrence risk using family history and population data
- Ethical issues, such as those concerning:
  - assisted reproduction
  - confidentiality
  - consent for postmortem examinations:
    - differing cultural beliefs about death
    - treatment of the body after death
  - 'duty to warn' and 'obligation to recontact'
  - financial implications of testing for the patient, including insurance implications
  - genetic testing of children, including predictive testing
  - informed consent
  - population screening for genetic disease
  - predictive genetic testing
  - preimplantation and prenatal genetic diagnosis
  - preimplantation and prenatal screening

---

[19] References to patients in the remainder of this document may include their families, whānau, and/or carers.

    - research ethics process
    - termination of pregnancy
- Familial implications of genetic diagnosis, such as:
    - asking questions in a sensitive and thoughtful manner
    - assisting patients with sharing relevant genetic information with other family members
    - communicating bad news in a caring and sensitive manner
    - engaging in realistic discussions regarding sensitive subjects
    - facilitating the discussion with empathy, honesty, respect, and sensitivity
- Financial, medical, personal, and psychological circumstances of the patient
- Genetic risk assessment of disease predisposition or carrier status, including complexities of incomplete penetrance and age-related penetrance
- Reproductive carrier screening and its limitations

LESS COMMON AND MORE COMPLEX PATIENT CONSIDERATIONS

Advanced Trainees will understand the resources that should be used to help manage patients.

- Unique challenges of genetic counselling in different scenarios, including:
    - acute care setting
    - in perinatal loss or sudden death
    - intensive care unit
    - the neonatal unit
    - the prenatal setting

UNDERTAKING THERAPY

Advanced Trainees will monitor the progress of patients during the therapy.

- Skills required for counselling, such as:
    - active listening
    - avoiding jargon, and using familiar language
    - encouraging questions
    - using open questions followed by appropriate closed questions

POST-THERAPY

Advanced Trainees will know how to monitor and manage patients post-therapy.

- Delivery of potentially distressing information about diagnosis and prognosis
- Provide accurate information regarding risk to family members of a known genetic condition
- Provide information in a timely manner, including pre- and post-test information and the outcome of testing

IMPORTANT SPECIFIC ISSUES

Advanced Trainees will identify important specialty-specific issues and the impact of these on diagnosis and management and integrate these into care.

- Complexities of communicating with patients and families around consent, result disclosure, and follow up
- Human Genetics Society of Australasia (HGSA) guidelines, and international equivalents
- Local and international policies on the indications and timing of genetic testing
- Uncertainty and emotional aspects of genetic testing and potential outcomes

Learning goal 17 – Metabolic genetics

KEY PRESENTATIONS AND CONDITIONS

Advanced Trainees will have a comprehensive depth of knowledge of these presentations and conditions.

- Key inborn errors of metabolism:
  - disorders of amino acid metabolism and transport, such as:
    - phenylketonuria (PKU)
    - maple syrup urine disease (MSUD)
    - homocystinuria
  - organic acidurias, such as:
    - methylmalonic aciduria (MMA)
    - propionic aciduria (PA)
    - glutaric aciduria type 1 (GA1)
  - urea cycle disorders and associated transporters, such as:
    - ornithine transcarbamylase (OTC) deficiency
  - disorders of fat and ketone metabolism, such as:
    - fatty acid oxidation defects
    - defects of ketogenesis and ketolysis
  - disorders of carbohydrate metabolism, such as:
    - galactosemia
    - glycogen storage disorders
  - lysosomal storage disorders, such as:
    - mucopolysaccharidoses
    - Fabry disease
    - Pompe disease
  - peroxisomal disorders, such as:
    - X-linked adrenoleukodystrophy (X-ALD)
    - Zellweger spectrum disorder (ZSD)
  - mitochondrial disorders

Presentations

- Metabolic acidosis
- Hypoglycaemia
- Hyperammonaemia
- Neuroregression
- Encephalopathy
- Cardiomyopathy

For each presentation and condition, Advanced Trainees will **know how to:**

Synthesise

- recognise the clinical presentation
- identify relevant epidemiology, prevalence, pathophysiology, and clinical science
- take a comprehensive clinical history
- conduct an appropriate examination
- establish a differential diagnosis
- plan and arrange appropriate investigations and appropriate referrals
- consider the impact of illness and disease on patients[20] and their quality of life when developing a management plan

Manage

- provide evidence-based management plans
- recognise potential complications of disease and its management, and initiate preventative strategies
- identify and link patients with appropriate clinical trials, registries and other research options.

Consider other factors

- identify individual and social factors including employment and lifestyle and the impact of these on diagnosis and management

---

[20] References to patients in the remainder of this document may include their families, whānau, and/or carers.

	• Liver failure or cholestasis • Hepatosplenomegaly • Rhabdomyolysis • Leukoencephalopathy • Positive newborn screen • Dysmorphic presentations	
LESS COMMON OR MORE COMPLEX PRESENTATIONS AND CONDITIONS Advanced Trainees will understand these presentations and conditions. Advanced Trainees will understand the resources that should be used to help manage patients with these presentations and conditions.	**Conditions** • Congenital disorders of glycosylation • Cerebral creatine deficiency syndromes • Disorders of neurotransmission • Disorders of vitamin and trace metal transport and metabolism • Disorders of lipid and bile acid metabolism • Porphyrias • Purine and pyrimidine disorders	
EPIDEMIOLOGY, PATHOPHYSIOLOGY, AND CLINICAL SCIENCES Advanced Trainees will have knowledge of the principles of the foundational sciences.	• Pathophysiology of clinical symptoms and signs of metabolic conditions and their corresponding common laboratory findings, including findings of newborn bloodspot screening and specialised biochemical tests • Presentation mechanisms at different stages of life, including: » prenatal » neonatal » childhood » puberty » adulthood » postmortem • Genetic and environmental factors that affect disease severity and outcomes • Metabolism under different conditions, such as: » pregnancy » fasting » exercise » other physiological stress • Familiarity with key metabolic pathways, such as: » amino acid metabolism » glucose homeostasis » fatty acid oxidation » urea cycle	
INVESTIGATIONS, PROCEDURES, AND CLINICAL ASSESSMENT TOOLS Advanced Trainees will know the scientific foundation of each	• Metabolic screening investigations, including: » acylcarnitine profile » plasma amino acids » urine metabolic screen • Newborn screening	

investigation and procedure, including relevant anatomy and physiology. They will be able to interpret the reported results of each investigation or procedure. Advanced Trainees will know how to explain the investigation or procedure to patients, families, and carers, and be able to explain procedural risk and obtain informed consent where applicable.	• Other relevant investigations, such as: » CSF neurotransmitters » enzyme assays » very long chain fatty acids » homocysteine • Nutritional assessment and investigations • Disease-specific rating scales
IMPORTANT SPECIFIC ISSUES Advanced Trainees will identify important specialty-specific issues and the impact of these on diagnosis and management and integrate these into care.	• Principles of acute and chronic metabolic disease management: » avoidance of catabolism » use of specialised metabolic diets » strategies to remove or sequester toxic metabolites, such as use of ammonia scavengers, haemofiltration, and l-carnitine » use of cofactors and alternative pathways • Specific treatments, such as: » enzyme replacement therapy » solid organ and haematopoietic stem cell transplantation » substrate reduction and chaperone therapy » gene therapy • Impact of metabolic disease on pregnancy • Role of the multidisciplinary team • Role of clinical trials, disease registries, and natural history studies in the management and development of new treatments