

Effective for residents who enter training on or after July 1, 2022.

DEFINITION

Medical Genetics and Genomics is the branch of medicine concerned with the study, diagnosis, management, and prevention of genetic, epigenetic, and genomic disorders in patients, families and communities, as well as with the effect of genetic variation on human development and health.

MEDICAL GENETICS AND GENOMICS PRACTICE

Medical geneticists provide care for patients of all ages who have a known or suspected personal or family history of a genetic, epigenetic, or genomic disorder, including inborn errors of metabolism. Genetic disorders are often rare and are typically complex with multi-system dysfunction.

Medical geneticists gather and synthesize clinical information from many sources and perform clinical assessments with a focus on dysmorphology and phenotyping. They request and interpret the results of laboratory, imaging, and genetic investigations. Establishing a diagnosis may include monitoring the evolving phenotype, incorporating information learned from testing other family members, or collaborating with researchers to investigate new genetic anomalies. Medical geneticists provide guidance to referring physicians, patients, and families regarding diagnosis; make recommendations regarding management; and provide anticipatory guidance. They provide long-term treatment and follow-up for patients with inborn errors of metabolism. As more targeted therapies are being developed, medical geneticists have an expanding role in the delivery of interventions to patients with a broader range of genetic disorders.

Medical geneticists apply their expertise in genetic and genomic testing to counsel individual patients, educate referring physicians, and advise the health care system. They counsel patients regarding the implications of genetic and genomic testing and about the findings and their implications for inheritance, treatment, and surveillance. They guide other physicians in their interpretation of the results of genetic and genomic testing, and help them use these tests and their results judiciously within their practice. Medical geneticists participate in the development of testing technologies and advise administrators, policy makers, and health care systems regarding the implementation of and resource allocation for new testing methods and practices.

Most medical geneticists work in academic settings where they have access to the technology required for diagnostic testing; the medical, surgical, and diagnostic specialists required for consultation and referral; and the genetic counsellors, genetic laboratories, and administrative support required for the care of their patient population. They also work remotely, providing their expertise via outreach clinics and telehealth consultations.

Medical geneticists work effectively within an interprofessional health care team comprised of genetic counsellors, genetic assistants, laboratory geneticists, nurses, dietitians, newborn screening teams, and social workers to address the complex needs of their patient population. With the impact of developments in the genetic understanding of pathophysiology and management in all aspects of medicine, medical geneticists collaborate with physicians within all specialties; this includes multidisciplinary specialty clinics focusing on specific systems or disorders.

MEDICAL GENETICS AND GENOMICS COMPETENCIES

Medical Expert

Definition:

As *Medical Experts*, medical geneticists integrate all of the CanMEDS Roles, applying medical knowledge, clinical skills, and professional values in their provision of high-quality and safe patient-centred care. Medical Expert is the central physician Role in the CanMEDS Framework and defines the physician's clinical scope of practice.

Key and Enabling Competencies: Medical geneticists are able to...

1. Practise medicine within their defined scope of practice and expertise

- 1.1. Demonstrate a commitment to high-quality care of their patients
- 1.2. Integrate the CanMEDS Intrinsic Roles into their practice of Medical Genetics and Genomics
- 1.3. Apply knowledge of the clinical and biomedical sciences relevant to Medical Genetics and Genomics

Basic principles of genetics and genomics

- 1.3.1. General structure and function of the human genome
- 1.3.2. Chromosome structure, morphology, and nomenclature
- 1.3.3. Principles of genomics
- 1.3.4. Processes underlying genomic variability, redundancy, and plasticity
- 1.3.5. Factors influencing gene expression
- 1.3.6. Normal and abnormal gene structure and function
- 1.3.7. Patterns of single gene inheritance
- 1.3.8. Genetics of common disorders with complex inheritance
- 1.3.9. Normal and abnormal cell function and division

MEDICAL GENETICS AND GENOMICS COMPETENCIES (2022)

- 1.3.9.1. DNA transcription and replication
- 1.3.9.2. RNA processing and translation
- 1.3.9.3. Pre- and post-translational factors influencing protein function

- 1.3.10. Basic principles of biochemistry applicable to Medical Genetics and Genomics
- 1.3.11. Epidemiology, including biostatistics, genetic epidemiology, and population genetics
- 1.3.12. Principles of and approaches to population screening

- Diagnostic approaches and techniques
- 1.3.13. Methods of assessment of phenotypic variation
- 1.3.14. Methods of syndrome identification and diagnosis, including the use of computer diagnostic aids
- 1.3.15. Principles and application of cytogenetic techniques
- 1.3.16. Principles and application of molecular genetic techniques and genomic analysis
 - 1.3.16.1. Variant interpretation using bioinformatics
 - 1.3.16.2. Genetic and genomic testing relevant to precision medicine
 - 1.3.16.2.1. Pharmacogenetics and pharmacogenomics
 - 1.3.16.3. Methods for evaluation of epigenetic modification and protein expression (i.e., proteomics, metabolomics)
- 1.3.17. Methods and applications of laboratory investigations relevant to inborn errors of metabolism

- Clinical disorders and therapeutics
- 1.3.18. Approach to the evaluation of patients with a suspected genetic disorder, with consideration to the following etiologies
 - 1.3.18.1. Environmental factors
 - 1.3.18.2. Chromosomal disorders
 - 1.3.18.3. Single gene disorders
 - 1.3.18.3.1. Dominant
 - 1.3.18.3.2. Recessive
 - 1.3.18.3.3. X-linked
 - 1.3.18.4. Disorders due to mutations in the mitochondrial genome
 - 1.3.18.5. Disorders due to mosaicism
 - 1.3.18.6. Imprinting disorders
 - 1.3.18.7. Disorders associated with unstable triplet repeats

MEDICAL GENETICS AND GENOMICS COMPETENCIES (2022)

- 1.3.18.8. Multifactorial genetic disorders

- 1.3.19. Clinical features, etiology, diagnosis, management, natural history, and prognosis of:
 - 1.3.19.1. Syndromes with well-defined dysmorphology
 - 1.3.19.2. Malformations, deformations, disruptions, sequences, and associations
 - 1.3.19.3. Cancer syndromes
 - 1.3.19.4. Cardiac genetic disorders
 - 1.3.19.5. Neurogenetic disorders
 - 1.3.19.6. Skeletal dysplasias
 - 1.3.19.7. Inborn errors of metabolism
 - 1.3.19.8. Other single gene disorders
 - 1.3.19.9. Imprinting disorders
 - 1.3.19.10. Chromosomal syndromes
 - 1.3.19.11. Genomic disorders associated with copy number variation

- 1.3.20. Genetic approach to prenatal patients, including screening, imaging, methods of further investigation, and genetic testing

- 1.3.21. Predictive testing and risk assessment for predisposition to monogenic or complex genetic diseases

- 1.3.22. Methods of action, indications, contraindications, availability and alternatives for therapeutic interventions, including supportive and disease modifying therapies

- 1.4. Apply knowledge of the basic and clinical sciences applicable to the medical care of patients of all ages, relevant to Medical Genetics and Genomics
 - 1.4.1. Normal anatomy of all body systems, including common variations
 - 1.4.2. Physiology as it applies to all body systems, and the changes that occur in the continuum from fetal life to adulthood
 - 1.4.3. Pathophysiology as it applies to cardiac, vascular, pulmonary, gastrointestinal and hepatobiliary, renal, endocrine, neurological, musculoskeletal, hematologic, and immunologic systems, as well as infection and shock
 - 1.4.4. Indicators of normal and abnormal psychomotor development, including the use and limitations of commonly used instruments for the assessment of behaviour and intelligence
 - 1.4.5. Feeding and nutrition
 - 1.4.5.1. Recommended nutritional requirements for all age groups

MEDICAL GENETICS AND GENOMICS COMPETENCIES (2022)

- 1.4.5.2. Nutritional assessment, including effect of disease states on nutritional requirements
- 1.4.5.3. Parenteral and enteral nutrition for all age groups, including special formula options
- 1.4.6. Considerations for investigations and tests in special patient populations (children, seniors, critically ill patients), including limits of blood volume and minimum requirements for testing
- 1.4.7. Medical imaging modalities and their indications, contraindications, and risks
- 1.4.8. Pharmacology as it relates to pharmacokinetics, pharmacodynamics, mechanism of action, routes of delivery and elimination, therapeutic monitoring, and adverse effects of commonly prescribed medications
 - 1.4.8.1. Modifications of drug dosing due to age, size, and altered pathophysiologic states, including renal and liver dysfunction
 - 1.4.8.2. Pharmacogenetic effects on drug response
- 1.4.9. Acute care, including emergencies and critical care
 - 1.4.9.1. Algorithms of cardiopulmonary resuscitation for patients of all ages
- 1.4.10. Epidemiology, clinical manifestations, approach to investigation, and management of common acute and chronic medical conditions
- 1.4.11. Management of patients requiring complex chronic care and/or technology dependence
- 1.4.12. Principles of palliative care, including advanced care planning and end-of-life care
- 1.4.13. Legal issues in medicine
 - 1.4.13.1. Assent and consent
 - 1.4.13.2. Capacity and medical decision-making
 - 1.4.13.3. Privacy and confidentiality
 - 1.4.13.4. Requirements for mandatory reporting
- Issues specific to peripartum care
- 1.4.14. Developmental biology as it relates to normal and abnormal human morphogenesis
- 1.4.15. Genetic and non-genetic (intrinsic and extrinsic) factors predisposing to fetal loss, infertility, and abnormalities of morphogenesis
- 1.4.16. Indications, limitations, and risks of techniques for preimplantation genetic testing and prenatal genetic screening and testing
 - 1.4.16.1. Complications of invasive procedures, including chorionic villus sampling and amniocentesis

MEDICAL GENETICS AND GENOMICS COMPETENCIES (2022)

- 1.4.17. Antepartum fetal surveillance in the normal and high-risk pregnancy, including appropriate use of obstetric ultrasound
- 1.4.18. Principles of antepartum care
 - 1.4.18.1. Effects of underlying medical, surgical, social, psychosocial, and mental health disorders on maternal and fetal health, and appropriate management of any implications for maternal or fetal health
 - 1.4.18.2. Outcomes of pregnancies complicated by fetal anomaly/anomalies or cytogenetic abnormalities, including aneuploidy
 - 1.4.18.3. Teratogenic agents and their effects
- 1.4.19. Placental transfer and breast milk excretion of drugs
- 1.4.20. Safety of pharmacotherapy in pregnancy, including knowledge of appropriate resources to obtain detailed information
- 1.5. Perform appropriately timed clinical assessments with recommendations that are presented in an organized manner
- 1.6. Carry out professional duties in the face of multiple competing demands
- 1.7. Recognize and respond to the complexity, uncertainty, and ambiguity inherent in Medical Genetics and Genomics practice

2. Perform a patient-centred clinical assessment and establish a management plan

- 2.1. Prioritize issues to be addressed in a patient encounter
 - 2.1.1. Ascertain the patient's and family's¹ perspective and priorities
- 2.2. Elicit a history, perform a physical exam, select appropriate investigations, and interpret their results for the purpose of diagnosis and management, disease prevention, and health promotion
 - 2.2.1. Elicit a comprehensive medical and family history
 - 2.2.2. Construct and interpret a standardized pedigree
 - 2.2.3. Carry out a comprehensive physical examination, applying special expertise in dysmorphology
 - 2.2.4. Formulate an appropriate differential diagnosis
 - 2.2.5. Select investigation strategies, demonstrating awareness of resource utilization
 - 2.2.6. Take the appropriate steps for syndrome identification, including the use of diagnostic aids, such as computer-assisted diagnosis and risk assessment tools
 - 2.2.7. Interpret the results of laboratory and imaging findings relevant to genetic disease, applying special expertise in cytogenetics, molecular genetics, genomics, and biochemical genetics
 - 2.2.8. Synthesize clinical, laboratory, and imaging findings to achieve or validate a diagnosis
- 2.3. Establish goals of care in collaboration with patients and their families which may include slowing disease progression, treating symptoms, achieving cure, improving function, and palliation
- 2.4. Establish a patient-centred management plan, for the following
 - 2.4.1. Pre-conception counselling
 - 2.4.2. Evaluation of a suspected genetic, genomic, or metabolic disorder
 - 2.4.3. Management of a genetic, genomic, or metabolic disorder
 - 2.4.4. Management of at-risk family members, including cascade testing

3. Plan and perform procedures and therapies for the purpose of assessment and/or management

- 3.1. Determine the most appropriate procedures or therapies
- 3.2. Obtain and document informed consent, explaining the risks and benefits of, and the rationale for, a proposed procedure or therapy

¹ Throughout this document, references to the patient's family are intended to include all those who are personally significant to the patient and are concerned with his or her care, including, according to the patient's circumstances, family members, partners, caregivers, legal guardians, and substitute decision-makers.

- 3.3. Prioritize procedures or therapies, taking into account clinical urgency and available resources
- 3.4. Perform procedures in a skilful and safe manner, adapting to unanticipated findings or changing clinical circumstances
 - 3.4.1. Skin biopsies

4. Establish plans for ongoing care and, when appropriate, timely consultation

- 4.1. Implement a patient-centred care plan that supports ongoing care, follow-up on investigations, response to treatment, and further consultation
 - 4.1.1. Provide continuity in care when indicated, and periodically assess the appropriateness of the care plan
 - 4.1.2. Assess and manage adherence to treatment
 - 4.1.3. Anticipate, recognize, and manage complications of therapy and/or the underlying condition of the patient
 - 4.1.4. Determine the need and timing of referral to other physicians and other health care professionals
 - 4.1.5. Incorporate appropriate recommendations from consultants and other health care professionals into diagnostic and treatment plans

5. Actively contribute, as an individual and as a member of a team providing care, to the continuous improvement of health care quality and patient safety

- 5.1. Recognize and respond to harm from health care delivery, including patient safety incidents
 - 5.1.1. Pre-analytic, analytic, and post-analytic errors
 - 5.1.1.1. Errors due to sample collection
 - 5.1.1.2. Errors due to process issues, including failed collection, and sorting and routing errors
 - 5.1.1.3. Contaminated tests
 - 5.1.1.4. False negatives and false positives of testing leading to incorrect diagnosis
 - 5.1.1.5. Errors in result tracking
 - 5.1.2. Medication errors
 - 5.1.3. Privacy breach
- 5.2. Adopt strategies that promote patient safety and address human and system factors

Communicator

Definition:

As *Communicators*, medical geneticists form relationships with patients and their families that facilitate the gathering and sharing of essential information for effective health care.

Key and Enabling Competencies: Medical geneticists are able to...

1. Establish professional therapeutic relationships with patients and their families

- 1.1. Communicate using a patient-centred approach that encourages patient and family trust and autonomy and is characterized by empathy, respect, and compassion
- 1.2. Optimize the physical or virtual environment for patient and family comfort, dignity, privacy, engagement, and safety
- 1.3. Recognize when the perspectives, values, or biases of patients, family members, physicians, or other health care professionals may have an impact on the quality of care, and modify the approach to the patient accordingly
 - 1.3.1. Recognize one's own biases, including ethno-cultural differences, and their impact on communication and patient care
 - 1.3.2. Identify how cultural background, age, gender, socioeconomic background, and spiritual values affect communication
- 1.4. Respond to a patient's or family member's non-verbal behaviours to enhance communication
- 1.5. Manage disagreements and emotionally charged conversations
 - 1.5.1. Recognize, explore, and address differences in perspective or goals of care when parents disagree with each other
- 1.6. Adapt to the unique needs and preferences of each patient and their family and to their clinical condition and circumstances
 - 1.6.1. Communicate effectively with patients when there are communication and language barriers
 - 1.6.1.1. Communicate effectively through the use of an interpreter
 - 1.6.2. Tailor communication to the patient's capacity and health literacy

2. Elicit and synthesize accurate and relevant information, incorporating the perspectives of patients and their families

- 2.1. Use patient-centred interviewing skills to effectively gather relevant biomedical and psychosocial information
- 2.2. Provide a clear structure for and manage the flow of an entire patient encounter
- 2.3. Seek and synthesize relevant information from other sources, including the patient's family, with the patient's consent

3. Share health care information and plans with patients and their families

3.1. Share information and explanations that are clear, accurate, and timely, while assessing for patient and family understanding

3.1.1. Use language and terminology that facilitates understanding and effective decision-making

3.1.2. Demonstrate empathy and compassion in the delivery of challenging or bad news

3.1.3. Convey the concept of uncertainty as it applies to genetic testing and outcomes

3.2. Disclose harmful patient safety incidents to patients and their families

4. Engage patients and their families in developing plans that reflect the patient's health care needs and goals

4.1. Facilitate discussions with patients and their families in a way that is respectful, non-judgmental, and culturally safe

4.1.1. Provide psychological support, including through referral when required

4.2. Assist patients and their families to identify, access, and make use of information and communication technologies to support their care and manage their health

4.3. Use communication skills and strategies that help patients and their families make informed decisions regarding their health

4.3.1. Help the patient and family choose a course of action, remaining non-directive but prepared to offer advice when appropriate

5. Document and share written and electronic information about the medical encounter to optimize clinical decision-making, patient safety, confidentiality, and privacy

5.1. Document clinical encounters in an accurate, complete, timely, and accessible manner, in compliance with regulatory and legal requirements

5.2. Communicate effectively using a written health record, electronic medical record, or other digital technology

5.2.1. Recognize and effectively address the challenges of providing care via telephone or digital technology

5.3. Share information with patients and others in a manner that enhances understanding and that respects patient privacy and confidentiality

Collaborator

Definition:

As *Collaborators*, medical geneticists work effectively with other health care professionals to provide safe, high-quality, patient-centred care.

Key and Enabling Competencies: Medical geneticists are able to...

1. Work effectively with physicians and other colleagues in the health care professions

- 1.1. Establish and maintain positive relationships with physicians and other colleagues in the health care professions to support relationship-centred collaborative care
 - 1.1.1. Receive feedback respectfully
- 1.2. Negotiate overlapping and shared responsibilities with physicians and other colleagues in the health care professions in episodic and ongoing care
 - 1.2.1. Apply knowledge of the roles and responsibilities of the professionals within the medical genetics health care team:
 - 1.2.1.1. Clinicians
 - 1.2.1.2. Genetic counsellors
 - 1.2.1.3. Laboratory-based geneticists
 - 1.2.1.4. Researchers
 - 1.2.1.5. Other health care professionals
 - 1.2.2. Work collaboratively with genetic counsellors to address patient issues and ensure appropriate medical care
- 1.3. Engage in respectful shared decision-making with physicians and other colleagues in the health care professions
 - 1.3.1. Provide advice to clinical colleagues regarding procurement, handling, and interpretation of genetic testing
 - 1.3.2. Convey information from the genetic consultation to other clinicians in a manner that enhances patient management
 - 1.3.2.1. Encourage discussion, questions, and interaction relevant to the case
 - 1.3.3. Contribute effectively at multidisciplinary rounds, by presenting and discussing findings
 - 1.3.4. Support clinical colleagues in the development and implementation of a management plan, when appropriate

2. Work with physicians and other colleagues in the health care professions to promote understanding, manage differences, and resolve conflicts

- 2.1. Show respect toward collaborators
- 2.2. Implement strategies to promote understanding, manage differences, and resolve conflict in a manner that supports a collaborative culture

3. Hand over the care of a patient to another health care professional to facilitate continuity of safe patient care

- 3.1. Determine when care should be transferred to another physician or health care professional
- 3.2. Demonstrate safe handover of care, using both oral and written communication, during a patient transition to a different health care professional, setting, or stage of care
 - 3.2.1. Provide anticipatory guidance for results of outstanding investigations and/or next steps for management

Leader

Definition:

As *Leaders*, medical geneticists engage with others to contribute to a vision of a high-quality health care system and take responsibility for the delivery of excellent patient care through their activities as clinicians, administrators, scholars, and teachers.

Key and Enabling Competencies: Medical geneticists are able to...

1. Contribute to the improvement of health care delivery in teams, organizations, and systems

- 1.1. Apply the science of quality improvement to systems of patient care
- 1.2. Contribute to a culture that promotes patient safety
- 1.3. Analyze patient safety incidents to enhance systems of care
 - 1.3.1. Participate in reviews of mortality, morbidity, and critical events
- 1.4. Use health informatics to improve the quality of patient care and optimize patient safety

2. Engage in the stewardship of health care resources

- 2.1. Allocate health care resources for optimal patient care
- 2.2. Apply evidence and management processes to achieve cost-appropriate care

3. Demonstrate leadership in health care systems

3.1. Demonstrate leadership skills to enhance health care

3.1.1. Assume responsibility for the organization and leadership of interprofessional teams, when appropriate

3.2. Facilitate change in health care to enhance services and outcomes

4. Manage career planning, finances, and health human resources in personal practice(s)

4.1. Set priorities and manage time to integrate practice and personal life

4.1.1. Develop strategies to achieve a balance between patient care, practice requirements, personal life, and career goals

4.2. Manage personal professional practice(s) and career

4.3. Implement processes to ensure personal practice improvement

Health Advocate

Definition:

As *Health Advocates*, medical geneticists contribute their expertise and influence as they work with communities or patient populations to improve health. They work with those they serve to determine and understand needs, speak on behalf of others when required, and support the mobilization of resources to effect change.

Key and Enabling Competencies: Medical geneticists are able to...

1. Respond to an individual patient's health needs by advocating with the patient and family within and beyond the clinical environment

1.1. Work with patients and families to address determinants of health that affect them and their access to needed health services or resources

1.1.1. Facilitate patient and family access to resources and services in the health and social systems

1.1.2. Work with patients and families, including those at risk for marginalization,² to ensure equity in access to and treatment within health care services

1.2. Work with patients and their families to increase opportunities to adopt healthy behaviours

1.2.1. Recommend patient and family education resources

1.3. Incorporate disease prevention, health promotion, and health surveillance into interactions with individual patients and families

² "Marginalized populations" are populations with decreased access to the social determinants of health. Examples include those excluded on the basis of race; ethnic or cultural origin; age; gender; sexuality; economic or housing status; and mental or physical illness and/or disability.

2. Respond to the needs of the communities or populations they serve by advocating with them for system-level change in a socially accountable manner

- 2.1. Work with a community or population to identify the determinants of health that affect them
- 2.2. Improve clinical practice by applying a process of continuous quality improvement to disease prevention, health promotion, and health surveillance activities
- 2.3. Contribute to a process to improve health in the community or population they serve
 - 2.3.1. Recognize and respond to situations where health advocacy is required, including improved access to testing and treatment for patients with a genetic disease
 - 2.3.2. Develop and apply health guidelines for anticipatory guidance for the care of patients with genetic disorders
 - 2.3.3. Advocate at the local, regional, and national level for patients with rare genetic disorders, including issues related to discrimination and access to treatment

Scholar

Definition:

As *Scholars*, medical geneticists demonstrate a lifelong commitment to excellence in practice through continuous learning, and by teaching others, evaluating evidence, and contributing to scholarship.

Key and Enabling Competencies: Medical geneticists are able to...

1. Engage in the continuous enhancement of their professional activities through ongoing learning

- 1.1. Develop, implement, monitor, and revise a personal learning plan to enhance professional practice
- 1.2. Identify opportunities for learning and improvement by regularly reflecting on and assessing their performance using various internal and external data sources
- 1.3. Engage in collaborative learning to continuously improve personal practice and contribute to collective improvements in practice

2. Teach students, residents, the public, and other health care professionals

- 2.1. Recognize the influence of role-modelling and the impact of the formal, informal, and hidden curriculum on learners
- 2.2. Promote a safe and respectful learning environment
- 2.3. Ensure patient safety is maintained when learners are involved

- 2.4. Plan and deliver learning activities
- 2.5. Provide feedback to enhance learning and performance
- 2.6. Assess and evaluate learners, teachers, and programs in an educationally appropriate manner

3. Integrate best available evidence into practice

- 3.1. Recognize practice uncertainty and knowledge gaps in clinical and other professional encounters and generate focused questions that can address them
- 3.2. Identify, select, and navigate pre-appraised resources
- 3.3. Critically evaluate the integrity, reliability, and applicability of health-related research and literature
 - 3.3.1. Develop strategies to identify and evaluate emerging developments in Medical Genetics and Genomics and related fields
- 3.4. Integrate evidence into decision-making in practice
 - 3.4.1. Incorporate the outcomes of trials testing therapeutic interventions into practice

4. Contribute to the creation and dissemination of knowledge and practices applicable to health

- 4.1. Demonstrate an understanding of the scientific principles of research and scholarly inquiry and the role of research evidence in health care
 - 4.1.1. Describe the roles of clinicians and research scientists in cooperatively advancing knowledge through research endeavours
- 4.2. Identify ethical principles for research and incorporate them into obtaining informed consent, considering potential harms and benefits and vulnerable populations
 - 4.2.1. Describe the role of a research ethics board
- 4.3. Contribute to the work of a research program
- 4.4. Pose questions amenable to scholarly investigation and select appropriate methods to address them
- 4.5. Summarize and communicate to professional and lay audiences, including patients and their families, the findings of relevant research and scholarly inquiry

Professional

Definition:

As *Professionals*, medical geneticists are committed to the health and well-being of individual patients and society through ethical practice, high personal standards of behaviour, accountability to the profession and society, physician-led regulation, and maintenance of personal health.

Key and Enabling Competencies: Medical geneticists are able to...

1. Demonstrate a commitment to patients by applying best practices and adhering to high ethical standards

- 1.1. Exhibit appropriate professional behaviours and relationships in all aspects of practice, demonstrating honesty, integrity, humility, commitment, compassion, respect, altruism, respect for diversity, and maintenance of confidentiality
 - 1.1.1. Recognize and respond appropriately to abuse, gender bias, discrimination, intimidation, and disrespect
 - 1.1.2. Recognize the limits of one's skill and expertise
- 1.2. Demonstrate a commitment to excellence in all aspects of practice
- 1.3. Recognize and respond to ethical issues encountered in practice
- 1.4. Recognize and manage conflicts of interest
- 1.5. Exhibit professional behaviours in the use of technology-enabled communication

2. Demonstrate a commitment to society by recognizing and responding to societal expectations in health care

- 2.1. Demonstrate accountability to patients, society, and the profession by responding to societal expectations of physicians
- 2.2. Demonstrate a commitment to patient safety and quality improvement

3. Demonstrate a commitment to the profession by adhering to standards and participating in physician-led regulation

- 3.1. Fulfil and adhere to professional and ethical codes, standards of practice, and laws governing practice
 - 3.1.1. Adhere to the laws and regulations related to Medical Genetics and Genomics, including reproductive options and technology
 - 3.1.2. Apply laws governing decision-making, including the identification and role of substitute decision-makers
 - 3.1.3. Apply knowledge of the legal and professional requirements relating to assent and informed consent by children and mature minors
 - 3.1.4. Adhere to requirements for mandatory reporting

3.2. Recognize and respond to unprofessional and unethical behaviours in physicians and other colleagues in the health care professions

3.3. Participate in peer assessment and standard setting

4. Demonstrate a commitment to physician health and well-being to foster optimal patient care

4.1. Exhibit self-awareness and manage influences on personal well-being and professional performance

4.2. Manage personal and professional demands for a sustainable practice throughout the physician life cycle

4.3. Promote a culture that recognizes, supports, and responds effectively to colleagues in need

This document is to be reviewed by the Specialty Committee in Medical Genetics and Genomics by January 31, 2024.

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