

2009

**EDITORIAL REVISION – MAY 2016
VERSION 1.2**

This document applies to those who begin training on or after July 1st, 2010.

DEFINITION

Medical Genetics and Genomics is that branch of medicine concerned with the effect of genetic variation on human development and health and also with the study, diagnosis, management, and prevention of genetic and related disorders in individuals, families, and communities.

GOALS

The practice of Medical Genetics and Genomics is based on an in-depth knowledge of basic genetic principles, knowledge of genetic disease as it affects all body systems and individuals of all ages, and a clear understanding of the principles of genetic counselling.

Upon completion of training, a resident is expected to be a competent specialist in Medical Genetics and Genomics capable of assuming a consultant's role in the specialty. The resident must acquire a working knowledge of the theoretical basis of the specialty, including its foundations in the basic medical sciences and research.

Residents must demonstrate the requisite knowledge, skills, and attitudes for effective patient-centred care and service to a diverse population. In all aspects of specialist practice, the graduate must be able to address ethical issues and issues of gender, sexual orientation, age, culture, and ethnicity in a professional manner.

MEDICAL GENETICS AND GENOMICS COMPETENCIES

At the completion of training, the resident will have acquired the following competencies and will function effectively as a:

Medical Expert

Definition:

As *Medical Experts*, Medical Genetics and Genomics specialists integrate all of the CanMEDS Roles, applying medical knowledge, clinical skills, and professional attitudes in their provision of patient-centred care. *Medical Expert* is the central physician Role in the CanMEDS framework.

Key and Enabling Competencies: Medical Genetics and Genomics specialists are able to...

1. Function effectively as consultants, integrating all of the CanMEDS Roles to provide optimal, ethical and patient-centred medical care

- 1.1. Perform a consultation effectively, including the presentation of well-documented assessments and recommendations in written and/or oral form in response to a request from another health care professional
- 1.2. Demonstrate use of all CanMEDS competencies relevant to Medical Genetics and Genomics
- 1.3. Identify and appropriately respond to relevant ethical issues arising in the care of patients, families, and populations
- 1.4. Demonstrate the ability to prioritize professional duties when faced with multiple patients and problems
- 1.5. Demonstrate compassionate patient and family-centred care
- 1.6. Recognize and respond to the ethical dimensions in medical decision-making
- 1.7. Demonstrate medical expertise in situations other than patient care, such as providing expert legal testimony or advising governments, as needed

2. Establish and maintain clinical knowledge, skills and attitudes appropriate to Medical Genetics and Genomics

- 2.1. Apply knowledge of the clinical, socio-behavioural, and fundamental biomedical sciences relevant to Medical Genetics and Genomics
 - 2.1.1. General structure of the human genome
 - 2.1.2. Information that can be obtained from an integrated assessment of genomic function at the ribonucleic acid (RNA) or protein level that cannot be obtained from the deoxyribonucleic acid (DNA) sequence alone
 - 2.1.3. Processes underlying genomic variability, redundancy, and plasticity
 - 2.1.4. Factors influencing gene expression
 - 2.1.5. Pre- and post-translational factors influencing protein function
 - 2.1.6. Normal and abnormal gene structure and function
 - 2.1.7. Normal and abnormal cell division
 - 2.1.8. Chromosome structure, morphology and nomenclature

OBJECTIVES OF TRAINING IN MEDICAL GENETICS AND GENOMICS (2009)

- 2.1.9. Principles and application of cytogenetic techniques
- 2.1.10. Principles and application of molecular genetic techniques
- 2.1.11. Principles of genomics, including the scope of genomic variability
- 2.1.12. Principles and application of techniques for genomic analysis
- 2.1.13. Basic principles of biochemistry applicable to Medical Genetics and Genomics
- 2.1.14. Principles and application of laboratory investigation relevant to inborn errors of metabolism
- 2.1.15. Monogenic and complex inheritance
- 2.1.16. Developmental biology as it relates to normal and abnormal human morphogenesis
- 2.1.17. Principles of epidemiology, including biostatistics, genetic epidemiology, and population genetics
- 2.1.18. Indications, limitations, and risks of techniques of pre-implantation and prenatal assessment, and options for reproductive intervention
- 2.1.19. Genetic and non-genetic (intrinsic and extrinsic) factors predisposing to fetal loss, infertility, and abnormalities of morphogenesis
- 2.1.20. Teratogenic agents and their effects
- 2.1.21. Phenotypic variation and specific methods of assessment
- 2.1.22. Principles underlying genomics enabling individualized medical care, including but not limited to pharmacogenetics
- 2.1.23. Methods of syndrome identification and diagnosis, including but not limited to the use of computer diagnostic aids
- 2.1.24. Clinical features, etiology, diagnosis, management, natural history, and prognosis of common conditions seen in a medical genetics service, including but not limited to the following:
 - 2.1.24.1. Syndromes with well-defined dysmorphology
 - 2.1.24.2. Common malformations, deformations, disruptions, sequences and associations
 - 2.1.24.3. Cancer syndromes
 - 2.1.24.4. Neurogenetic conditions
 - 2.1.24.5. Cardiac genetic conditions
 - 2.1.24.6. Inborn errors of metabolism
 - 2.1.24.7. Skeletal dysplasias
 - 2.1.24.8. Other single gene disorders
 - 2.1.24.9. Imprinting disorders
 - 2.1.24.10. Chromosomal syndromes

- 2.1.24.11. Genomic disorders characterized by recurrent microdeletions and microduplications
- 2.1.25. Approach to the evaluation of patients commonly referred to Medical Genetics and Genomics with conditions that may have a significant genetic etiology, including but not limited to intellectual disability, autism, deafness, and short or tall stature
- 2.1.26. Indicators of normal and abnormal psychomotor development, including the use and limitations of commonly used instruments for the assessment of behaviour and intelligence
- 2.1.27. Principles and approaches of population screening
- 2.1.28. Principles of genetic testing for the diagnosis of disease and predictive testing to assess risk for predisposition to monogenic or complex genetic diseases
- 2.2. Describe the CanMEDS framework of competencies relevant to Medical Genetics and Genomics
- 2.3. Apply lifelong learning skills of the Scholar Role to implement a personal program to keep up-to-date and enhance areas of professional competence
- 2.4. Contribute to the enhancement of quality care and patient safety in Medical Genetics and Genomics, integrating the available best evidence and best practices

3. Perform a complete and appropriate assessment of a patient

- 3.1. Identify and explore issues to be addressed in a patient encounter, including the patient's context and preferences
- 3.2. Elicit a history that is relevant, concise, and accurate to context and preferences, for the purposes of prevention and health promotion, diagnosis and management
 - 3.2.1. Elicit a comprehensive medical and family history
 - 3.2.2. Construct and interpret a standardized pedigree
- 3.3. Perform a physical examination that is relevant and accurate for the purposes of prevention and health promotion, diagnosis and/or management of genetic and related conditions
 - 3.3.1. Carry out a comprehensive physical examination, applying special expertise in phenotypic variation (dysmorphology)
- 3.4. Select medically appropriate investigative methods in a resource-effective and ethical manner
- 3.5. Demonstrate effective clinical problem solving and judgment to address patient problems, including interpreting available data and integrating information to generate differential diagnoses and management plans
 - 3.5.1. Formulate an appropriate differential diagnosis and plan an appropriate

course of investigation in a resource-effective and ethical manner

- 3.5.2. Take the appropriate steps for syndrome identification, including the use of diagnostic aids, including but not limited to computer assisted diagnosis
- 3.5.3. Interpret laboratory and imaging findings relevant to genetic disease, applying special expertise in cytogenetics, molecular genetics, genomics, and biochemical genetics
- 3.5.4. Synthesize clinical, laboratory, and imaging data to achieve or validate a diagnosis

4. Use preventive and therapeutic interventions effectively

- 4.1. Implement a management plan in collaboration with a patient and the patient's family
- 4.2. Demonstrate appropriate and timely application of primary, secondary, and tertiary preventive interventions relevant to Medical Genetics and Genomics
- 4.3. Demonstrate appropriate and timely application of therapeutic interventions relevant to Medical Genetics and Genomics, including metabolic diseases
- 4.4. Ensure appropriate informed consent is obtained for investigations and therapies
- 4.5. Ensure patients and their families receive appropriate, compassionate, and ethical end-of-life care

5. Demonstrate proficient and appropriate use of procedural skills

- 5.1. Demonstrate effective, appropriate, and timely performance of diagnostic procedures relevant to Medical Genetics and Genomics, including but not limited to:
 - 5.1.1. Skin biopsies
- 5.2. Ensure informed consent is obtained for procedures, investigations, and therapies
- 5.3. Document and disseminate information related to procedures performed and their outcomes
- 5.4. Ensure adequate follow-up is arranged for procedures performed

6. Seek appropriate consultation from other health professionals, recognizing the limits of their own expertise

- 6.1. Demonstrate insight into their own limits of expertise
- 6.2. Demonstrate effective, appropriate, and timely consultation of another health professional as needed for optimal patient care
- 6.3. Arrange appropriate follow-up care services for patients and their families
 - 6.3.1. Provide continuity in care when indicated, and periodically assess the appropriateness of the care plan

Communicator

Definition:

As *Communicators*, Medical Genetics and Genomics specialists effectively facilitate the doctor-patient relationship and the dynamic exchanges that occur before, during, and after the medical encounter.

Key and Enabling Competencies: Medical Genetics and Genomics specialists are able to...

1. Develop rapport, trust, and ethical therapeutic relationships with patients and families

- 1.1. Recognize that being a good communicator is a core clinical skill for physicians, and that effective physician-patient communication can foster patient satisfaction, physician satisfaction, adherence and improved clinical outcomes
- 1.2. Establish positive therapeutic relationships with patients and their families that are characterized by understanding, trust, respect, honesty and empathy
- 1.3. Respect patient confidentiality, privacy and autonomy
- 1.4. Listen effectively
- 1.5. Be aware of and responsive to nonverbal cues
- 1.6. Demonstrate effective use of non-verbal communication
- 1.7. Conduct a structured clinical encounter effectively

2. Accurately elicit and synthesize relevant information and perspectives of patients and families, colleagues, and other professionals

- 2.1. Gather information about a patient's concerns, expectations, and illness experience, while considering the influence of the patient's age, gender, spiritual values, and ethnic, cultural and socioeconomic background
- 2.2. Seek out and synthesize relevant information from other sources, such as a patient's family, caregivers, and other professionals

3. Convey relevant information and explanations accurately to patients and families, colleagues, and other professionals

- 3.1. Deliver information to a patient and family, colleagues, and other professionals in a humane and understandable manner that encourages discussion and participation in decision-making
 - 3.1.1. Provide genetic counselling with empathy and compassion, especially when delivering bad news
 - 3.1.2. Remain objective and impartial
 - 3.1.3. Help the individual and family choose a course of action, remaining non-directive but prepared to offer advice when appropriate

- 3.1.4. Provide psychological support, either personally or through referral
- 3.1.5. Communicate information concerning the medical diagnosis and prognosis, the applicable risks, and the management options available, at a level appropriate to the referring provider
- 3.1.6. Communicate information regarding results of genomic testing, including incidental findings and findings of unknown significance, as appropriate

4. Develop a common understanding on issues, problems and plans with patients, families, and other professionals to develop a shared plan of care

- 4.1. Identify and explore problems to be addressed effectively, including the patient's context, responses, expectations, concerns, and preferences
- 4.2. Respect diversity and difference, including but not limited to the impact of gender, religion, and cultural beliefs on decision-making
 - 4.2.1. Recognize one's own biases, including ethno-cultural differences, and their impact on communication and patient care
 - 4.2.2. Identify how cultural background, age, gender, socioeconomic background and spiritual values affect communication
 - 4.2.3. Recognize and effectively address the challenges of a language barrier
 - 4.2.3.1. Communicate effectively through the use of an interpreter
- 4.3. Encourage discussion, questions, and interaction in the encounter
- 4.4. Engage patients, families, and relevant health professionals in shared decision-making to develop a plan of care
- 4.5. Address challenging communication issues effectively, such as obtaining informed consent, delivering bad news, and addressing anger, confusion and misunderstanding

5. Convey effective oral and written information about a medical encounter

- 5.1. Maintain clear, accurate, and appropriate records of clinical encounters and plans
- 5.2. Present oral reports of clinical encounters and plans
- 5.3. Summarize findings, consultation notes and counselling, for referring physicians, agencies, and families
- 5.4. Present medical information to the public or media about a medical issue

Collaborator

Definition:

As *Collaborators*, Medical Genetics and Genomics specialists effectively work within a health care team to achieve optimal patient care.

Key and Enabling Competencies: Medical Genetics and Genomics specialists are able to...

1. Participate effectively and appropriately in an interprofessional health care team

- 1.1. Describe the Medical Genetics and Genomics specialist's roles and responsibilities to other professionals
- 1.2. Describe the roles and responsibilities of the following professionals within the medical genetics health care team in cooperatively working towards solving questions directly related to patient diagnosis and management:
 - 1.2.1. Clinicians
 - 1.2.2. Genetic counselors
 - 1.2.3. Laboratory based clinicians
 - 1.2.4. Laboratory based researchers
 - 1.2.5. Other health care professionals
- 1.3. Recognize and respect the diversity of roles, responsibilities, and competences of other professionals in relation to their own
- 1.4. Work with others to assess, plan, provide, and integrate care for individuals and groups of patients
 - 1.4.1. Plan and coordinate care in collaboration with patients and their families
- 1.5. Collaborate to carry out academic, educational and administrative activities
- 1.6. Demonstrate personal and professional attitudes consistent with a consultant physician role
- 1.7. Identify the factors influencing team dynamics
- 1.8. Respect team ethics, including confidentiality, resource allocation, and professionalism
- 1.9. Demonstrate leadership in a health care team, as appropriate

2. Work with other health professionals effectively to prevent, negotiate, and resolve interprofessional conflict

- 2.1. Demonstrate a respectful attitude towards colleagues and members of an interprofessional team
- 2.2. Work with other professionals to prevent conflicts

- 2.3. Employ collaborative negotiation to resolve conflicts
- 2.4. Respect differences and address misunderstandings and limitations in other professionals
- 2.5. Recognize one's own differences, misunderstanding and limitations that may contribute to interprofessional tension
- 2.6. Reflect on interprofessional team function

Manager

Definition:

As *Managers*, Medical Genetics and Genomics specialists are integral participants in health care organizations, organizing sustainable practices, making decisions about allocating resources, and contributing to the effectiveness of the health care system.

Key and Enabling Competencies: Medical Genetics and Genomics specialists are able to...

1. Participate in activities that contribute to the effectiveness of their health care organizations and systems

- 1.1. Work collaboratively with others in their organizations
- 1.2. Apply the principles of quality assurance to clinical care, laboratory data, and education
- 1.3. Demonstrate knowledge of planning, evaluation, and assessment of outcomes of a health care program
- 1.4. Describe the structure and function of the health care system as it relates to Medical Genetics and Genomics, including the roles of physicians
- 1.5. Describe principles of health care financing, including physician remuneration, budgeting and organizational funding

2. Manage their practice and career effectively

- 2.1. Set priorities and manage time to balance patient care, practice requirements, outside activities, and personal life
- 2.2. Identify factors associated with time management:
 - 2.2.1. Recognize that the use of time requires punctuality, planning, efficiency, and establishment of routines for carrying out regular activities
- 2.3. Describe funding models operating in the delivery of medical genetics services
- 2.4. Describe the principles and application of practice management, including finances and human resources
- 2.5. Implement processes to ensure personal practice improvement

- 2.6. Employ information technology and medical databases appropriately for patient care
- 2.7. Demonstrate commitment to the protection of medical records

3. Allocate finite health care resources appropriately

- 3.1. Recognize the importance of just allocation of health care resources, balancing effectiveness, efficiency, and access with optimal patient care
- 3.2. Apply evidence and management processes for cost-appropriate care
- 3.3. Recognize the issues arising from emerging technologies in the context of a publically funded health care system including but not limited to direct to consumer testing and self-pay options

4. Serve in administration and leadership roles, as appropriate

- 4.1. Chair or participate effectively in committees and meetings
- 4.2. Lead or implement change in health care
- 4.3. Plan relevant elements of health care delivery (e.g., work schedules)
 - 4.3.1. Demonstrate knowledge of employment policies and procedures
 - 4.3.2. Demonstrate the ability to effectively coordinate the work of the health care team

Health Advocate

Definition:

As *Health Advocates*, Medical Genetics and Genomics specialists responsibly use their expertise and influence to advance the health and well-being of individual patients, communities, and populations.

Key and Enabling Competencies: Medical Genetics and Genomics specialists are able to...

1. Respond to individual patient health needs and issues as part of patient care

- 1.1. Identify the health and support needs of individual patients and families
- 1.2. Recognize the potential benefits to patients and families of involvement with support organizations for genetic disorders
- 1.3. Refer patients to community, national, and international resources, as appropriate
- 1.4. Identify opportunities for health promotion and disease prevention pertaining to patients and families

- 1.5. Demonstrate understanding of the importance of advocating on the individual's behalf for access to resources to prevent, diagnose, and treat genetic and related conditions

2. Respond to the health needs of the communities they serve

- 2.1. Identify the characteristics of the communities they serve
- 2.2. Identify opportunities for advocacy, health promotion and disease prevention in the communities they serve, and respond appropriately
- 2.3. Appreciate the possibility of competing interests of different groups within the community served
- 2.4. Appreciate the possibility of competing interests between the community served and other populations

3. Identify the determinants of health for the population they serve

- 3.1. Identify the determinants of health of the communities within the population, including barriers to access to care and resources
- 3.2. Identify vulnerable or marginalized groups within those served and respond appropriately

4. Promote the health of individual patients, communities, and population

- 4.1. Describe an approach to implementing a change in a determinant of health of the population they serve
- 4.2. Describe the importance of participating actively in public policy regarding the application and implementation of strategies for the prevention, diagnosis, and treatment of genetic and related conditions
- 4.3. Identify points of influence in the health care system and its structure
- 4.4. Describe the roles of national and international organizations in the promotion of genetic health and the prevention, detection, and treatment of genetic and related conditions
- 4.5. Describe the ethical and professional issues inherent in health advocacy, including altruism, social justice, autonomy, integrity, and idealism
- 4.6. Appreciate the possibility of conflict between the role of health advocate for a patient or community with that of manager or gatekeeper
- 4.7. Describe the role of the medical profession in advocating collectively for health and patient safety

Scholar

Definition:

As *Scholars*, Medical Genetics and Genomics specialists demonstrate a lifelong commitment to reflective learning, as well as the creation, dissemination, application, and translation of medical knowledge.

Key and Enabling Competencies: Medical Genetics and Genomics specialists are able to...

1. Maintain and enhance professional activities through ongoing learning

- 1.1. Describe the principles of maintenance of competence
- 1.2. Describe the principles and strategies for implementing a personal knowledge management system
- 1.3. Recognize and reflect on learning issues in practice
- 1.4. Conduct a personal practice audit
- 1.5. Pose an appropriate learning question
- 1.6. Access and interpret the relevant evidence
- 1.7. Integrate new learning into practice
- 1.8. Evaluate the impact of any change in practice
- 1.9. Document the learning process

2. Critically evaluate medical information and its sources, and apply this appropriately to practice decisions

- 2.1. Describe the principles of critical appraisal
- 2.2. Critically appraise retrieved evidence in order to address a clinical question
- 2.3. Integrate critical appraisal conclusions into clinical care

3. Facilitate the learning of patients, families, students, residents, other health professionals, the public, and others

- 3.1. Describe principles of learning relevant to medical education
- 3.2. Identify collaboratively the learning needs and desired learning outcomes of others
- 3.3. Select effective teaching strategies and content to facilitate others' learning
- 3.4. Deliver an effective lecture or presentation
- 3.5. Assess and reflect on a teaching encounter
- 3.6. Provide effective feedback
- 3.7. Describe the principles of ethics with respect to teaching

4. Contribute to the development, dissemination, and translation of new knowledge and practices

- 4.1. Describe the principles of research and scholarly inquiry
 - 4.1.1. Describe the roles of clinicians and research scientists in cooperatively advancing knowledge through genetics research endeavours
- 4.2. Describe the principles of research ethics
 - 4.2.1. Describe the role of a research ethics board
- 4.3. Participate in clinical or basic science studies as a member of a research team
- 4.4. Pose a scholarly question
- 4.5. Conduct a systematic search for evidence
- 4.6. Identify strategies for obtaining research funding, including identifying appropriate funding sources and preparing a grant application
- 4.7. Select and apply appropriate methods to address the question
- 4.8. Disseminate the findings of a study
- 4.9. Describe the scientific rationale and ethical considerations for studying additional family members (affected and unaffected) and members of the larger community in research studies
- 4.10. Complete a scholarly project relevant to Medical Genetics and Genomics suitable for submission for peer-reviewed publication

Professional

Definition:

As *Professionals*, Medical Genetics and Genomics specialists are committed to the health and well-being of individuals and society through ethical practice, profession-led regulation, and high personal standards of behaviour.

Key and Enabling Competencies: Medical Genetics and Genomics specialists are able to...

1. Demonstrate a commitment to their patients, profession, and society through ethical practice

- 1.1. Exhibit appropriate professional behaviors in practice, including honesty, integrity, commitment, compassion, respect, and altruism
- 1.2. Demonstrate a commitment to delivering the highest quality care and maintenance of competence
 - 1.2.1. Recognize the limitations of their skills and expertise and seek consultation whenever indicated

OBJECTIVES OF TRAINING IN MEDICAL GENETICS AND GENOMICS (2009)

- 1.2.2. Accept feedback respectfully
 - 1.3. Recognize and appropriately respond to ethical issues encountered in practice
 - 1.3.1. Recognize and respond appropriately to abuse, gender bias, discrimination, intimidation, and disrespect
 - 1.4. Manage conflicts of interest
 - 1.5. Recognize the principles and limits of patient confidentiality as defined by professional practice standards and the law
 - 1.5.1. Describe the challenges imposed by patient confidentiality in the rare instances in which relatives are at risk for a serious and potentially preventable disease
 - 1.6. Recognize the laws and regulations related to autonomy, disclosure, privacy, discrimination and issues of patient competence
 - 1.7. Maintain appropriate boundaries with patients
- 2. Demonstrate a commitment to their patients, profession, and society through participation in profession-led regulation**
- 2.1. Demonstrate knowledge and understanding of the professional, legal, and ethical codes of practice
 - 2.1.1. Describe the laws and regulations related to Medical Genetics and Genomics, including reproductive options and technology
 - 2.1.2. Identify factors associated with litigation in the practice of Medical Genetics and Genomics
 - 2.2. Fulfil the regulatory and legal obligations required of current practice
 - 2.3. Demonstrate accountability to professional regulatory bodies
 - 2.4. Recognize and respond to others' unprofessional behaviours in practice
 - 2.5. Participate in peer review
- 3. Demonstrate a commitment to physician health and sustainable practice**
- 3.1. Balance personal and professional priorities to ensure personal health and a sustainable practice
 - 3.2. Strive to heighten personal and professional awareness and insight
 - 3.3. Recognize other professionals in need and respond appropriately

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