

This document is to be used in conjunction with the *Entrustable Professional Activity User Guide*, which is available on the Royal College's website.

*This document applies to those who begin training on or after July 1, 2022.*

## **Medical Genetics and Genomics: Transition to Discipline EPA #1**

### **Gathering and summarizing a history and physical examination**

#### Key Features:

- This EPA focuses on verifying clinical assessment skills obtained in medical school.
- This includes gathering a complete history relevant to a genetic evaluation, including a prenatal, basic developmental and family history, drawing a pedigree, and demonstrating an approach to a dysmorphism exam.
- It also includes formulating a basic differential diagnosis and summarizing the findings for oral presentation to a supervisor.
- This EPA does not include making management decisions or communicating a management plan with the patient and/or family.

#### Assessment Plan:

Direct observation, case presentation, and documentation review by medical geneticist

Use form 1. Form collects information on:

- Patient age: under 6 months; under 10; adult
- Setting: inpatient; outpatient
- Activity observed (select all that apply): history; physical
- Presentation (write in):

Collect 3 observations of achievement.

- At least 1 observation of a complete history and physical
- At least 1 physical exam for each age group
- A variety of clinical presentations
- At least 2 different observers

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Relevant CanMEDS milestones:

- 1 ME 2.1 Ascertain the patient's/family's understanding of the reason for the visit**
- 2 COM 1.1 Communicate in a manner that encourages trust, and is characterized by empathy, respect and compassion**
- 3 ME 2.2 Elicit an appropriate history, including prenatal, birth, developmental and medical history, and a three-generation family history**
- 4 ME 2.2 Construct a standardized pedigree**
- 5 ME 2.2 Perform a physical exam, applying an approach to the main elements of a dysmorphology exam**
- 6 ME 2.2 Develop a basic differential diagnosis relevant to the patient's presentation**
- 7 ME 2.2 Synthesize and organize clinical information for clear and succinct presentation to a supervisor**

## **Medical Genetics and Genomics: Transition to Discipline EPA #2**

### **Selecting first line genetic testing**

#### Key Features:

- This EPA focuses on appropriate selection of first line genetic testing, demonstrating resource stewardship.
- It also includes basic pre-test counselling.

#### Assessment Plan:

Direct observation by medical geneticist or Core or TTP resident

Use form 1. Form collects information on:

- Type of genetic testing: cytogenetic; molecular; other

Collect 2 observations of achievement.

- At least 1 cytogenetic
- At least 1 molecular
- At least 2 different observers

#### Relevant CanMEDS milestones:

- 1 ME 1.3 Apply knowledge of the principles and broad categories of genetic testing (i.e., single gene vs chromosome-level testing)**
- 2 ME 2.2 Select the appropriate investigation**
- 3 ME 3.1 Describe the advantages and limitations of genetic testing**
- 4 COM 3.1 Use plain language and avoid medical jargon**
- 5 COM 3.1 Use strategies to verify and validate the patient's and/or family's understanding**
- 6 COM 4.3 Answer questions from the patient and/or family**

## **Medical Genetics and Genomics: Transition to Discipline EPA #3**

### **Interpreting the results of first line genetic testing**

#### Key Features:

- This EPA focuses on interpreting broad categories of results (e.g., pathogenic, benign, variants of uncertain significance) and their implications.
- It does not include verifying lab interpretation.

#### Assessment Plan:

Direct observation by medical geneticist or Core or TTP resident

Use form 1. Form collects information on:

- Type of genetic testing: cytogenetic; molecular; other
- Result: benign; pathogenic; variant of uncertain significance

Collect 2 observations of achievement.

- At least 2 different observers

#### Relevant CanMEDS Milestones:

- 1 ME 1.3 Apply knowledge of genetic testing for diagnosis and for assessment of risk for genetic conditions**
- 2 ME 2.2 Categorize results of first line genetic testing as benign, pathogenic or variants of uncertain significance**
- 3 ME 2.2 Describe the direct implications of the results of first line genetic testing**
- 4 S 3.4 Integrate best evidence into the interpretation of the results of first line genetic testing**

## **Medical Genetics and Genomics: Foundations EPA #1**

### **Recognizing the acuity of a patient's illness, initiating stabilization and management, and consulting as needed**

#### Key Features:

- This EPA focuses on recognizing that a patient is, or is likely to become, critically ill, and responding appropriately.
- An important aspect of this EPA is the timely and appropriate recognition of the need for assistance and/or definitive management.
- This includes prioritizing actions, facilitating the primary priorities of management, and providing targeted treatment.
- The observation of this EPA must include pediatric and adult patients.
- This EPA may be observed in a simulation setting.

#### Assessment Plan:

Direct and indirect observation by supervisor (most responsible physician, other consulting staff, or Core or TTP resident)

Use form 1. Form collects information on:

- Patient demographic: neonate; pediatric; adult
- Setting: clinical; simulation

Collect 2 observations of achievement.

- At least 1 pediatric or neonatal patient
- At least 1 adult patient
- At least 1 observation in the clinical setting

#### Relevant CanMEDS milestones:

- 1 ME 2.1 Determine the acuity of the issue and the priorities for patient care**
- 2 P 1.1 Work within personal limits, asking for assistance as needed**
- 3 ME 2.2 Provide assessment and initial stabilization of ABCs**
- 4 ME 2.2 Perform a history and physical exam relevant to the patient presentation, in a time-effective manner**
- 5 ME 2.4 Develop and implement a plan for initial management**
- 6 COL 1.3 Work effectively with other members of the health care team**
- 7 P 4.1 Maintain capacity for professional clinical performance in stressful situations**

## Medical Genetics and Genomics: Foundations EPA #2

### Assessing and initiating management for patients with a common medical presentation

#### Key Features:

- This EPA includes performing a history and physical exam, selecting investigations and interpreting the results, and developing a differential diagnosis for patients with common, non-complex presentations.
- It also includes the development of an initial treatment or management plan.
- The observation of this EPA must include patients in maternal-fetal medicine, neonatal, pediatric, and adult demographics, and a range of clinical conditions and clinical settings.

#### Assessment Plan:

Direct or indirect observation (e.g., case discussion, review of consult letter or other documents) by supervisor (staff and/or Core or TTP resident)

Use form 1. Form collects information on:

- Patient demographic: maternal-fetal medicine; neonate; pediatric; adult
- Setting: inpatient; outpatient
- Type of observation: direct; indirect
- Category of main condition (select all that apply): allergy/immunology; cardiac; dermatologic; developmental/behavioural/psychosocial; endocrine; gastrointestinal; hematology/oncology; infectious disease; mental health; neurology; otolaryngology/ophthalmology; renal/genitourinary; respiratory; rheumatic/musculoskeletal; other (write in)

Collect 12 observations of achievement.

- At least 5 pediatric patients
- At least 5 adult patients
- At least 2 maternal-fetal medicine patients
- At least 1 direct observation by staff for each of the maternal-fetal medicine, pediatric, and adult populations

#### Relevant CanMEDS milestones:

- 1 ME 2.2 Elicit a history relevant to the patient presentation**
- 2 ME 2.2 Perform a physical examination**
- 3 ME 2.2 Generate a differential diagnosis**
- 4 ME 2.2 Select and interpret investigations**
- 5 ME 2.2 Synthesize and interpret information from the clinical assessment**
- 6 ME 2.4 Develop and implement a plan for initial management**
- 7 ME 1.7 Seek assistance as needed**

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- 8 COM 5.1 Document the clinical encounter to convey the clinical assessment and the rationale for decisions and/or recommendations**

### Medical Genetics and Genomics: Foundations EPA #3

#### Providing ongoing care for patients with common medical problems, and advancing their care plans

##### Key Features:

- This EPA focuses on the ongoing care of patients with common, non-complex conditions.
- It includes ongoing reassessment of clinical status and management of the evolving clinical course including further investigations, response to treatment and possible complications.
- The observation of this EPA includes pediatric and adult patients, and a range of clinical conditions.

##### Assessment Plan:

Direct or indirect observation by supervising staff or Core, TTP or subspecialty resident

Use form 1. Form collects information on:

- Patient demographic: neonate; pediatric; adult
- Setting: inpatient; outpatient
- Type of observation: direct; indirect
- Category of main condition (select all that apply): allergy/immunology; cardiac; dermatologic; developmental/behavioural/psychosocial; endocrine; gastrointestinal; hematology/oncology; infectious disease; mental health; neurology; otolaryngology/ophthalmology; renal/genitourinary; respiratory; rheumatic/musculoskeletal; other (write in)

Collect 10 observations of achievement.

- At least 1 neonate
- At least 4 pediatric patients
- At least 5 adult patients
- At least 2 observations by staff for each of the pediatric and adult populations
- At least 1 direct observation by staff for each of the pediatric and adult populations

##### Relevant CanMEDS milestones:

- 1 ME 2.2 Gather clinical information to assess health status and symptoms**
- 2 ME 2.2 Select and interpret investigations**
- 3 ME 2.2 Assess patients for complications, response to therapy, and evolution of the clinical course**
- 4 ME 2.4 Adapt management plans to the clinical course**
- 5 ME 1.7 Seek assistance as needed**
- 6 COL 1.3 Integrate the patient's perspective and context into the care plan**



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- 7 HA 1.2 Work with the patient and/or family to increase their understanding of their illness and health care needs**
- 8 COM 5.1 Document the clinical encounter to convey the clinical assessment and the rationale for decisions and/or recommendations**

## **Medical Genetics and Genomics: Foundations EPA #4**

### **Consulting specialists and other health professionals, synthesizing recommendations, and integrating these into the care plan**

#### Key Features:

- This EPA focuses on appropriate utilization, communication, and collaboration with other health care professionals.
- It includes recognizing the need for consultation, developing a well-defined question for the consultant, and incorporating recommendations into the care plan.
- It may include coordinating the involvement of multiple consultants and other health care professionals but does not include balancing differing recommendations or identifying specialty consultation needs for complex presentations.

#### Assessment Plan:

Direct or indirect observation by supervisor (staff and/or supervising resident) or individual being consulted (physician or other health professional)

Use form 1. Form collects information on:

- Patient demographic: neonate; pediatric; adult
- Setting: inpatient; outpatient; emergency department

Collect 2 observations of achievement.

- At least 1 neonate or pediatric patient
- At least 1 adult patient

#### Relevant CanMEDS milestones

- 1 ME 4.1 Develop and prioritize well defined questions to be addressed with a consultant or other health care professional**
- 2 COL 1.2 Apply knowledge of the roles and scopes of practice of other health care professionals**
- 3 COL 1.3 Communicate effectively with physicians and other health care professionals**
- 4 ME 2.4 Incorporate consultant recommendations into the diagnostic or treatment plan**
- 5 ME 4.1 Coordinate investigation, treatment, and follow-up plans when multiple physicians and healthcare professionals are involved**
- 6 ME 4.1 Ensure follow-up on the results of consultation requests and/or recommendations**

## **Medical Genetics and Genomics: Foundations EPA #5**

### **Communicating clinical findings, investigations, and management plans to patients and/or families**

#### Key Features:

- This EPA focuses on the application of communication skills and strategies to convey information and engage the patient and family in shared decision making.
- This EPA includes situations commonly presenting in Pediatrics and Internal Medicine.

#### Assessment Plan:

Direct observation by supervising staff or Core, TTP or subspecialty resident

Use form 1. Form collects information on:

- Patient demographic: maternal-fetal medicine; neonate; pediatric; adult
- Setting: inpatient; outpatient
- Topic discussed (write in):

Collect 5 observations achievement.

- At least 1 adult patient
- At least 1 pediatric patient
- At least 2 inpatients
- A variety of topics
- A variety of observers

#### Relevant CanMEDS milestones:

- 1 COM 3.1 Provide accurate information about the medical condition and plans for investigation and/or management**
- 2 COM 3.1 Use plain language and avoid medical jargon**
- 3 COM 3.1 Use strategies to verify and validate the patient's and/or family's understanding**
- 4 COM 4.3 Answer questions from the patient and/or family**
- 5 COM 4.3 Use communication skills and strategies that help the patient and/or family make informed decisions**
- 6 COM 4.1 Communicate in a manner that is respectful, non-judgmental and culturally aware**
- 7 HA 1.3 Incorporate disease prevention, health promotion, and health surveillance activities into interactions with patients**

## **Medical Genetics and Genomics: Foundations EPA #6**

### **Documenting clinical encounters**

#### Key Features:

- This EPA focuses on the application of written communication skills in a variety of formats: discharge summaries; consultations; progress notes.
- This includes a synthesis of the pertinent clinical findings, investigations and management plan.
- The documents submitted for review must be the sole work of the resident.
- This EPA may be observed in any care setting and any patient presentation.

#### Assessment Plan:

Document review by supervising staff or Core, TTP or subspecialty resident

Use form 1. Form collects information on:

- Patient demographic: maternal-fetal medicine; neonate; pediatric; adult
- Setting: inpatient; outpatient
- Type of documentation: admission note; clinic note; consult report; discharge letter; progress note; other (write in)

Collect 9 observations of achievement.

- A variety of clinical settings
- At least 2 observations in pediatric inpatient setting
- At least 2 observations in adult inpatient setting
- A variety of types of documentation

#### Relevant CanMEDS milestones

- 1 ME 2.2 Synthesize and interpret information from the clinical assessment**
- 2 COM 5.1 Organize information in appropriate sections**
- 3 COM 5.1 Document all relevant findings and investigations**
- 4 COM 5.1 Convey clinical reasoning and the rationale for decisions**
- 5 COM 5.1 Provide a clear plan for ongoing management**
- 6 COM 5.1 Complete clinical documentation in a timely manner**

## **Medical Genetics and Genomics: Foundations EPA #7**

### **Providing handover**

#### Key Features:

- This EPA focuses on communicating patient information between health care providers at transitions in care and prioritizing key clinical features.
- It includes oral handover and/or written documentation when going off duty (end of day, end of call) and at the end of a rotation.
- This EPA is observed in the inpatient ward setting.

#### Assessment Plan:

Direct or indirect observation by supervising staff or Core, TTP or subspecialty resident

Use form 1. Form collects information on:

- Patient demographic: neonate; pediatric; adult
- Observation type (select all that apply): direct; indirect

Collect 6 observations of achievement.

- At least 1 observation in neonate demographic
- At least 2 observations in pediatric demographic
- At least 3 observations in adult demographic
- At least 1 direct observation in the adult demographic
- At least 1 direct observation in the neonatal or pediatric demographic
- A variety of observers

#### Relevant CanMEDS milestones:

- 1 ME 5.2 Use structured communication tools and strategies to enhance patient safety**
- 2 COL 3.2 Summarize and prioritize patient issues providing rationale for key decisions**
- 3 COL 3.2 Provide anticipatory guidance for ongoing management, such as results of outstanding investigations and/or anticipated events/outcomes**
- 4 COL 3.2 Clarify issues with the receiving physician(s), as needed**
- 5 COL 1.3 Integrate the patient's perspective and context into the care plan**
- 6 P 1.1 Complete assigned responsibilities in a timely manner**

## **Medical Genetics and Genomics: Core EPA#1**

### **Assessing, diagnosing, and managing patients with an acute medical presentation of a genetic disorder**

#### Key Features:

- This EPA focuses on providing medical care to patients presenting with an acute medical complication of a genetic disorder at any time point from initial presentation to stabilization.
- This EPA includes providing consultation to other clinical services and/or primary inpatient management.
- This EPA may be observed in a simulation setting.

#### Assessment Plan:

Direct and indirect (e.g. case discussion, review of consult letter or other documents)  
observation by attending physician or TTP resident

Use form 1. Form collects information on:

- Setting: clinical; simulation
- Inborn error of metabolism: yes; no

Collect 5 observations of achievement.

- At least 3 in a clinical setting
- At least 4 patients with an inborn error of metabolism

#### Relevant CanMEDS milestones

- 1 COL 2.1 Respond to requests for consultation in a timely manner**
- 2 ME 2.2 Elicit a relevant comprehensive medical and family history**
- 3 ME 2.2 Perform a focused physical exam**
- 4 ME 2.2 Develop and prioritize the differential diagnosis**
- 5 ME 2.2 Select, sequence and prioritize investigations**
- 6 ME 2.2 Synthesize clinical, laboratory and/or imaging data to achieve or validate a diagnosis**
- 7 ME 2.4 Develop and implement a management plan**
- 8 COL 1.3 Support clinical colleagues in the implementation of a management plan**
- 9 ME 4.1 Monitor the evolution of the clinical course and/or the patient's response to intervention**
- 10 COM 5.1 Document the clinical encounter to convey the clinical assessment and the rationale for decisions and/or recommendations**

## Medical Genetics and Genomics: Core EPA#2

### Assessing, diagnosing, and managing stable patients with a known or suspected genetic disorder

#### Key Features:

- This EPA includes clinical assessment and evidence-informed decision making.
- This requires a patient-centred approach, considering the patient and family's goals and expectations, and considering multisystem manifestations.
- It also includes judicious use of health care resources.

#### Assessment Plan:

Direct observation, case discussion, and/or review of consult letter or other documents by attending physician, genetic counsellor or TTP resident

Use form 1. Form collects information on:

- Patient demographic: prenatal; pediatric; adult
- Category of disorder (select all that apply): cancer; congenital anomaly and malformation; connective tissue; cardiovascular; disorder of growth; endocrine; genodermatosis; hematologic; immunologic; inborn error of metabolism; neurodevelopmental; neurologic; renal; skeletal dysplasia; other (please specify)

Collect 15 observations of achievement.

- At least 5 prenatal patients
- At least 5 pediatric patients
- At least 5 adult patients
- A variety of observers
- A variety of categories of disorder
- No more than 3 observations by a genetic counsellor

#### Relevant CanMEDs milestones

- 1 P 1.2 Prepare for the consultation, reviewing clinical information and medical literature as needed**
- 2 ME 2.2 Elicit a relevant comprehensive medical and family history**
- 3 ME 2.2 Perform a physical exam with a focus on dysmorphology, where relevant**
- 4 ME 2.2 Develop and prioritize the differential diagnosis**
- 5 ME 2.2 Select and interpret investigations**
- 6 L 2.1 Demonstrate judicious use of resources**
- 7 ME 2.2 Synthesize clinical, laboratory and/or imaging data to achieve or validate a diagnosis**

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- 8 ME 2.4 Develop and implement a management plan**
- 9 COM 5.1 Document the clinical encounter to convey the clinical assessment and the rationale for decisions and/or recommendations**



## Medical Genetics and Genomics: Core EPA#3

### Providing pre-test counselling for genetic and/or genomic testing

#### Key Features:

- The focus of this EPA is the application of communication skills to counsel patients regarding genetic and/or genomic testing.
- Pre-test counselling includes discussing the indications for testing, the possible outcomes and limitations of the test, and the ethical, legal and social implications of testing.
- This EPA includes assessing the patient's and/or family's understanding of the elements of consent and obtaining informed consent.
- This EPA may be observed in simulation.

#### Assessment Plan:

Direct observation by attending physician, genetic counsellor, or TTP resident

Use form 1. Form collects information on:

- Patient demographic: prenatal; pediatric; adult
- Setting: clinical; simulation
- Indications for testing: diagnostic testing; presymptomatic testing; predictive testing; carrier testing; prenatal testing; newborn screening; preimplantation testing
- Testing modality: cytogenetic; functional assays; targeted molecular; genome wide sequencing; other (write in)

Collect 5 observations of achievement.

- At least 1 prenatal patient
- At least 1 genome wide sequencing
- At least 1 presymptomatic or predictive testing

#### Relevant CanMEDS milestones:

- 1 ME 3.2 Describe the indications, possible outcomes and limitations of genetic testing**
- 2 ME 3.2 Describe the ethical, legal and social implications of testing**
- 3 COM 3.1 Share information and explanations that are clear and accurate**
- 4 COM 3.1 Verify understanding of information conveyed**
- 5 COM 1.5 Recognize when strong emotions (such as, anger, fear, anxiety, or sadness) are affecting an interaction and respond appropriately**
- 6 COM 4.3 Use communication skills and strategies that help the patient and/or family make informed decisions**
- 7 COM 4.1 Communicate in a manner that is respectful, non-judgmental and culturally aware**

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**8 COM 1.6 Tailor approaches to decision-making to patient capacity, values, and preferences**

## **Medical Genetics and Genomics: Core EPA#4**

### **Counselling patients and families regarding the implications of a genetic diagnosis**

#### Key Features:

- This EPA focuses on the application of communication skills to counsel patients regarding the personal, medical, reproductive and psychosocial implications of a genetic diagnosis.
- This includes counselling and/or providing recommendations regarding management and health promotion, and anticipatory guidance to enhance the quality of life for patients and families with a genetic condition.
- This EPA also includes discussing implications for family members including recurrence risk assessment and testing of at-risk relatives (e.g., cascade screening).

#### Assessment Plan:

Direct observation by attending physician, TTP resident, or genetic counsellor; this may be informed by review of documentation, including counselling letter.

Use form 1.

Collect 5 observations of achievement.

- A variety of observers
- At least 4 observations by a physician

#### Relevant CanMEDS milestones:

- 1 COM 3.1 Convey the personal, medical, reproductive and psychosocial implications of a genetic diagnosis**
- 2 COM 3.1 Share information and explanations that are clear and accurate**
- 3 COM 1.5 Recognize when strong emotions (such as, anger, fear, anxiety, or sadness) are affecting an interaction and respond appropriately**
- 4 ME 2.4 Provide recommendations regarding management and anticipatory guidance regarding the evolution of the condition**
- 5 HA 1.3 Discuss implications for family members including recurrence risk assessment and testing of at-risk relatives (e.g., cascade screening).**
- 6 HA 1.2 Recommend patient and family education resources**
- 7 HA 1.1 Facilitate timely patient access to services and resources**
- 8 COM 4.1 Provide psychological support, either personally or through referral**
- 9 COM 4.1 Communicate in a manner that is respectful, non-judgmental and culturally aware**

## **Medical Genetics and Genomics: Core EPA#5**

### **Leading discussions with patients, families and/or other health care professionals in challenging situations**

#### Key Features:

- This EPA focuses on the application of advanced communication and conflict resolution skills to address difficult situations that involve patients, families, and/or members of the health care team.
- This may include emotionally challenging situations, ethical dilemmas, and disagreements about the care plan amongst the team and the patient and family.
- This EPA may be observed in the simulation setting.

#### Assessment Plan:

Direct observation by attending physician, TTP resident, or other health professional

Use form 1. Form collects information on:

- Setting: clinical; simulation

Collect 2 observations of achievement.

#### Relevant CanMEDS milestones:

- 1 COM 1.2 Ensure the physical environment is suitable for the nature of the situation (e.g., privacy, safety)**
- 2 ME 2.1 Ascertain participants' understanding of the situation and the reason for the encounter**
- 3 COM 4.1 Communicate in a manner that is respectful, non-judgmental and culturally aware**
- 4 COM 3.1 Verify understanding of information conveyed**
- 5 COM 1.5 Recognize when strong emotions (such as, anger, fear, anxiety, or sadness) are affecting an interaction and respond appropriately**
- 6 COM 1.4 Identify, verify and validate non-verbal cues**
- 7 COM 2.2 Summarize and close the encounter effectively**

## **Medical Genetics and Genomics: Core EPA#6**

### **Working with other health professionals and contributing expertise in interdisciplinary teams**

#### Key Features:

- This EPA focuses on the role of the medical geneticist in coordinating the care needs of patients with known or suspected genetic disease.
- This includes working effectively with members of the interprofessional team, including genetic counselors, metabolic dieticians, nurses and administrative staff.
- It also includes shared decision making with other services and specialty physicians, responding to requests for guidance, and supporting the team to address complex patient issues.
- This EPA is typically based on observation over the course of at least one block.

#### Assessment Plan:

Direct and indirect observation by supervisor, with input from other health professionals which may include other physicians, laboratory geneticists, nurses, dieticians, genetic counsellors, and other residents

Use form 3. Form collects information on

- Input provided by (select all that apply): medical geneticist; laboratory geneticist; nurse; dietician; genetic counsellor; resident; other

Collect an observation at least 3 times during the Core stage.

- Observations should include a variety of observer roles

#### Relevant CanMEDS milestones:

- 1 P 1.1 Behave in a professional manner**
- 2 COL 1.2 Demonstrate an understanding of the scope and expertise of other health care professionals**
- 3 COL 1.3 Communicate effectively with physicians and other health care professionals**
- 4 P 1.1 Respond punctually to requests from other health care professionals**
- 5 COL 1.3 Provide guidance to resolve clinical challenges**
- 6 HA 1.1 Facilitate timely patient access to services and resources**

## **Medical Genetics and Genomics: Core EPA#7**

### **Advising on the indications and limitations of laboratory testing relevant to Medical Genetics and Genomics**

#### Key Features:

- This EPA includes appropriate test selection, demonstrating resource stewardship.
- It also includes assessing the quality of the test, selecting ancillary studies as appropriate, and interpreting the findings in the context of clinical history, specimen type, and other diagnostic results.
- This EPA includes cytogenetic, metabolic and molecular testing.

#### Assessment Plan:

Direct and indirect (e.g., review of clinical or laboratory cases) by attending physician, laboratory geneticist, genetic counselor, TTP resident, or lab fellow

Use form 1. Form collects information on:

- Setting: clinic; lab
- Testing modality: cytogenetic; metabolic; molecular

Collect 5 observations of achievement.

- At least 3 in the lab setting
- At least 1 of each testing modality
- At least 3 by a laboratory geneticist

#### Relevant CanMEDS milestones:

- 1 ME 2.2 Synthesize patient information to establish the clinical question**
- 2 ME 2.2 Determine that the selected investigations are appropriate to the clinical indication**
- 3 ME 2.2 Assess the quality and validity of the study, and any impact on the diagnostic interpretation**
- 4 ME 2.2 Interpret genetic and genomic results together with available clinical and laboratory data**
- 5 ME 2.2 Select additional testing based on an appreciation of the diagnostic possibilities, the clinical context, and the relevance and capabilities of available technologies**
- 6 L 2.1 Utilize genetic testing resources effectively to balance costs with potential utility of results**
- 7 ME 4.1 Coordinate the use of multiple diagnostic investigations so as to ensure complementarity and efficiency**
- 8 COL 1.3 Advise referring physicians on the most appropriate selection, sequence and/or timing of further investigation(s)**

## **Medical Genetics and Genomics: Core EPA#8**

### **Providing interpretation of cytogenetic, molecular, and biochemical results**

#### Key Features:

- This EPA focuses on synthesizing diverse sources of information to interpret cytogenetic, molecular, and biochemical test results, and convey that information to other health care professionals.
- Evidence includes scientific literature and publicly accessible raw data (e.g., UCSC Genome Browser, gnomAD).

#### Assessment Plan:

Direct observation or case discussion with attending physician, laboratory geneticist or CCMG lab fellow

Use form 1. Form collects information on:

- Lab discipline: cytogenetic; metabolic; molecular
- Setting: clinic; lab

Collect 10 observations of achievement.

- At least 5 in the lab setting
- At least 1 of each lab discipline

#### Relevant CanMEDS milestones:

- 1 ME 5.1 Recognize sources of analytical error for various genetic tests**
- 2 ME 2.2 Interpret genetic and genomic results together with available clinical and laboratory data**
- 3 S 3.4 Integrate best evidence and clinical expertise into decision-making**
- 4 COM 5.1 Synthesize the findings of genetic testing for inclusion in a comprehensive and clinically meaningful laboratory report**
- 5 COL 1.3 Work effectively with clinical colleagues to assist in the interpretation of laboratory findings in the clinical context**

**Medical Genetics and Genomics: Core EPA #9**

**Delivering scholarly teaching to a variety of audiences**

Key Features:

- This EPA focuses on the skills of critical appraisal as well as presentation and teaching skills.
- This EPA may be observed in a variety of settings. Examples include grand rounds or divisional rounds, lectures to students or other residents, journal club, and educational sessions for patients and families.

Assessment Plan:

Direct observation by attending physician or TTP resident

Use form 1.

Collect 2 observations of achievement.

- At least 2 different occasions
- At least 2 observers

Relevant CanMEDS milestones:

- 1 S 2.4 Identify the learning needs and desired learning outcomes of others**
- 2 S 2.4 Develop learning objectives for a teaching activity adapting to the audience and setting**
- 3 S 3.3 Critically evaluate the integrity, reliability and applicability of health-related research and literature**
- 4 S 3.4 Integrate best evidence and clinical expertise**
- 5 S 2.4 Present the information in an organized manner**
- 6 S 2.4 Use audiovisual aids effectively**
- 7 S 2.4 Provide adequate time for questions and discussion**



**Medical Genetics and Genomics: Core EPA#10**

**Supervising junior learners in the clinical setting**

Key Features:

- This EPA focuses on the informal teaching that occurs in the clinical (bedside) setting, and includes ensuring safe patient care, teaching and providing feedback.
- This EPA includes delegation of tasks to other residents and students and may include administrative duties relevant to organization of the medical team.

Assessment Plan:

Direct observation by attending physician or TTP resident

Use form 1.

Collect 3 observations of achievement.

Relevant CanMEDS milestones:

- 1 COL 2.1 Delegate tasks and responsibilities in an appropriate and respectful manner**
- 2 S 2.2 Create a positive learning environment**
- 3 S 2.3 Be available and accessible to junior learners**
- 4 S 2.4 Identify the learning needs and desired learning outcomes of others**
- 5 S 2.4 Provide clinical teaching and/or other informal learning activities**
- 6 S 2.5 Provide feedback to enhance learning and performance**
- 7 S 3.4 Integrate best evidence and clinical expertise into teaching**
- 8 P 1.1 Respond appropriately when behaviours toward colleagues and/or learners undermine a respectful environment**

## **Medical Genetics and Genomics: Core EPA#11**

### **Integrating scientific evidence to inform clinical practice**

#### Key Features:

- This EPA focuses on resolving questions that arise in the context of clinical work by selecting, accessing and critically appraising scientific evidence.
- This EPA includes gathering, synthesizing and interpreting scientific evidence.
- Evidence includes the primary scientific literature and curated databases (e.g., OMIM, GeneReviews).
- This EPA may be observed in the laboratory (i.e., lab-based issue) or clinical setting.

#### Assessment Plan:

Direct or indirect observation by attending physician, lab geneticist, TTP resident, or genetic counsellor

Use form 1. Form collects information on:

- Setting: clinic; lab

Collect 10 observations of achievement.

- At least 8 by physician or lab geneticist

#### Relevant CanMEDS milestones:

- 1 S 3.1 Formulate the knowledge gap arising from a clinical situation**
- 2 S 3.3 Apply knowledge of available resources to select appropriate sources of information**
- 3 S 3.3 Develop and execute an effective search strategy**
- 4 S 3.3 Critically evaluate the reliability and quality of resources**
- 5 S 3.4 Integrate findings to address the clinical question**

## **Medical Genetics and Genomics: Transition to Practice EPA #1**

### **Leading consultation and ongoing management of inpatients with known or suspected genetic conditions**

#### Key Features:

- This EPA focuses on leading the provision of care for inpatients in the role of the junior attending.
- This EPA includes assuming responsibility for the medical care decisions and working effectively with the interprofessional team.
- It also includes integrating teaching and supervision of junior trainees and delegation of tasks at the level of junior attending.
- The observation of this EPA is based on a period of at least one week.

#### Assessment Plan:

Direct and/or indirect observation by attending physician

Use form 1.

Collect 3 observations of achievement.

#### Relevant CanMEDS milestones:

- 1 ME 1.1 Demonstrate responsibility and accountability for decisions regarding patient care, acting in the role of most responsible physician**
- 2 ME 2.4 Establish patient-centred management plans**
- 3 S 3.4 Integrate best evidence and clinical expertise into decision-making**
- 4 L 4.1 Set priorities and manage time to fulfil diverse responsibilities**
- 5 COL 1.3 Communicate effectively with a consulting service**
- 6 L 4.1 Integrate supervisory and teaching responsibilities into the overall management of the clinical service**
- 7 L 4.2 Run the service efficiently, safely, and effectively**
- 8 P 4.2 Manage competing personal and professional priorities**

## Medical Genetics and Genomics: Transition to Practice EPA#2

### Managing a genetic practice

#### Key Features:

- This EPA focuses on the management of a longitudinal outpatient clinic in the role of the physician most responsible for patient care.
- This includes responsibility for medical care decisions, follow-up on investigations and accessibility in between clinic visits.
- It also includes time management, practice management, and the judicious use of resources in the outpatient setting.
- This EPA may be observed during earlier stages of training in the continuity clinic setting.
- This EPA is observed over the course of at least one month.

#### Assessment Plan:

Indirect observation by supervisor, with input from genetic counsellors, administrative assistants, and/or other health professionals

Use form 3.

Collect 2 observations of achievement.

#### Relevant CanMEDS milestones:

- 1 ME 1.1 Demonstrate responsibility and accountability for decisions regarding patient care, acting in the role of most responsible physician**
- 2 ME 1.6 Prioritize patients based on the urgency of clinical presentations**
- 3 S 3.4 Integrate best evidence and clinical expertise into decision-making**
- 4 COL 1.3 Provide accurate, timely and relevant written information to the referring/primary care physician**
- 5 L 4.2 Oversee clinic booking and scheduling**
- 6 L 4.1 Manage time effectively to maintain clinic flow**
- 7 COL 1.2 Work effectively with outpatient clinic staff**
- 8 L 4.1 Review and act on test results in a timely manner**
- 9 HA 1.1 Facilitate timely patient access to services and resources**

### **Medical Genetics and Genomics: Transition to Practice EPA#3**

#### **Working collaboratively with the health care team for coordinated patient care**

Key Features:

- This EPA focuses on the role of the geneticist in leading the health care team and delegating tasks appropriately to other health professionals.
- It includes reviewing the care plans and documentation of other members of the health care team.
- It also includes advocating effectively for patient needs.

Assessment Plan:

Direct and/or indirect observation by supervisor with input from other health care professionals

Use form 3. Form collects information on:

- Input provided by (select all that apply): geneticist; other physician; genetic counsellor; nurse; social worker; dietician; support staff; other health professional (please specify)

Collect 2 observations of achievement.

- At least 3 observers representing 3 different roles on each occasion

Relevant CanMEDS milestones:

- 1 ME 1.1 Demonstrate responsibility and accountability for decisions regarding patient care, acting in the role of most responsible physician**
- 2 COL 1.2 Make effective use of the scope and expertise of other health care professionals**
- 3 COL 2.1 Delegate tasks and responsibilities in an appropriate and respectful manner**
- 4 COL 1.1 Respond appropriately to input from other health care professionals**
- 5 COL 1.3 Communicate effectively with physicians and other health care professionals**
- 6 P 1.1 Behave in a professional manner**

## **Medical Genetics and Genomics: Transition to Practice EPA#4**

### **Developing a plan to transition to practice**

#### Key Features:

- This EPA may include a variety of scenarios. Examples include: a plan to act on identified performance gaps; a plan to correct gaps in areas of practice not experienced in training; a plan to prepare for practice in a specific setting and/or a practice requiring distinct skills.
- Achievement of this EPA includes developing a learning plan adapted to their practice profile. It includes a) providing the rationale for a learning plan, b) engaging in self-reflection, c) performing a personal needs assessment, d) managing time and e) identifying the methods to achieve the personal learning plan such as literature review, clinical training, mentorship, conference attendance and/or rounds attendance.

#### Assessment Plan:

Review of the resident's submission of a learning plan by the Competence Committee

Use form 4.

Collect 1 observation of achievement.

#### Relevant CanMEDS milestones:

- 1 P 2.1** Demonstrate a commitment to maintaining and enhancing competence
- 2 S 1.2** Interpret data on personal performance to identify opportunities for learning and improvement
- 3 L 4.2** Examine personal interests and career goals
- 4 S 1.1** Define learning needs related to personal practice and/or career goals
- 5 S 1.1** Create a learning plan that is feasible, includes clear deliverables and a plan for monitoring ongoing achievement
- 6 S 1.1** Identify resources required to implement a personal learning plan
- 7 L 4.2** Adjust educational experiences to gain competencies necessary for future practice