



Canadian College of Medical Geneticists Collège canadien des généticiens médicaux

CCMG Training and Speciality Requirements in Clinical Genetics

Revised 2007

Preamble

The aim of the Canadian College of Medical Geneticists (CCMG) is to produce a physician who is competent to assume responsibility for consultation service in genetic diagnosis, counseling and management of genetic disease.

The practice of clinical genetics is based on an in depth knowledge of basic genetic principles, a broad range of knowledge of genetic disease as it affects all body systems and individuals of all ages, and a clear understanding of the principles of genetic counseling.

On completion of the educational program, the graduate physician will be competent to function as a consultant in Clinical Genetics. It is understood that fellows successfully completing the program are regarded as fully competent, undifferentiated consultants in the speciality.

The CCMG training guidelines are modeled after the CanMEDS framework¹. This framework includes the competencies required of specialists and the role of the specialist beyond that of the specialty medical expert. The other roles of the specialist are now recognized as that of communicator, collaborator, manager, health advocate, scholar, and professional. The detailed objectives describe minimal standards and in no way exclude the necessity for mastery of additional knowledge, skills or attitudes necessary for the most effective management of patients with genetic disorders.

Required Background

Trainees must have an MD and at least three years postdoctoral training in a Royal College of Physicians and Surgeons of Canada (RCPSC) accredited residency program and/or the Collège des Médecins du Québec (CMQ), or equivalent training. It is assumed that the fellow will have had some exposure to either pediatrics and/or internal medicine. When planning their training, candidates are urged to consider the requirements for certification by the RCPS/CMQ as well as their provincial licensing requirements.

Administrative Aspects

1. Supervisory committee:

- a. Each trainee's program will be supervised by a committee, headed by a Fellow of the CCMG in clinical genetics who takes primary responsibility for the training.
- b. The committee will consist of the head and a minimum of two additional members. Other members might consist of molecular geneticists,

cytogeneticists and/or biochemical geneticists although the structure of the committee can vary depending on the background of the trainee.

- c. The committee ensures the trainee is registered with the CCMG by submitting registration form to the chair of Credentials by August 1 of the first year of training.
- d. The committee takes responsibility for ensuring the training program is in keeping with CCMG guidelines, and submits an outline of completed and planned training with the trainee's application for credentialing.
- e. The committee meets every six months with the candidate, and ensures that in-training evaluation forms are completed and discussed with the trainee. If remedial work is needed by the trainee, the committee must ensure that this is provided.
- f. The committee completes and submits the Final In-training evaluation (FITER).

2. Location of training

- a. Clinical genetics training must take place in a center accredited by the CCMG for training in Clinical Genetics.
- b. Elective training may be done at non-accredited centers at the discretion of the supervisory committee.
- c. In the event of termination of accreditation of a center during the candidate's training, the trainee will be allowed a maximum of six months to move to an accredited center for completion of training.

3. Training in foreign centers

- a. Training in American centers accredited by the American Board of Medical Genetics (ABMG) is recognized by the CCMG.
- b. As the ABMG and CCMG have different credentialing requirements, it is the responsibility of the trainee to ensure completion of all requirements of the CCMG .

4. Part time training

- a. Part-time training is recognized by the CCMG, provided it conforms to all requirements in this document and the trainee spends a minimum of 50% of time in the program.
- b. The total amount of time must equal two complete years in training.

Content of Training

This is a minimum 3-year training program. Trainees should be exposed to genetic diagnostic problems in both adults and children. The trainee must receive training in techniques of genetic counseling with regard to psychological dynamics, stress management and crisis intervention. The Supervisory Committee must ensure that the service load does not interfere with the educational goals of the program.

Mandatory training:

1. **15 months** on a clinical/medical genetics service supervised by a CCMG accredited clinical geneticist. During this time the fellow should gain experience in multiple aspects of clinical genetics including pediatric and adult genetics, prenatal diagnosis and cancer genetics.

2. **3 months** on a metabolic clinical service supervised by an expert in the management of metabolic disorders whose credentials are acceptable to the CCMG. The fellow should gain experience in the management of metabolic disorders in both children and adults.
3. **2 months** in high-risk obstetrics/fetal assessment unit.
4. **1 month** in a counseling and communication theory/practice area outside the actual genetics unit.
5. **2 months** in a cytogenetics laboratory supervised by a CCMG accredited cytogenetics.
6. **2 months** in a molecular genetics laboratory supervised by a CCMG accredited molecular geneticist.
7. **2 months** in a biochemical genetics laboratory supervised by a CCMG accredited biochemical geneticist or a similarly qualified individual such a clinical chemist expert in metabolic disorders whose credentials are acceptable to the CCMG.
8. **6 months** research. The trainee must participate in clinical or laboratory based research in an area relevant to the field of Medical Genetics. The period of research training may be obtained at the training centre or another hospital or university centre in Canada or abroad as approved by the candidate's supervisory committee and Program Director.
9. Courses/conferences (all the below activities should be documented in a **logbook**)
 - a) Documented participation in educational events and courses prescribed by the trainee's supervisory committee. All trainees are expected to take part in regular conferences, seminars and journal club sessions in medical genetics.
 - b) Documented attendance at one national or international genetics meeting each year.
 - c) Most trainees will have had graduate or undergraduate courses in genetics, biochemistry, molecular biology, and cell biology necessary for a four year degree in genetics, biochemistry, biology, or life-science programs. However, if course work (or an equivalent education experience) that approximates that required for an MSc in genetics and molecular biology, or the laboratory specialty are lacking, the trainee must gain this knowledge through suitable courses or private studies.

In addition, the trainee's logbook must document at least **200** patients or families for whom he/she has assumed responsibility and has completed a consultation. This should reflect a variety of problems including inpatient and outpatient

consultations, for prenatal diagnosis, dysmorphology, cancer genetics, adult onset genetic disorders, teratogen exposure, metabolic disorders and the interpretation of laboratory results.

Elective Training:

1. 3 months of electives relevant to medical genetics.

Clinical Genetics Training Guidelines

Key and Enabling Competency Statements

Note:

The 7 *Roles* are the thematic groups of competencies that organize the CanMEDS format (Medical Expert, Communicator, Collaborator, Manager, Health Advocate, Scholar, Professional)

The *Key Competencies* are the overall culminating objectives of the training. They are meant to be summative and cumulative, while also being observable and measurable.

The *Enabling Competencies* are the skills that allow the *Key Competencies* to be achieved. The *Enabling Competencies* break-down the *Key Competencies* into observable and measurable statements.

MEDICAL EXPERT/ CLINICAL DECISION-MAKER

Key Competencies

By the end of training, the Clinical Genetics Trainees will demonstrate the ability to:

1. Explain general and advanced concepts in human genetics and molecular biology;
2. Use diagnostic and therapeutic skills for ethical and effective patient care, and access and apply relevant information to clinical practice;
3. Demonstrate effective consultation services with respect to patient care, education and legal opinions.

Enabling Competencies

1. Explain general and advanced concepts in human genetics and molecular biology.

To achieve this, the Clinical Genetics Trainee will be able to:

- 1.1 Describe and discuss general concepts of human genetics and molecular biology, including:
 - 1.1.1 Normal and abnormal gene structure and function;
 - 1.1.2 The general structure of the human genome;
 - 1.1.3 The information that can be obtained from an integrated assessment of genomic function at the RNA or protein level, that cannot be obtained from the DNA sequence alone;
 - 1.1.4 How processes such as gene duplication and divergence, exon shuffling, and the activity of transposable elements help to explain genomic variability, redundancy, and plasticity;
 - 1.1.5 How gene expression is affected by differences in coding and non-coding regions, effects of trans-acting factors, and the structure of chromatin;
 - 1.1.6 How protein function is influenced by mRNA and polypeptide processing, targeting, and interactions;
 - 1.1.7 The effects of epigenetic processes;
 - 1.1.8 Normal and abnormal cell division;
 - 1.1.9 Chromosome structure, morphology and nomenclature, including the principles and application of the various cytogenetic techniques;
 - 1.1.10 Principles and application of somatic cell genetics;
 - 1.1.11 Principles and application of molecular genetic techniques;
 - 1.1.12 Basic principles of biochemistry and principles and application of laboratory investigation relevant to inborn errors of metabolism;
 - 1.1.13 Monogenic and complex inheritance;
 - 1.1.14 Developmental biology as it relates to normal and abnormal human morphogenesis;
 - 1.1.15 Principles of epidemiology, including biostatistics, genetic epidemiology, and population genetics.

2. Use diagnostic and therapeutic skills for ethical and effective patient care, and access and apply relevant information to clinical practice.

To achieve this, the Clinical Genetics Trainee will:

- 2.1 Demonstrate an understanding of the following:
 - 2.1.1 The indications, limitations and risks of techniques of fetal assessment and options for reproductive intervention;
 - 2.1.2 Genetic and non-genetic (intrinsic and extrinsic) factors predisposing to fetal loss, infertility, and abnormalities of morphogenesis;
 - 2.1.3 Teratogenic agents and their effects;
 - 2.1.4 Phenotypic variation and specific methods of assessment;
 - 2.1.5 Methods of syndrome identification and diagnosis, including the use of computer diagnostic aids;
 - 2.1.6 Etiology, diagnosis, management, natural history, and prognosis of well-defined genetic syndromes and diseases;
 - 2.1.7 The use and limitations of commonly used instruments for the assessment of behavior and intelligence;
 - 2.1.8 Indicators of normal and abnormal psychomotor development;
 - 2.1.9 Characteristic behavioral phenotypes of well-defined genetic syndromes and disorders;
 - 2.1.10 Community services and resources available to help patients and their families;
 - 2.1.11 Genetic screening and genetic testing;
 - 2.1.12 Federal and provincial laws related to genetic diseases, reproductive options and technology;
 - 2.1.13 Federal and provincial laws related to confidentiality, autonomy, disclosure, privacy, and issues of competence;
 - 2.1.14 The distinction between genetic testing for the diagnosis of disease and predictive testing to assess risk for predisposition to monogenic or complex genetic diseases as well as their applications and limitations.
- 2.2 Be able to perform a complete evaluation of physiological and pathological states relevant to the specific expertise of human medical genetics, including prenatal diagnosis, teratology, chromosomal abnormalities, disorders of morphogenesis, inborn errors of metabolism, and monogenic and complex genetic disorders. In order to do this the trainee must be able to:
 - 2.2.1 Elicit a comprehensive medical history and an appropriate family history, and to construct and interpret a standardized pedigree, and calculate risks;
 - 2.2.2 Carry out a comprehensive physical examination with special expertise in phenotypic variation;
 - 2.2.3 Formulate an appropriate differential diagnosis, and plan an appropriate course of investigation with respect to genetic disease;
 - 2.2.4 Perform special expertise in syndrome identification, including the use of diagnostic aids (e.g. computer assisted diagnosis).

3. Demonstrate effective consultation services with respect to patient care and education and legal opinions.

To achieve this, the Clinical Genetics Trainee will be able to:

- 3.1 Recognize, describe, and interpret laboratory and imaging findings relevant to genetic disease with special expertise in cytogenetics, molecular genetics and biochemical genetics;
- 3.2 Synthesize these clinical, laboratory, and imaging data to achieve or validate a diagnosis;
- 3.3 Plan and coordinate the care of individuals affected with genetic conditions;
- 3.4 Provide continuity in care and to periodically assess the appropriateness of the care plan;
- 3.5 Provide effective genetic counseling for patients and their families.

COMMUNICATOR

Key Competencies

By the end of training, the Clinical Genetics Trainees will demonstrate the ability to:

1. Listen effectively and establish therapeutic relationships with patients and families;
2. Obtain and synthesize relevant history from patients and families and their communities;
3. Discuss appropriate information with patients and families;
4. Provide consultation for clinical genetics cases to other health care providers.

Enabling Competencies

1. Listen effectively and establish therapeutic relationships with patients and families.

To achieve this, the Clinical Genetics trainee will be able to:

- 1.1 Recognize one's own biases, including ethno-cultural differences, and their impact on communication and patient care;
- 1.2 Understand how cultural background, age, gender, socioeconomic background and spiritual values influence communication;
- 1.3 Use, appropriately, non-verbal communication.

3. Obtain and synthesize relevant history from patients and families and their communities

To achieve this, the Clinical Genetics trainee will be able to:

- 2.1 Gather information not only about the disease but also about the patient's beliefs, concerns and expectations about the disorder, while considering the influence of factors such as the patient's age, gender, ethnic, cultural, and socioeconomic background, and spiritual values.

3. Discuss appropriate information with patients and families.

To achieve this, the Clinical Genetics trainee will be able to:

- 3.1 Provide genetic counseling: displaying empathy and compassion, especially in delivering bad news, remaining objective and impartial, remaining appropriately non-directive, but being prepared to advise in certain situations and to provide psychological support either personally or through referral employing active listening skills, delivering information to the patient and family in a manner that is understandable, encouraging discussion, and promoting patient and family participation in decision-making;
- 3.2 Help the individual and family choose an appropriate course of action for themselves;
- 3.3 Advise patients and families about support agencies.

4. Provide consultation for clinical genetics cases to other health care providers

To achieve this, the Clinical Genetics trainee will be able to:

- 4.1 Communicate, at a level appropriate to the consultant or the referring physician, information concerning the medical implications and prognosis, the risks that apply, and the options available;
- 4.2 Summarize findings, consultation notes and counseling, for referring physicians, agencies and families.

COLLABORATOR

Key Competencies

By the end of training, the Clinical Genetics Trainees will demonstrate the ability to:

1. Consult effectively with other physicians and health care professionals;
2. Contribute effectively to other interdisciplinary team activities.

Enabling Competencies

1. Consult effectively with other physicians and health care professionals.

To achieve this, the Clinical Genetics Trainee will be able to:

- 1.1 Demonstrate the ability to participate in collaborative decision making with other healthcare providers (including family physicians, other specialists and genetic counselors).

2. Contribute effectively to other interdisciplinary team activities.

To achieve this, the Clinical Genetics Trainee will be able to:

- 2.1 Participate in an interdisciplinary team meeting, and demonstrate the ability to accept, consider and respect the opinions of other team members, while contributing medical genetics-specific expertise him/herself;
- 2.2 Communicate effectively with the members of an interdisciplinary team in the resolution of conflicts, provision of feedback, and where appropriate, be able to assume a leadership role;
- 2.3 Describe how health care governance influences patient care, research and educational activities at a local, provincial, regional, and national level;
- 2.4 Demonstrate an understanding of the roles of clinicians and research scientists cooperatively to advance knowledge of human genetics in genetics research endeavors.

MANAGER

Key Competencies

By the end of training, the Molecular Genetics Trainees will demonstrate the ability to:

1. Utilize resources effectively to balance patient care, learning needs, and outside activities;
2. Allocate finite health care resources wisely;
3. Work effectively and efficiently in a health care organization;

Enabling Competencies

1. Utilize resources effectively to balance patient care, learning needs, and outside activities.

To achieve this, the Clinical Genetics Trainee will be able to:

- 1.1 Demonstrate the ability to balance personal and professional demands on activities of daily living;
- 1.2 Demonstrate understanding of the following professional skills in time management: recognition that the effective use of time depends on punctuality, requires planning, depends on development of speed as well as accuracy in clinical skills, on reservation of time for reading and keeping current with the genetics literature and on the establishment of routines for carrying out regular activities and adhere to them;
- 1.3 Utilize information technology to optimize patient care, life-long learning and other activities.

2. Allocate finite health care resources wisely.

To achieve this, the Clinical Genetics Trainee will be able to:

- 2.1 Demonstrate knowledge of planning, evaluation, and assessment of outcome of a health care program.

3. Work effectively and efficiently in a health care organization.

To achieve this, the Clinical Genetics Trainee will be able to:

- 3.1 Demonstrate understanding of the importance of quality assurance as it relates to clinical care, laboratory data, and education;
- 3.2 Demonstrate understanding of issues involving potential litigation;
- 3.3 Demonstrate commitment to the maintenance of complete and accurate medical records, and recognize the uses and limitations of electronic patient databases;
- 3.4 Demonstrate knowledge of how to identify employment policies and procedures;
- 3.5 Demonstrate the ability to effectively coordinate the work of the health care team.

HEALTH ADVOCATE

Key Competencies

By the end of training, the Clinical Genetics Trainee will demonstrate the ability to:

1. Identify the important determinants of health affecting patients and contribute effectively to the improved health of patients and communities;
2. Recognize and respond to those issues where advocacy is appropriate.

Enabling Competencies

- 1. Identify the important determinants of health affecting patients and contribute effectively to the improved health of patients and communities.**

To achieve this, the Clinical Genetics Trainee will be able to:

- 1.1 Demonstrate an awareness of, and a willingness to refer patients to, community and national resources.

- 2. Recognize and respond to those issues where advocacy is appropriate.**

To achieve this, the Clinical Genetics Trainee will be able to:

- 2.1 Demonstrate understanding of the importance of participating actively in public policy discussions and decision-making regarding the application of new genetic technologies;
- 2.2 Demonstrate understanding of the roles of national and international agencies in the promotion of genetic health and the prevention, detection, and treatment of genetic disorders.

SCHOLAR

Key Competencies

By the end of training, the Clinical Genetics Trainees will demonstrate the ability to:

1. Conduct ongoing learning activities to maintain and advance professional knowledge;
2. Facilitate the learning of other health care professionals, students, laboratory colleagues, the public and others regarding molecular diagnostic testing;
3. Conduct research projects for the advancement of the field of medical genetics.

Enabling Competencies

1. Conduct ongoing learning activities to maintain and advance professional knowledge.

To achieve this, the Clinical Genetics Trainee will be able to:

- 1.1. Critically assess the literature as related to human genetics, patient diagnosis and management;
- 1.2. Attend continuing education events, including conferences, rounds, clinical and research seminars, patient conferences;
- 1.3. Recognize limitations of current knowledge base and seek appropriate continuing educational activities;
- 1.4. Demonstrate the ability to utilize technology for continuing education.

2. Facilitate the learning of other health care professionals, students, laboratory colleagues, the public and others regarding medical genetics.

To achieve this, the Clinical Genetics Trainee will be able to:

- 2.1. Deliver effective lectures and presentations on clinical genetics and molecular biology, cytogenetic and biochemical genetic concepts;
- 2.2. Present concise and audience appropriate summaries of patient evaluations/ case reports.

3. Conduct research projects for the advancement of the field of medical genetics.

To achieve this, the Clinical Genetics Trainee will be able to:

- 3.1. Plan and conduct a minimum six month clinical or laboratory based research project in an area relevant to medical genetics;
- 3.2. Summarize results in a format suitable for publication in a referred journal;
- 3.3. Identify personal limitations with respect to previous research experience and conduct appropriate activities to address these limitations.

PROFESSIONAL

Key Competencies

By the end of training, the Clinical Genetics Trainees will demonstrate the ability to:

1. Deliver highest quality care with integrity, honesty and compassion;
2. Exhibit appropriate personal and interpersonal professional behaviors.

Enabling Competencies

1. Deliver highest quality care with integrity, honesty and compassion.

To achieve this, the Clinical Genetics Trainee will be able to:

- 1.1. Demonstrate understanding of the importance of confidentiality and the difficulties it poses in the rare instances where relatives are at risk for a serious and potentially preventable disease;
- 1.2. Recognize the sensitive nature of genetic testing and information and act to minimize potential harms;
- 1.3. Recognize ethical issues in clinical genetics, including but not limited to testing of minors, impact of molecular test results on extended family members, testing for late onset disorders and prenatal testing;
- 1.4. Identify personal limitations and the necessity of seeking the opinions of colleagues or other professionals when required;
- 1.5. Demonstrate the willingness and ability to appraise accurately his/her own professional performance.

2. Exhibit appropriate personal and interpersonal professional behaviors.

To achieve this, the Clinical Genetics Trainee will be able to:

- 2.1 Display personal and professional attitudes consistent with a consulting physician role by: periodically reviewing his/her own personal and professional performance against national standards set for the specialty, showing willingness to include the patient in discussions concerning appropriate diagnostic and management procedures, showing appropriate respect for the opinions of fellow consultants and referring physicians in the management of patient problems and being willing to provide means whereby differences of opinion can be discussed and resolved;
- 2.2 Demonstrate the ability to recognize and respond appropriately to abuse, gender bias, discrimination, intimidation, and disrespect;
- 2.3 Demonstrate the knowledge of how to sustain career satisfaction.

REFERENCE

¹ Frank, JR. (Ed). 2005. The CanMEDS 2005 physician competency framework. Better standards. Better physicians. Better care. Ottawa: The Royal College of Physicians and Surgeons of Canada.

4 Cataraqui Street, Suite 310, Kingston, ON K7K 1Z7
info@ccmg-ccgm.org | www.ccmg-ccgm.org | 613-507-8345

Updated – December 2013