CCMG Training and Specialty Requirements in Molecular Genetics  
Revised 2007

Preamble
The aim of the Molecular Genetics Training Program is to produce scientific specialists with the competence to effectively apply molecular diagnostic testing for disease diagnosis. Competence implies the individual has the knowledge, skills and attitudes to:

1) Identify and interpret molecular abnormalities.
2) Participate in the management of patients and their families with molecular genetic disorders.
3) Assume the day-to-day responsibilities for the operation and standards of a molecular genetics diagnostic laboratory.

The Molecular Geneticist will have a thorough grounding in the theory, methodology and techniques of human molecular genetics, and will be familiar with a broad spectrum of disorders representing all modes of inheritance typically encountered in the molecular diagnostic setting.

The CCMG training guidelines are modeled after the CanMEDS framework. 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 practice of molecular genetics.

Required Background
Trainees in CCMG-accredited programs must have either a PhD or an MD degree.

A candidate’s PhD must have a strong genetics content. The required educational experience would approximate that required for an MSc in genetics and molecular biology, or the laboratory specialty. If this experience is lacking, the trainee must gain this knowledge through suitable courses and/or private study.

Individuals with an MD degree must have completed at least 3 years of residency training in a program accredited by the Royal College of Physicians and Surgeons of Canada (RCPSC) and/or Collège des Médecins du Québec (CMQ).

Administrative Aspects
1. Supervisory committee:
   a. Each trainee’s program will be supervised by a committee, headed by a fellow of the CCMG in molecular 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 clinical geneticists and cytogeneticists, 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 Credentials Committee by submitting a registration form to the CCMG Secretariat 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, including graduation of responsibility in the laboratory and clinical setting. The committee must submit 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 Report (FITER) to the CCMG secretariat by August 1 of the final year of training.

2. **Location of training**
   a. Molecular genetics training must take place in a centre accredited by the CCMG in Molecular genetics.
   b. Elective training may be done at non-accredited centres at the discretion of the supervisory committee.
   c. In the event of termination of accreditation of a centre during the candidate’s training, the trainee will be allowed a maximum of six months to move to an accredited centre.

3. **Training in foreign centres**
   a. Training in American centres 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**
Two year program, including:

**Mandatory training:**
1. Minimum of **12 months** in a CCMG-accredited laboratory that provides molecular diagnostic services. Training will include:
   a. **Technical skills:** Bench experience in molecular diagnostic techniques, with a logbook recording involvement in a minimum of **100 cases**. It is the
responsibility of the local fellowship committee to determine if the trainee requires more cases to become familiarized with all technical aspects.

b. **Interpretative and consultative skills**: Experience in interpreting results and communicating to others, with a logbook recording a minimum involvement in **200 cases**. It is the sole responsibility of the local fellowship committee to determine the total number of cases to be reviewed by the candidate to ensure a high level of competence.

c. **Management skills**: Experience in management of a molecular diagnostic laboratory.

2. Rotations in other medical genetics specialties, including:
   a. **Cytogenetic laboratory training**: a minimum of **three months** to be spent in a CCMG-accredited laboratory providing cytogenetic diagnostic services.
   b. **Clinical genetics training**: a **minimum participation in 25 clinical genetics counseling sessions**, with a CCMG certified clinical geneticist, but not necessarily in a CCMG accredited centre. This may be reduced if the local fellowship committee deems that the candidate has sufficient relevant experience. The trainee should keep a logbook recording participation in a minimum of 25 counselling sessions, representing a variety of clinical scenarios. Participation must include developing an understanding of the issues through researching the literature and discussions with clinical colleagues.

3. Courses/conferences
   a. Documented participation in educational events and courses prescribed by the trainee’s supervisory committee.
   b. Documented attendance at one national or international genetics meeting each year.

4. Research training
   a. Minimum **six months** involvement in a research project related to molecular genetics or diagnostics, ideally culminating in submission of a publication to a refereed journal. The supervisory committee must approve the area of research. Given that promotion and career development will depend not only on clinical service responsibilities, but also on published research, research training should be encouraged to include clinical aspects of molecular genetics, in addition to basic science. Trainees with little or no previous research experience should be encouraged to take an extra year in research. The extra year need not be in an approved CCMG centre.

**Elective training:**
1. Rotations in related fields, such as molecular pathology, biochemical genetics, embryopathology, developmental genetics, relevant aspects of obstetrics.
2. Visits to other molecular diagnostic service laboratories or CCMG training centres.
Molecular 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.
Key Competencies

By the end of training, Molecular Genetics Trainees will demonstrate the ability to:
1. Explain advanced concepts in human molecular biology and genetics;
2. Define the pathobiology of human genetic disorders and their molecular genetic causes;
3. Design and implement effective molecular genetic testing;
4. Relate molecular genetic testing for human inherited disease to other molecular diagnostic testing applications;
5. Demonstrate expertise with standard and advanced molecular biology techniques.

Enabling Competencies

1. Explain advanced concepts in human molecular biology and genetics.
To achieve this, the Molecular Genetics Trainee will be able to:
   1.1. Describe general concepts of molecular biology and genetics;
   1.2. Explain details of the human genome and the structure of genetic material;
   1.3. Describe the progress and resources of the Human Genome project and molecular approaches used to address genome initiatives.

2. Define the pathobiology of human genetic disorders and their molecular genetic causes.
To achieve this, the Molecular Genetics Trainee will be able to:
   2.1. Explain the pathophysiology of inherited genetic disorders, particularly those amenable to molecular diagnosis;
   2.2. Explain the nature of human variation, including mutations, polymorphism and genome variation;
   2.3. Correlate genotype and phenotype for human genetic variation;
   2.4. Use mutation databases as a tool in interpretation of human genetic variation.

3. Design and implement effective molecular genetic testing.
To achieve this, the Molecular Genetics Trainee will be able to:
   3.1. Plan and interpret direct and indirect analysis for a variety of mutation types, including but not limited to:
       3.1.1. Recurrent point mutations (alteration of restriction sites polymorphisms, allele specific oligonucleotides, heteroduplexes, allele specific amplification, etc.);
       3.1.2. Deletions and duplication (semi-quantitative PCR, quantitative PCR, multiple ligation-dependent probe amplification etc.);
       3.1.3. Dynamic mutations (triplet repeat mutations, other repeat mutations etc);
       3.1.4. Imprinting mutations and other epigenetic modifications;
       3.1.5. Indirect molecular analyses based on linked markers.
   3.2. Compare the technical benefits and limitations of available genetic analysis methods;
3.3. Recognize and interpret artifacts or unusual results in molecular genetic test results and conduct appropriate investigations;

3.4. Demonstrate awareness of the variables that contribute to the quality of results and an ability to trouble shoot successfully;

3.5. Calculate genetic risk by inferential methods including linkage analysis, Bayesian probability, pedigree analysis and risk calculation in familial or potential new mutation situations;

3.6. Utilize electronic databases and resources to obtain information on genetic material and genetic variation;

3.7. Assign correct mutation nomenclature to genetic variations according to currently approved CCMG guidelines.

4. Relate molecular genetic testing for human inherited disease to other molecular diagnostic testing applications.

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

4.1. Describe variations detectable by molecular cytogenetic techniques and the significance of molecular cytogenetic mutations in human disease;

4.2. Explain the technical basis of molecular cytogenetic techniques;

4.3. Describe the difference between inherited and somatic mutations and the significance of somatic mutations in human disease;

4.4. Describe methods for detection of somatic mutations (loss of heterozygosity, gene dosage abnormalities, fusion gene expression etc);

4.5. Explain the complementary nature of different molecular diagnostic approaches.

5. Demonstrate expertise with standard and advanced molecular biology techniques.

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

5.1. Demonstrate expertise with standard molecular biology techniques including but not limited to:

5.1.1. Tissue culture, including sterile technique and culture of relevant cell types (i.e. amniocytes, chorionic villi, fibroblasts, lymphoblasts);

5.1.2. DNA and RNA isolation in full scale from sources such as blood, tissue, cultured cells; in small scale, from sources such as archival materials or blood spots;

5.1.3. Propagation and manipulation of organisms used as tools in molecular analysis;

5.1.4. Labeling techniques including isotopic and non-isotopic methods;

5.1.5. Southern blotting or equivalent.

5.2. Demonstrate expertise with advanced molecular biology techniques including but not limited to:

5.2.1. Polymerase Chain Reaction including real time PCR, quantitative PCR, and other technical applications of PCR;

5.2.2. Direct mutation scanning techniques, such as single strand confirmation polymorphisms, denaturing high performance liquid chromatography (DHPLC), protein truncation test etc;

5.2.3. DNA sequencing, including basic principles and use of automation.
COMMUNICATOR

Key Competencies

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

1. Provide consultation for molecular diagnostic cases to other health care providers, laboratory staff, patients and others;
2. Collate required clinical and other laboratory information for decisions regarding appropriate molecular genetic test utilization;
3. Report results and implications of molecular diagnostic testing to relevant individuals.

Enabling Competencies

1. Provide consultation for molecular diagnostic cases to other health care providers, laboratory staff, patients and others.
   To achieve this, the Molecular Genetics Trainee will be able to:
   1.1. Communicate with referring health care providers or other individuals to compile information required to assess appropriate molecular diagnostic investigations for clinical cases;
   1.2. Convey relevant information regarding molecular genetic testing possibilities to relevant individuals.

2. Collate required clinical and other laboratory information for decisions regarding appropriate molecular genetic test utilization.
   To achieve this, the Molecular Genetics Trainee will be able to:
   2.1. Recognize the importance of clinical or other laboratory information for cases referred for molecular diagnostic testing;
   2.2. Communicate effectively with relevant health care providers to obtain required information;
   2.3. Utilize clinical and other laboratory information to make decisions regarding appropriate molecular diagnostic testing to be performed.

3. Report results and implications of molecular diagnostic testing to relevant individuals.
   To achieve this, the Molecular Genetics Trainee will be able to:
   3.1. Correlate results with clinical or other laboratory information and provide relevant risk calculations in reports;
   3.2. Communicate molecular diagnostic results and implications in both oral and written forms;
   3.3. Maintain patient confidentiality and privacy in the reporting of results;
   3.4. Provide consultative services regarding implications of molecular testing results and additional recommended investigations.
Key Competencies

By the end of training, Molecular Genetics Trainees will demonstrate the ability to:
1. Participate effectively as a team member with relevant health care providers in collaborative decision making for molecular diagnostic cases;
2. Act to resolve conflict in interprofessional teams.

Enabling Competencies

1. Participate effectively as a team member with relevant health care providers in collaborative decision making for molecular diagnostic cases.
To achieve this, the Molecular Genetics Trainee will be able to:
1.1. Describe the role and responsibilities of a clinical molecular genetics professional to other health care providers;
1.2. Participate effectively as a team member in activities related to molecular genetic testing, including education, research and clinical care;
1.3. Demonstrate respect for other health care professionals and their role in health care teams.

2. Act to resolve conflict in interprofessional teams.
To achieve this, the Molecular Genetics Trainee will be able to:
2.1. Demonstrate appropriate conflict resolution skills;
2.2. Act to prevent and resolve conflicts with other health care professionals.
Key Competencies

By the end of training, Molecular Genetics Trainees will demonstrate the ability to:
1. Utilize molecular genetic testing resources effectively;
2. Manage staff, equipment and sample resources in a molecular diagnostic setting;
3. Promote quality control and quality assurance in molecular diagnostic testing;
4. Maintain complete and accurate records of molecular diagnostic testing;
5. Manage time effectively and prioritize required activities.

Enabling Competencies

1. Utilize molecular genetic testing resources effectively.
   To achieve this, the Molecular Genetics Trainee will be able to:
   1.1. Use molecular testing resources in a manner that balances costs with potential implications of results;
   1.2. Organize multiple molecular diagnostic investigations in an appropriate concurrent or sequential manner.

2. Manage staff, equipment and sample resources in a molecular diagnostic setting.
   To achieve this, the Molecular Genetics Trainee will be able to:
   2.1. Describe equipment and supplies used in molecular testing and their costs;
   2.2. Demonstrate the ability to prepare a laboratory budget;
   2.3. Demonstrate familiarity with bids and service contracts for laboratory equipment;
   2.4. Explain the technical training requirements for laboratory technologists;
   2.5. Develop and prepare appropriate laboratory protocols for molecular diagnostic laboratory staff;
   2.6. Describe the Workplace Hazardous Materials Information System (WHMIS) biohazard regulations and safe laboratory operating procedures;
   2.7. Recognize the sensitive nature of genetic samples and act to minimize potential harms.

3. Promote quality assurance in molecular diagnostic testing.
   To achieve this, the Molecular Genetics Trainee will be able to:
   3.1. Explain the concepts of laboratory quality assurance programs, including methods of implementation and monitoring;
   3.2. Describe issues in quality assurance that are unique to molecular diagnostic testing.

4. Maintain complete and accurate records of molecular diagnostic testing.
   To achieve this, the Molecular Genetics Trainee will be able to:
   4.1. Describe methods to implement and maintain an efficient system to manage laboratory information, data and reports;
   4.2. Maintain complete records for all cases, including both written and oral information;
4.3. Maintain confidentiality for all cases, including both oral and written communication;
4.4. Explain the medico legal implications in the practice of molecular genetics and appropriate use of medical records.

5. Manage time effectively and prioritize required activities.

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

5.1. Set, prioritize and manage time to balance required activities;
5.2. Recognize critical aspects of certain activities and allocate time appropriately.
HEALTH ADVOCATE

Key Competencies

By the end of training, Molecular Genetics Trainees will demonstrate the ability to:
1. Describe specific public health practices or policies that affect provision of molecular genetic testing services;
2. Respond to the health needs of individuals, communities and populations served by molecular diagnostic testing.

Enabling Competencies

1. Describe specific public health practices or policies that affect provision of molecular genetic testing services.
   To achieve this, the Molecular Genetics Trainee will be able to:
   1.1. Explain how health care governance influences resource allocation for molecular genetic services at the local, regional, provincial and national level;
   1.2. Describe the roles of national and international organizations in the determination of guidelines affecting molecular diagnostic testing;
   1.3. Participate in discussion regarding public policy and decision making processes with respect to current and future molecular diagnostic testing.

2. Respond to the health needs of individuals, communities and populations served by molecular diagnostic testing.
   To achieve this, the Molecular Genetics Trainee will be able to:
   2.1. Become informed about community resources and related patient support groups for individuals served by molecular diagnostic testing;
   2.2. Liaise effectively with individuals, communities and populations on issues applicable to molecular diagnostic testing;
   2.3. Act as a resource and information source regarding molecular diagnostic testing for individuals, communities and populations.
Key Competencies

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

1. Conduct ongoing learning activities to maintain and advance their professional knowledge;
2. Facilitate the learning of other health care professionals, students, laboratory staff, the public and other regarding molecular diagnostic testing;
3. Conduct research projects and publish findings for advancement of knowledge.

Enabling Competencies

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

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

1.1. Critically assess the literature as related to human genetics, molecular biology and diagnostics;
1.2. Demonstrate commitment to continuing education events, including conferences, rounds, clinical and research seminars, and patient conferences;
1.3. Recognize limitations of current knowledge and plan appropriate additional educational activities.

2. Facilitate the learning of other health care professionals, students, laboratory staff, the public and others regarding molecular diagnostic testing.

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

2.1. Deliver effective lectures and presentations on human genetics and molecular biology concepts;
2.2. Present concise and audience appropriate summaries of molecular diagnostic methodologies, clinical situations related to molecular diagnostic testing and case reports or presentations.

3. Conduct research projects and publish findings for advancement of knowledge.

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

3.1. Plan and conduct a minimum six month research project related to human molecular genetics or molecular diagnostic testing;
3.2. Summarize results and submit for publication in a referred journal;
3.3. Identify possible personal limitations with respect to previous research experience and recognize that an additional year of research may be beneficial in certain situations for promotion and career development.
PROFESSIONAL

Key Competencies

By the end of training, Molecular Genetics Trainees will be able to:

1. Demonstrate ethical practices and a sense of responsibility in molecular diagnostic testing;
2. Demonstrate appropriate respectful behavior consistent with a clinical molecular diagnostician role.

Enabling Competencies

1. Demonstrate ethical practices and a sense of responsibility in molecular diagnostic testing.
   To achieve this, the Molecular Genetics Trainee will be able to:
   1.1. Maintain confidentiality and ensure appropriate release of molecular diagnostic samples, data and reports;
   1.2. Recognize ethical issues in molecular diagnostic testing, 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;
   1.3. Identify personal limitations and the necessity of seeking the opinions of colleagues or other professionals when required;
   1.4. Identify technical laboratory limitations requiring referral to other laboratories.

2. Demonstrate appropriate respectful behavior consistent with a clinical molecular diagnostician role.
   To achieve this, the Molecular Genetics Trainee will be able to:
   2.1. Demonstrate a professional attitude to clinical and laboratory colleagues, to laboratory staff, students and trainees, and patients.

REFERENCE