Application Form for Biochemical Laboratory Genetics Fellowship

Name of Institution: McGill University Health Centre
Location: Montreal, QC
Number of positions: One every two years
Length: 2 years (maximum) (Length of training to be assessed by the Canadian College of Medical Geneticists (CCMG), which certifies the training

Program Information (append description):
Number of fellowship positions requested: One every two years
Academic affiliation: McGill University Health Centre (MUHC)

- Name of hospitals involved in training: insert % time spent by the fellow in each institution

McGill University Health Centre (MUHC) – 75% (minimum)
Biochemical Genetics Laboratory, Cytogenetics and Molecular Laboratory, Clinical Genetics and Clinical Biochemical Genetics

Specific rotations in laboratories of non-affiliated hospitals:
CHU Ste-Justine -4%
CHU Sherbrooke- 8%
Children's Hospital of Eastern Ontario- 4%
CHU Quebec -1%
Electives and Research – MUHC or other laboratories -9%

- % time spent by the fellow in each institution

Background:

McGill University has long been a training site for clinical genetics and laboratory genetics fellowships that are certified by the CCMG. We would like for these fellowships to be recognized by the Postgraduate Medical Education Office.

The Laboratory Biochemical Genetics Fellowship is **2-year** training program that is certified by the Canadian College of Medical Geneticists (CCMG). Trainees are expected to participate fully in all aspects of laboratory medicine as it relates to biochemical genetics, multi-disciplinary case discussion, rounds, seminars and meetings related to clinical and biochemical genetics. There is increasing responsibility over the 2 year period to include more independence in result interpretation, test development, lab management, quality assurance and other competencies as outlined in the Training Objectives below.

Research: The trainee will participate in a research project that will be identified in the second year of training. Research expertise in the Department of Medical Genetics includes Peroxisomal Disorders (Dr. Nancy Braverman), B12 and folate disorders (Dr. David Rosenblatt). There are other biochemical genetics related research opportunities in other departments.

Mission: To provide the trainees with a consultant level of expertise in biochemical genetic test interpretation, test development, lab management, quality assurance and other competencies.

Outline: This fellowship will enhance the residency training of the fellow by providing training in provision of specialized laboratory techniques that will allow the fellow to supervise technicians in a biochemical genetics laboratory or to establish their own biochemical genetics laboratory.

Name of the Fellowship Program Director: Dr. Daniela Buhas

Names of the Teaching Faculty

- D. Buhas, N. Braverman, Y. Trakadis, F. Parente, B. Gilfix, D. Rosenblatt: biochemical laboratory test methods and interpretation
- J. Lavoie and M. Blumenkrantz : Cytogenetics laboratory training and result interpretation
- A. Ruchon and I. DeBie: Molecular Genetics laboratory training and result interpretation
- D. Buhas, N. Braverman, J. Mitchell ,Y. Trakadis, I. DeBie, L. Russell, D. D'Agostino: clinical and biochemical genetics
- CHU Sherbrooke--P. Waters and C. Aurais-Blais: biochemical laboratory test methods and interpretation
- CHU Ste-Justine--P. Allard and C. Brunel-Guitton: biochemical laboratory test methods and interpretation
- Chu Quebec--Y. Giguere and M-T. Berthier: biochemical laboratory test methods and interpretation
- CHEO--N. Lepage and O. Al-Dirbashi: biochemical laboratory test methods and interpretation

The teaching faculty has a broad range of expertise in a number of different types of laboratory biochemical genetics, molecular genetics and cytogenetics. A number of the teaching faculty also have extensive experience in pediatric and adult clinical and biochemical genetics. The clinical and laboratory faculty collaborate closely with respect to patient care and training of residents and fellows.

Academic Facilities

The MUHC offers a variety of biochemical genetic laboratory investigations. The clinical investigations include plasma and urine amino acids, acylcarnitine profile, transferrin isoelectric focusing, vitamin B12 and folate disorders, homocysteine and disorders of porphyrin metabolism. As well, there is research laboratory for peroxisomal disorders. These laboratories maintain a collection of teaching cases, consisting of patient clinical presentation and laboratory abnormalities.

The MUHC also has laboratories for molecular genetics and cytogenetics. As well, the MUHC Cell Bank serves as repository for fibroblasts and other tissues obtained from patients with a variety of disorders. In addition to these laboratory facilities, the clinical teams (physician, genetic counsellors, nurse and metabolic dietician) of the MUHC Department of Medical Genetics hold outpatient clinics a minimum of four days a week. These clinics serve patients of all ages with known or suspected genetic conditions, including those with inborn errors of metabolism. The care of the latter patients is discussed weekly at a case review conference. A weekly Academic Half-Day includes Basic Science seminars, Clinical Case Presentation, Journal Club and clinically oriented Resident Teaching.

The fellows and other trainees have full time electronic access to the McGill University libraries. As well, the Department of Medical Genetics maintains its own small library of books relevant to many aspects of medical genetics. Finally, the Department offers full time access to POSSUM, a database of physical traits and syndromes.

Fellow Duties and Responsibilities

- Call responsibilities to cover service: *None*
- The fellow is the senior supervisor of residents: *Only during the residents' biochemical laboratory rotations and the fellow's second year of training.*
- Outline whether there are fixed rotations at the various institutions: *Yes*
- Outpatient clinic responsibilities need to be outlined: *The fellow will observe in outpatient clinics but will have no direct clinical responsibilities.*
- Outline the role of the fellow towards residents on service: *The fellow will discuss abnormal biochemical laboratory results with residents on service, under the supervision of staff in Biochemical Genetics*
- Teaching responsibilities towards residents: *Residents take a two-month Biochemical Genetics laboratory rotation. The fellow will teach these residents the technical and analytic aspects of biochemical laboratory tests.*
- Outline participation in academic activities involving the residents: seminars, outcome assessment (morbidity and mortality rounds etc). *The fellow will attend the weekly Academic Half Day and will give Journal Club at least once per year. He will also attend the weekly case discussion of patients with inborn errors of metabolism; as part of his participation in this meeting, he will develop and present one or more topics relative to his interests. At the end of the fellowship, he will present a seminar that summarizes his fellowship project.*
- Describe any support staff available to the fellow: program coordinator, nurse clinician, secretarial *The fellow has support from the technicians in the various laboratories, including the Biochemical Genetics laboratory. Once the fellowship is approved, the fellow will have the support of a program coordinator.*
- Proposed meetings to be attended by the fellow *The fellow will attend the weekly case discussion of patients with inborn errors of metabolism and the weekly Academic Half Day. He will also attend the weekly discussion of abnormal biochemical laboratory test results that are pertinent to his training. The fellow will also attend an annual genetics meeting of his choice, such as the Garrod Association or the Society for Inborn Metabolic Disorders.*
- Research productivity and publications expected by the Fellow. *The fellow will develop a laboratory research project under the supervision of his training committee. The fellow will be encouraged to present this research project at a scientific meeting or to prepare a manuscript that is suitable for publication.*

Curriculum

- Intended case load: *The fellow will maintain a logbook of hands-on laboratory experience and methods.*
  The fellow will maintain a separate logbook of interpretation patient diagnostic laboratory tests, including teaching cases. A minimum of 200 such cases covering a broad range of inborn errors of metabolism is required.
  The fellow will observe a minimum of 40 clinical cases covering a broad range of inborn errors of metabolism and other genetic conditions and will record these cases in a logbook.

The fellow will observe a minimum of 10 cases of newborn screening, following the case from initial referral to the metabolic service to the final evaluation and counselling. These cases will be recorded in the fellow's logbook.

- Intended Percentage of varieties of cases. *Not specified but exposure should be broad.*
  Regular reading materials provided (if any) :

1. *Techniques in diagnostic human biochemical genetics A Laboratory Manual, edited by Frits A. Hommes, Wiley-Liss New York 1991.*
2. *Physician's guide to the Laboratory diagnosis of metabolic diseases, edited by N Blau*
3. *Inborn metabolic diseases - diagnosis and treatment, edited by J Fernandes, J-M Saudubray and Van den Berghe*
4. *A clinical guide to inherited metabolic diseases, edited by JTR Clarke (second edition)*
5. *Inherited Metabolic Diseases, edited by GF Hofmann (a copy will be provided)*
6. *Scriver's Online Metabolic and Molecular Bases of Inherited Disease,edited by Valle, Beaudet,Vogelstein, Kinzler, Antonarakis,and Ballabio(online access provided)*

- Conference weekly schedules *The fellow will attend the weekly case discussion of patients with inborn errors of metabolism and the weekly Academic Half Day. He will also attend the weekly discussion of abnormal biochemical laboratory test results that are pertinent to his training.*
- Role of the fellow in attending, presenting, supervising, organization *See above under Fellow Duties and Responsibilities*

Canadian College *of* Medical Geneticists
Collège canadien *de* généticiens médicaux

CCMG Laboratory Biochemical Genetics Training Guideline and Specialty Requirements

Preamble

The aim of the Laboratory Biochemical Genetics Training Program is to produce scientific specialists who are competent to effectively apply biochemical genetic testing for the diagnosis and management of individuals of all ages with inherited metabolic disease. Competence implies the individual has the knowledge, skills and attitudes to:

1) Identify and interpret investigations of inborn errors of metabolism.
2) Participate in the management of patients and their families with metabolic diseases.
3) Assume the day-to-day responsibilities for the operation and standards of a biochemical genetics laboratory.

The Laboratory Biochemical geneticist will have a thorough grounding in the theory, methodology and techniques of biochemical genetic analysis, and will be familiar with a broad spectrum of disorders representing all modes of inheritance and indications typically encountered in the biochemical genetic setting.

Trainees are expected to participate fully in all aspects of laboratory medicine as it relates to biochemical genetics, multi-disciplinary case discussion, rounds, seminars and meetings related to clinical and biochemical genetics. There is increasing responsibility over the 2 year training period to include more independence in result interpretation, test development, lab management, quality assurance and other competencies as outlined here in the Training Guideline.

The CCMG training guidelines are modeled after the CanMEDS framework[1]. 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 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 laboratory practice of biochemical genetics.

Required Background

Trainees must have a PhD and/or an MD degree.

A candidate's PhD must be in a relevant basic science discipline (e.g. biochemistry, chemistry or human genetics) or Clinical Chemistry (FCACB qualified or Board eligible). A candidate's MD specialty must be in Medical Biochemistry (FRCPC qualified or Board eligible).

PhD trainees must have successfully defended and submitted the final version of their PhD thesis before beginning CCMG training. The start date for training can not be before submission of the final version, with all revisions (if any) approved by the relevant university officer(s).

Clinical Chemists and Medical Biochemists are not eligible for credit and are required to complete the 2 year CCMG Laboratory Biochemical Genetics fellowship program.

Administrative Aspects

1. **Supervisory committee:**
   a. Each trainee's program will be supervised by a committee, headed by a Biochemical Genetics Laboratory Director who must be a fellow of the CCMG certified in Biochemical Genetics or Laboratory Biochemical Genetics.
   b. Biochemical Genetics Laboratory Director who must be a fellow of the CCMG certified in Biochemical Genetics or Laboratory Biochemical Genetics.
   c. The committee will consist of the head and a minimum of two additional members. Other members might consist of clinical specialists with experience in care of patients with inborn errors of metabolism, clinical geneticists, or clinical chemists, although the structure of the committee can vary depending on the background of the trainee.
   d. The program director or supervisor on behalf of the committee ensures the trainee is registered with the CCMG Credentials Committee by submitting a registration form to the CCMG Secretariat within the first three months of commencing training.
   e. The committee takes responsibility for ensuring the training program is meeting the needs of the trainee and 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.
   f. The committee meets every six months with the candidate, and ensures that in-training evaluation forms (ITER; one for each rotation and one for every 6 months for longer rotations) are completed and discussed with the trainee. If remedial work is needed by the trainee, the committee must ensure that this is provided.
   g. The committee completes and submits the Final In-Training Evaluation Report (FITER) to the CCMG secretariat prior to oral examination. Please note that the FITER is additional to the ITER covering the last 6 months of training. The committee must also submit a Completion of Training form, if the FITER does not coincide with the completion of training.

2. **Location of training**
   a. Laboratory Biochemical Genetics training must take place in a center accredited by the CCMG for training in laboratory Biochemical Genetics.
   b. Elective training may be done at non-accredited centres at the discretion of the supervisory committee.

c. In the event that accreditation of a center is terminated 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 centres accredited by the American Board of Medical Genetics (ABMG) is recognized by the CCMG.
b. As the ABMG and CCMG have different training and credentialing requirements, it is the responsibility of the trainee to ensure completion of all requirements if the trainee's intention is to sit both ABMG and CCMG examinations.

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.

5. Second specialty training

CCMG fellows qualified or currently in clinical Biochemical Genetics who want to certify in laboratory Biochemical Genetics must complete a minimum of 16 months training in a CCMG-accredited Biochemical Genetics laboratory. All training as outlined below in Mandatory training item 1 must be completed. The Supervisory Committee may recommend additional training depending upon previous training and experience

6. Credentialing

Candidates are advised to review the Credentialing requirements on the website early in their training to facilitate Credentials submission and review.

Content of Training

Two year program, including:

Mandatory training:

1. **Minimum of 16 months in a CCMG-accredited laboratory directed by a fellow of the CCMG certified in Biochemical Genetics.** *Training sites must ensure compliance with their laboratory accreditation programs. This may require bench work performed by a trainee be performed using non-clinical samples or under supervision of appropriately certified individuals at the discretion of the training center and supervised result reporting (co-signed) or the use of archival cases.*

   It is recognized that not all training centres will be able to provide a fully-comprehensive training program. Those responsible for training at a given center must identify any 'gaps' and encourage trainees to obtain the appropriate training at other CCMG- or ABMG-accredited centres to meet the overall training objectives of the program.

   Training will include:

a. **Technical skills**: Hands-on experience with all wet and dry bench biochemical genetic techniques, with a logbook recording **150 cases** (maximum 20 cases for any specific test) in

which there was significant participation of the trainee in the laboratory. Cases must demonstrate experience with a variety of techniques including:

- Cell culture (e.g. skin fibroblast)
- Cell isolation (WBC preparation)
- Fluorometry
- Electrophoresis or related techniques (e.g. isoelectric focusing)
- Chromatography (thin layer, ion exchange, HPLC)
    - Amino acid analysis
    - Urine glycosaminoglycans
    - Oligosaccharide analysis
- Mass spectrometry (GC-MS, MS/MS)
    - Organic acids
    - Acylcarnitines
    - Carnitine, total and free
    - Very long chain fatty acids / phytanic acids
- Enzyme activity assay (lysosomal 4-MU assays)

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. Interpretive and consultative skills: Experience in interpreting results and communicating to others, with a logbook recording involvement in **200 cases** (though it is likely that more than 200 cases will be reviewed). 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.

Case distribution: The cases documented must demonstrate a variety of indications, analysis of different specimen types, and biochemical genetic techniques.

Abnormal/Illustrative cases: At least 120 of the cases must represent abnormal results. Abnormal results include both IEM diagnostic patterns and biochemical abnormalities that might suggest secondary biochemical/nutritional disturbances or artefactual findings. In order to meet the abnormal/illustrative minimum, archival material (inactive cases illustrative of a rare abnormal that is unlikely to be seen during the training period) or external proficiency results may be used (to a maximum of 40 archival cases). Abnormal results observed during technical experience can also be logged.

c. Management skills: Experience in management of a biochemical genetics diagnostic laboratory.

2. Rotations in other medical genetics specialties, including:

a. **Cytogenetic and Molecular genetic laboratory training**: a minimum of **two months** to be spent in CCMG-accredited laboratories providing cytogenetic and molecular diagnostic services.
b. **Clinical Biochemical Genetics training**: a minimum participation in **40 patient encounters** (counseling sessions), with a metabolic physician, CCMG/ABMG-certified clinical geneticist or CAGC/ABMG-certified genetic counselor, but not necessarily in a CCMG-accredited centre. This rotation can occur as a block or throughout the training program. The trainee should keep a logbook recording participation in a minimum of 40 patient encounters, representing at least 25 metabolic cases, 5 prenatal cases (not necessarily metabolic) and 10 newborn screening follow up cases. Participation must include developing an understanding of the issues through researching the literature and discussions with clinical colleagues.

3. Courses/conferences
   a. Participation in educational events and courses prescribed by the trainee's supervisory committee is to be documented in an education logbook.
   b. Documented attendance at one local (e.g. departmental annual research day, a research institute research day), national or international genetics meeting during the training period.

4. Research training
   a. **The trainee must participate in clinical or laboratory-based research for a period equivalent to 6 months full time research**; this may be accomplished through a dedicated 6 month block of time or distributed throughout the training period. 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. The research can be applied or translational in nature such as development of a new test, test validation, test improvement, quality improvement, case follow up and cost-benefit analysis.
   b. A research supervisor must be identified and the proposed research objectives and methodology are to be submitted to the supervisory committee for review and approval. The research supervisor is responsible for completing an evaluation (ITER) of the trainee. The trainee is responsible for a written summary of completed research to be submitted to Program director and dissemination to the appropriate audience (laboratory staff, clinicians, publication).

5. Logbooks
   CCMG Logbook templates available on the CCMG website **must** be used. Patient confidentiality must be guarded. Therefore, before submitting to the CCMG, cases must have all identifiers removed so as not to be traceable. Each Logbook is in an Excel format with tabs for documentation of:
   a. Technical cases (tab: Technical Log)
   b. Consultative/interpretive cases (tab: Consultative Log)
   c. Clinical experience (tab: Clinical Log)
   d. Education experience (tab: Education Activities)
   e. Research experience (tab: Research Activities)

f. Training program (tab: Training Outline)

A logbook should not only be viewed as a mechanism for tracking the number of cases/experiences accumulated but as a means for documenting learning, illustrative cases and approaches/troubleshooting undertaken that can be reflected upon and recalled as you study for the CCMG exam and as you start your career as a laboratory geneticist. The logbook should be reviewed regularly and discussed by the supervisor and trainee to ensure they represent the breadth of testing required and acquisition of competencies.

Biochemical Genetics Specialty Requirements

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 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

Key Competencies

By the end of training, the Laboratory Biochemical Genetics trainee will be able to:

1. Explain advanced concepts in cell biology, human biochemistry and medical genetics;
2. Define the pathobiology of metabolic disorders, their biochemical genetic causes and treatment;
3. Relate biochemical genetic testing for human inherited disease to other biochemical genetic testing applications;
4. Demonstrate expertise with standard and advanced biochemical genetic techniques.
5. Demonstrate the ability to implement effective biochemical genetic testing;

Enabling Competencies

1. Explain advanced concepts in cell biology, human biochemistry and human genetics
To achieve this, the Laboratory Biochemical Genetics traineewill be able to:

1.1 Discuss the general concepts of medical genetics outlined in the CCMG General Knowledge Guidelines.

1.2. Describe and discuss general concepts of human biochemistry and molecular biology, including:

1.2.1. The principles of enzyme kinetics and measurement of enzyme activity;

1.2.2. Regulation of intermediary metabolism including biochemical and hormonal regulation, and tissue compartmentalization.

1.3. Describe and discuss general concepts of human physiology and biochemistry including:

1.3.1. Fluid and electrolyte balance, acid-base regulation, intermediary metabolism and metabolic response to fasting

1.3.2. The function, and functional anatomy, of major organ systems

1.4. Understand principles of normal nutrition and consequences of under-nutrition and specific nutritional deficiencies

2. Define the pathobiology of metabolic disorders, their biochemical genetic causes and treatment;

To achieve this, the Laboratory Biochemical Genetics trainee will:

2.1. Demonstrate an understanding of the pathological and biochemical changes, clinical symptoms, investigations and management in metabolic disorders of the following pathways:

- 2.1.1. Disorders of amino acid metabolism
- 2.1.2. Disorders of organic acid metabolism
- 2.1.3. Hyperammonaemia and urea cycle disorders
- 2.1.4. Disorders of carbohydrate metabolism: glycogen storage disease, galactosemia, etc.
- 2.1.5. Disorders of fatty acid oxidation and carnitine metabolism
- 2.1.6. Disorders of ketone body metabolism
- 2.1.7. Lysosomal storage disorders
- 2.1.8. Peroxisomal disorders
- 2.1.9. Disorders of purine and pyrimidine metabolism
- 2.1.10. Respiratory chain disorders and disorders of pyruvate metabolism
- 2.1.11. Porphyrias
- 2.1.12. Disorders of cholesterol, sterol and bile acid metabolism
- 2.1.13. Disorders of vitamin metabolism (e.g., cobalamin)
- 2.1.14. Disorders of creatine metabolism
- 2.1.15. Defects of membrane transport: cystinuria, lysinuric protein intolerance, etc.
- 2.1.16. Disorders of glycosylation
- 2.1.17. Disorders of neurotransmitters

2.2. Understand the principles of treatment related to inborn errors of metabolism

- 2.2.1. Demonstrate knowledge of the general principles of dietary management in inborn errors of metabolism
- 2.2.2. Demonstrate knowledge of the appropriate indications for emergency /crisis management of metabolic disorders
- 2.2.3. Demonstrate knowledge of the underlying principles of various dietary or pharmacological treatment strategies employed for metabolic disorders including substrate reduction, correcting co-factor or product deficiency, providing alternative substrates/promoting alternative pathways, blocking effects of toxic metabolites, stimulating residual enzyme activity, enzyme replacement, organ transplantation, gene therapy.

2.3. Describe normal and abnormal biochemical phenotypic variation including secondary causes of abnormal biochemical phenotypes (e.g., secondary causes of lactic acidemia).

3. Relate biochemical genetic testing for human inherited disease to other biochemical genetic testing applications;

To achieve this, the Laboratory Biochemical Genetics trainee will:

3.1. Understand principles of newborn screening and demonstrate the ability to interpret results and select appropriate follow-up testing algorithms;

3.2. Describe the principles and practice of carrier testing: Ashkenazi Jewish population, other ethnic groups where appropriate;

3.3. Describe the use of biochemical genetic testing for the monitoring of metabolic disease.

4. Demonstrate expertise with standard and advanced biochemical genetic techniques.

To achieve this, the Laboratory Biochemical Genetics trainee will:

4.1. Understand methodological basis and demonstrate proficiency with the following techniques:

- 4.1.1. Reagent preparation, storage and handling
- 4.1.2. Sample preparation, isolation, concentration, extraction, purification
- 4.1.3. Stability/storage of patient tissues and other specimens
- 4.1.4. Cell isolation (WBC preparation)
- 4.1.5. Spectrophotometry
- 4.1.6. Fluorometry
- 4.1.7. Electrophoresis or related techniques (isoelectric focusing)
- 4.1.8. Chromatography (thin layer, ion exchange, HPLC)
- 4.1.9. Mass spectrometry (GC-MS, MS/MS)

4.2. Understand the methodological basis and demonstrate proficiency with the biochemical genetics analysis of:

- 4.2.1. Carnitine, total and free
- 4.2.2. Urine glycosaminoglycans and oligosaccharide analysis
- 4.2.3. Organic acids: gas chromatography/mass spectrometry
- 4.2.4. Very long chain fatty acids /phytanic acids
- 4.2.5. Amino acid analysis
- 4.2.6. Acylcarnitines
- 4.2.7. Enzyme activity (lysosomal 4-MU assays)
- 4.2.8. Tissue culture

4.3. Recognize the limitations of each biochemical genetic technique;

4.4. Troubleshoot biochemical genetic techniques.

5. Demonstrate the ability to implement effective biochemical genetic testing;

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

5.1. Describe the indications for performing biochemical genetic testing. This should be evidence-based and best practice, taking into consideration current CCMG guidelines and scientific literature;

5.2. Formulate an appropriate metabolic differential diagnosis, and plan an appropriate course of investigation with respect to the following:

- 5.2.1. Acute encephalopathy
- 5.2.2. Chronic encephalopathy, including leukodystrophies

5.2.3. Liver disease including acute liver failure, intrahepatic cholestasis
5.2.4. Organomegaly
5.2.5. Cardiomyopathy
5.2.6. Eye disease: corneal clouding, cataract, retinal changes, optic neuropathy
5.2.7. Specific dysmorphic syndromes associated with inborn errors of metabolism
5.2.8. Skeletal abnormalities of metabolic disorders (e.g., dysostosis multiplex, rhizomelic chondrodysplasia punctata, frequent fractures)
5.2.9. Hair and skin manifestations of metabolic disorders (e.g., ichthyosis, angiokeratoma, lipomatosis)
5.2.10. Malodour (e.g., trimethylaminuria)
5.2.11. Renal disorders: Fanconi syndrome, recurrent renal calculi
5.2.12. Muscle disease: myopathy, exercise induced rhabdomyolysis
5.2.13. Sudden Unexpected death
5.2.14. Hypoglycemia
5.2.15. Metabolic acidosis
5.2.16. Hyperammonemia
5.2.17. Lactic acidemia

5.3. Demonstrate proficiency in the interpretation of results from:
5.3.1. Amino acids analysis
5.3.2. Organic acids analysis
5.3.3. Intermediary metabolites (glucose, ammonia, lactate, pyruvate, free fatty acids, ketones) analysis
5.3.4. Carnitine / acylcarnitines analysis
5.3.5. Enzymes studies: specific and non-specific assays (flux studies), respiratory chain enzymes
5.3.6. Glycosaminoglycans and oligosaccharides analysis
5.3.7. Transferrin isoform analysis
5.3.8. Peroxisomal metabolites (VLCFA, phytanic, plasmalogens, bile acids, pipecolic acid) analysis
5.3.9. Neurotransmitter and biogenic amine metabolite analysis from CSF and other fluids
5.3.10. Purines and pyrimidines analysis
5.3.11. Creatine and creatine metabolites (guanidinoacetate) analysis

5.4. Advise on indications, patient preparation, and result interpretation of:
5.4.1. Loading, fasting, and other challenge tests for disorders of intermediary metabolism
5.4.2. Metabolic autopsy

5.5. Advise on the influence of clinical context and pre-analytical variables on test results: fasting, nutritional status, medications, age, gender, pregnancy, sampling procedure, etc.

5.6. Understand the appropriate use of testing for long-term monitoring of metabolic disorders

5.7. Demonstrate an understanding of the role of screening tests vs. diagnostic tests

5.8. Advise on appropriate selection of reference labs for testing not offered on-site and demonstrate an understanding of the interpretation of results from reference labs

5.9. Recommend other genetic/non-genetic laboratory testing that would be of clinical benefit;
5.10. Use the medical/scientific literature when compiling a report;
5.11. Develop and validate a diagnostic test according to accreditation standards using standard test development procedures (e.g., CLSI document EP10-A3, "Preliminary Evaluation of Quantitative Clinical Laboratory Measurement Procedures; Approved Guideline, 3ed. 2006). This will include the appropriate interpretation of results during the course of test development, planning for subsequent experiments, and documentation of all experiments in a final report according to a standard laboratory format.

COMMUNICATOR

Key Competencies

By the end of training, the Laboratory Biochemical Genetics trainees will demonstrate the ability to:

1. Provide consultation for biochemical genetic cases to health care providers, laboratory staff, patients and others;
2. Integrate clinical and laboratory information to assist with result interpretation and decision making for appropriate biochemical genetic test utilization;
3. Report results and interpretation of metabolic testing to relevant individuals.

Enabling Competencies

1. Provide consultation for biochemical genetic cases to health care providers, laboratory staff, patients and others.

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

1.1 Communicate with referring health care providers or other individuals to compile information required to assess appropriate metabolic diagnostic investigations for clinical cases;

1.2 Convey relevant information regarding biochemical genetic testing possibilities to relevant individuals.

2. Integrate clinical and laboratory information to assist with result interpretation and decision making for appropriate biochemical genetic tests.

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

2.1. Recognize the importance of clinical or other laboratory information for cases referred for biochemical genetic 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 biochemical genetic testing to be performed.

3. **Report results and interpretation of biochemical genetic testing to relevant healthcare providers.**
   To achieve this, the Laboratory Biochemical Genetics trainee will be able to:
   3.1. Correlate results with clinical and/or other laboratory information;
   3.2. Communicate biochemical genetic results and interpretation in both oral and written forms;
   3.3. Use standard formats and include clinically relevant comments in biochemical genetic reports;
   3.3 Maintain patient confidentiality and privacy in the reporting of results;
   3.4 Provide consultative services regarding the appropriate biochemical genetic tests and any other additional investigations.

COLLABORATOR

Key Competencies

By the end of training, the Laboratory Biochemical Genetics trainees will demonstrate the ability to:

1. Participate effectively as a team member with relevant health care providers in collaborative decision-making for metabolic cases;
2. Mediate decision-making in inter-professional teams;
3. Contribute effectively to other interdisciplinary team activities.

Enabling Competencies

1. **Participate effectively as a team member with relevant health care providers in collaborative decision making for metabolic cases.**
   To achieve this, the Laboratory Biochemical Genetics trainee will be able to:
   1.1. Describe the role and responsibilities of a biochemical genetics professional to other health care providers;
   1.2. Develop rapport, trust and ethical relationships with the health care team;
   1.3. Participate effectively as a team member in activities related to biochemical genetic testing, including education, research and clinical care;
   1.4. Demonstrate respect for other health care professionals and their role in health care teams;
   1.5. Network with other biochemical genetic laboratories.

2. **Mediate decision-making in inter-professional teams.**
   To achieve this, the Laboratory Biochemical Genetics trainee will be able to:
   2.1. Demonstrate appropriate conflict resolution skills;
   2.2. Facilitate communication within inter-professional teams to prevent and resolve conflicts.

3. **Contribute effectively to other interdisciplinary team activities.**

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

3.1. Participate in an interdisciplinary team meeting, and demonstrate the ability to consider and respect the opinions of other team members, while contributing biochemical genetics-specific expertise him/herself;

3.2. 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.

MANAGER

Key Competencies:

By the end of training, the Laboratory Biochemical Genetics trainee will demonstrate the ability to:

1. Understand and apply the essential elements of Quality Management system within the laboratory;
2. Utilize biochemical genetic testing resources effectively;
3. Manage staff and equipment to work effectively and efficiently in a health care organization;
4. Manage time effectively and prioritize required activities.

Enabling Competencies

1. Understand and apply the essential elements of a Quality Management system within the laboratory.

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

1.1. Explain the concepts of laboratory quality assurance programs, including methods of implementation and monitoring;

1.2. Understand the principles of quality control (including, but not limited to, statistical quality control), and be able to develop an effective quality control program, to interpret quality control data and to recommend appropriate corrective action;

1.3. Demonstrate understanding of Lab standards and guidelines such as external proficiency programs, turnaround times, etc.;

1.4. Understand the administrative, legal and physical requirements for the operation of a hospital biochemical genetics laboratory including:

1.4.1. The provincial requirements for accreditation of biochemical genetic laboratories and general hospital laboratories;

1.4.2. The biochemical genetic requirements for CCMG-accreditation of centres.

1.5. Maintain complete and accurate records of biochemical genetic testing.

1.5.1. Describe methods to implement and maintain an efficient system to manage laboratory information, data and reports;

1.5.2 Maintain complete records for all cases, including written, electronic and oral information;

1.5.3 Demonstrate knowledge of laboratory information systems for patient tracking and record maintenance;

1.5.4 Maintain confidentiality for all cases, including both oral and written communication;

1.5.5. Explain the medico-legal implications in the practice of biochemical genetics and appropriate use of medical records.

1.6. Understand the importance of Continuous Quality Improvement (CQI) as it applies to lab policies, processes and procedures;

1.7. Describe issues in quality assurance that are unique to biochemical genetic testing.

1.8. Demonstrate the ability to respond effectively to laboratory-related complaints.

2. Utilize biochemical genetic testing resources effectively.

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

2.1. Use biochemical genetic testing resources in a manner that balances costs with potential implications of results;

2.2. Organize multiple biochemical genetic investigations in an appropriate concurrent or sequential manner;

2.3. Coordinate biochemical genetic testing with other diagnostic investigations;

2.4. Determine what constitutes an unacceptable or suboptimal specimen or result;

2.5. Determine what constitutes the most appropriate specimen for a specific diagnostic question and how such a specimen should be procured and processed in the biochemical genetics laboratory;

2.6. Recognize the sensitive nature of genetic samples and act to minimize potential harms;

2.7. Advise on the principles of establishing new technologies in the biochemical genetics laboratory including but not limited to the cost/benefit ratio, sensitivity and specificity of the test and the validation of the test method and the results;

2.8. Demonstrate knowledge of the CCMG Biochemical Genetics Practice Guidelines.

3. Manage staff and equipment to work effectively and efficiently in a health care organization.

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

3.1. Describe equipment and supplies used in biochemical genetic testing and their costs;

3.2. Demonstrate the ability to prepare a laboratory budget;

3.3. Demonstrate familiarity with bids and service contracts for laboratory equipment;

3.4. Explain the technical training requirements for laboratory technologists;

3.5. Develop appropriate laboratory protocols for laboratory staff;

3.6. Demonstrate the ability to effectively prioritize the work of laboratory staff;

3.7. Describe the Workplace Hazardous Materials Information System (WHMIS) biohazard regulations and safe laboratory operating procedures;

3.8. Demonstrate understanding of the process of staff recruitment and interview skills;

3.9. Demonstrate understanding of the accreditation process and understand the process for responding to reviewers recommendations.

4. Manage time effectively and prioritize required activities.

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

4.1. Set, prioritize and manage time to balance required activities;

4.2. Recognize critical aspects of certain activities and allocate time appropriately.

HEALTH ADVOCATE

Key Competencies

By the end of training, Laboratory Biochemical Genetics trainees will demonstrate the ability to:

1. Describe specific public health practices or policies that affect provision of biochemical genetic testing services;
2. Respond to the health needs of individuals, communities and populations served by biochemical genetic testing.

Enabling Competencies

1. Describe specific public health practices or policies that affect provision of biochemical genetic testing services.

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

1.1. Explain how health care governance influences resource allocation for biochemical 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 biochemical genetic testing;

1.3. Participate in discussion regarding public policy and decision-making processes with respect to current and future biochemical genetic testing (i.e. introduction of new tests, new platforms).

2. Respond to the health needs of individuals, communities and populations served by biochemical genetic testing.

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

2.1. Recognize and respond to those medical genetic issues where advocacy is appropriate;

2.2 Become informed about community resources and related patient support groups for individuals and families served by biochemical genetic testing;

2.3. Liaise effectively with individuals, communities and populations on issues applicable to biochemical genetic testing;

2.4. Act as a resource and information source regarding biochemical genetic testing for individuals, communities and populations.

SCHOLAR

Key Competencies

By the end of training, the Laboratory Biochemical 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 biochemical genetic testing;
3. Conduct research projects and publish findings for advancement of knowledge.

Enabling Competencies

1. **Conduct ongoing learning activities to maintain and advance professional knowledge**
   To achieve this, the Laboratory Biochemical Genetics trainee will be able to:
   1.1. Critically assess the literature as it relates to human genetics, biochemical genetics 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 base and seek appropriate continuing educational activities;
   1.4. Be aware of and maintain accreditation through ongoing CME programs (eg RCPC / CCMG MOC program).

2. **Facilitate the learning of other health care professionals, students, laboratory colleagues, the public and others regarding biochemical genetics.**
   To achieve this, the Laboratory Biochemical Genetics trainee will be able to:
   2.1 Demonstrate the willingness and ability to enhance and apply teaching skills in the education of colleagues, undergraduate and postgraduate trainees, and other health care professionals;
   2.2. Deliver effective lectures and presentations on human genetics and biochemical genetics concepts;
   2.3. Present concise and audience appropriate summaries of biochemical genetic test methodologies, clinical situations related to biochemical genetic testing and case reports or presentations.

3. **Conduct research projects and publish findings for advancement of knowledge.**
   To achieve this, the Laboratory Biochemical Genetics trainee will be able to:
   3.1. Plan and conduct a minimum six month research project related to metabolism or biochemical genetic testing;
   3.2. Summarize results and submit and present the results to an appropriate audience (e.g. publication in a referred journal, presentation at a conference or rounds, presentation to laboratory staff);
   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, Laboratory Biochemical Genetics trainees will be able to:

1. Demonstrate ethical practices and a sense of responsibility in biochemical genetic testing;
2. Demonstrate appropriate respectful behaviour consistent with a clinical biochemical genetics diagnostician.

Enabling Competencies

1. Demonstrate ethical practices and a sense of responsibility in biochemical genetic diagnostic testing.

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

1.1. Demonstrate understanding of the regulatory framework governing the practice of medicine. These include the Legal system as well as local Medical Advisory Councils, or Appointment Boards, Provincial and Territorial licensing bodies and Federal guidelines;

1.2. Maintain confidentiality and ensure appropriate release of biochemical genetic samples, data and reports;

1.3. Recognize ethical issues in biochemical genetic testing, including but not limited to the process of consent, the needs of the patient vs. the family, the impact of cultural and personal beliefs, end of life decisions, testing of minors, impact of biochemical genetic results on extended family members, testing for late onset disorders, prenatal testing and the use of human subjects for research;

1.4. Identify personal limitations and the necessity of seeking the opinions of colleagues or other professionals when required;

1.5. Identify technical laboratory limitations requiring referral to other laboratories.

2. Demonstrate appropriate respectful behavior consistent with a clinical biochemical genetic diagnostician

To achieve this, the Laboratory Biochemical 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;

2.2. Respect the opinions of fellow consultants and referring physicians in the management of patients and be willing to accept differences of opinion;

2.3. Demonstrate the ability to recognize and respond appropriately to abuse, gender bias, discrimination, intimidation, and disrespect;

2.4. 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.