

Application Form for Biochemical Clinical Genetics Fellowship

Name of Institution: McGill University Health Centre

Location: Montreal, QC

Number of positions: One every two years or one every year, if a Medical Genetics trainee

Length: 2 years (maximum) (Length of training to be assessed by the Canadian College of Medical Geneticists (CCMG), which certifies the training. Trainees must have a MD degree. A candidate's MD specialty must be in Pediatrics (and subspecialties) or Internal Medicine (and subspecialties).

1 year – if trainee has completed a specialty in Medical Genetics

Program Information (append description):

Number of fellowship positions requested: One every two years, or one every year, if a Medical Genetics trainee

Academic affiliation: McGill University Health Centre (MUHC)

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

2 year training

McGill University Health Centre (MUHC) – 92% (minimum)

Clinical Biochemical Genetics, Cytogenetics and Molecular Laboratory, Clinical Genetics And Biochemical Genetics Laboratory

Specific rotations in laboratories of non-affiliated hospitals:

CHU Ste-Justine -1%

CHU Sherbrooke- 1%

Children's Hospital of Eastern Ontario- 1%

CHU Quebec -1%

Electives and Research – 4%

1 year training

McGill University Health Centre (MUHC)- 100%

Clinical Biochemical Genetics and Biochemical Genetics Laboratory

- % 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 Clinical Biochemical Genetics Fellowship is 1 or **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 clinical 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 training period to include more independence in patient encounters, counseling and provision of patient/family support, treatment and management, the interpretation of laboratory results 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. Lysosomal, mitochondrial, neuropsychiatric- are just a few of clinical fields where clinical research could be completed. Laboratory research expertise in the Division of Medical Genetics includes Peroxisomal Disorders (Dr. Nancy Braverman), B12 and folate disorders (Dr. David Rosenblatt) and lysosomal disorders (Dr John Mitchell). There are other biochemical genetics related research opportunities in other departments.

Mission: To produce clinical specialists who are competent to effectively diagnose and manage individuals of all ages with inherited metabolic disease.

Outline: This fellowship will enhance the residency training of the fellow by providing training in provision of having a thorough grounding in the diagnosis, investigation and management of a broad spectrum of disorders typically encountered in the biochemical genetics setting.

Name of the Fellowship Program Director: Dr. Daniela Buhas

Names of the Teaching Faculty

- D. Buhas, N. Braverman, J. Mitchell ,Y. Trakadis, I. DeBie, L. Russell, D. D’Agostino, B.Mucha, and T.Kitzler: clinical and biochemical genetics
- D. Buhas, N. Braverman, Y. Trakadis, 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
- CHU Sherbrooke--P. Waters and C. Aurais-Blais: biochemical laboratory test methods and interpretation
- CHU Ste-Justine--P. Allard and F.Parente: biochemical laboratory test methods and interpretation
- Chu Quebec--Y. Giguere and M-T. Berthier: biochemical laboratory test methods and interpretation
- CHEO--N. Lepage: biochemical laboratory test methods and interpretation

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

Academic Facilities

The MUHC Biochemical Clinical Genetics (BCG) team includes physicians, nurse and metabolic dietician. The pediatric BCG clinics, scheduled twice a week, serve patients with known or suspected inborn errors of metabolism. Once a month, there is a BCG Adult clinic, where known metabolic patients or under

investigations are evaluated. The inpatient service is actively involved in the diagnosis and the management of patients with biochemical genetic phenotypes.

The care of these patients is discussed weekly at a case review conference.

The MUHC offers a variety of biochemical genetic laboratory investigations. The clinical investigations include plasma, CSF and urine amino acids, acylcarnitine profile, transferrin isoelectric focusing, vitamin B12 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 Division of Medical Genetics hold outpatient clinics a minimum of four days a week. 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 Division of Medical Genetics maintains its own small library of books relevant to many aspects of medical genetics. Finally, the Division offers full time access to London Database, a database of physical traits and syndromes.

Fellow Duties and Responsibilities

- Call responsibilities to cover service: *the fellow will be involved in the evaluation of patients with known/suspected metabolic condition while covering call (shared schedule with the Medical Genetics Residents); could also be involved in an "availability" mode if these patients are evaluated in ER, Medical Day or Newborn screening* The fellow is the senior supervisor of residents: Yes- for fellows with Medical Genetics training ; *Only during the fellow's second year of training (for 2 years training). In the last clinical BCG rotation the fellow will be acting as junior staff (with minimal supervision)*
- Outline whether there are fixed rotations at the various institutions: *Yes*
- Outpatient clinic responsibilities need to be outlined: *The fellow is expected to gain experience in all aspects of diagnosis and management of metabolic disorders in both children and adults*
- Outline the role of the fellow towards residents on service: *The fellow will actively participate as a team member in the evaluation and management of metabolic patients. In the second year of training, his supervision role will increase gradually.*
- Teaching responsibilities towards residents: *The fellow will apply teaching skills with junior residents, under the supervision of staff in Biochemical Genetics. The fellow will deliver effective lectures and presentations on the concepts of inborn errors of metabolism.*
- 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 nurse, metabolic dietician and 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 weekly discussions of specific metabolic topics or 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 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
A logbook documenting at least 150 cases for whom the fellow has assumed consultant level of responsibility. This should reflect a broad range of metabolic disorders including inpatient and outpatient consultation in both the pediatric and adult setting.
The fellow is expected to gain experience in interpreting results and communicating to others, with a logbook recording involvement in 50 cases.
At least 120 of the 200 (combined clinical and laboratory) cases should represent known or newly diagnosed IEM. The remainder may include clinical cases where a diagnosis of an IEM may reasonably be considered part of the differential diagnosis. Cases may include telehealth and/or telephone consultations but the fellow involvement should clearly indicate discussion of history, exam and initial investigation, formulation of a differential diagnosis in that particular case as well as advising on diagnostic workup and plan for management.*

- *Intended Percentage of varieties of cases. Not specified but exposure should be broad.*

Regular reading materials provided (if any) :

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

- *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 weekly discussions of specific metabolic topics or 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*