FELLOWSHIP IN PEDIATRIC NEUROGENETICS
2-YEAR PROGRAM

Fellowship Director: Dr. Myriam Srour

Length of fellowship : 2 years

Requirements: Residency in Pediatric Neurology

Goal:

- This post-graduate fellowship program aims to provide pediatric neurologists clinical and research training in neurogenetics.

The specific goals for the Fellowship Program are outlined in the attached document.

In addition to the varied clinics, the fellow will have access to the following teaching rounds:

- Neurogenetics rounds every 4-8 weeks at the MCH
- Pediatric Neurology rounds every week
- Pediatric Neurology protected teaching (Neurogenetics topics)
- Neurology grand rounds every week
- Neuromuscular multidisciplinary rounds at the MNH every two weeks

The basics in genetic laboratory methods and techniques will be introduced such as Sanger sequencing, Next Generation sequencing such as whole exome and whole genome sequencing, chromosomal microarray, PCR, linkage analysis, SNP genotyping and homozygosity mapping.

It is also strongly recommended that the post-graduate fellow undertake clinical or laboratory research projects.

Faculty:

- Genevieve Bernard
- Bernard Brais
- Colin Chalk
- Genevieve Legault
- Rami Massie
- Ken Myers
- Erin O'Farrall
- Chantal Poulin
- Guy Rouleau
- Michael Shevell
- Eric Shoubridge
- Myriam Srour

Institutions:

- Montreal Children's Hospital, CUSM (Base institution)
- Montreal Neurological Hospital and Institute, CUSM
- Montreal General Hospital, CUSM

Affiliated Institutions:

- CHU Sainte-Justine
- Clinique Neuromusculaire, Centre de réadaptation Marie-Enfant, CHU-Sainte-Justine
- Clinique Neuromusculaire, Centre de réadaptation Lucie-Bruneau
- Clinique Neuromusculaire, CSSS de Jonquière

Subspecialty Clinics:

The fellow in Neurogenetics will have privilege access to following CUSM sub-specialised clinics:

- Neurogenetics clinic at the MNH (B. Brais, Dr. Rouleau and Dr Srour)
- Ataxia and adult leukoencephalopathy clinic at the MNH (B. Brais and G. Bernard)
- Pediatric Movement disorders and ataxia clinic at the MCH (G. Bernard)
- Pediatric Neurogenetics clinic at MCH (M. Srour)
- Developmental delay clinic at the MCH (M Shevell)
- Neurofibromatosis Clinic (G. Legault, and J. Ortenberg)
- Brain malformations clinic MCH (M. Srour)
- Adult Neuromuscular clinic MNH (B. Brais, R. Massie and E. O'Ferrall)
- Pediatric Neuromuscular clinic at MCH (C. Poulin, M. Oskoui)

- Peripheral nerve and myasthenia clinics at the MGH (C. Chalk, R. Massie and E. O'Ferrall)

APPLICATION FORM FOR FELLOWSHIPS
Pediatric Neurogenetics Fellowship- 2 year program

Program Information:

Number of fellowship positions requested: 2/year

Academic affiliation: McGill University Health Center (MUHC)

Name of hospitals involved in training and percentage of time spent by the fellow in each institution:
Montreal Children's Hospital (MCH), CUSM: This is the base hospital of the fellowship program. Between 75-100% of the fellow's time will be spent at the MCH.

The other sites where the fellow may rotate through or attend clinics include:
- The Montreal Neurological Hospital and Institute, CUSM
- The Montreal General Hospital, CUSM

In particular, fellows with a particular interest in the neurogenetics of Neuromuscular diseases may chose to spend more time the Neuromuscular and Neuropathy clinics at the MNI and MGH.

Background:
The field of Neurogenetics is rapidly expanding, especially given the technological advances in the past decades and the continued progress and developments in the field. The genetic etiology of many disorders has been identified, enabling improved diagnosis, prognositication and genetic counseling regarding familial recurrent risk. The identification of the underlying genetic etiology also allows improvement understanding of the pathogenesis of the underlying disorder, and in some cases the institution or development of treatments targeting the specific genetic defect. With increased frequency, neurologists have to organize and order genetic tests for their patients, and in turn, interpret the results.

Research activity:
The fellow in Pediatic Neurogenetics will have access to multiple clinicians and researchers, and will have the opportunity to chose a clinical or basic science research topic in the subspecialized field of his/her interest such as: Neuromuscular, Neurodegenerative, Metabolics, Movement disorders, Ataxia, Developmental Delay, Brain malformations, Leukoencephalopathies, Mitochondrial disorders…
For the two-year Pediatric Neurogenetic fellowship program, it will be expected that the fellow be involved in at least one research project during his/her fellowship with at least 6 dedicated months.

Publications:
It will be expected that the fellow submit at least one paper as first author, and will also likely be involved in the writing of a review or book chapter.

Mission:
This fellowship in Pediatric Neurogenetics aims to provide the Pediatric Neurologist with the knowledge, experience and training required to be able to identify a patient with a potentially heritable or genetic disorder, obtain a family history and collateral information, think of a differential diagnosis, order the appropriate work-up and genetic testing, interpret the results and explain to the proband and family the implications of the results. In addition, this fellowship aims to make the fellow familiar with the commonly used Neurogenetic laboratory techniques and strategies such as Sanger sequence analysis, Next Generation methods such as whole-exome/genome sequencing analysis, chromosomal microarray, PCR, linkage analysis, SNP genotyping analysis and homozygosity mapping.

Outline how intended fellowship will enhance residency training:
This fellowship will enhance the residency training in multiple ways:

- Provide instructions to residents and medical students at a level appropriate to their clinical education and professional competence.
- Willingly share knowledge with others with whom they are associated, thus ensuring the most effective delivery of health care to patients.
- Be involved in complex Neurogenetic cases, which will lead to discussion and case-based teaching by the fellow with the Neurology residents.
- Take part in Neurogenetics Rounds, where neurologists, medical geneticists genetic counselors, residents and allied professional are present.
- Teach at Neurology Residents' Academic half day.

Name of the Fellowship Program Director: Dr. Myriam Srour

Names of the Teaching Faculty:

Genevieve Bernard: Runs the Pediatric Movement disorders and ataxia clinic at the MCH, Leukodystrophy and Neurometabolic clinic at MCH and part of the Ataxia and adult leukoencephalopathy clinic at the MNH. Dr Bernard's laboratory focuses on the genetic causes of pediatric neurodegenerative disorders, and more specifically on leukodystrophies /inherited white matter disorders and pediatric movement disorders.

Bernard Brais: Dr Brais is the co-director of the neuromuscular group of the MNH. He is part of the Neurogenetic Clinic and the Ataxia and adult leukoencephalopathy clinic at the MNH, as well as the medical director of the Neuromuscular clinic at the Centre de Réadaptation Marie-Enfant of the CHU-Sainte-Justine and the Neuromuscular clinic of the Centre de réadaptation Lucie-Bruneau. He investigates the genetic basis of neurogenetic disorders with founder effects in Quebec, with an increasing focus on disorders with ataxic manifestations such as Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS).

Colin Chalk: Dr Chalk is a Neurologist at the MGH and specializes in Peripheral Neuromuscular disorders. He is part of the Peripheral Nerve and Mysthenia clinics at the MGH.

Genevieve Legault; Dr Legault is a pediatric neurologist with a specialty in neuro-onclogy. She is the neurologist of the neurofibromatosis clinic.

Rami Massie: Dr Massie is an adult neurologist with a subspecialty in Neuromuscular disorders. He sees patients in the EMG laboratory, in the ALS clinic and in the neuromuscular clinic, with a particular interest for disorders of peripheral nerves. He also works in the Montreal General Hospital Neuropathy Clinic.

Ken Myers: Dr Myers is a pediatric neurologist and clinician researcher at the MCH. He specializes in epilepsy, with a focus on the phenotyping of the genetic epileptic syndromes.

Erin O'Ferrall: Dr O'Ferrall is an adult Neurologist with a subspecialty in Neuromuscular disorders. She is part of the Neuromuscular clinic at the MNH, the Peripheral Nerve clinic and Myasthenia clinic at the MGH and the Neuromuscular clinic of the Centre de réadaptation Lucie-Bruneau.

Maryam Oskoui : Dr Oskoui is a pediatric Neurologist with a focus on Neuromuscular disorders. She is part of the Neuromuscular clinic at the MCH. Her research focuses on the transition of patients from pediatric to adult medical institutions.

Chantal Poulin: Dr Poulin is a pediatric Neurologist with a focus on Neuromuscular disorders. She is part of the Neuromuscular clinic at the MCH.

Guy Rouleau: Dr. Rouleau is an adult neurologist and geneticist. He is a clinician researcher. His laboratory is focussed on understanding the genetic basis for diseases, and to identify genes that cause neurological and psychiatric diseases like amyotrophic lateral sclerosis, stroke, essential tremor, familial aneurysms, cavernous angiomas, epilepsy, spinocerebellar ataxia, spastic paraplegia, autism, Tourette syndrome, restless legs syndrome, schizophrenia and bipolar disorder. He has a Neurogenetics Clinic at the MNI where he assesses patients with ALS, movement disorders, NF, hereditary spastic paraparesis and various other neurogenetic disorders.

Eric Shoubridge: Dr. Eric Shoubridge's laboratory focuses on the molecular genetics of mitochondrial diseases.

Michael Shevell: Dr Shevell is a pediatric Neurologist at the MCH with a focus on developmental delay and cerebral palsy. He is part of the Developmental Delay clinic at the MCH. His research focuses on neurodevelopmental disabilities.

Myriam Srour: Dr Srour is a pediatric Neurologist and Clinician Scientist at the MCH. She is

the director of the Neurogenetics and Brain malformations clinics at the MCH, and also has a Neurogenetics clinic with a focus on epilepsy at the MNI. Her research focuses on the genetic basis of neurodevelopmental disorders and structural brain malformations.

Academic Facilities

Outline facilities for clinical and academic pursuit

The Neurology clinical spaces at the MCH will be accessible and available to the fellow. The residents' rooms at the MCH and the MNH that have access to shared computers, as well as a very complete neurology library will be available to the Fellow.

Library access, materials relevant to fellowship training

The fellow will have access to the library at the MCH, as well as the MNH and the MGH. The MUHC Glen site and MNI libraries have a large collection of neurogenetics reference books and medical journals. The McGill library has a large access to neurogenetic journals on line. The Resident's room at the MCH also has a large collection of reference materials.

Availability of a skills lab if applicable

Several physicians and researchers have neurogenetics laboratories. If the fellow choses to be involved in a research project with a laboratory component, he or she can have access to the laboratory facilities.

Fellow Duties and Responsibilities

- The fellow will be responsible to attend McGill out-patient clinics affiliated to the program during the first 6 months of the fellowship. For the remaining 6 months period clinics will be selected by the fellow according to interests or selected research project. The total number of outpatient clinics per week will be determined in advance in consultation with the fellowship director (between 3 to 6 half-day clinics per week). This will depend on the fellow's focus and research project. The fellow is required to inform the attending physician of his absence in advance to allow for rescheduling of patients.
- The fellow is expected to write complete and thorough consultations letters on patients seen in clinic. These letters are expected to be sent to the referring physician in a timely manner. In addition, with the guidance and supervision of the staff physician, the fellow is expected to organize the patient's genetic testing, the interpretation of the results and the discussion of the genetics results with the patient/family.
- The fellow will be expected to attend the Neurogenetics rounds every 4-8 weeks at the MCH.
- The fellow will be encouraged to attend the Pediatric Neurology protected teaching (Neurogenetics topics), Neurology grand rounds every week and the Neuromuscular multidisciplinary rounds at the MNH every two weeks.

- The fellow will be expected to provide case-based and case-focused teaching to residents. The fellow will be expected to give formal teaching to the adult and pediatric neurology residents at their academic half day once per year.
- The fellow will be encouraged to attend one international meeting and present a poster/platform (example: American Academy of Neurology, American Society of Human Genetics, Child Neurology Society…).

Evaluation: The fellow will meet on regular basis (3/year) with the Fellowship Program Director to review performance, progress and go over any issues and challenges. The fellow will also meet on a regular basis with his/her specific research supervisor. The research supervisor will be asked to report to the fellowship director at least once a year on the performance of the fellow.

Pediatric Neurogenetics Fellowship
General and Specific Goals & Objectives

1. Medical Expert/Clinical Decision-Maker

General Requirements

- Demonstrate diagnostic and therapeutic skills for ethical and effective patient care.
- Access and apply relevant information to clinical practice.
- Demonstrate effective consultation services with respect to patient care, education and legal opinions.

Specific Requirements

- Provide scientifically based, comprehensive and effective diagnosis and management for patients with Neurogenetic disorders.
  The fellow will have access to a number of sub-specialized McGill primary neurogenetics clinics such as:
  - Neurogenetics clinic at the MNH (B. Brais, Dr. Rouleau and Dr Srour)
  - Ataxia and adult leukoencephalopathy clinic at the MNH (B. Brais and G. Bernard)
  - Pediatric Movement disorders and ataxia clinic at the MCH (G. Bernard)
  - Pediatric Neurogenetics clinic at MCH (M. Srour)
  - Developmental delay clinic at the MCH (M Shevell)
  - Neurofibromatosis Clinic (G. Legault, and J. Ortenberg)
  - Brain malformations clinic MCH (M. Srour)
  - Adult Neuromuscular clinic MNH (B. Brais, R. Massie and E. O'Ferrall)
  - Pediatric Neuromuscular clinic at MCH (C. Poulin, M. Oskoui)
  - Leukodystrophy and Neurometabolic clinic at MCH (G. Bernard)
  - Peripheral nerve and myasthenia clinics at the MGH (C. Chalk, R. Massie and E. O'Ferrall)

  The fellow will also be encouraged to do electives in a number of Montreal clinics that care for specific neurogenetics clienteles such as:
  - Neuromuscular clinic – Centre de Réadaptation Marie-Enfant of the CHU-Sainte-Justine (B. Brais)
  - Neuromuscular clinic of the Centre de réadaptation Lucie-Bruneau (B. Brais and E. O'Ferrall)

Clinical:

- For a patient with a Neurogenetic disorder, the fellow will be able to:
  - Obtain a complete neurological history from adults and children obtaining a collateral history when necessary.
  - Perform an appropriate physical examination.
  - Determine whether a patient's symptoms and signs may be the result of a Neurogenetic disorder.
  - Take a detailed family history and draw a pedigree using computer programs.
  - Know when to suspect a neurogenetic disorder.
  - Formulate an appropriate anatomical localization, differential and provisional diagnosis if appropriate.

- Outline an appropriate plan of laboratory investigations, including learning the procedures to have genetic tests approved and samples sent to appropriate laboratories.
- Outline an appropriate therapeutic plan.
- Exhibit appropriate clinical judgment in outlining a differential diagnosis and an investigative and therapeutic plan, taking into account matters such as the patient's age, family history, general health, risk and cost of investigative procedures, risk and cost of therapeutic interventions, and epidemiology of the disease.
- Be familiar with genetic counselling of families regarding recurrence risk to future pregnancies or to progeny of other family members.

Technical Skills

- If the fellow is interested, genetic laboratory techniques can be introduced such as PCR, linkage analysis, sequence analysis, whole-exome/genome sequencing analysis, SNP genotyping analysis, homozygosity mapping, and functional assessment of gene mutations.

Knowledge

- To understand the general principles related to the main cytogenetic and molecular genetic techniques such as Fluorescence in situ Hybridization (FISH), Chromosomal microarray, karyotype, linkage analysis, molecular sequencing, deletion/duplication analysis, SNP genotyping, whole exome/genome sequencing.
  - To understand when and how to order clinical genetic investigations.
  - To interpret genetic results.
  - To outline strategy to clarify unclear genetic results.
  - To know when to refer patients and family members for genetic counselling.

2. Communicator

General Requirements

- Establish therapeutic relationships with patients/families.
- Obtain and synthesize relevant history from patients/families/communities.
- Listen effectively.
- Discuss appropriate information with patients/families and the health care team.
- Explain genetic results to patients and families, along with implications regarding recurrence risks in future pregnancies and risks for other family members.

Specific Requirements

- Communicate effectively with patients, their families and medical colleagues (particularly referring physicians, medical geneticists and genetic counsellors) and other health care professionals in both the inpatient and outpatient settings.
- The fellow will:
  - Communicate effectively and regularly with patients and their families.
  - Be considerate and compassionate in communicating with patients and families, willingly provide accurate information appropriate to the clinical situation, with a reasonable attempt at prognosis.
  - Learn to write concise reports of the clinical findings with conclusions and recommendations comprehensible to the non-specialist.
  - Communicate effectively and appropriately with the nurses and paramedical personnel.

- When ordering investigative procedures, ensure there has been adequate communication about the patient with the person who will actually be doing and/or reporting the diagnostic study.

3. Collaborator

General Requirements

- Consult effectively with other physicians and health care professionals in particular medical geneticists and genetic counsellors.
- Contribute effectively to other interdisciplinary team activities.

Specific Requirements

- Be an effective teacher of other physicians (including medical students and house officers), other health care personnel, and patients. The fellow will:
  - Provide instructions to medical students and more junior physicians at a level appropriate to their clinical education and professional competence.
  - Willingly share knowledge with others with whom they are associated, thus ensuring the most effective delivery of health care to patients.

4. Leader

General Requirements

- Utilize resources effectively to balance patient care, learning needs, and outside activities.
- Allocate finite health care resources wisely.
- Work effectively and efficiently in a health care organization.
- Utilize information technology to optimize patient care, life-long learning and other activities.

Specific Requirements

- Be proficient in professional skills related to the diagnosis and treatment of Neurogenetic disorders.
- Demonstrate the following professional skills in time management:
  - Recognize that effective use of time depends upon punctuality.
  - Recognize that effective use of time requires planning.
  - Develop speed as well as accuracy in clinical skills.
  - Reserve time for reading and keeping current with the neurogenetics literature.
  - Establish routines for carrying out regular activities and adhere to them.
- Maintain complete and accurate medical records:
  - Record and maintain a complete and accurate medical record for every patient seen; this record will include the patient's history and the findings on physical examination (including the neurological examination), a differential diagnosis, a provisional diagnosis
  - Effectively coordinate the work of the health care team.
  - Indicate, in the treatment plan, that for the optimal treatment of many patients with neurogenenic disorders, a team approach is necessary -- members of the team may include other physicians such medical geneticists, genetic counsellors, nurses, rehabilitation personnel (physiotherapists, occupational therapists, speech therapists, psychologists, social workers, etc..
  - Identify where an important role(s) can be played by disease focused lay groups with regard to helping the patient and/or *f*amily and to facilitate its happening.

5. Health Advocate

General Requirements

- Identify the important determinants of health affecting patients.
- Contribute effectively to improved health of patients and communities.

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

Specific Requirements

- Learn about medical genetics and genetic counselling services offered.
- Learn about community resources and related patient support groups; provide assistance to access programs (e.g. home care, occupational and physiotherapy, drug plans, application for nursing homes etc.) and participate in their activities.
- Educate, be able to generate and access information (e.g. printed material, videotapes, websites) and be available as a resource person to counsel patients effectively on neurologic disorders.
- Counsel patients on the importance of taking responsibility for their own well-being and recognize the important determinants predisposing to worsening of neurological status.
- Understand the role of national and international bodies (e.g. AAN, ASHG) in the promotion of neurological health, and the prevention, detection, and treatment of nervous system disorders.

6. <u>Scholar</u>

General Requirements

- Develop, implement and monitor a personal continuing education strategy.
- Critically appraise sources of medical information.
- Facilitate learning of patients, house staff/students and other health professionals.
- Contribute to development of new knowledge.

Specific Requirements

- Be able to critically assess the neurological literature as it relates to neurogenetics diagnosis, investigations and treatment:
  - Develop criteria for evaluating neurologic literature.
  - Critically assess the neurologic literature using these criteria.
  - Be familiar with the design of experimental and observational, familial and population genetics studies.
  - Be able to calculate absolute risk of transmission.
- Be able to participate in neurogenetics clinical or basic science studies as a member of a research team:
  - Be able to describe principles of good research.
  - Use the above principles, and be able to judge whether a research project is properly designed.
  - Be prepared to present research findings to peers at local, national and international conferences.

7. <u>Professional</u>

General Requirements

- Deliver highest quality care with integrity, honesty and compassion.
- Exhibit appropriate personal and interpersonal professional behaviours with patients/families, peer residents and other health care professionals.
- Practice medicine ethically consistent with obligations of a physician.

Specific Requirements

- Demonstrate personal and professional attitudes consistent with a consulting neurogeneticists role:
  - Periodically review his/her own personal and professional performance against national standards set for the specialty.
  - Be willing to include the patient in discussions concerning appropriate diagnostic and management procedures.

- Show appropriate respect for the opinions of fellow consultants and referring physicians in the management of patient problems and be willing to provide means whereby differences of opinion can be discussed and resolved.

- Be willing and able to appraise accurately his/her own professional performances and show that he/she recognizes his/her own limitations with regard to skills and knowledge by appropriately consulting other physicians and paramedical personnel when caring for the patient.
- Be willing and able to keep his/her practice current through reading and other modes of continuing medical education and develop a habit of maintaining current his/her clinical skills and knowledge base through continuing medical education.