APPLICATION FORM FOR FELLOWSHIPS

Name of Institution: Shriners Hospital for Children

Location: Shriners Hospital for Children
1529 Cedar Avenue
Montreal, Quebec
H3G 1A6 2C7
Tel: (514) 842-5964
Fax: (514) 842-5581
Email: frauch@shriners.mcgill.ca

Type of Fellowship: Pediatric Metabolic and Genetic Bone Disorders

Program Information:

Number of fellowship positions requested: 1

Academic affiliation: McGill University

Name of hospitals involved in training and % of time spent in each institution:
Shriners Hospital for Children 100%

Background:

The Genetics Unit at the Shriners Hospital for Children in Montreal offers a fellowship training program in pediatric metabolic and genetic bone disorders. The fellowship is suitable for MDs who have completed their pediatric or genetic training and who wish to specialize in pediatric bone diseases.

The Montreal Shriners Hospital for Children is one of 22 Shriners Hospitals for Children in the U.S., Canada and Mexico that provide specialized care for children. Services are provided at no charge, regardless of the country of origin of the patients. The Montreal Shriners Hospital is a 25-bed inpatient facility that provides the full spectrum of care for pediatric orthopedic and metabolic and genetic bone disorders. An interdisciplinary team approach is used in patient care programs. Patients are followed and treated up to the age of 21 years.

Osteogenesis imperfecta is a major focus of the clinical care and clinical research activity at the Shriners Hospital. More than 400 children and adolescents with osteogenesis imperfecta are followed at the Shriners Hospital. The clinical part of the program is run by the multidisciplinary osteogenesis imperfecta team. Team members deal with all aspects of osteogenesis imperfecta patient care in both the inpatient and the outpatient setting. In particular, team members run a
common clinic, the multidisciplinary osteogenesis imperfecta clinic, which attracts osteogenesis imperfecta patients not only from North America, but also South America, Europe, and the Middle East.

The fellow is a key member of the team that cares for patients with metabolic and genetic bone disorders. The fellow sees all patients that are followed in this highly specialized environment and discusses each case with a staff member. Prior to the onset of the fellowship, the fellow and the program director will together predetermine an individualized set of training goals. Goals of fellowship and trainee performance will be reviewed quarterly. The trainee has reciprocal opportunities to assess their supervisors and the training program.

**Research activities:**

Apart from providing family-oriented clinical care, the Shriners Hospital includes one of the major research centers in the pediatric bone field. The research center, named Genetics Unit, supports both clinical and basic science research. Founded by Francis Glorieux, the Genetics Unit has about 30 full-time staff. It is part of an intramural research program sponsored by Shriners Hospitals for Children. The focus of the basic research component is on cell and developmental biology of bone and cartilage development. It includes 7 programs led by 5 Principal Investigators. The clinical research laboratory includes the pediatric bone histomorphometry unit (quantitative evaluation of bone histology) which is unique worldwide, as well as state of the art radiological methods for measuring bone density and bone geometry.

Since 1992, studies have assessed the use of bisphosphonate treatment in children with osteogenesis imperfecta. The results obtained in this program have made intravenous pamidronate treatment the worldwide standard of care for children and adolescents with moderate to severe osteogenesis imperfecta. The program also has led to the discovery of three new types of osteogenesis imperfecta (osteogenesis imperfecta types V, VI and VII). Recently, mechanical treatment approaches using vibration therapy have been added to the spectrum of research activity at the Shriners Hospital.

The fellow chooses research topics and performs research projects in close collaboration with the Fellowship Program Director. Research projects depend on the fellow’s interest and prior knowledge and can include any of the clinical or laboratory methodologies available at the Shriners Hospital. Fellows are expected to spend about 50% of their time on research-related activities.

**Publications:**

This is the list of publications from the Metabolic and Genetic Bone Disease Program since 2005 (publications that were first authored by a clinical fellow are indicated by an asterisk):


Höglér W, Blimkie CJ, Cowell CT, Inglis D, Rauch F, Kemp AF, Wiebe P, Duncan CS, Farpour-Lambert N, Woodhead HJ. Sex-specific developmental changes in muscle size and bone


**Mission:**

The goal of the Fellowship in Pediatric Metabolic and Genetic Bone Disorders is to form individuals who will become fully independent in caring for children and adolescents with metabolic and genetic bone disorders. In addition, this fellowship in one of the world’s top institutions for pediatric bone research provides the foundation for an academic career as a clinical researcher in the area of pediatric bone disorders.

**Major strengths:**

The Genetics Unit at the Shriners Hospital is widely recognized as one of the major centers for clinical care as well as for clinical and basic science research in the field of bone development and pediatric bone disorders. Most previous fellows have successfully established themselves as independent academic investigators and have become key players in the field.

**Fellowship Program Director:** Frank Rauch, MD

Dr. Rauch completed his pediatric residency training at the Children’s Hospital of the University of Cologne in Cologne, Germany. He then performed a research fellowship in the Genetics Unit laboratory of the Shriners Hospital for Children in Montreal. Since 2001 he has been taking care of children with metabolic and genetic bone disorders at the Shriners Hospital. Dr. Rauch is also the Director of Clinical Laboratories at the Shriners Hospital. During the past five years he has supervised 5 clinical fellows who took part in the Shriners fellowship program. Dr Rauch is an Associate Professor at the Department of Pediatrics of McGill University. His research interest mainly involves the diagnosis and treatment of children with metabolic and genetic bone disorders, in particular osteogenesis imperfecta as well as the contribution of mechanical influences on common bone disorders. Dr Rauch is currently Editor in Chief of the Journal of Musculoskeletal and Neuronal Interactions. He has published 100 original articles, mostly on pediatric bone disorders. In 17 of these publications, the first author was one of the previous clinical fellows.
Teaching Faculty

Francis H Glorieux, MD, PhD, OC

Francis H Glorieux is a pediatrician and geneticist with 35 years experience in the treatment and investigation of genetic and metabolic bone diseases in children. His clinical studies have led to the delineation of new treatment programs for familial hypophosphatemic rickets and osteogenesis imperfecta. He is the Director of Research of the Genetics Unit, which is one of the six Centers of Excellence within the Shriners Hospital Network (22 Institutions). Dr Glorieux has long been recognized as one of the leading clinical scientists in the pediatric bone field and has been awarded an Order of Canada award for his lifetime achievements.

Dr Leanne Ward, MD, FRCPC

Dr Ward is a pediatric endocrinologist who trained in pediatric bone diseases at the Montreal Shriners Hospital. She is now the Director of the Pediatric Bone Health Clinical and Research Programs at the Children’s Hospital of Eastern Ontario in Ottawa, with collaborative links to the Montreal Shriners Hospital on a part-time basis. She has extensive experience with conducting multicentre trials in the field of pediatric metabolic bone disorders.

Academic Facilities

The metabolic and genetic bone disease program runs two to three outpatients clinics per week as well as the intravenous bisphosphonate infusion program in a day care setting. All activities take place at the Shriners Hospital. Apart from the Program Director and Teaching Faculty mentioned above, our clinical team consists two full-time clinical nurses specialized in pediatric bone disorders as well as seven technical and administrative full-time staff. We are equipped with four examining rooms and two treatment rooms for the administration of intravenous therapy.

Our electronic database uses the Filemaker system and records all patient visit data and medication prescriptions. This database can be easily queried for research purposes. All computers of the Genetics Unit have access to the McGill University electronic library system.

In addition, the following specialized laboratories at the Shriners Hospital are at the disposal of the team for both clinical care and clinical research:

- Bone Densitometry Laboratory: Fully staffed with three certified radiology technicians and equipped with a Hologic Discovery® dual-energy X-ray absorptiometry and a Stratec XCT-2000 peripheral quantitative computed tomography system. This constantly updated laboratory has performed pediatric bone densitometry since 1990 and has provided centralized bone density assessment services for various multicenter studies in the past.

- Clinical Biomedical Laboratory: Laboratory for the determination of serum and urine parameters of bone and mineral metabolism. The results obtained in this laboratory have contributed to numerous original publications, including the most comprehensive assessment of bone metabolism in OI that is available to date. It is staffed with two biomedical technologists.
• Molecular Diagnostics Laboratory: Laboratory for the sequence analysis of genes that are involved in causing heritable bone disorders. The laboratory includes one technician and is supervised by Peter Roughley, Ph.D.

• Pediatric Bone Histomorphometry Laboratory: Specialized laboratory for the quantitative histological analysis of pediatric bone samples. This laboratory has been instrumental in delineating new types of osteogenesis imperfecta and in evaluating the skeletal effects of new treatment modalities.

Interdisciplinary Communication

1 Orthopedic Surgery: There is close collaboration with the orthopedic department which includes four full-time orthopedic surgeons as well as a clinical orthopedic fellow. There are about 12000 visits per year to orthopedic clinics at the outpatients department of the Shriners Hospital. François Fassier, MD, the current Chief of Staff at the Shriners Hospital, is an orthopedic surgeon who has introduced several new orthopedic procedures for osteogenesis imperfecta. Dr Fassier has developed a telescoping nail, the Fassier-Duval nail, for intramedullary rodding of long bones. The Fassier-Duval nail is now worldwide the most widely used elongating nail for intramedullary rodding of children with osteogenesis imperfecta and other pediatric bone fragility disorders.

2 Physiotherapy/Occupational Therapy: Patients with bone fragility disorders are regularly evaluated in the departments for physiotherapy and occupational therapy. These two departments actively take part in all clinical trials that are carried out at the Shriners Hospital for Children.

Fellow Duties and Responsibilities

• Outpatient clinic: The fellow runs two to three clinics per week, with six to ten patients per clinic. Each patient is subsequently discussed with a staff member.

• Day care unit: The fellow provides medical care for patients who receive intravenous bisphosphonates at the Shriners Hospital (typically between 5 and 8 patients per week)

• The fellow prepares the multidisciplinary osteogenesis imperfecta clinic and presents each case to the entire team during the clinic.

• Support staff available to the fellow: The fellow’s outpatient clinics as well as the intravenous infusion programs are organized by a clinical nurse and an administrative coordinator. The fellow’s research activities are supported by a research assistant.

• Call responsibilities to cover service: None

• Include whether the fellow is the senior supervisor of residents: No

• Outline whether there are fixed rotations at various institutions: No

• Outline role of the fellow towards residents: Residents in the Medical Genetics Program are on site during two to three months per year. The fellow introduces residents to the procedures of the service and assigns patients to them. The fellow does not have further
teaching responsibilities towards residents.

The trainee should complete the following objectives:

- Become familiar with advanced diagnostic methodologies (indications of use, interpretation, pitfalls, and methodological issues) that are used to assess bone. These methodologies include:
  - Dual-energy X-ray absorptiometry
  - Peripheral quantitative computed tomography
  - Mechanography
  - Biochemical markers of bone metabolism
  - Quantitative computerized bone histomorphometry.
- Use of bisphosphonate treatment in children with bone fragility disorders: Indications, treatment protocols, follow up, criteria for stopping treatment
- Learn to perform transiliac bone biopsies.
- Interpret sequence analysis results of genes that are commonly involved in pediatric bone disorders
- Proposed meetings to be attended by the fellow: The fellow is expected to attend the annual meeting of the American Society for Bone and Mineral Research as well as the biannual International Conference for Children’s Bone Health.
- Research productivity and publications expected by the fellow: It is expected that the fellow will complete two research projects per year and that each project leads to a publication.

Curriculum

- Intended case load and varieties of cases: There are between 800 and 900 patient visits to the metabolic and genetic bone clinic per year. Patients with osteogenesis imperfecta make up about 60% of these visits. The remainder of the visits are by patients affected with a wide range of metabolic and genetic bone disorders, such as familial hypophosphatemic rickets, pseudovitamin D deficiency rickets, fibrous dysplasia and a large number of rarer skeletal dysplasias. All of these visits are primarily handled by the clinical fellow.
- Schedule of clinical activities:
  - Outpatient clinics are scheduled each week on Tuesdays and Wednesdays from 8 am to 1 pm. The multidisciplinary osteogenesis imperfecta clinic takes place once or twice per month on Tuesday afternoons (1.30 pm to 4.30 pm).
  - The intravenous bisphosphonate infusion program runs each week from Monday to Wednesday (8 am to 2 pm).
  - On Thursdays and Fridays there are no planned patient contacts.
- The fellow is expected to perform consults for minor medical issues that arise in patients who are hospitalized in the orthopedic department. Typically, there are one to two consults per month.

- A molecular diagnostics meeting is held once per month (Thursdays 10am to 12 am) to review the results of the molecular diagnostics laboratory for our clinics patients. The fellow is expected to briefly present the cases during these meetings.

- Depending on the fellow’s interest, there is the opportunity to participate in the Skeletal Dysplasia Clinic and the Skeletal Radiology Rounds that are both held at the Medical Genetics Department, located at the Montreal Children’s Hospital.

- The fellow is expected to attend weekly Genetics Unit laboratory meeting (Fridays, 11 am to 12 pm) where clinical and basic research projects are presented. The fellow typically gives two such on-hour presentations twice per year during the laboratory meeting.

- Finally the fellow is encouraged to take charge of his/her own education depending on his intended future site and style of practice. The specific needs of each fellow will be assessed by the Program Director and Teaching Faculties and training will be tailored to best suit each individual fellow’s needs.