

# Nephrogenetics Fellowship

## **Section I:**

**Fellowship Director(s):** Dr. Ahsan Alam and Dr. Thomas Kitzler

**Fellowship Location (s):** MUHC (Royal Victoria Hospital, Montreal Children's Hospital, Montreal General Hospital)

**Fellowship Type:** Clinical= 50-75% and Research = 25-50%

**Number of Fellowship Positions:** 1-2

**Length of Training:** 2 year

**Funding:** Candidates must secure independent funding to support their Fellowship. Potential sources include institutional funding, departmental funding, external fellowships, or foundation support. For internal funding opportunities that are available, please contact the fellowship co-directors ([ahsan.alam@mcgill.ca](mailto:ahsan.alam@mcgill.ca) or [thomas.kitzler@mcgill.ca](mailto:thomas.kitzler@mcgill.ca)).

## **Fellowship Eligibility Requirements:**

Adult Nephrology, Pediatric Nephrology or Medical Genetics accredited by the Royal College of Physicians and Surgeons of Canada (RCPSC) or an accredited training program outside Canada

## **Admission Requirements:**

All candidates should have certification of a 3-year residency training in Internal Medicine or Pediatrics and completing 2-years of residency training in Adult or Pediatric Nephrology in Canada or abroad. Alternatively, candidates can apply at completion of a 5-year primary residency specialty program in Medical Genetics.

## **Section II:**

### **Program Information:**

#### **Mission:**

The principal objective of this 2-year fellowship is to train highly competent, independent consultants in nephrology genetics. The second year of training will place emphasis on engaging in scholarly work as well as focusing on specific clinical domains of interest. Graduates will be proficient across all CanMEDS roles, equipped to provide expert clinical care, engage in meaningful scholarly activity, contribute to the education of others, and lead quality improvement initiatives pertinent to genetic kidney disease within the Canadian healthcare system.

## **Background:**

Nephrology genetics is an evolving subspecialty that integrates expertise in nephrology and medical genetics. Its primary focus encompasses the comprehensive diagnostic evaluation, clinical management, and appropriate counselling for individuals and families affected by, or at risk of, inherited kidney diseases. The MUHC Nephrogenetics Fellowship aims to equip trainees with the specialized knowledge and skills necessary to function as expert consultants in this increasingly important and expanding field.

The impetus for establishing a dedicated Nephrogenetics fellowship at the MUHC stems from the growing recognition of the substantial role genetic factors play in the etiology of chronic kidney disease (CKD). It is estimated that genetic mutations are responsible for 10-20% of CKD cases in both adult and pediatric populations within Canada. This genetic contribution significantly impacts the overall burden of CKD and end-stage kidney disease. Despite this, a notable gap exists in the current training and comfort level among many nephrologists regarding the integration of genetic assessment and testing into routine clinical practice.

The possibility of establishing a precise genetic diagnosis in patients with a kidney disease has become increasingly evident and valuable. Genetic understanding can refine diagnoses and risk prediction, which can significantly inform prognosis and disease surveillance. Genetic information can guide the use of targeted therapies in specific conditions, such as in ADPKD or Fabry disease. Furthermore, a confirmed genetic diagnosis facilitates cascade screening for at-risk family members, enabling early detection and preventative measures, and can potentially obviate the need for invasive procedures like kidney biopsies in certain clinical scenarios. It may also be a critical step in ensuring the safety of living kidney donors with a known or suspected genetic condition.

Fellows will develop a comprehensive diagnostic approach central to nephrology genetics practice including:

**Clinical Assessment:** including obtaining a detailed multi-generational family history and constructing an accurate pedigree, a thorough review of systems to identify potential extra-renal manifestations suggestive of syndromic conditions, and a focused physical examination, including assessment for dysmorphic features. Fellows will understand the role and limitations of imaging (ultrasound, CT and MRI) for structural information and quantitative measurements like total kidney volume

**Genetic Testing:** This is a core diagnostic tool in renal genetics. Training will cover the appropriate selection, ordering, and interpretation of various genetic tests, including targeted gene panels (testing multiple genes associated with a specific phenotype, like CKD or cystic disease) and broader approaches like exome sequencing (sequencing the protein-coding regions of the genome). Interpreting genetic test results is a critical and

complex skill. Fellows will understand the clinical significance of identified genetic variants, including the challenging category of Variants of Uncertain Significance (VUS), which require careful correlation with the patient's phenotype. This often necessitates a multidisciplinary approach involving collaboration between clinicians, genetic counsellors, and laboratory geneticists. Training will also address the evolving landscape of variant re-classification and the potential need for periodic re-analysis of genomic data or re-contacting patients with updated information.

Personalized Care Plans: The identification of a specific genetic cause allows for tailoring management strategies. This may involve initiating gene-specific therapies (e.g., tolvaptan for eligible ADPKD patients), implementing targeted surveillance protocols for associated complications (e.g., screening for aneurysms in ADPKD or tumors in VHL), avoiding potentially harmful medications or interventions, or adjusting prognostic expectations.

Genetic Counselling: This is an indispensable aspect of care for patients and families with genetic kidney disease. Fellows will develop competence in providing comprehensive counselling, which includes explaining the diagnosis, mode of inheritance, recurrence risks, natural history of the condition, and management options. It also involves discussing the complex implications of genetic testing (benefits, limitations, potential psychological impact), addressing psychosocial concerns, exploring family planning options (including preimplantation genetic diagnosis where applicable), and facilitating informed decision-making.

Multidisciplinary Team Approach: Effective care requires collaboration among various specialists. This includes nephrologists (adult and pediatric), medical geneticists, genetic counsellors, laboratory scientists, pathologists, radiologists, nurses, dietitians, social workers, and primary care physicians, working together to provide integrated care.

CKD Management: General principles of CKD management remain crucial, including optimal blood pressure control, reduction of proteinuria, management of renal complications, lifestyle modifications, and timely planning for renal replacement therapy (dialysis or transplantation) when indicated. These strategies are applied within the context of the specific genetic diagnosis.

### **Fellowship Highlights/Aspirations**

The MUHC Nephrogenetics fellowship is critically positioned to mobilize specialized expertise, address the unmet needs of patients with genetic kidney diseases, and propel the advancement of precision nephrology within the Canadian healthcare landscape. The MUHC Nephrogenetics fellowship program benefits from a unique integration of nephrology and medical genetics within a single academic health centre, with access to specialized clinics, transplant programs, and advanced genomic and functional research

platforms.

### **How is this Fellowship unique to the province?**

The MUHC has the advantage of integrating adult and pediatric nephrology and medical genetics care in a common institution and offering specialized genetic clinics. The MUHC is also a centre for advanced CKD, the largest kidney transplant centre in the province, and provides a corridor of health service to a large indigenous population in Quebec.

### **Outline how this fellowship will enhance the residency training/program**

Nephrogenetics training is an area that is not well represented in current nephrology residency training programs across Canada. Fellows will be a resource and mentor to existing trainees in nephrology (adult and pediatric), medical genetics and internal medicine residency programs. They will have the opportunity to provide academic didactic sessions, present journal club, and provide clinical teaching in the in-patient and ambulatory setting.

### **List of main supervisors/site/area of expertise**

#### Montreal General Hospital (30%):

Dr. Ahsan Alam – Polycystic kidney disease clinic

Dr. Ratna Samanta – Glomerulonephritis clinic

#### Royal Victoria Hospital (40%):

Dr. Ahsan Alam – Living kidney donor clinic, post-kidney transplant clinic

Dr. Andrey Cybulsky – Pediatric to adult transition clinic

Dr. Tomoko Takano – Glomerulonephritis/CKD clinic

#### Montreal Children's Hospital (30%):

Dr. Thomas Kitzler – Medical genetics clinic, in-patient service, renal genetic oncology clinic

Felicia Russo – Genetic Counselling clinic

Dr. Indra Gupta – Pediatric nephrology clinic (CAKUT)

Asynchronous clinical care may be provided depending on the nephrogenetic referral. Fellows may provide consultative care for patients outside of the ambulatory clinic setting (e.g. in-patient setting, hemodialysis units, transplant unit, etc).

## **Section III**

### **Training Resources:**

Fellows will have access to extensive nephrogenetics research infrastructure, including clinical genomic datasets, specialized renal genetics clinics, and functional research

platforms including cellular and zebrafish models. Access to shared desk space with a computer will be provided to fellows. Online access to journals and educational resources will be available through the McGill Library or PGME.

### **Reading Material N/A**

### **Is research a mandatory component? Yes**

#### **Research Requirements:**

Fellows are expected to undertake at least one research project (e.g. clinical research study, quality improvement/quality assurance project, case report, etc.). The expectation is that the project will be presented at a relevant scientific meeting (local, national, or international) and/or prepared as a manuscript for potential publication.

#### **Research Support:**

Fellows will be assigned a faculty supervisor with relevant expertise to guide their scholarly project. Fellows will have protected research time corresponding to 25-50% of their training scheduled (i.e., 6-13 research blocks).

### **List of clinical rotations:**

The first 12-month, full-time clinical fellowship program will provide intensive, specialized training in nephrology genetics. The curriculum is structured as thirteen 4-week blocks, designed to integrate diverse learning experiences including direct patient care, laboratory exposure, didactic sessions, and dedicated time for scholarly activity. A degree of flexibility to accommodate the fellows' prior training background (e.g., adult versus pediatric nephrology) and specific career aspirations will be considered.

Ambulatory Clinic Rotations (4-6 blocks) form the cornerstone of the fellowship, providing hands-on experience under the supervision of expert faculty. Fellows will participate in a variety of longitudinal clinics where they will see new referrals and provide ongoing management of patients with various genetic renal conditions.

Genetic Counselling Rotation (1-2 blocks) involves working closely with certified genetic counsellor(s). Fellows will observe and actively participate in various counselling scenarios, including pre-test counselling (discussing implications, limitations, potential outcomes), post-test disclosure of results (pathogenic, VUS, negative), and counselling for families regarding recurrence risks and reproductive options. This experience aims to deepen the fellows' understanding of genetic counselling principles, communication strategies for sensitive information, addressing psychosocial impacts, navigating ethical dilemmas, and supporting patient autonomy in decision-making.

Clinical Laboratory Rotation (1 block): Fellows will have dedicated exposure to clinical genomic laboratory workflows and will gain familiarity with various sequencing technologies, including targeted gene panels, exome sequencing, and genome sequencing. Fellows will develop competency in variant interpretation and classification using established guidelines (e.g., ACMG/AMP criteria), including the evaluation of pathogenic, likely pathogenic, and variants of uncertain significance. This training will emphasize the integration of genomic data with clinical phenotypes in collaboration with medical geneticists, genetic counsellors, and laboratory specialists.

Research/Scholarly Project (1-3 blocks): Active participation in scholarly activity is a mandatory component of the fellowship, fostering critical thinking, research skills, and contribution to the field. Each fellow must complete a scholarly project during the year. This could take the form of clinical research (e.g., analyzing registry data, genotype-phenotype correlations), a quality improvement project focused on renal genetics care pathways, or a medical education project. Fellows will be guided through the process of developing a feasible project proposal, obtaining ethics approval if necessary, conducting the project work (data collection, analysis), and preparing the findings for dissemination.

The second year will be adapted to interests of the Fellow, with strong emphasis on working on an independent scholarly activity. In addition to QI/QA or clinical research, Fellows may also consider basic science research in the 2-year Fellowship program. Clinical exposure will also be continued with a focus on clinics that align with the interest and career development of the Fellow. On call duties will be maintained throughout the Fellowship training.

**Will your program involve elective time?**

Elective Rotation (1 block per year) can be taken to provide exposure to related specialized areas. Options might include rotations in Prenatal Diagnosis/Maternal-Fetal Medicine, advanced CKD clinics, transplant clinics, or clinics with a focus on cardiovascular or oncology genetics.

**Will your program involve outside-McGill training?**

Outside McGill training is optional and limited to elective rotations designed to complement training objectives not available at MUHC. Fellows may organize up to 1 block to complement training objectives not readily provided at McGill. Any outside-McGill rotations would require approval of the fellowship program director(s) to ensure the site is accredited.

**Exposure and Case Types:**

Examples include, but are not limited to: Cystic Kidney Diseases, Hereditary Glomerular

Diseases, Tubulointerstitial Diseases, Metabolic and Systemic Disorders with Renal Involvement, and Congenital Anomalies of the Kidney and Urinary Tract.

Fellows will be expected to keep a log book of their cases to ensure they have appropriate sampling of the above-mentioned conditions.

#### **Section IV:**

##### **Clinical Responsibilities:**

Depending on the clinical rotation, fellows will be expected to provide safe, effective and compassionate patient care under supervision, while transitioning towards independent practice as a sub-specialist. Fellows are responsible for patient assessment, diagnostic evaluations, treatment, reassessment and modification of clinical care plans. They will be responsible for completing consultations and maintaining accurate and timely clinical notes. They will be expected to collaborate within and across multidisciplinary teams including other specialists, nursing staff, and the patient. They will act as senior members of the clinical team.

##### **Call Duty Expectations:**

Fellows will participate in home call responsibilities aligned with their primary specialty (nephrology or medical genetics), under faculty supervision and in accordance with FMRQ regulations. Call frequency will be comparable to other fellows within the respective specialty and structured to support educational objectives.

##### **Supervision responsibilities:**

Developing skills as an educator is an integral part of the CanMEDS Scholar role. Fellows will be expected to teach, supervise and provide feedback to junior trainees, such as medical students and residents rotating through nephrology or genetics clinics, during clinical activities or didactic sessions. They may be given the opportunity to do small group teaching of undergraduate medical students in their renal block.

##### **Academic Responsibilities (per academic year):**

Fellows will be expected to present at least 1 time at divisional rounds.

Fellows will present their research/scholarly activity at least 1 time at divisional rounds.

Fellows will present at least 2 times at the divisional journal club.

Fellows will be expected to participate in national academic forums, such as the Canadian Nephro-genetic Consortium (CaN-Gen) virtual case conference.

##### **Additional Responsibilities:**

Fellows may be expected to oversee the site's fellow call schedule.

**Meetings and Conferences:**

Fellows are strongly encouraged to submit abstracts based on their scholarly work for presentation at national or international conferences relevant to nephrology or genetics. Funding (in keeping with the nephrology residency program) will be provided for fellows invited to present their work at a conference at least once per training, in keeping with the policy of the nephrology residency program.

**Vacation:**

Fellows will be provided annual leave similar to FMRQ collective agreement. Currently, residents are entitled to four (4) calendar weeks of paid annual vacation during the year of residency, in addition to statutory hospital holidays. This vacation shall be divisible.

**Section V:****Fellowship Training Objectives (at the end of this fellowship, the trainee should be able to):****Medical Expert:**

- Elicit, document, and interpret a comprehensive, multi-generational family history, constructing a standardized pedigree for patients with suspected genetic kidney disease.
- Perform and interpret a comprehensive physical examination, recognizing syndromic features and dysmorphology relevant to inherited kidney disease syndromes.
- Integrate clinical findings, family history, imaging, and laboratory data to formulate appropriate differential diagnoses for various genetic kidney disease presentations.
- Select, order, and justify appropriate genetic testing strategies (e.g., gene panels, exome sequencing) based on clinical presentation and available resources. (*and Leader*)
- Critically evaluate and interpret genetic test reports, including assessing the clinical significance of variants (pathogenic, likely pathogenic, VUS, benign). (*and Scholar*)
- Apply knowledge of inheritance patterns (autosomal dominant/recessive, X-linked, mitochondrial) to risk assessment and counselling.
- Develop evidence-based, personalized management plans for common genetic kidney diseases, incorporating targeted therapies where applicable. (*and Scholar*)

**Communicator:**

- Counsel patients and families effectively regarding genetic diagnoses, prognosis, inheritance patterns, risks/benefits of testing, and management options, demonstrating empathy.

- Communicate complex genetic information clearly and concisely in written (e.g., consultation letters, chart notes) and oral formats to referring physicians and colleagues.

**Collaborator:**

- Collaborate effectively with medical geneticists, genetic counsellors, laboratory personnel, nephrologists, and other healthcare professionals in a multidisciplinary team.
- Manage transitions of care safely and effectively (e.g., pediatric to adult care, initiation of RRT).

**Leader:**

- Contribute to quality improvement activities related to the care pathway for patients with genetic kidney disease within the local health system.

**Health Advocate:**

- Advocate for patient access to appropriate genetic testing, counselling, specialized care, and novel therapies, addressing potential barriers.

**Scholar:**

- Critically appraise scientific literature relevant to renal genetics and integrate new findings into clinical practice and teaching.
- Design, conduct, and present a scholarly project (research, QI, or educational) relevant to renal genetics.
- Facilitate the learning of patients, families, medical students, residents, and other health professionals about renal genetics topics.
- Develop skills as a clinical educator.

**Professional:**

- Demonstrate adherence to high ethical standards in practice, including informed consent, confidentiality, management of VUS and incidental findings.
- Recognize and manage potential conflicts of interest in clinical practice and research.
- Exhibit self-awareness regarding personal well-being and implement strategies for sustainable practice.
- Engage in reflective practice and respond constructively to feedback for continuous professional development. (*and Scholar*)

**Assessment Process:**

The MUHC NephroGenetics Fellowship program utilizes a variety of assessment tools to enhance learner education in building competency. Some of the tools utilized include ITER

forms (for clinical and research blocks), EPAs, and a summative oral/OSCE exam. Fellows will have at least a bi-annual in-person feedback sessions. A review of the fellow's logbook will be expected to ensure an adequate scope of exposure. Additional research-specific assessments will be scheduled as required.