# Curriculum Vitae
# Grant A. Mitchell, M.D.


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| **Telephone**: | (514) 345-4727; fax (514) 345 4766 |
| **Birth date**: | 31 January 1954 |
| **Birth place**: | Regina, Saskatchewan, Canada |
| **Citizenship**: | Canadian |
| **Languages spoken, written**: | English, French |


## OVERVIEW

I have always been fascinated by how gene mutations affect function and health. I specialized in biochemical genetics, a field that is devoted to this question. I was fortunate to train with outstanding mentors in Canada and abroad in clinical and molecular genetics of metabolic diseases and then to be offered employment at the largest university center in Quebec that follows patients with inborn errors of metabolism. This has allowed me to perform fundamental research in various areas of metabolism. Thanks to the enormous potential of Quebec population genetics and to collaborators from around the province I have gained in-depth experience with many inborn errors that are Quebec genetic diseases, more frequent in Quebec than elsewhere because of genetic founder effects. For several such conditions I have contributed to genetic discoveries, to offering precise molecular diagnosis, to description of the clinical spectrum and to therapeutic trials.

From 2000 to 2011 I served as Division Head fo Medical Genetics. These 10 years were a diversion from my principal activities but came at a crucial juncture for genetics in Québec during which our Division took on its modern form. During this time our physician staff more than doubled, genetic counsellors were introduced to our center, we developed a training program for counsellors, we gained Canadian College of Medical Genetics certification for molecular and biochemical genetic testing and also were granted the privilege of training CCMG fellows in all four disciplines (molecular, biochemical, cyto- and clinical genetics). This period started shortly after the recognition of Medical Genetics as a specialty by the Royal College and at the provincial level in Québec. Our center obtained certification for residency training. There was a critical shortage of medical geneticists and of qualified PhD laboratory directors at the time; many of our trainees are now leaders in medical genetics in Québec and elsewhere. A major time commitment was necessary. The training programs began to alleviate the shortage of medical geneticists in Quebec. Our institution made important contributions to these major changes in Medical Genetics in Québec during this period. I emphasize that these results represent a team effort that relied on many colleagues in Medical Genetics.

During my tenure as Division head, I could not devote the customary effort to research and academic clinical medicine. When it became possible to step down as Division head in favor of an energetic young colleague, I was able to return to full time work as a physician-researcher, concentrating on patient care, molecular diagnostics and restarting my research programs.

We are again starting to make fundamental contributions to the field of lipid energy metabolism. In clinical research we are continuing to exploit the great possibilities of Quebec-wide cohorts for French-Canadian and other founder diseases, with colleagues from across Quebec. This provides many advantages both for science and for patient care, including the use of the largest possible and most homogeneous cohorts to yield the most complete and precise descriptions of disease spectra, optimized power on therapeutic trials and rapid precise molecular diagnosis using Quebec founder mutations.

## TRAINING AND CERTIFICATIONS

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| 1976 | B.Sc (Med), Biochemistry, U. of Saskatchewan |
| 1978 | M.D., U. of Saskatchewan. Gold medal in Medicine. |
| 1978-79 | Rotating internship, Toronto General Hospital. (One month elective in genetics, Hospital for Sick Children, Toronto. Drs Roderick McInnes, and Geoff Sherwood supervisors). |
| 1979-80 | Pediatric residency (R1), Hospital for Sick Children, Toronto. (One mo in genetics). |
| 1980-82 | Pediatric residency (R2-3), Hôpital Sainte-Justine, Montréal. (3-mo in genetics, Drs Serge B. Melançon and Louis Dallaire, supervisors; and 1-mo genetics elective at the Montreal Children's Hospital, Clark Fraser & Charles Scriver, supervisors.). |
| 1983-84 | Bursar, programme d'échange France-Québec and Pediatric residency (year IV), Hôpital Necker-Enfants Malades, Paris, France. Basic and clinical research on inborn errors of metabolism. Prof. Jean-Marie Saudubray, supervisor.<br>Canadian Pediatrics certification (FRCP), November 21, 1983.<br>Bursar, Fondation pour la Recherche Médicale de France. Continuation of research at l'Hôpital Necker-Enfants Malades. Prof. Jean-Marie Saudubray, supervisor. |
| 1984 | Fellowship in clinical genetics, Hôpital Sainte-Justine, Montréal. Serge Melançon and Louis Dallaire, supervisors.<br>Quebec pediatrics certification. |
| 1985-88 | Bursar, Fonds de Recherche en Santé du Québec : Research fellow, Departments of Pediatric Genetics and of Molecular Biology and Genetics, Howard Hughes Medical Institute, Johns Hopkins Hospital, Baltimore, Maryland. Dr David Valle, supervisor.<br>Research topic: The molecular biology of ornithine-delta-aminotransferase. |
| 1987 | Certification, Canadian College of Medical Geneticists (Clinical). |
| 1988-92 | Professeur adjoint de recherche, Département de pédiatrie, U de Montréal. |
| 1989-2007 | Investigator, Federal Center of Excellence "Genetic Basis of Human Disease". |
| 1990- | Certification, American College of Human Genetics (clinical, biochem, molecular). |
| 1991- | Professeur associé, Département de Biochimie et Dépt de Nutrition, U de Montréal |
| 1992 | Royal College Certification in Medical Genetics (June 26, 1992) |
| 1992-95 | Professeur adjoint, Département de Pédiatrie, Université de Montréal. |
| 1995-2002 | Professeur agrégé PTG, Département de Pédiatrie, Université de Montréal. |
| 1995- | Membre du CMDP du Centre hospitalier de l'Université de Montréal (CHUM) |
| 1997- | Québec Medical Genetics Certification (creation of a new specialty) |
| 2000- | Certification in Molecular Genetics, Canadian College of Medical Genetics |
| 2002-06-01 - | Professeur titulaire, Département de Pédiatrie, Université de Montréal |

## AWARDS

Society for Pediatric Research Young Investigator Award, Washington, DC, 1989.
Royal College of Physicians and Surgeons of Canada Medal in Medicine, Ottawa, Ontario, 1992.
Prix d'Excellence du Conseil des Clubs de Service, Montréal, Québec, 16 October 1993.
Prix de Carrière, Canadian Pediatric Society, Charlottetown, PEI, June 23, 2016.

## TEACHING

### University Teaching

- Since 2000, I supervise the course SMC6061, Génétique médicale avancée (three credits). After a brief introduction, professors present recent, medically-important discoveries.

- In the Department of Biochemistry, my niche is to teach students about the medical applications of molecular genetics. For over 10 years I was the only physician and only geneticist to do this. I teach in 2 courses, BCM 1503 (first year) and BCM 6021 (Masters).
- In the Programme du conseil génétique of the Pediatrics Dept, I teach in two courses, CGE6001 and CGE6010. I teach molecular analysis in 6001 and metabolism in 6010.
- The course MMD1030, Génétique médicale, is given to first-year students of medicine. I teach about inborn errors of metabolism (3h).

**Clinical Teaching (Patient-related)**

This is the biggest part of my teaching activities. In addition to impromptu teaching at moments of patient contact, there are 3 more formal teaching moments. (1) The Wednesday morning metabolic meetings. (2) Molecular diagnostics and exome rounds (Thursday afternoons). (3) The academic meetings of the Division of Medical Genetics (Friday afternoons, 2h).

**Diagnostic molecular genetics teaching (Clinical laboratory)**.

*Residents* (Since 2012, I share this responsibility with a young colleague who has taken the lead.) The residency training period is at least 2 months and is accompanied by directed readings, discussions at laboratory rounds and a series of written questions. *Genetic counsellors*: 1 month training in the molecular laboratory. *CCMG fellows* in molecular genetics and other branches of genetics. Our first fellow started in 2012, we currently have two others in training.

**CLINICAL ACTIVITIES** Activities with direct patient contact are the largest sector of my work and teaching although they occupy little space in this CV. The small team of metabolic nurses, dieticians and physicians work closely with the metabolic laboratory to follow and treat children and adults who suffer from inborn errors of metabolism. Many conditions are subject to acute decompensations and close monitoring is necessary. I also co-direct the Genetics section of the molecular diagnostic lab. These activities provide the initial ideas for most of my research.

**ADMINISTRATIVE ACTIVITIES** From 2001-2011 when I was head of the Division of Medical Genetics, my work on committees and reports was extensive. During much of this time I was vice-president of the Association des médecins généticiens du Québec, and I represented the specialty at the meetings and other activities of the Quebec Federation of Medical Specialists. Currently, I am co-directeur of the Axe santé métabolique in the Research Center of CHU Sainte-Justine. I serve on the provincial committee for neonatal screening. I am corresponding member for Canada, Society for the Study of Inborn Metabolic Diseases (SSIEM), 2005-present. I am an editor of the *Metabolic and Molecular Bases of Inherited Disease*.

**RESEARCH:**

**Salary support from research organizations**. From 1988 to 2004 I was funded by the FRSQ (Québec) or the MRC (now CIHR, Federal) as a Bursar, Scholar, and Senior Resarcher-bursar (the final level of funding). **Research Funds**: Currently I am funded by private sources for research on tyrosinemia. I have submitted CIHR applications for work on acyl-CoA metabolism and energy metabolism.

**PUBLICATIONS.** My fundamental research publications concentrate mainly on lipid energy metabolism and my clinical publications, on Quebec genetic metabolic diseases. I am an editor and contributor for a standard text of inborn errors (*Online Molecular and Metabolic Bases of Inherited Disease*), and a contributor to standard texts in pediatrics (Nelson) and pediatric hepatology (Suchy). See annex.