



[Position for a M.Sc. or Ph.D. student](#)

**Site :** Faculty of Pharmacy, Université de Montréal, Pavillon Jean Coutu

**Laboratory of:** Dr. Yan Burelle

**Web site:** <http://bio.pharm.umontreal.ca/burelle>

**Project description:** Leigh Syndrome French Canadian (LSFC): from gene discovery to treatment for patients.

**BACKGROUND:** LSFC is a monogenic mitochondrial disease characterized by Leigh syndrome (a subacute neurodegeneration of the brainstem and basal ganglia) and recurrent episodes of acute and often fatal metabolic acidosis and coma, referred as lactic acidosis "crisis". At the molecular level, our group discovered that mutation of the LRPPRC gene encoding a protein involved in the processing of messengers for mtDNA-encoded respiratory chain subunits is responsible for the disease. However, despite this major advance, translation of this discovery into therapeutic options (which are currently non-existent) for these patients remains a major challenge. This is due in part to our lack of understanding of the biological function of the LRPPRC protein, and of the pathogenic mechanisms, particularly factors that precipitate death following a lactic acidosis "crisis". Our research consortium funded by a CIHR emerging team grant has access to patient fibroblasts and has generated two mouse models with tissue specific knockout of LRPPRC which will allow addressing these questions.

**AIMS:** The project proposed will aim to 1) determine the impact of LRPPRC knockout on mitochondrial function and cellular fate in the two mouse models developed and 2) corroborate findings in cultured fibroblasts from human patients and control donors.

**RESEARCH ENVIROMENT:** This research program offer the opportunity to train in the collaborative and inter-disciplinary environment of the LSFC consortium which is composed of several laboratories with internationally-recognized expertise in the field of genomics, metabolomics, mitochondrial molecular genetics of mitochondrial functional phenotyping. The laboratory of Dr. Yan Burelle, located in the Pavillon Jean Coutu Université de Montréal, benefits from modern infrastructures and of the proximity of several technological platform.

**References :**

- 1- Picard M, Hepple RT, **Burelle Y.** Am J Physiol Cell Physiol. 2011 Oct 26.
- 2- Daussin FN, Godin R, Ascah A, Deschênes S, **Burelle Y.** J Physiol. 2011 Feb 15;589(Pt 4):855-61. 2011 Jan 4.
- 3- Ascah A, Khairallah M, Daussin F, Bourcier-Lucas C, Godin R, Allen BG, Petrof BJ, Des Rosiers C, **Burelle Y.** Am J Physiol Heart Circ Physiol. 2011 Jan;300(1):H144-53.
- 4- **Burelle Y,** Khairallah M, Ascah A, Allen BG, Deschepper CF, Petrof BJ, Des Rosiers C. J Mol Cell Cardiol. 2010 Feb;48(2):310-21. 2009 Sep 18. Review.

**Qualifications:**

*We would like to add to our team a highly motivated graduate student, with lab experience and good bench skill. Applicants should hold an undergraduate degree in biomedical sciences, biochemistry, physiology, cellular or molecular biology (or related topics). Students could be registered in the biochemical sciences or pharmaceutical sciences programs. French and English knowledge is an asset.*

**Contact:**

Applicants should submit by email a resume, a list of publications, university records and the names, with contact information, of two references that could comments on your academic and scientific achievements if possible in one .pdf document.

**Available:** Winter 2012, open until filled. Successful candidates will be supported by research grants (salary based on CIHR guidelines) and will have the opportunities to apply at various competitions for studentship.

**Contact info:** Yan Burelle Ph.D. Associate Professor, Chercheur Boursier Junior II FRQS. <mailto:Yan.burelle@umontreal.ca>.