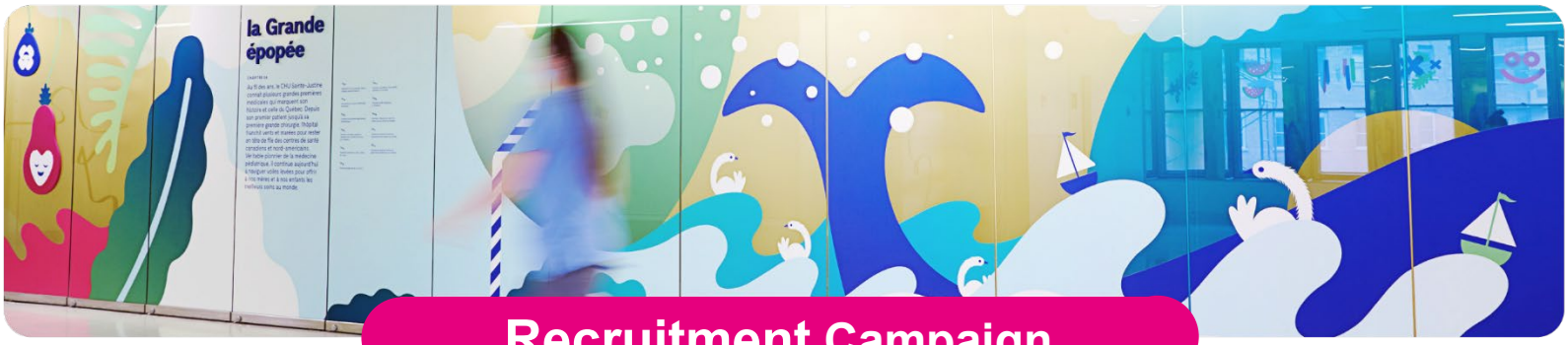

Recruitment Campaign

00 – Characterization and treatment of rare diseases

Type of project : ☒ M.Sc. ☒ Ph. D. ☐ Postdoctoral Fellowship

Research Team

Philippe Campeau, MD
Clinical Professor, Department of Pediatrics, University of Montreal
Researcher, Musculoskeletal Health, Rehabilitation and Medical Technologies Axis, Azrieli Research Center
PCampeauLab.org

Research Interests

We study epilepsy, epigenetic diseases and skeletal dysplasias. We identify disease-causing genes, decipher disease pathophysiology, and improve the management of children affected by these conditions.

Description of the research project

Sub-project 1 : Proactive Identification of Genetic Variants Potentially Amenable to Antisense Oligonucleotide Treatments

There is a promising new approach to treat genetic conditions using “ASOs” (antisense oligonucleotides). A unique advantage of ASOs is that they can be custom designed for a given child’s rare condition. One major challenge to expanding access is in identifying diagnostic DNA variants that could be amenable to an ASO approach, in a timely manner. ASO CONNECT will trial the prospective assessment for ASO amenability of all genetic diagnoses made at SickKids and CHU Sainte Justine over a two year period. The study will also compare rates of potential ASO amenability to other genetic medicine approaches.

Sub-project 2 : Functional analysis of human mutations in Mendelian disease genes using bioluminescence assays

Centre de recherche Azrieli
CHU Sainte-Justine
Le centre hospitalier universitaire mère-enfant
Université de Montréal

In this project, we will study a broader set of diseases using a defined set of medium throuput assays. As part of a genome sequencing project in our hospital, we will identify candidate mutations to explain the manifestations of children suspected to have genetic diseases. The candidate variants will then be assessed using plasmids and lentivirus transfected and transduced in human cell lines. The effects of the variants on splicing, protein stability and protein-protein interactions will be assessed using bioluminescence assays. This will allow us to rapidly provide genetic counselling to affected families, and could help in the development of new assays, such as RNA therapies which modulate protein expression and splicing.

Sub-project 3 : Role of fibronectin in skeletal development and associated disease
We will characterize a newly generated knock-in mouse model for SMD (p.C260G). This mouse model and two complementary already existing cell culture model systems (CRISPR/Cas9 knock-in mutants in ATDC5 cells) will be used to delineate potential therapeutic strategies focusing on ER stress, lysosomal protein degradation, and growth factor signaling.

Candidates are invited to specify their preference for one or other of these sub-projects in their cover letter.

Study programs

Individuals who have completed or are about to complete training in the following areas are invited to apply:

- Bachelor of Science in Biochemistry and Molecular Medicine
- Bachelor of Biological Sciences
- Bachelor of Neuroscience
- Bachelor of Biomedical Sciences
- Related fields

The selected person will have to apply for admission to the University of Montreal, to the master's or doctoral program of the Faculty of Medicine either in Biomedical Sciences, or in Biochemistry, or in Molecular Biology.

Required expertise and skills

- Molecular biology, cell culture, bioinformatics skills.

Funding

Masters (M.Sc.): Minimum funding of $ 24,600 CAD per year will be granted to the candidate, until they receive their own financial support through granting agencies or foundations.

Doctorate (Ph.D.): Minimum funding of $ 27,100 CAD per year will be granted to the candidate, until they receive their own financial support through granting agencies or foundations.

Keywords

Genetic diseases
Epigenetics
Gene therapy
RNA

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