ANNUAL REPORT
2019/2020

THE HESS B. AND DIANE FINESTONE LABORATORY
IN MEMORY OF
JACOB AND JENNY FINESTONE

Submitted by: Dr. David S. Rosenblatt, Holder, Dodd Q. Chu and Family Chair in Medical Genetics, Professor of Human Genetics, Medicine, Pediatrics and Biology, McGill University
MEMBERSHIP

PHYSICIANS AND SCIENTISTS
Gawa Bidla
Brian Gilfix (Medical Biochemistry)
David S. Rosenblatt
David Watkins

CLINICAL SUPPORT STAFF
Maria Galvez
Leah Ladores
Keo Phommarinh
Sina Yak

RESEARCH SUPPORT STAFF
GRADUATE STUDENTS
Lina Sobhy Abdrabo

UNDERGRADUATE AND SUMMER STUDENTS
Marilou Charron (Co-supervise with Yann Joly)
Shira Perton (Observer from New York)
Krithika Ragupathi (Co-supervised with Jean-Baptiste Riviere)
Mark Sorin
ANNUAL REPORT 2019/2020

The Hess B. and Diane Finestone Laboratory in Memory of Jacob and Jenny Finestone was established in 1988. The intent of the donor was to honour the memory of Jacob and Jenny Finestone, and the 80th birthday of Mr. Hess B. Finestone. A permanent endowment was created at McGill University devoted to the advancement of medical genetics. The specific objectives of the endowment are to a) fund research projects related to the field of medical genetics; b) fund trainees in the field of medical genetics; and c) publicize the field of medical genetics through the support of special lectures, visiting professorships and other appropriate means. Dr. David S. Rosenblatt has been Director of the laboratory since its inception, and this annual report describes the activity of his laboratory.

RESEARCH

The Hess B. and Diane Finestone Laboratory in Memory of Jacob and Jenny Finestone is located at the Research Institute of the McGill University Health Centre (MUHC). Our facility is one of two major international referral laboratories for the diagnosis of patients with inherited disorders of folate and vitamin B₁₂ transport and metabolism. It is involved in studying the biochemical and molecular bases of these diseases. Since the MUH has a CLIA certified cellular and molecular diagnostic laboratory, advances in knowledge from research can be immediately translated to clinical diagnostics.

Ongoing projects in the laboratory:

Using genome and RNA sequencing in patients with undiagnosed inborn errors of vitamin B₁₂ metabolism following somatic cell studies

Assay of MTHFD1 specific activity is patients with variants in the MTHFD1 gene

The hunt for causal mutations in patients with known inborn errors of cobalamin metabolism in whom both causal mutations have not been found.

In addition, ongoing collaborations continue with groups at the NIH (Charles Venditti) and at Baylor College of Medicine (Ross Poché) on mouse and zebra fish models of inborn errors of cobalamin metabolism. Our laboratory provides specialized assays for vitamin B₁₂ function in cell lines derived from mice with defects in the pathway.

Other ongoing projects (Brian Gilfix) include the development of high resolution melting for targeted mutation testing in alpha-1-antitypsin deficiency. In addition, Dr. Gilfix is collaborating with Dr. Paul Goodyer to develop an in vitro model to study the effects of aminoglycoside antibiotics on certain classes of detrimental mutations in genes associated inborn errors of vitamin B₁₂ metabolism. This is with the view to develop possible treatments for a number of inborn errors.
RESEARCH OPERATING FUNDS

CIHR Operating Grant, PI – 2016-2019: This grant is for the use of next generation sequencing to discover disorders of vitamin B₁₂ metabolism. Funding Extension until 2021.

NIH, Co-Investigator-2019-2024: Ronin (Thap11) in Neural Crest Development. PI is Ross Poché, Baylor College of Medicine

ORIGINAL PUBLICATIONS

v3 Molecular Test in the Surgical Management of Thyroid Nodules in the Canadian Public Healthcare Setting. Thyroid doi: 10.1089/thy.2019.0539, 2020

GRADUATE STUDENT SUPERVISED

Lina Sobhy Abdrabo  M.Sc.  2016-2019
Title: Next generation sequencing to discover genes underlying methylmalonic aciduria
# FINANCIAL REPORT – 2019/2020

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<tr>
<th>Description</th>
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<tr>
<td>Starting Balance</td>
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<td>*Salary Support and Benefits</td>
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<td>Phones, Computers, Printing, Couriers</td>
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<td><strong>Balance</strong></td>
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*Research Associate: David Watkins (adjusted to include retro salary/benefits expense of $9117)

(For FY20) transferred to 252879 June 1, 2020*