ANNUAL REPORT
2015/2016

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

Submitted by: Dr. David S. Rosenblatt
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MEMBERSHIP

PHYSICIANS AND SCIENTISTS
David S. Rosenblatt
David Watkins

ADMINISTRATIVE SUPPORT STAFF
Danielle Piazzoli (Veyre)

CLINICAL SUPPORT STAFF
Maria Galvez
Leah Ladores

RESEARCH SUPPORT STAFF
GRADUATE STUDENTS
Jordan Chu
Mihaela Pupavac
ANNUAL REPORT 2015/2016

The Hess B. and Diane Finestone Laboratory in Memory of Jacob and Jenny Finestone, was first 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. Material relating to members of the Department of Human Genetics at McGill and the Departments of Medical Genetics at the McGill University Health Centre and the Jewish General Hospital should be sought in the respective university or hospital annual reports.

RESEARCH

The Hess B. and Diane Finestone Laboratory in Memory of Jacob and Jenny Finestone, had been located on Livingston 3 of the Montreal General Hospital Site of the McGill University Health Centre. This is the first year that it has been located at the new Glen Site. The new site has the advantage of grouping many leading scientists from the hospital communities on one site. 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 MUHC has a CLIA certified cellular and molecular diagnostic laboratory, advances in knowledge from research can be immediately translated to clinical diagnostics.

2015-2016 has seen a number of scientific highlights:

1) Dr. Rosenblatt has continued to champion the concept of RaDiCAL (Rare Disease Collaboration for Autosomal Loci). He has proposed the use of single patients with Mendelian disorders to discover new genes. With colleagues at McGill he has continued to use exome sequencing for new gene discovery.

2) The group has compared the results that have been obtained over decades using a somatic cell genetic approach, with those that can now be obtained using next generation sequencing panels. This has confirmed the utility of both techniques.
RESEARCH OPERATING FUNDS

CIHR Operating Grant, PI – 2009-2014. A one year no cost extension was granted until October 2015. Much of the current year was structured in anticipation of being successful in the Project Grants competition in 2016.

ORIGINAL PUBLICATIONS


ABSTRACTS


TEACHING

**David Watkins**

**Biology 575**

Department: Biology/Human Genetics  
Format: Lecture  
**Title:** *Inborn Errors of Cobalamin Transport and Metabolism*  
Role: Lecturer  
Level: Undergraduate/Graduate  
Time: 2 1.5-hour class lectures

**David Rosenblatt**

**Unit 8**

Department: Human Genetics  
Format: Lectures  
Role: Lecturer-2 sessions  
**Titles:** *Introduction to Medical Genetics*  
*Huntington Disease*  
Level: Medical Students  
Time: 2 lectures -1 hour each
## FINANCIAL REPORT – 2015/2016

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<th>Description</th>
<th>Amount</th>
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<td>*Salary Support and Benefits</td>
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<td>Materials and Supplies</td>
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<td>Conferences, Travel, Special Events</td>
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<td>Phones, Pagers, Computer, Printing, Couriers</td>
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<td>Miscellaneous</td>
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<td><strong>Total Expenses</strong></td>
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<td><strong>Balance</strong></td>
<td><strong>$ 23,912</strong></td>
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* Chu, Pupavac, Rosenblatt, Veyre, Watkins