ANNUAL REPORT
2013/2014

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

Submitted by: Dr. David S. Rosenblatt
# TABLE OF CONTENTS

MEMBERSHIP ........................................................................................................................................ 2
ANNUAL REPORT 2013/2014 .................................................................................................................. 3
RESEARCH ........................................................................................................................................... 3
RESEARCH OPERATING FUNDS ............................................................................................................. 4
INVITED PRESENTATIONS ..................................................................................................................... 4
ORIGINAL PUBLICATIONS .................................................................................................................... 4
ABSTRACTS .......................................................................................................................................... 6
TEACHING ............................................................................................................................................ 6
FINANCIAL REPORT – 2013/2014 ........................................................................................................ 8
MEMBERSHIP

PHYSICIANS AND SCIENTISTS
David S. Rosenblatt
David Watkins

ADMINISTRATION
Tracy Wang
Danielle Veyre

CLINICAL SUPPORT STAFF
Maria Galvez
Leah Ladores

RESEARCH SUPPORT STAFF
Wayne Mah (with James Coulton)
Maira Moreno (with Loydie Jerome Majewski)
Tania Cruz

GRADUATE STUDENTS
Alison Brebner
Justin Deme (with James Coulton)
Mihaela Pupavac

UNDERGRADUATE AND SUMMER STUDENTS
Francis Petrella
Tracy Wang (with Dr. Chantal Seguin)
ANNUAL REPORT 2013/2014

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, is located on Livingston 3 of the Montreal General Hospital Site of the McGill University Health Centre. It 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 Dr. Rosenblatt directs a certified molecular diagnostic laboratory adjacent to the research laboratory, advances in knowledge from research can be immediately translated to clinical diagnostics.

2013-2014 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) With colleagues at the University of Colorado, the NIH and the University of Zurich, we described mutations in the HCFC1 gene that codes for a restriction factor. These mutations result in combined homocystinuria and methylmalonic aciduria, a phenocopy of the cblC inborn error. Of great importance, because the HCFC1 gene is located on the X chromosome, the genetic counseling of this disorder differs from that of the other inborn errors of cobalamin metabolism.
3) Together with James Coulton in the Department of Microbiology and Immunology, we continued studies on the intracellular function of proteins involved in intracellular cobalamin metabolism.

4) With Loydie Jerome-Majewska, we determined that the \textit{Mmache} gene is required for implantation in the mouse.

---

**RESEARCH OPERATING FUNDS**

CIHR Operating Grant, PI – 2009-2014

---

**INVITED PRESENTATIONS**

November 5, 2013
Genomics in Metabolism
Copenhagen, Denmark
Title: \textit{Using next generation sequencing to discover novel steps in one-carbon metabolism}

December 13, 2013
Journee Francophones de Nutition
Bordeaux, France
Title: \textit{Les maladie hereditaires du metabolism de la vitamine B\textsubscript{12}: contribution a la connaissance du metabolism cellulaire.}

---

**ORIGINAL PUBLICATIONS**


Fischer S, Huemer M, Deodato F, Ballhausen D, Baumgartner M, Boneh A, Burlina AB, Cerone R,

ABSTRACTS


TEACHING

David Watkins

Biology 575

Department: Biology/Human Genetics
Format: Lecture
Title: Inborn Errors of Folate and Cobalamin Transport and Metabolism
Role: Lecturer
Level: Undergraduate/Graduate
Time: 3 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 – 2013/14**

Starting Balance $94,625

<table>
<thead>
<tr>
<th>Description</th>
<th>Amount</th>
</tr>
</thead>
<tbody>
<tr>
<td>*Salary Support and Benefits</td>
<td>$ 50,558</td>
</tr>
<tr>
<td>Materials and Supplies</td>
<td>$ 4,287</td>
</tr>
<tr>
<td>Conferences, Travel, Special Events</td>
<td>$ 17,100</td>
</tr>
<tr>
<td>Memberships</td>
<td>$ 2,281</td>
</tr>
<tr>
<td>Phones, Pagers, Computer, Printing, Couriers</td>
<td>$ 1,619</td>
</tr>
<tr>
<td>Miscellaneous</td>
<td>$ 597</td>
</tr>
</tbody>
</table>

Total Expenses $76,442

**Balance** $18,183

*Michaud, Notte, Rosenblatt, Veyre, Wang*