ANNUAL REPORT
2009/2010

THE HESS B. AND DIANE FINESTONE LABORATORY
IN MEMORY OF
JACOB AND JENNY FINESTONE
http://www.mcgill.ca/finestone

Submitted by: Dr. David S. Rosenblatt
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MEMBERSHIP

PHYSICIANS AND SCIENTISTS
David S. Rosenblatt
David Watkins

ADMINISTRATION
Laura Benner

CLINICAL SUPPORT STAFF
Maria Galvez
Jocelyne Lavallée

RESEARCH SUPPORT STAFF
Suzanne Dufrasne
Maria Plesa

GRADUATE STUDENTS
Natacscia Anastasio
Junhui Liu
Isabelle Racine-Miousse
Mihaela Pupavac

SUMMER STUDENTS
Simon Zhu

INDEPENDENT STUDIES STUDENTS
Lara Reichman
Laura Nuñez Dempsey
The Hess B. and Diane Finestone Laboratory in Memory of Jacob and Jenny Finestone was established in 1988 to honour the memory of Jacob and Jenny Finestone and the 80th birthday of Mr. Hess B. Finestone by providing a permanent endowment 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. This Finestone report will restrict itself to the activities of the Director. Material previously found in this report 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 at 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 B_{12} 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 diagnosis.

2009-2010 has seen a number of scientific accomplishments:

1. With colleagues in California, we have shown that with the advent of expanded newborn screening, there will be unusual clinical presentations of patients. We published, in the Journal of Pediatrics, a report of an asymptomatic mother, who was diagnosed as having the \textit{cblC} inborn error of vitamin B_{12} metabolism, based on an abnormal screening result in her baby.

2. With colleagues at the Cleveland Clinic, we showed that the product of the \textit{MMACHC} gene that is responsible for \textit{cblC} is capable of processing alkylcobalamins. This work was published in Molecular Genetics and Metabolism.

3. With Fei Li, a McGill undergraduate student, we published, in Molecular Genetics and Metabolism, a systematic review of the role of vitamin B_{12} in birth defects.

4. With David Watkins, four chapters relating to folate and vitamin B_{12} metabolism were published in major texts.

5. Dr. Rosenblatt presented a plenary session at the International Congress of Inborn Errors of Metabolism (ICIEM) in San Diego on the role of homocysteine in heart disease.
RESEARCH OPERATING FUNDS

CIHR, Operating Grant, PI – 2009-2014
CIHR, Group Grant, Co-Investigator – 1975-2009

MEETINGS AND PRESENTATIONS

August 31, 2009
International Congress of Inborn Errors of Metabolism (ICIEM)
Title: Homocysteine and Coronary Heart Disease
San Diego, CA

March 10, 2010
Medical Grand Rounds
Title: Homocysteine and Coronary Heart Disease
MUHC – Montreal General Hospital

ORIGINAL PUBLICATIONS


**CHAPTERS**


**TEACHING**

**Biology 575**

Department: Biology/Human Genetics  
Format: Lecture  
**Title:** *Inborn Errors of Folate and Cobalamin Transport and Metabolism*  
Role: Lecturer and Course Coordinator  
Level: Undergraduate/Graduate  
Time: 6 hours

**Unit 8**

Department: Human Genetics  
Format: Lecture and Small Group Teaching  
Role: Lecturer-2 sessions  
**Title:** *Introduction to Medical Genetics*  
**Huntington Disease**  
Level: Medical Students  
Time: 2 lectures plus 3 2-hour sessions, 8 hours in total

Starting Balance $105,028

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<td>*Salary Support and Benefits</td>
<td>$58,111</td>
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<td>Computer Charges</td>
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<td>Materials, Supplies, Phones &amp; Pagers</td>
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<td>Miscellaneous</td>
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Total Expenses $85,295

**Balance $19,733

Notes

* Rosenblatt, Dufrasne, Veyre, Leslie, Benner, Anastasio

** Balance was capitalized