ANNUAL REPORT
2011/2012

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 (Maternity Leave)
Danielle Veyre
Sandra Anastasio

CLINICAL SUPPORT STAFF
Maria Galvez
Leah Ladores

RESEARCH SUPPORT STAFF
Wayne Mah
Maira Moreno
Maria Plesa

GRADUATE STUDENTS
Isabelle Racine-Miousse
Laura Dempsey Nuñez
Peg Illson
Jaeseung Kim

UNDERGRADUATE AND SUMMER STUDENTS
Alison Brebner
Selim Fares (Visiting Student from Paris)
Francis Petrella
ANNUAL REPORT 2011/2012

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 honor 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 the third floor of Livingston Hall 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 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 immediate translated to clinical diagnosis.

2011-2012 has seen a number of major scientific highlights:

1) Dr. Rosenblatt has championed the concept of RaDiCAL (Rare Disease Collaboration for Autosomal Loci). He proposed the use of single patients with Mendelian disorders to discover new genes.

2) With Jacek Majewski at the McGill University-Genome Quebec Innovation Centre, and with colleagues at McGill and around the world, we used next generation sequencing technology to find the gene responsible for three different diseases. This has been a proof of principle that McGill can compete with the best in the world in gene discovery using this advanced technology.

3) With the group of Don Jacobsen at the Cleveland Clinic, we described the proteome of cblC cell lines.

4) With Loydie Jerome-Majewska, we showed different expression patterns of MMACHC and MMADHC in the developing mouse. It was particularly important to show MMACHC expression in the developing heart, since some patients with the cblC disorder have congenital heart disease.
5) We described novel mutations in patients with the *cblF* form of combined homocystinuria and methylmalonic aciduria.

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**RESEARCH OPERATING FUNDS**

CIHR, Operating Grant, PI – 2009-2014

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**INVITED PRESENTATIONS**

August 30 - September 2, 2011
Annual Symposium of the Society for the Study of Inborn Errors of Metabolism
Geneva, Switzerland

**Title: Disorders of Folate Transport and Metabolism**

March 31 - April 3, 2012
SIMD Annual Meeting
Westin Charlotte
Charlotte, NC

**Title: Combined Homocystinuria and Methylmalonic aciduria (cblC)**

April 27, 2012
Harvey L Levy Symposium
Beyond Archibald Garrod: Understanding and Treating Metabolic Disorders
Children’s Hospital Boston Division of Genetics and Metabolism
Harvard Medical School
Boston, Massachusetts

**Title: Combined Homocystinuria and Methylmalonic Aciduria (cblC): From Patient to Gene to Function-and Beyond-RaDiCAL Thoughts**
ORIGINAL PUBLICATIONS


CHAPTERS


Shevell M, Watkins D, Rosenblatt DS. Disorders of Cobalamin and Folate Metabolism and Epilepsy. In: The Causes of Epilepsy: Common and Uncommon Causes in Adults and Children,


ABSTRACTS


Mioussse IR, Plesa M, Rosenblatt DS, Coulton JW. Phage display predicts regions on MMACHC that bind to its partner MMADHC in human cobalamin metabolism. ICHG. American Society for Human Genetics, Montreal, October 11-15, 2011.


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**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  
Course Co-Supervisor with Dr. Nancy Braverman

**Unit 1**  
Department: Human Genetics  
Format: Small Group Teaching  
Role: Group Leader  
*Title*: Genetics: Problem Solving  
Level: Medical Students-First Year  
Time: 1 2-hour group session
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
## FINANCIAL REPORT – 2011/2012

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**Notes**  

* Rosenblatt, Veyre, Benner, Kim, Dempsey-Nunez, Illson, Fares.*