



# ANNUAL REPORT

## 2006/2007

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, Director*

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## **MEMBERSHIP**

### **PHYSICIANS AND SCIENTISTS**

Ken Dewar  
Eleanor Elstein  
William D. Foulkes  
T. Mary Fujiwara  
Brian M. Gilfix

Kenneth Morgan  
David S. Rosenblatt  
Marc Tischkowitz  
Patricia N. Tonin  
David Watkins

### **ADMINISTRATION**

Maryse Bibeau  
Razia Chanda  
Maria Fiorito  
Vanessa Flannery  
Claire Goguen

Yasmin Karim  
Jenny Koulis  
Morgen Patterson  
Danielle Veyre

### **CLINICAL SUPPORT STAFF**

Suzanne Dufrasne  
Maria Fiorito  
Jennifer Fitzpatrick  
Stephanie Fox  
Maria Galvez  
Angela Hosack

Maria Lalous (maternity leave Dec/06)  
Jocelyne Lavallée  
Laura Palma  
Laura Robb  
Nora Wong  
Sonya Zaor

### **RESEARCH SUPPORT STAFF**

Suzanna Arcand  
Joana Dias  
Nancy Hamel  
Kimberley Kotar  
Mathieu Lemire  
Lili Li  
I-Ching Lu

Kevin McKee  
Nicole Roslin  
Nelly Sabbagian  
Zhen Shen  
Jessica Wasserscheid  
Daniel Yavin

### **POST-DOCTORAL FELLOWS/CLINICAL FELLOWS**

Yohan Bosse  
Sarah Gail Buxbaum  
Luca Cavallone  
P. Campeau, MD  
Patrick Dion

Nicholas Dupre  
Li Fan  
Claudia Gaspar  
Karen Gambaro  
Richard Le Blanc

Alexandre Montpetit  
Laura Oksanen  
Matthew Oughton  
Tomi Pastinen

Michael Quinn  
David Serre  
André Toulous  
Ahmet Yilmaz

## **GRADUATE STUDENTS**

Faith Au-Yeung  
Amanpreet Badhwar  
Ashley Birch  
Neal Cody  
Patrick Cossette  
Daniel Darmond  
Adrian Diaz  
Qingling Duan  
Vincenzo Forgetta  
Julie Gauthier  
Abigail Gradinger  
Heidi Howard  
Margaret Illson  
Tayma Khalil  
Kathleen Klein  
Peter Lee

Jordan Lerner-Ellis  
Anastasia Levechenko  
Amanda Loewy  
Junhui Liu  
Alexandre Marcil  
Christianne Messaed  
Emmanuel Mongin  
Emily Moras  
Emil Nashi  
Nicole Palmour  
Guillaume Pare  
Isabelle Racine-Miousse  
Saravanan Sundararajan  
Paulina Wojnarowicz  
Lama Yamani

## **SUMMER STUDENTS AND INDEPENDENT STUDIES STUDENTS**

Emily Brown  
Marie Caudrelier  
Joanna Grater

Carter Li  
David Novak  
Rebecca Shapiro

## ANNUAL REPORT 2006/2007

The Hess B. and Diane Finestone Laboratory in Memory of Jacob and Jenny Finestone exists in order to promote the field of medical genetics at McGill University. Dr. David S. Rosenblatt has been Director of the laboratory since its inception. The laboratory was established with the help of an endowment to McGill University and funding is used to advance the academic goals of the Division of Medical Genetics in the Department of Medicine of the McGill University Health Centre (MUHC). As such, this report also serves as the Annual Report of the Division of Medical Genetics of the Department of Medicine of the MUHC. It is available on the Internet (<http://www.mcgill.ca/finestone>). Since the University Division in Medicine also has included the Division at the Jewish General Hospital, this report also encompasses parts of that activity. Within the past few years, major advances have occurred with the creation of the Department of Medical Genetics at the Jewish General Hospital, and more recently, the Department of Medical Genetics at the MUHC.

### Highlights: Research

**Dr. William Foulkes and Dr. Marc Tischkowitz** demonstrated the involvement of *PALB2/FANCN* in hereditary breast cancer families.

**Dr. David Rosenblatt** and his collaborators have shown that mutations in the *MCEE* gene can result in elevated methylmalonic acid excretion. With colleagues in the New York, he has described mutations in the *PCFT* gene in patients with Hereditary Folate Malabsorption, and with colleagues in Nebraska, he has shown that the polymorphic background of the *MTRR* gene affects the phenotype of a disease-causing mutation.

**Dr. Patricia Tonin** developed a derivative ovarian cancer cell line through the transfer of chromosome 3 fragments, using a novel modification of an established technique involving whole chromosome transfer. This cell line is now being used to identify the underlying chromosome 3p gene(s) involved in tumour suppression. A positive outcome has been the modulation of transcriptome networks, which will enable them to prioritize gene candidates for an independent project aimed at elucidating genes involved in ovarian tumourigenesis.

### Highlights: Awards

**Dr. William Foulkes** was awarded a highly competitive Chercheur Nationaux Award from the FRSQ.

**Dr. Marc Tischkowitz** was awarded a Chercheur Clinicien Award from the FRSQ.

## **Highlights: Teaching**

Dr. Nicholas AhMew completed the RV year of his training program and successfully passed the examinations of the Royal College in Medical Genetics. He plans to spend next year in China before continuing with fellowship training. Dr. Philippe Campeau completed his RIV year and served as Chief Resident. He is to be congratulated on an excellent job. Dr. Khalid Al-Tihli, Dr. Maha Al-Awadi, and Dr. Mouna Ben Amor completed their RIII year and Dr. Daniela D'Agostino and Dr. Ahmad Alfares their RI year.

## **Highlights: Clinical**

After more than five years, a Department of Medical Genetics was finally established at the MUHC. Dr. Teresa Costa has been appointed the first Chief of this hospital department. Dr. Costa has held positions in the past in Montreal, Halifax and Toronto.

In keeping with the new RUIS structure mandated by the Government of Quebec, a provincial committee and a local McGill committee have been established to examine how to best serve the interest of the Quebec population in the area of Medical Genetics.

## **Clinical Statistics:**

### **Suzanne Dufrasne, M.Ps, Psychologist:**

#### *Huntington Disease*

Number of new patients seen: 16

Number of sessions in person: 28

Number of counselling phone calls with patients (pre-session, follow-up): 48

Number of counselling phone calls with health professionals: 20

Total number of interventions with patients: 76

#### *Other Diseases (Creutzfeldt-Jakob, Familial adenomatous polyposis)*

Number of patients seen: 2

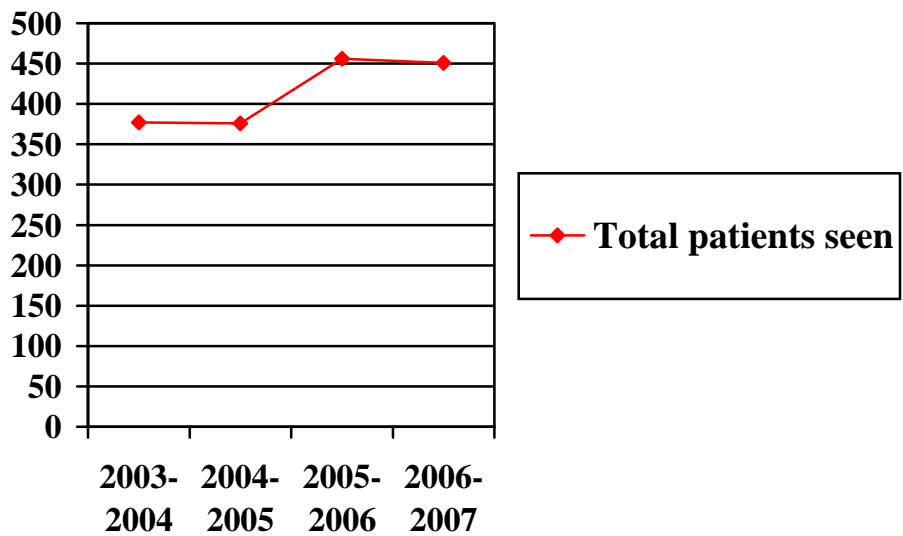
### **Stephanie Fox, MSc, MS, Genetic Counsellor:**

#### ***Montreal General Hospital Hereditary Cancer Clinic – April 1 to December 4, 2006:***

Number of new patients seen: 75

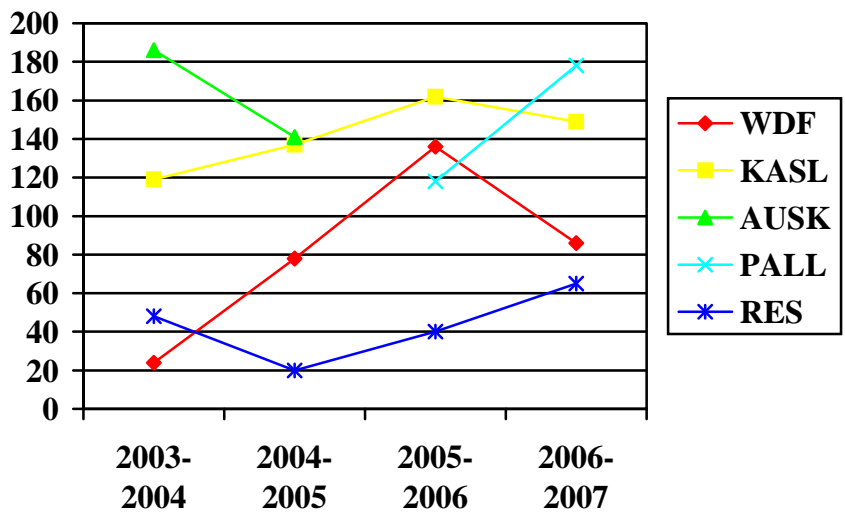
Number of follow-up patients seen: 80

Total cases seen: 155



**Total Patients Seen at MUHC**

2003-2004: 377  
 2004-2005: 376  
 2005-2006: 456  
 2006-2007: 451 ~20% increase since 2004-2005



**NOTES FOR LEGEND:**

**Fiscal periods are calculated from April 1<sup>st</sup>- March 31<sup>st</sup>.**

- Totals for **KASL** (Lidia Kasprzak) take into account all patients seen by KASL and/or her replacements (**SFOX** [Stephanie Fox], **MLAL** [Maria Lalous], **LROB** [Laura Robb]) over each financial period.
- For 2005-2006 fiscal period, **PALL** (Laura Palma) totals are calculated from time of start date **July 1, 2005** and therefore, do not include the statistics for April 1<sup>st</sup>- June 30<sup>th</sup>, 2005).
- Stats for **RES** include the total number of patients seen by either genetics residents and/or **FITZJ** (Jennifer Fitzpatrick)

## Research Interests and Accomplishments of Individual Members:

**Dr. Eleanor Elstein** has an active clinic in the area of cardiovascular genetics, which evaluates inherited cardiac diseases as well as systemic genetic diseases that have cardiac manifestations.

**Dr. Ken Dewar** and his laboratory are using genomics and bioinformatics technologies to study genome structure and variation. One focus of the lab is to develop genetic mapping tools for investigating complex trait mapping in a nonhuman primate, the vervet monkey. Using comparative genomics approaches they have leveraged other genome projects (human, chimpanzee, rhesus monkey) to streamline the discovery of markers of genetic variation (SNPs). They have also embarked upon the generation of corresponding genome wide physical map, entailing the paired-end sequencing and genome alignments for >200,000 BAC clones. The vervet BAC map is being used to delineate chromosomal breakpoints and to identify vervet BAC clones associated with evolutionary recently derived centromeres and pericentromeric regions.

His group is also interested in understanding genome structure and virulence factors in the human bacterial pathogen *Clostridium difficile*. *C. difficile* remains a serious health risk in Quebec, North America, and Europe, in part due to an epidemic strain of increased virulence which has emerged in the last several years. The laboratory has performed the sequencing of the entire 4 Mb genome of a virulent strain from Montreal, and is now generating the genome sequences of other isolates with important clinical phenotypes. Cross-genome comparisons of gene content and organization will be used to identify additional candidate genes involved in pathogenicity.

**Dr. William Foulkes** and colleagues have been characterizing mutations in the new breast cancer gene PALB2. In addition, he has worked with BRCA1-mutated cell lines to try to develop novel treatments. With George Chong, he has described several novel mutations in genes associated with hereditary colorectal cancer. As mentioned above, in recognition of the excellence of his work, he has been awarded a Chercheur Nationaux award from the FRSQ.

**Mary Fujiwara** studies the distribution and maintenance of genetic variability, including deleterious alleles, in populations. During the current reporting period, she continued collaborations on the Hutterite population of North America, an inbred population isolate, to map and further delineate the clinical variability of a Joubert-related cerebello-oculo-renal syndrome. In addition, she participated in the mapping of two other rare Mendelian diseases, Sanfilippo C syndrome and methylmalonic aciduria and homocystinuria (cblC type), in patients and families collected from around the world. She also participated in collaborations on identifying underlying genetic susceptibility for complex diseases including kidney stone formation and early-onset coronary heart disease in the French-Canadian population.

She also collaborate with Daniel Bichet (Hôpital Sacré-Coeur de Montréal) on the genetics of nephrogenic and neurohypophyseal diabetes insipidus. The study of mutations in three different genes has shown that the mode of inheritance can differ based on the particular mutation. They maintain locus specific mutation databases for these genes which encode the hormone arginine vasopressin, its receptor, and a water channel (*AVP*, *AVPR2*, and *AQP2*, respectively).



**Dr. Brian Gilfix** focuses on two main areas of research: 1) Homocysteine and its Metabolism/Inborn Errors of Metabolism (Homocystinuria)-they have a large cohort of patients in the Adult Genetics Clinic with homocystinuria. He is using this opportunity to explore new treatments of homocystinuria and investigate the effect of elevated homocysteine on other risk factors for cardiovascular disease; 2) Development of Laboratory Methods in Molecular Diagnostics and HPLC-he is developing and implementing assays based on hybridization probes to replace standard assays based on restriction fragment length polymorphism used to genotype single nucleotide polymorphisms in the clinical laboratory. The benefit of this is decreased net cost and faster turnaround time.

**Dr. Ken Morgan** has a major interest in human population genetics and genetic epidemiology. He is involved in the genetic analysis of Mendelian and complex traits in humans and mice. Accomplishments related to human genetics include mapping rare Mendelian diseases. Ongoing collaborations include segregation and linkage analysis of intermediate phenotypes contributing to calcium kidney stone formation; genetic susceptibility to inflammatory bowel disease in children; and reducing the complexity of pedigrees in founder populations such that statistical analysis is computationally feasible.

He is a member of the CIHR Institute of Genetics Priorities and Planning committee for genetic epidemiology and statistical genetics and was on the organizing committee of the “Second Annual Canadian Genetic Epidemiology and Statistical Genetics Meeting”, held in Toronto, 15-17 April 2007.

**Dr. David Rosenblatt** and his laboratory continue to be the major international referral source for the diagnosis of patients with inherited disorders of folate and vitamin B<sub>12</sub>. They are involved in studying the biochemical and molecular bases of these diseases. This year they have expanded mutation analysis among patients with combined homocystinuria and methylmalonic aciduria due to mutations in MMACHC. They have also shown that mutations in the MCEE gene can result in elevated methylmalonic acid excretion. With colleagues in the New York, they have described mutations in the *PCFT* gene in patients with Hereditary Folate Malabsorption, and with colleagues in Nebraska, they have shown that the polymorphic background of the *MTRR* gene affects the phenotype of a disease-causing mutation. Dr. Rosenblatt continues to serve as Chairman of the Department of Human Genetics. In August 2006, he served as the Chairman of the FASEB Summer Research Conference on One-Carbon Metabolism in Indian Wells, California.

**Dr Marc Tischkowitz** has been establishing a research program at the Segal Centre, Jewish General Hospital to undertake innovative and effective research in the field of hereditary predisposition to cancer, with a focus on the genetic links between breast cancer and Fanconi Anemia. He was part of an international collaboration that identified PALB2/FANCN as a new Fanconi Anemia gene and together with Dr Foulkes he helped to establish that PALB2 is a breast cancer predisposition gene.

**Patricia Tonin** works in two principal areas of research; these are described on the web site: [www.toninlab.mcgill.ca](http://www.toninlab.mcgill.ca).

A. The Molecular study of Human Epithelial Ovarian Cancer

More than 70% of women diagnosed with ovarian cancer die of the disease. Our knowledge of the molecular events associated with the development and progression of epithelial ovarian cancer has been limited by the lack of a suitable model system. Also, since the disease is often diagnosed at a late stage when numerous complex chromosomal changes have already taken place, the early molecular events remain largely unknown. Research in the lab is focused on the identification of tumour suppressor genes, particularly those physically associated with chromosomes 3p and 17q. Various molecular genetic techniques are used to identify them, which include, allelic content analysis such loss of heterozygosity studies and single nucleotide (SNP) polymorphism analyses, large-scale gene expression assays (Affymetrix platform), and more recently functional approaches based on chromosome transfer fragment and gene complementation. A major accomplishment of the last year was development of a derivative ovarian cancer cell line through the transfer of chromosome 3 fragments. This cell line was developed using a novel modification of an established technique involving whole chromosome transfer. This cell line is now being used to identify the underlying chromosome 3p gene(s) involved in tumour suppression. A positive outcome has been the modulation of transcriptome networks, which will enable them to prioritize gene candidates for an independent project aimed at elucidating genes involved in ovarian tumourigenesis. [See News item at [www.toninlab.mcgill.ca](http://www.toninlab.mcgill.ca) for Press Release.]

In collaboration with colleagues at the CHUM-Notre Dame, they investigate gene expression profiles of ovarian cancer samples with the aim of identifying signature patterns of gene expression in order to elucidate molecular pathways important in ovarian tumourigenesis.

B. Breast and Ovarian Cancer Susceptibility Genes

Hereditary breast and ovarian cancer accounts for approximately 5-10% of all breast and ovarian cancers. A large majority of cancer families are attributed to germline mutations in BRCA1 and BRCA2. However, about 40% of cancer families are negative for mutations in these known genes. Dr. Tonin's group is also focused on determining the contribution of known and unknown cancer susceptibility genes to inherited predisposition to breast and ovarian cancer. They focus on the founder French Canadian population of Quebec as reviewed in a special issue of the *Bulletin du Cancer*, which commemorated the 100<sup>th</sup> anniversary of *Société Française du Cancer* in France. In 2006 they described a new founder BRCA2 mutation found to recur in the French Canadian population. Moreover, they have extended their analysis to further refine those cancer families most likely to harbour BRCA1 or BRCA2 mutations.

## MEETINGS AND PRESENTATIONS

### KEN DEWAR

- February 23, 2006 Department of Physiology Seminar Series,  
McGill University, Montreal, QC  
**Title:** Comparative Genomics: Comparing and contrasting aspects of  
genome evolution
- November 26, 2006 Academic Tribute and Colloquium in Honor of Dr. Abraham Fuks  
McGill University, Montreal QC  
**Title:** I have 50 % of my mother's genes, yet am 99% identical to a  
chimpanzee

### SUZANNE DUFRASNE

- November 21, 2006 Seminar for Genetic Counselling students, with Dr. Sylvain Chouinard.  
Ste-Justine Hospital, Montreal, QC  
**Title:** Predictive testing for Huntington Disease

### WILLIAM FOULKES

- February 2, 2006 5<sup>th</sup> International "From Gene to Cure" Congress  
Vrije Universiteit Amsterdam  
Amsterdam, Netherlands  
**Title:** Genetic Risk Assessment
- February 2, 2006 5<sup>th</sup> International "From Gene to Cure" Congress  
Vrije Universiteit Amsterdam  
Amsterdam, Netherlands  
**Title:** Prevention of hereditary breast cancer
- June 15, 2006 Toronto Breast Cancer Symposium 2006  
Metro Toronto Convention Center  
Toronto, Ontario  
**Title:** Genetics and Breast Cancer: An Update
- April 20, 2006 "Basal-like and BRCA1-associated Breast Cancer" meeting  
Harvard Club  
Boston, MA, USA  
**Title:** Clinico-pathological features of basal-like/BRCA1 tumors
- August 16, 2006 Australian Ovarian Cancer Study and the Family Cancer Clinics of  
Australia  
Couran Cove Island Resort  
Stradbroke Island, Australia

**Title:** Recent advances in understanding of the inherited susceptibility to cancers of the prostate, pancreas, stomach and colorectum

August 18, 2006 Australian Ovarian Cancer Study and the Family Cancer Clinics of Australia  
Couran Cove Island Resort  
Stradbroke Island, Australia  
**Title:** Hereditary breast cancer: from pathology to treatment and beyond

October 28, 2006 10<sup>th</sup> Annual Cincinnati Comprehensive Breast Cancer Conference  
Cutting Edge Strategies in Breast Cancer: The next decade  
Cincinnati, OH, USA  
**Title:** Breakthrough treatments for BRCA1 and BRCA2 mutation carriers

November 9, 2006 Cancer Colloquia IV: Cell and Molecular Biology of Breast Cancer  
University of St-Andrews  
St-Andrews, Scotland  
**Title:** Hereditary breast cancer: from pathology to treatment and beyond

#### **BRIAN GILFIX**

October 6, 2006 McGill Neurology Conference  
Montreal General Hospital  
Montréal, QC  
**Title:** Myelopathy due to vitamin B12 deficiency

November 14, 2006 Speaker, Life as a Physician Seminar Series  
McGill Faculty of Medicine  
Montréal, QC

#### **KENNETH MORGAN**

August 3-5, 2006 Invited speaker in workshop: The Genomic Revolution and the Origin of  
of Humanity.  
Origins Institute, McMaster University  
Hamilton, Ontario  
**Title:** Challenges and approaches to understanding complex diseases.

October 19, 2006 Invited speaker at conference: Innovations in Genomics Research:  
Implications for Health Services and Policy  
Newfoundland and Labrador Centre for Health Services and Policy  
St. John's, Newfoundland  
**Title:** Population history, structure and genetics of the Hutterites in North  
America.

## **LAURA PALMA**

- October 9-13, 2006 The American Society of Human Genetics, 56th Annual Meeting  
Ernest N. Morial Convention Center, New Orleans, LA  
Attendee
- January 12, 2007 Multidisciplinary Urology/Genetics Rounds  
Montréal, QC  
**Title:** Genetics of Pheochromocytoma and other Paragangliomas
- May 4, 2007 Medical Genetics Journal Club  
Montréal, QC  
Title: Assisted reproduction for inherited predisposition to cancer:  
Challenging the barriers of preventive medicine?

## **DAVID ROSENBLATT**

- August 5-10, 2006 FASEB Summer Research Conference  
One Carbon Metabolism  
Indian Wells, California.  
Chairman
- September 11, 2006 National Council of Jewish Women of Canada  
Gelber Conference Centre, Montreal, QC  
Moderator: Women and Health: Unravelling the Mysteries of Hormones
- Sept 26-29, 2006 Canadian Academy of Health Sciences  
2<sup>nd</sup> Annual Meeting  
Ottawa, ON
- October 6-7, 2006 Genetic Testing  
Center for Disease Control (CDC)  
Atlanta, GA
- April 19-22, 2007 Canadian Genetic Diseases Network  
16<sup>th</sup> Annual Meeting  
Saint-Sauveur, Quebec  
**Title:** Inborn errors of vitamin B<sub>12</sub>: From clinical phenotypic variability  
to novel metabolic steps.

## MARK TISCHKOWITZ

- May 2006 CBCRA Reasons for Hope Conference, Presentation with Jean-Sébastien Brunet, Louis R Bégin, David Huntsman, Maggie CU Cheang, Torsten Nielsen, Lars Akslen and William D Foulkes  
Montréal, QC  
**Title:** The *BRCA1*-associated “core basal phenotype” (ER-, HER2-, CK5/6+) of breast cancer is associated with a poor prognosis.
- May 2, 2006 Réseau de Médecine Génétique Appliquées du FRSQ Journées Génétiques Meeting  
Montréal, QC  
**Title:** BRCA1-associated “core basal phenotype” (ER-, HER2-, CK5/6+) of breast cancer is associated with a poor prognosis
- May 6, 2006 Montreal Children’s Hospital Friday Lecture and Seminar Series  
Montréal, QC  
**Title:** Chromosome Breakage Syndromes and DNA repair
- May 8, 2006 Polymnia Galiatsatos, Adrian Gologan, Marc Tischkowitz  
Jewish General Hospital Medical Rounds  
**Title:** HNPCC: Diagnostic criteria and more...
- September 22, 2006 Hôpital Charles LeMoyne Oncology Rounds  
Montréal, QC  
**Title:** An update on Hereditary Breast Cancer
- October 26, 2006 Montreal Centre for Experimental Therapeutics in Cancer, 4th Annual Meeting  
Montréal, QC  
**Title:** What’s new in DNA Repair?
- October 19-22, 2006 Speaker, 18<sup>th</sup> Annual Fanconi Anemia Scientific Symposium  
North Bethesda, Maryland, USA
- November 10, 2006 Jewish General Hospital Obstetrics and Gynecology Rounds  
Montréal, QC  
**Title:** Screening for genetic diseases in the Ashkenazi Jewish population
- December 14, 2006 McGill Thursday Evening Lecture Series  
Montréal, QC  
**Title:** Genetic factors in breast and ovarian cancer: a primer for family doctors

- February 15, 2007 Genetic Counselling Students Seminar  
McGill University  
Montreal, QC  
**Title:** Pathology in Cancer Genetics
- February 16, 2007 Montreal Children's Hospital Friday Lecture and Seminar Series  
Montreal, QC  
**Title:** Screening for genetic diseases in the Ashkenazi Jewish population
- February 26, 2007 Jewish General Hospital Medical Rounds (also presented at Montreal General Hospital and Royal Victoria Hospital)  
Montréal, QC  
**Title:** A stitch in time? Management issues in hereditary gastric cancer
- March 6, 2007 Environmental Carcinogenesis, MSc Course EXMD 614  
McGill University  
Montreal, QC  
**Title:** It's a dangerous world out there: DNA repair and environmental toxins
- March 23, 2007 Montreal Children's Hospital Genetics Rounds, with Sonya Zaor  
Montreal, QC  
**Title:** Dilemmas in Hereditary Gastric Cancer - a new case and an old case revisited
- April 13, 2007 McGill University Hospitals Research Institute Genetics Axis  
McGill Department of Human Genetics Seminar Series  
Montreal, QC  
**Title:** PALB2/FANCN – another link between Fanconi Anemia and breast cancer
- April 13, 2007 Montreal Children's Hospital Neonatal Rounds  
Montreal, QC  
**Title:** Fanconi anemia - an important cause of congenital malformations
- May 16, 2007 "Lets Talk about Colorectal Cancer" - Free Public Forum: Chevra Kadisha B'nai Jacob - Beit Hazikaron and Colorectal Association of Canada  
**Title:** Genetics of Colorectal Cancer
- May 27, 2007 30-minute interview, Radio 940  
Montreal, QC  
**Title:** Hereditary predisposition to breast cancer

May 27, 2007      Public Seminar, Salon des Baby Boomers Plus, Palais des congrès  
Montréal, QC  
**Title:** Cancer- Are you at risk? Can you prevent it?

### **PATRICIA N. TONIN**

May 18, 2006      CHUM-Hopital Hotel Dieu - Service de Médecine Génique  
Montreal, Quebec  
**Title:** The importance of chromosome 3 genes in ovarian cancer  
revealed by integrating genomic and functional strategies

May 1-2, 2006      Réseau de médecine génétique appliquée (RGMA) du FRSQ  
6<sup>èmes</sup> Journée Génétiques  
**Title:** The influence of the BRCA2 ovarian cancer cluster region  
(OCCR) in French Canadian breast and/ovarian cancer families of Quebec

May 13-16, 2006      Canadian Conference on Ovarian Cancer Research  
Third National Conference On Ovarian Cancer Research,  
Vancouver, British Columbia  
**Title:** Histopathological subtypes of ovarian cancers in familial  
breast-ovarian cancer families: Are there novel breast-ovarian cancer  
susceptibility genes?

March 20, 2007      Cancer Research Laboratory Program, London Regional Cancer Program  
London Health Sciences Centre  
London, Ontario  
**Title:** Integrating genomic, gene expression and functional  
complementation strategies to identify genes implicated in ovarian cancer

### **NORA WONG**

April 18, 2007      Public Service Announcement  
Minimed-SMBD-Jewish General Hospital  
April 11, 18, 25 & May 2, 9, 16  
Montréal, QC  
**Title:** Medical Genetics-Genetic risk and cancer

May 31, 2007      *Reasons to Hope, Knowledge to Cope: Innovations in Cancer Patient  
Education: Cancer Patient Education Network (CPEN)-Canada-5<sup>th</sup> Annual  
Conference, May 31-June 2, 2007*



Session A: Research in cancer patient education  
Marriott Château Champlain  
Montreal, QC

**Title:** Cancer prevention Education: Insights from genetics and lifestyle changes

**SONYA ZAOR**

March 23, 2007

McGill Medical Genetic Grand Rounds with Marc Tischkowitz  
Montréal, QC

**Title:** Dilemmas in Hereditary Gastric Cancer - a new case and an old case revisited.

## ORIGINAL PUBLICATIONS

Baker K, Chong G, **Foulkes WD**, Jass JR. Transforming growth factor-pathway disruption and infiltration of colorectal cancers by intraepithelial lymphocytes. *Histopathol* 49(4): 371-380, 2006.

Barker KT, **Foulkes WD**, Schwartz CE, Labadie C, Monsell F, Houlston RS, Harper J. Is the E133K allele of VG5Q associated with Klippel-Trenaunay and other overgrowth syndromes? *J Med Genet* 43(7): 613-614, 2006.

Bergwitz, C, Roslin NM, Tieder M, Loredó-Osti JC, Bastepe M, Abu-Zahra H, Frappier D, Burkett K, Carpenter TO, Anderson D, Garabédian M, Sermet I, **Fujiwara TM**, **Morgan K**, Tenenhouse HS, and Jüppner H. *SLC34A3* mutations in patients with hereditary hypophosphatemic rickets with hypercalciuria predict a key role for the sodium-phosphate cotransporter NaPi-IIc in maintaining phosphate homeostasis. *Am J Hum Genet* 78:179-192, 2006.

Bikker H, Bakker HD, Abeling NGGM, Poll-The BT, Kleijer WJ, **Rosenblatt DS**, Waterham HR, Wanders RJA, Duran M. A homozygous nonsense mutation in the methylmalonyl-CoA epimerase gene results in mild methylmalonic aciduria. *Hum Mutat* 27(7): 640-643, 2006.

Cody, N, Ouellet V, Manderson EN, Quinn M, Filali-Mouhim A, Tellis, P, Provencher D, Mes-Masson A-M, Chevrette M, **Tonin PN**. Transfer of chromosome 3 fragments identified candidate regions involved in growth suppression of a human ovarian cancer cell line monoallelic for chromosome 3p. *Oncogene* 26(4): 618-32, 2007.

Dean NL, Loredó-Osti JC, **Fujiwara TM**, **Morgan K**, Tan SL, Naumova AK, and Ao A. Transmission ratio distortion in the myotonic dystrophy locus in human preimplantation embryos. *Eur J Hum Genet* 14:299-306, 2006.

Dobson CM, Gradinger AB, Longo N, Wu X, Leclerc D, Lerner-Ellis JP, Lemieux M, Belair C, Watkins D, **Rosenblatt DS**, Gravel RA. Homozygous nonsense mutation in the *MCEE* gene and siRNA suppression of methylmalonyl-CoA epimerase expression: A novel cause of mild methylmalonic aciduria. *Mol Genet Metab* 88: 327-333, 2006.

Ernest S, Carter M, Shao H, Hosack A, Lerner N, Colmenares C, **Rosenblatt DS**, Yoh-Han Pao, Ross ME, Nadeau J. Parallel changes in metabolite and expression profiles in *Crooked-tail* mutant and folate-reduced wild-type mice. *Hum Mol Genet* 15(23): 3387- 93, 2006.

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## RESEARCH OPERATING FUNDS – 2006-2007

### AGENCY

**Dewar, Ken**                      Genome Canada, Group Grant – 2006-2009  
Genome Canada, PI – 2005-2009  
CIHR, Group Grant – 2005-2008  
National Institutes of Health, PI, Group Grant – 2004-2008  
Genome Canada (Emerging Issues), PI – 2006-2007  
Canada Foundation for Innovation, PI, Group Grant – 2006-2007

**Brian Gilfix**                      Gustav Levinschi Foundation Award, PI – 2007  
March of Dimes Birth Defects Foundation, PI – 2006-2008

**Foulkes, William**                      CBCRA, IDEA, PI – 2006-2007  
CBCRA, Operating Grant, PI – 2006-2009  
CBCRA, Operating Grant, PI – 2005-2008  
CIHR, Operating Grant, (PI as of 12/18/06) –  
2003-2006, extended 2007  
Susan G Komen BrCa Found, Co-Investigator –2006-2009  
NIH, Group Grant, Co-Investigator – 2002-2006, extended 2007  
CIHR, Group Grant, Co-Investigator – 2003-2007  
PCRFC, Operating Grant, Co-Investigator – 2005-2007  
CBCRI, Operating Grant, Co-Investigator – 2004-2009

**Morgan, Kenneth**                      Networks of Centres of Excellence, Operating, Investigator –  
1999-2009  
CIHR, Group Grant, Investigator – 2003-2008  
CIHR, Operating Grant, Co-PI – 2006-2009  
CIHR, Operating Grant, Co-application – 2007-2009

**Rosenblatt, David S.**                      CIHR, Operating Grant, PI – 2006-2009  
CIHR, Group Grant, Co-Investigator – 2007-2012

**Tischkowitz, Mark**

Rethink Breast Cancer, Operating Grant, PI – 2006-2008

**Tonin, Patricia N.**

Cancer Research Society, Inc., Strategic Grant, Co-PI – 2005-2007

CIHR, Operating Grant, Investigator – 2005-2008

Quebec Breast Cancer Foundation, Group Operating Grant –  
2004-2007

CIHR, Operating Grant, Investigator – 2004-2006

Fonds de la Recherche en Santé du Québec: Réseau cancerologie,  
Group Operating Grant – 2000-2007

VRQ, Group Operating Grant – 2002-2006

Canadian Foundation for Innovation, Innovation Fund –  
2002-2006

Montreal Breast Cancer Foundation, Group Operating Grant –  
2003-2006

## FINANCIAL REPORT – 2006/2007

Starting Balance	\$ 87,772
Deficit from 2005-2006	(\$18,637)

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**\$ 69,135**

Salary Support and Benefits (Dufrasne, Flannery, Rosenblatt, Valancy, Veyre)	\$ 48,909
Conferences, Seminars, Travel	\$ 18,195
Catering and Events	\$ 2,699
Library and Membership	\$ 6,320
Board Accreditation (Fox, Palma, Secord)	\$ 6,955
Materials and Supplies	\$ 1,321
Miscellaneous	\$ 1,941

**Total Expenses:** **\$ 86,340**

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**Balance:** **(\$17,205)**