Department of Medical Genetics Annual Report: April 1, 2009-May 31, 2010
Division of Medical Genetics, Department of Medicine, Jewish General Hospital

I. Highlights

Medical Genetics has been functioning as a full Department at the Jewish General Hospital in its own right since 2005, while maintaining its status as a Division within the McGill Department of Medicine. This document serves as the annual report for both entities.

The major clinical focus of the department continues to be in the areas of Hereditary Cancer and Prenatal Diagnosis. Dr. Marc Tischkowitz has been on site at the JGH since September 2005 and he practices as a full time medical geneticist. Nora Wong and Sonya Zaor are full time counselors, primarily responsible for cancer genetics. Maria Lalous is a full time genetic counsellor in the area of prenatal diagnosis. Razia Chanda, Lefki Athanasatos and Danielle Veyre provide administrative support for the Department.

Laura Hayes was hired with funds from the Hereditary Breast and Ovarian Foundation (HBOC) to help patients who carry mutations in cancer susceptibility genes navigate the medical system and obtain the follow-up care they require. Carly Pouchet was initially hired in 2008 to work as a research Genetic Counsellor to recruit and coordinate ongoing research studies in hereditary cancer families. In 2010 her role has been modified so that she is working half time in the clinical area and half time in research.

Nora Wong continued her commitment to the Hereditary Breast and Ovarian Foundation by forming and chairing a committee to create the first national conference aimed at keeping BRCA1/2 families up to date regarding current management options. She continues her research on psychosocial aspects in HBOC families. During the last year, she has worked closely with Dr. Alicia Navarro de Souza, a Brazilian psychiatrist and Dr. Danielle Groleau, PhD, medical anthropologist (McGill) on a project examining the experience of women living with HBOC.

Dr. Marc Tischkowitz trained in both medical oncology and clinical genetics and his main clinical and research interest is hereditary cancer predisposition. In addition to his work in hereditary cancer, he supervises the prenatal diagnostic service and runs a monthly general adult genetics clinic in response to a clear need within the McGill RUIS.
Dr. William Foulkes also continues to play a major role in clinical and research aspects of hereditary cancer predisposition at the JGH. In his role as Director of the Program in Cancer Genetics (www.mcgill.ca/cancergenetics/) housed in the Departments of Human Genetics and Oncology at McGill, he has played a major role in bringing expertise together from these two different but crucially important disciplines.

On the eighth floor of the A Pavilion, the department has administrative space and a dedicated counselling room, primarily used for prenatal diagnosis. Much of the clinical work in cancer genetics takes place in the Cancer Prevention Centre in the Segal Cancer Centre and provides state of the art facilities for managing individuals and families with a hereditary predisposition to cancer.

Following the successful evaluation of the service by the Government’s Program de la lutte contre le cancer, we continue to expand our services within the McGill RUIS. In collaboration with Dr Barbara Young, Dr Tischkowitz and Dr Foulkes have developed a genetics service for the Nunavik region, and now do outreach clinics there on a periodic basis.

We are continuing to make improvements to the Cancer Genetics module of the Segal Oncology System electronic health record, and over the last 12 months we have focused on the development of a custom pedigree drawing program which allows us to link up different individuals within the same family. Our current objective is to make the clinical service paperless, commencing with Cancer Genetics.

II. Evaluation of past academic year

a) Faculty

Dr. David Rosenblatt continues to be Director of the University Division of Medical Genetics in the McGill Department of Medicine; this enhances integrated service delivery and allows the Department to share in the larger talent pool of the Department of Medicine. He is also Chair of the McGill Department of Human Genetics. The report of his academic activities for the past year can be obtained at www.mcgill.ca/finestone/. He has played a major role in assuring the full involvement of the JGH Department in all aspect of the McGill RUIS.

Dr. Marc Tischkowitz is primarily responsible for the day-to-day clinical activity in the department. He shares cancer genetics clinics with Dr. Foulkes and runs a monthly multidisciplinary hereditary gastrointestinal cancer clinic with Dr. Polymnia Galiatsatos, a gastroenterologist at the JGH. In addition he runs a monthly general genetics clinic where he sees referrals for all types of genetic disorder. He also sees ward referrals (including neonatal referrals) where a genetic disorder is suspected and is the medical supervisor for the prenatal diagnosis service.

Dr. William Foulkes continues to run the cancer genetics service, dividing his time between the MUHC and the JGH. There are close links to the Departments of Oncology and Pathology, and to a number of surgical divisions, reflecting the multidisciplinary nature of the clinical service.
Please also refer to the annual report of the Program in Cancer Genetics for additional information: www.mcgill.ca/cancergenetics/.

b) Clinical Activities

Our clinical activity across all areas continues to rise at a rapid pace as illustrated in the table and graph below.

<table>
<thead>
<tr>
<th>Service Type</th>
<th>2005</th>
<th>2006</th>
<th>2007</th>
<th>2008</th>
<th>2009</th>
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<tbody>
<tr>
<td>CANCER GENETICS</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>new consultations</td>
<td>283</td>
<td>351</td>
<td>395</td>
<td>438</td>
<td>500</td>
</tr>
<tr>
<td>return visits</td>
<td>289</td>
<td>250</td>
<td>261</td>
<td>255</td>
<td>332</td>
</tr>
<tr>
<td>ward consultations</td>
<td>4</td>
<td>5</td>
<td>4</td>
<td>6</td>
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</tr>
<tr>
<td>phone consultations</td>
<td>40</td>
<td>39</td>
<td>42</td>
<td>99</td>
<td>59</td>
</tr>
<tr>
<td>total (excluding phone cons)</td>
<td>576</td>
<td>606</td>
<td>660</td>
<td>693</td>
<td>828</td>
</tr>
<tr>
<td>total</td>
<td>616</td>
<td>645</td>
<td>702</td>
<td>798</td>
<td>887</td>
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<tr>
<td>year-on-year increase</td>
<td>5%</td>
<td>8%</td>
<td>14%</td>
<td>11%</td>
<td></td>
</tr>
<tr>
<td>PRENATAL DIAGNOSIS</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>new consultations</td>
<td>245</td>
<td>281</td>
<td>260</td>
<td>203</td>
<td>304</td>
</tr>
<tr>
<td>return visits</td>
<td>75</td>
<td>162</td>
<td>112</td>
<td>306</td>
<td>156</td>
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<tr>
<td>total</td>
<td>320</td>
<td>443</td>
<td>372*</td>
<td>509</td>
<td>460</td>
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<tr>
<td>year-on-year increase</td>
<td>38%</td>
<td>-16%</td>
<td>37%</td>
<td>-10%</td>
<td></td>
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<tr>
<td>GENERAL GENETICS</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>new+ward consultations</td>
<td>49</td>
<td>34</td>
<td>71</td>
<td>66</td>
<td></td>
</tr>
<tr>
<td>return visits</td>
<td>11</td>
<td>23</td>
<td>29</td>
<td>30</td>
<td></td>
</tr>
<tr>
<td>total</td>
<td>62</td>
<td>57</td>
<td>100</td>
<td>96</td>
<td></td>
</tr>
<tr>
<td>TOTAL FOR ALL AREAS</td>
<td>936</td>
<td>1150</td>
<td>1131</td>
<td>1407</td>
<td>1443</td>
</tr>
<tr>
<td>year-on-year increase</td>
<td>23%</td>
<td>-2%</td>
<td>24%</td>
<td>3%</td>
<td></td>
</tr>
<tr>
<td>INCREASE IN CASELOAD PER CLINICAL STAFF MEMBER SINCE 2005</td>
<td>23%</td>
<td>21%*</td>
<td>50%</td>
<td>54%</td>
<td></td>
</tr>
</tbody>
</table>

c) Honours and Awards

Dr. William Foulkes is a Chercheur-National at the SMBD-Jewish General Hospital. This award was given to him by the Fonds de la recherche en Santé (FRSQ). He has also been promoted to the rank of Full Professor of Human Genetics, Medicine and Oncology at McGill and is a James McGill Professor.

Dr. Marc Tischkowitz holds a Chercheur-Boursier Clinicien Junior 1 award from the FRSQ.

d) Teaching Activities – 2009-2010

GRADUATE AND UNDERGRADUATE COURSES
### William Foulkes

**Inherited Susceptibility to Cancer**  
**516-614B Environmental Carcinogenesis**  
**Department:** Medicine (Div. Experimental Medicine)  
**Format:** Lecture  
**Title:** Cancer Genetics/Prevention  
**Role:** Lecturer  
**Level:** Graduate students  
**Time:** One 2 hour session  
**Years:** 1999-present

**516-0635D Experimental and Clinical Oncology**  
**Department:** Medicine  
**Format:** Lecture  
**Title:** Cancer Genetics  
**Role:** Lecturer  
**Level:** Graduate students  
**Time:** One 1.5 hour session  
**Years:** 2002-present

**Medical Genetics**  
**Unit 8 small group teaching in medical genetics**  
**Format:** Lecture  
**Role:** Lecturer  
**Level:** Medical students  
**Time:** One 2 hour lecture and 4 small group sessions, 3 hours each  
**Years:** 1997-present

### David Rosenblatt

**Biology 575**  
**Department:** Biology/Human Genetics  
**Format:** Lecture  
**Title:** *Inborn Errors of Folate and Cobalamin Transport and Metabolism*  
**Role:** Lecturer and Course Co-ordinator  
**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

**Marc Tischkowitz**

**HGEN 690**
Department: Human Genetics  
Format: Lecture and student presentations  
Title: *Cancer genetics*  
Role: Lecturer  
Level: MSc program  
Time: 2 x three-hour sessions

**HGEN 692**
Department: Human Genetics  
Format: Lecture  
Title: DNA repair and pediatric cancer syndromes  
Adult cancer predisposition syndromes  
Role: Lecturer  
Level: MSc program  
Time: 2 x two-hour sessions

**516 – 614B Environmental Carcinogenesis**
Department: Medicine (Div. Experimental Medicine)  
Format: Lecture  
Title: *It's a dangerous world out there: DNA repair and environmental toxins*  
Role: Lecturer  
Level: MSc program  
Time: One two-hour session

**516–0635D Experimental and Clinical Oncology**
Format: Lecture  
Title: *Clinical Issues in Hereditary Cancer Genetics*  
Role: Lecturer  
Level: MSc program  
Time: 1.5-hour seminar

**Unit 8 Small Groups**
Department: Human Genetics  
Format: Lecture and Small Group Teaching  
Role: Lecturer-2 sessions:  
Titles: Cancer, Prenatal, Ethics, Screening, Developmental Delay  
Level: Medical Students  
Time: 5 x Two hour sessions, ten hours in total

**Genetic Counseling HGEN650**
Department: Human Genetics, Oncology
Format: Lecture and Small Group Teaching  
Role: Lecturer  
Titles: Using Pathology in Cancer Genetics  
Level: MSc  
Time: 1.5 hours

**BIOL370 Applied Genetics**  
Department: Biology  
Format: Lecture  
Role: Lecturer  
*Title:* *An introduction to Cancer Genetics*  
Level: Undergraduate  
Time: 2 hours

**Maria Lalous**

**McGill Genetic Counselling MSc Program**  
Clinical Supervision:

<table>
<thead>
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<th>Length Of Clinical Rotation (wks)</th>
<th>Number</th>
<th>Total time (wks)</th>
</tr>
</thead>
<tbody>
<tr>
<td>First Year Student</td>
<td>4</td>
<td>1</td>
<td>4</td>
</tr>
<tr>
<td>Second year Student</td>
<td>6</td>
<td>1</td>
<td>6</td>
</tr>
<tr>
<td>Total</td>
<td>-</td>
<td>2</td>
<td>10</td>
</tr>
</tbody>
</table>

**Medical Genetics**  
**Unit 8 small group teaching in medical genetics**  
Format: One 2 hour lecture and 4 small group sessions, 2 hours each  
Role: Lecturer  
Level: Medical students

**Nora Wong**

**HGEN601**  
Department: Human Genetics  
Format: Lecture  
Title: Decision-making under uncertainty  
Role: Lecturer  
Level: Graduate  
Time: 1 X 3 hour session

**Unit 8 Small Groups**  
Department: Human Genetics  
Format: Lecture and Small Group Teaching  
Role: Lecturer-2 sessions:  
Titles: Cancer Genetics
Level: Medical Students
Time: 1 X 2 hour session

**HGEN 610, HGEN 611**

**Department:** Human Genetics
**Format:** Independent research project for Shenela Lakhani
**Title:** Hereditary Breast and Ovarian Cancer Lay Conference, *Quality Assurance Results: What are the needs of BRCA1/2 mutation carriers and where do we go from here?*
**Role:** Project advisor
**Level:** Graduate

**Department:** Human Genetics
**Format:** Independent research project for Lynn Macrae
**Title:** *The experience of being BRCA1/2 mutation-negative in women who learned of their family’s mutation status in childhood or adolescence*
**Role:** Project advisor
**Level:** Graduate

**Workshop 2 - Cancer Risk Assessment**

**Title:** *Methods used to evaluate cancer risk*
**Format:** One small group session
**Role:** Lecturer
**Level:** Genetic counseling students
**Time:** 1 X 4 hour session

**McGill Genetic Counseling MSc Program**

**Clinical Supervision:**

<table>
<thead>
<tr>
<th></th>
<th>Length Of Clinical Rotation (wks)</th>
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<tbody>
<tr>
<td>First Year Student</td>
<td>4</td>
<td>1</td>
<td>4</td>
</tr>
<tr>
<td>Second Year Student</td>
<td>8</td>
<td>1</td>
<td>6</td>
</tr>
</tbody>
</table>

**Sonya Zaor**

**McGill Genetic Counselling MSc Program**

**Unit 8 Small Groups**

**Department:** Human Genetics
**Format:** Lecture and Small Group Teaching
**Role:** Lecturer and small group leader-2 sessions:
**Titles:** Cancer Genetics
**Level:** Medical Students
**Time:** Two hour sessions
Clinical Supervision:

<table>
<thead>
<tr>
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<th>Length Of Clinical Rotation (wks)</th>
<th>Number</th>
<th>Total time (wks)</th>
</tr>
</thead>
<tbody>
<tr>
<td>First Year Student</td>
<td>4</td>
<td>1</td>
<td>4</td>
</tr>
<tr>
<td>Second Year Student</td>
<td>6</td>
<td>1</td>
<td>6</td>
</tr>
</tbody>
</table>

**Workshop 1 - Psychosocial Issues in Cancer Genetics: a patient testimony**

Format: One small group session, 2 hours  
Role: Leader  
Level: 1st-year Genetic counselling students

**INVITED LECTURES, TALKS, PRESENTATIONS**

**William Foulkes**

April 17th 2009  
Title: *Breast cancer research: Updates for basal-like breast cancer and PALB2*  
CNIO Seminar Series 2008-2009  
Madrid, Spain

May 1st 2009  
Title: *Breast and ovarian cancer*  
XXXIVth Annual AMHOQ meeting  
Pointe à Carcy Terminal, Quebec City, Qc

May 27-29th 2009  
Title: *Dual recognition of authentic and de novo splice sites in individuals homozygous for a novel founder mutation in PMS2 is associated with an attenuated phenotype*  
2nd Annual Canadian Human Genetics Conferences  
Harrison Hot Springs, BC

August 12th 2009  
Title: *BRCA2 mutation carriers – what the medical oncologist needs to know*  
MOGA Annual Scientific Meeting 2009 Plenary Session  
Canberra, Australia

August 13th 2009  
Title: *Choosing your parents to minimize the risk of cancer and BRCA1*  
MOGA Annual Scientific Meeting 2009 Plenary Session  
Canberra, Australia

August 14th 2009  
Titles: *Triggers for referral to a genetics service*
MOGA Annual Scientific Meeting 2009
Canberra, Australia

October 20th 2009
Title: *Genetic Mechanisms underlying inherited susceptibility to cancer*
Grand Rounds - Memorial Sloan Kettering Cancer Center
New York, New York

November 4th 2009
Title: *Inherited Susceptibility to the two most prevalent cancers in the Western world*
Rural Multimedia Webcast
Lady Meredith House, McGill University
Montreal, Quebec

November 14th 2009
Title: *The Value of Genetic Testing in NETs*
CNETs Canada CarcinoidNeuroendocrine International Conference and Symposium
Montreal, Quebec

November 26th 2009
Title: *Ovarian Cancer: What can we do to make a real difference?*
CIHR Café Scientifique - DOVE Project
McIntyre Medical Building, McGill University, Montreal, Quebec

February 18th 2010
Title: Ovarian Cancer: *What can we do to make a real difference?*
CIHR Café Scientifique - DOVE Project
Café de Musée des Beaux-Arts, Montreal, Quebec

April 29, 2010
Title: *Inherited Susceptibility to Common Cancers*
CAMO 2010 Annual Scientific Symposium
Montreal, Quebec

May 5th 2010
Title: Ovarian Cancer: *What can we do to make a real difference?*
CIHR Café Scientifique - DOVE Project
Café de Musée des Beaux-Arts, Montreal, Quebec

**David Rosenblatt**

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

**Marc Tischkowitz**

June 2\(^{nd}\), 2009
*Everything you need to know about hereditary breast cancer*
Hôpital Sacré-Cœur, Montreal

September 11\(^{th}\), 2009
*An update on the PALB2 gene – going beyond Fanconi Anemia and breast cancer*
Department of Genetics, Montreal University Health Centre.

October 15\(^{th}\), 2009
*Mutation screening strategies and testing for non BRCA1/2 breast cancer genes as a clinical service – what should we be offering?*
Presentation and chair of panel, The Third International Symposium on Hereditary Breast and Ovarian Cancer, Montreal

October 16\(^{th}\), 2009
*PALB2, BRIP1 and the breast cancer-Fanconi connection*
The Third International Symposium on Hereditary Breast and Ovarian Cancer, Montreal

October 17\(^{th}\), 2009
*Molecular Diagnosis and Counselling of Endocrine Neoplasia Syndromes*
Meet the Professor session, Canadian Society of Endocrinology and Metabolism Annual Conference, Montreal

November 27\(^{th}\), 2009
*Café au lait macules – not just NF1*
Department of Genetics, Hôpital St Justine, Montreal

February 24\(^{th}\), 2010
*Hereditary diseases in the Ashkenazim: Are you at risk & what can you do about it?*
Public lecture, Congregation Shaar Hashomayim, Montreal

February 25\(^{th}\), 2010
*Cancer Risks - Myths and Realities*
McGill Women's Networking Group, Montreal

February 25\(^{th}\), 2010
*BRCA, TNBC and PARP – what’s the link?*
Montreal Biomarkers Forum, Montreal

May 4th, 2010
The characterisation of Genomic Instability in Cell Lines Derived from Heterozygous PALB2 mutation carriers

Réseau de Médicine Génétique Appliquées du FRSQ Journées Génétiques Meeting, Montreal

May 20th, 2010

Investigating the PALB2 Hereditary Breast Cancer Gene in Quebec Families

Québec-Flandre partenaires en recherche et innovation Brussels

e) Service to Academic Community and other Contributions

Members of the Division are very active in the academic activities of the Department of Human Genetics at McGill. In addition to being Chair of the Department, Dr. Rosenblatt is a Past President of the Association of Medical Geneticists of Quebec. Dr. Foulkes is Director of the McGill Program in Cancer Genetics and plays a key role on the Board of Directors of the Hereditary Breast and Ovarian Cancer Foundation. Dr Tischkowitz is a member of the Clinical Practice Committee, Canadian College of Medical Genetics and a member of Editorial board for the journal Clinical & Investigative Medicine. He is also the Director of the Department of Oncology Visiting Speaker Program (www.medicine.mcgill.ca/oncology/speakers/speakers_visitingSpeakers.asp). Nora Wong is the editor of the Hereditary Breast and Ovarian Cancer Foundation newsletter, an annual publication for individuals with BRCA1 or BRCA2 mutations. She is also headed the planning committee for the HBOC’s first Lay conference day in 2009.

f) Research Operating Funds – 2008

-Susan G Komen BrCa Found, Research Advisory Council, 2010-2011
-CCSRI, Bridge Funding, 2010-2011 (PI)
-CGCF, Operating, 2009-2010 (PI)
-CIHR, APOGEE, Emerging Team Grant, 2009-2014 (Co-PI)
-WEBC, Operating, 2009-2010 (PI)
-PCC, Operating, 2009-2010 (PI)
-Marsha Rivkin Center for Ovarian Cancer Research, 2009-2010 (PI)
-US ARMY, Synergistic, 2008-2010 (PI)
-WEBC, Operating, 2008-2009
-CBCRA, Operating, 2006-2009, extended to 2010 (PI)
-CBCRA, Operating, 2005-2008, extended 2009 (PI)
-Susan G KomenBrCa Found, 2008-2011 (Co-I)
-Susan G KomenBrCa Found, 2007-2010 (Co-I)
-Susan G KomenBrCa Found, 2006-2009 (Co-I)
-NIH – Group Grant, 2002-2006, extended 2009 (Co-I)
-CBCRA, Operating, 2004-2009, extended to 2010 (Co-I)

David Rosenblatt
- CIHR, Operating Grant, PI – 2009-2014
- CIHR, Group Grant, Co-Investigator – 1975-2009
**Marc Tischkowitz**  
-Quebec Ministry of Economic Development, Innovation and Export (PI)  
-Weekend to End Breast Cancer Operating (PI)  
-Komen Career Catalyst (PI)

**Nora Wong**  
Mapping the decisional trajectory in breast cancer risk management among BRCA1 and BRCA2 mutation carriers WEBC Ideal grant (PI)

**g) Publications for 2009**

Peer Reviewed Papers


--Novak DJ, Sabbaghian N, Maillet P, Chappuis PO, **Foulkes WD**, **Tischkowitz M**. Analysis of the genes coding for the BRCA1-interacting proteins, RAP80 and Abraxas (CCDC98), in high-risk, non-BRCA1/2, multiethnic breast cancer cases. Breast Cancer Res Treat. 2009 Sep;117(2):453-9


--Vigano A, Trutschnigg B, Kilgour RD, Hamel N, Hornby L, Luca E, **Foulkes W**, Tremblay ML, Morais JA. Relationship between angiotensin-converting enzyme gene polymorphism and


**Chapters 2009**


**III. Objectives and Priorities**

Our **objective** remains to provide comprehensive service to our clientele. Our **priority** is to make it clear that further resources must be made available for prenatal, cancer and other general genetics services at the Jewish General Hospital. The Department of Medical Genetics at the JGH aspires to making the needs of both individuals and families in the area of medical genetics better known to the hospital and the community at large.

David S. Rosenblatt, MD  
Chief, Department of Medical Genetics, JGH  
Chief, Division of Medical Genetics, Department of Medicine, McGill & JGH  
Chair, Department of Human Genetics, McGill