Department of Medical Genetics Annual Report: April 1, 2010-May 31, 2011
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. Carly Pouchet, originally hired as a research genetic counsellor, currently works clinically in both cancer genetics and in prenatal diagnosis. Razia Chanda, Martah Urquilla 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.

Nora Wong continues her research on psychosocial aspects in HBOC families. She has continued her work with Dr. Alicia Navarro de Souza, a Brazilian psychiatrist on a project examining the experience of women living with HBOC. She is also working on a project looking at the risk experience of young women from HBOC families who were subsequently found to be non-carriers as well as another project looking at the attitudes and preferences for genetic testing in women newly diagnosed with breast or ovarian cancer with Dr. Foulkes and Dr. Carmen Loiselle.

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 counseling 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, with Dr. Foulkes visiting in April 2010.

II. Evaluation of past academic year

1) Teaching Activities - 2010-2011

Graduate and Undergraduate Courses:

**William Foulkes**

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

**516-6148 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
Medical Genetics
Unit 1 small group teaching in probability in genetics
Format: Small group
Role: Facilitator
Level: Medical students
Time: 1 small group session, 3 hours
Years: 2010

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-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

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

Biol 370 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>
<tr>
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<th>Length Of Clinical Rotation (wks)</th>
<th>Number</th>
<th>Total time (wks)</th>
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<tbody>
<tr>
<td>First Year Student</td>
<td>4</td>
<td>1</td>
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<tr>
<td>Second year Student</td>
<td>2</td>
<td>1</td>
<td>2</td>
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<tr>
<td>Total</td>
<td>-</td>
<td>2</td>
<td>6</td>
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</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 M Sc Program**
Clinical Supervision:

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

**Sonya Zaor**

**McGill Genetic Counselling M Sc 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>
<th></th>
<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>
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</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

**Carly Pouchet**

**Unit 8 Small Groups** Department: Human Genetics  
Format: Small Group Teaching  
Role: Facilitator-4 sessions:  
Titles: Problem Solving, Cancer Genetics, Prenatal Diagnosis, Ethics  
Level: Medical Students  
Time: 4 X 2 hour session

**INVITED LECTURES, TALKS, PRESENTATIONS**

**William Foulkes**

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

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

October 8, 2010  
**Title:** Tumour Size and Survival - a reappraisal  
Ninth Annual Controversies in Breast Cancer: Adjuvant and Neoadjuvant Therapy New York, New York

December 7, 2010  
**Title:** Basal-Like Tumor Correlations  
Triple-Negative Breast Cancer Symposium San Antonio, Texas

**David Rosenblatt**

March 10, 2010  
Medical Grand Rounds  
**Title:** Homocysteine and Coronary Heart Disease McGill University Health Centre
October 28, 2010
2010-2011 NHGRI Division of Intramural Research Seminar Series

Title: Novel Biological Insights into Vitamin B12 Transport and Metabolism: Lessons from the Clinic Bethesda, MD

March 1, 2011
SIMD Annual Meeting

Title: Clinical characterization of patients with various forms of homocysteinuria Asilomar, CA

Marc Tischkowitz

May 20th, 2010 Investigating the PALB2 Hereditary Breast Cancer Gene in Quebec Families, Quebec—Flandre partenaires en recherche et innovation Brussels, 20 May 2010

November 11th, 2010 Hereditary Breast Cancer Department of Radio-Oncology, L'Hôpital Notre-Dame, CHUM, Montreal.

November 25th 2010 Moving from the lab to the clinic: personalized interpretation of cancer risk and treatment response. Goodman Cancer Research Public Lecture Series, McGill University, Montreal

March 14th 2011 DICER1: The link between microRNAs, multinodular and rare tumors. Jewish General Hospital Medical Rounds

March 31st 2011: What can Rare Gynecological Tumors Tell Us About Gene Regulation? Jewish General Hospital Obstetrics and Gynecology Rounds.

Nora Wong

January 27, 2011
Thursday Evening Lecture Series, McGill University
Title: Who to refer for a genetic evaluation for breast cancer and why

2) Research Operating Funds

William Foulkes

–Susan G Komen BrCa Fund, 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)
–WEBRC, Operating, 2009-2010 (PI)
–Marsha Rivkin Center for Ovarian Cancer Research, 2009-2010 (PI)
–US ARMY, Synergistic, 2008-2010 (PI)
–WEBRC, Operating, 2008-2009
–CBCRA, IDEA grant, 2009-2010 (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, 2002-2011, (Co-I)
3) Clinical Activities

Our overall clinical activity continues to increase as illustrated in the table below. This increase is particularly strong in Cancer Genetics. There was a reduction in the PND cases because of medical leave of FT counsellor for 10 weeks with a PT counsellor seeing a reduced load during this time.

<table>
<thead>
<tr>
<th>Service Type</th>
<th>2005</th>
<th>2006</th>
<th>2007</th>
<th>2008</th>
<th>2009</th>
<th>2010</th>
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<tr>
<td><strong>Cancer Genetics</strong></td>
<td></td>
<td></td>
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<td></td>
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<tr>
<td>new consultations</td>
<td>283</td>
<td>351</td>
<td>395</td>
<td>438</td>
<td>500</td>
<td>596</td>
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<tr>
<td>return visits</td>
<td>289</td>
<td>250</td>
<td>261</td>
<td>255</td>
<td>332</td>
<td>403</td>
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<tr>
<td>ward consultations</td>
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<td>5</td>
<td>4</td>
<td>6</td>
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<tr>
<td>phone consultations</td>
<td>40</td>
<td>39</td>
<td>42</td>
<td>99</td>
<td>59</td>
<td>75</td>
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<tr>
<td>total (excluding phone consults)</td>
<td>576</td>
<td>606</td>
<td>660</td>
<td>693</td>
<td>828</td>
<td>1004</td>
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<tr>
<td>year-on-year increase</td>
<td>616</td>
<td>645</td>
<td>702</td>
<td>798</td>
<td>887</td>
<td>1079</td>
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<tr>
<td><strong>Prenatal Diagnosis</strong></td>
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<td></td>
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<tr>
<td>new consultations</td>
<td>245</td>
<td>281</td>
<td>260</td>
<td>203</td>
<td>304</td>
<td>283</td>
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<tr>
<td>return visits</td>
<td>75</td>
<td>162</td>
<td>112</td>
<td>306</td>
<td>156</td>
<td>97</td>
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<tr>
<td>total</td>
<td>320</td>
<td>443</td>
<td>372*</td>
<td>509</td>
<td>460</td>
<td>380</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>-12%</td>
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<td><strong>General Genetics</strong></td>
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<tr>
<td>new+ward consultations</td>
<td>49</td>
<td>34</td>
<td>71</td>
<td>66</td>
<td>69</td>
<td></td>
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<tr>
<td>return visits</td>
<td>11</td>
<td>23</td>
<td>29</td>
<td>30</td>
<td>27</td>
<td></td>
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<tr>
<td>total</td>
<td>62</td>
<td>57</td>
<td>100</td>
<td>96</td>
<td>96</td>
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<tr>
<td><strong>Total for all areas</strong></td>
<td>936</td>
<td>1150</td>
<td>1131</td>
<td>1407</td>
<td>1443</td>
<td>1555</td>
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<tr>
<td>year-on-year increase</td>
<td>23%</td>
<td>-2%</td>
<td>24%</td>
<td>3%</td>
<td>7%</td>
<td></td>
</tr>
</tbody>
</table>

4) Academic Staff

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 and his term is expected to be completed at the end of August 2011. It is hoped that the next University Chair of the McGill Department of
Human Genetics will also be Chief of the Departments of Medical Genetics at the JGH and the MUHC. The report of the academic activities of Dr. Rosenblatt for the past year can be obtained at www.mcgill.ca/finestone/. Dr. Rosenblatt has assured 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/.

6) Honours and Awards
Dr. William Foulkes holds a Chercheur-National award from the FRSQ. He is also James McGill professor. Dr. Marc Tischkowitz holds a Chercheur-Boursier Clinicien Junior 1 award from the FRSQ.

5) Consulting Activities: None

6) Other

- 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, the Canadian Society for Clinical Investigation and the Society for Inherited Metabolic Diseases (USA). He serves on the editorial board of Human Mutation and Molecular Genetics and Metabolism. Dr. Foulkes is Director of the McGill Program in Cancer Genetics and is the scientific director of the biennial HBOC conference, sponsored by the Hereditary Breast and Ovarian Cancer Foundation. He is section editor for Current Oncology, and sits on the editorial board of the Journal of Medical Genetics. 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.

- Publications for 2010

Peer Reviewed Papers


28. Marc Tischkowitz and Bing Xia. PALB2/FANCN - recombining cancer and Fanconi anemia Cancer Research 2010;70(19)7353-9


**Book co-editorship**


**Chapters**


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.

Respectfully Submitted,

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