PREAMBLE

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 academic activities of Dr. David Rosenblatt can be found in the annual report of the Finestone laboratory: www.mcgill.ca/finestone. Information on academic and research activities of all McGill faculty members can also be found in the annual report of the McGill Department of Human Genetics. This report serves as the annual report of both the Department of the Jewish General Hospital and the Division of Medical Genetics in the Department of Medicine.

Clinical Activities

The overall clinical activity is relatively stable since 2013, but there have been some new initiatives. In the hereditary cancer clinic, there were 382 follow-up visits and 523 new visits in 2013. In the Prenatal Diagnosis clinic there were 217 follow-up visits and 316 new visits in 2013. Specialized clinic have been created. In the Urology-Genetics Clinic, which is led by Nassim Taherian, 20 new patients were seen with 19 follow-up visits. In the Hereditary Colorectal Cancer Registry, 6 new patients and 51 follow-up visits or telephone consults have occurred.

The advent of next generation sequencing and the venue of new multiplex genes panel testing, is significantly impacting on current counselling models. The genetic counsellors are currently evaluating new counselling approaches to ensure patients benefit from state of the art genetic services, while ensuring education and patient guided-care. Genetic counsellors share their knowledge with many McGill University students registered in the Genetic Counselling master program, the medical faculty or residents, and fellows from McGill and other universities.

SUMMARY

1. Research and Publications:

   The major clinical focus of the department continues to be in the areas of Hereditary Cancer and Prenatal Diagnosis. Prenatal diagnosis activities mainly involve patient care and clinical teaching, whereas Hereditary Cancer has a major research focus at the Jewish
General Hospital and Lady Davis Research Institute. The most exciting work from Dr. Foulkes’ group in 2013 continues to be the work on DICER1 mutations in cancer, but they have also commenced several whole exome sequencing projects that should bear fruit in 2014. The research laboratory of Dr. Rosenblatt is located at the Montreal General Hospital site of the McGill University Health Centre. He has medical responsibility for the prenatal diagnosis program at the Jewish General Hospital.

2. **Teaching and learning (undergraduate and graduate):**

Dr. Foulkes and Dr. Rosenblatt are involved in teaching at all levels—undergraduate, graduate and residency training. The genetic counsellors are actively involved in teaching in the M.Sc. program in Genetic Counselling and with residents in several specialty programs, in particular Medical Genetics and Obstetrics and Gynecology.

**McGill Genetic Counselling MSc Program: Role of Genetic Counsellors**

**Lynn Macrae**—on maternity leave in 2013

**Carly Pouchet**—on Maternity leave in 2013

Prenatal Diagnosis II-HGEN 600-Genetic Counselling  
Department: Human Genetics  
Format: One group teaching  
Role: Facilitator  
Title: Maternal Serum Screening  
Time: 1.5 hours

**Laurence Baret**

Unit 8 Small Groups  
Department: Human Genetics  
Format: Lecture and Small Group Teaching  
Role: Facilitator/ Lecturer  
Titles: Ethical issues in genetics  
Level: Medical Students  
Time: 2 hour sessions

Unit 8 Small Groups  
Department: Human Genetics  
Format: Lecture and Small Group Teaching  
Role: Facilitator/ Lecturer  
Titles: Cancer Genetics  
Level: Medical Students  
Time: 2 hour sessions

Genetic counselling Case discussion  
Department: Human Genetics  
Format: Seminar/ Lecture  
Role: Lecturer
Titles: Case discussion and psychosocial issues  
Level: Master students  
Time: 1.5 hours  

**Nassim Taherian**

Unit 8 Small Groups Department:  
Human Genetics  
Format: Small Group Teaching  
Role: Facilitator  
Titles: Genetic Testing in Families and Populations  
Level: Medical Students  
Time: 2 hour sessions  

Unit 8 Small Groups Department:  
Human Genetics  
Format: Small Group Teaching  
Role: Facilitator  
Titles: Cancer Genetics  
Level: Medical Students  
Time: 2 hour sessions  

Fundamentals of Medicine and Dentistry  
Department: Human Genetics  
Format: Small Group Teaching  
Role: Facilitator  
Titles: Family History Taking and Risk Assessment  
Level: Medical Students  
Time: 3 hour sessions  

Introduction to Research Ethics-HGEN 600-Genetic Counselling  
Department: Human Genetics  
Format: One Group Teaching  
Role: Lecturer  
Titles: Consent Form Design, and the IRB Process  
Time: 1.5 hours  

**Nora Wong**

HGEN601  
Department: Human Genetics  
Format: Lecture  
Title: Psychosocial aspects of living with hereditary cancer risk and 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

Unit 8  Small Groups  
Department: Human Genetics  
Format: Lecture and Small Group Teaching  
Role: Teaching assistant  
Titles: Family history taking  
Level: Medical Students  
Time: 1 X 3 hour session

HGEN 610, HGEN 611  
Department: Human Genetics  
Format: Independent research project for Laura Dempsey-Nunez  
Title: RAD51C/D mutations in women with synchronous cancers of the ovary and endometrium: a hospital cohort  
Role: Project advisor  
Level: Graduate

Department: Psychiatry  
Format: MSc Theses  
Title: Psychosocial determinants of decision-making in women newly diagnosed with breast and ovarian cancer  
Role: co-supervisor  
Level: Graduate

Sonya Zaor  
First year Practicum  
Department: Human Genetics  
Format: Workshop  
Title: Psychosocial Issues in Cancer Genetics: a patient testimony  
Role: Leader  
Level: 1st yr Genetic Counselling student  
Time: 2 hours

Summary of McGill Clinical Supervision by Genetic Counsellors

<table>
<thead>
<tr>
<th>Laurence Baret</th>
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<tbody>
<tr>
<td>Clinical Supervision</td>
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<tr>
<td>First Year GC</td>
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<tr>
<td></td>
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<tr>
<td>---------------------</td>
</tr>
<tr>
<td>Visiting fellow</td>
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<tr>
<td>Medical Student</td>
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**Maria Lalous**

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<tr>
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**Heidi Rothenmund**

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<tbody>
<tr>
<td>Second Year GC</td>
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<td>2*</td>
<td>6</td>
</tr>
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</table>

Co-supervised with Nassim Taherian and Nora Wong

*Please note: one student did not complete her rotation due to medical leave

**Nassim Taherian**

<table>
<thead>
<tr>
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<th>Total time</th>
</tr>
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<tbody>
<tr>
<td>Second Year GC</td>
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<td>2*</td>
<td>6</td>
</tr>
</tbody>
</table>

Co-supervised with Nora Wong and Heidi Rothenmund

*Please note: one student did not complete her rotation due to medical leave

**Nora Wong**

<table>
<thead>
<tr>
<th>Clinical Supervision</th>
<th>Length of Rotation (wks)</th>
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<th>Total time</th>
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<tr>
<td>Second Year GC</td>
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<td>2*</td>
<td>6</td>
</tr>
</tbody>
</table>

Co-supervised with Nassim Taherian and Heidi Rothenmund

*Please note: one student did not complete her rotation due to medical leave

**Sonya Zaor**

<table>
<thead>
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<th>Clinical Supervision</th>
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<th>Number</th>
<th>Total time (wks)</th>
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<td>5</td>
</tr>
<tr>
<td>Second Year GC</td>
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<td>3</td>
</tr>
<tr>
<td>Visiting fellow</td>
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<tr>
<td>Medical resident</td>
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<td>2</td>
<td>12</td>
</tr>
<tr>
<td><strong>TOTAL</strong></td>
<td></td>
<td><strong>5</strong></td>
<td><strong>21</strong></td>
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</table>

*3 ad hoc McGill medical students, resident in radiology (CHUM), resident in pathology have not been included
3. **Involvement in the community:**

Dr. Foulkes has been involved with many activities relating to hereditary cancer. He is often solicited as a resource for the media when expert opinion in this area is required. Nora Wong and Laurence Baret have been involved with many activities relating to hereditary breast and ovarian cancer leading information booth at many events in the community.

4. **Partnerships:**

The faculty of the JGH Department has close working relationships with the hospital and university Departments of Medicine, Oncology, Pediatrics, and Obstetrics.

5. **Milestones:**

Dr. Rosenblatt completed twelve years as Chair of the Department of Human Genetics at McGill. Dr. Eric Shoubridge has been appointed as the new Chair.

6. **Honours, awards and prizes:**

Dr. Rosenblatt is the Chair holder of the Dodd Q. Chu and Family Chair in Medical Genetics at McGill. Dr. Foulkes is a James McGill Professor of Medicine at McGill University. In 2013, Dr. Rosenblatt was named a Champion of Genetics by the Canadian Gene Cure Foundation.

7. **Fundraising:**

Laura Hayes is supported by funds from the Hereditary Breast and Ovarian Foundation (HBOC).

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**SECTION I – DEPARTMENT/DIVISION STATUS UPDATE**

1. **Mission and objectives of the Division**

Medical Genetics at the JGH is committed to the best possible patient care and teaching in the area of prenatal diagnosis. It is committed to excellence patient care, teaching and research in Hereditary Cancer. In the area of Hereditary Cancer Research, it is committed to being amongst the best centres in the world. The Division strives to provide comprehensive service to our clientele. Its 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 Jewish General Hospital 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.
2. **A nominative list of academic staff, their academic rank**

<table>
<thead>
<tr>
<th>Name</th>
<th>Academic Rank</th>
</tr>
</thead>
<tbody>
<tr>
<td>William Foulkes</td>
<td>Professor</td>
</tr>
<tr>
<td>Laurence Baret</td>
<td>Faculty Lecturer</td>
</tr>
<tr>
<td>Maria Lalous</td>
<td>Faculty Lecturer</td>
</tr>
<tr>
<td>Lynn Macrae</td>
<td>Faculty Lecturer-maternity leave</td>
</tr>
<tr>
<td>Carly Pouchet</td>
<td>Faculty Lecturer-maternity leave</td>
</tr>
<tr>
<td>David Rosenblatt</td>
<td>Professor</td>
</tr>
<tr>
<td>Heidi Rothenmund</td>
<td>Faculty Lecturer</td>
</tr>
<tr>
<td>Nassim Taherian</td>
<td>Academic Associate</td>
</tr>
<tr>
<td>Nora Wong</td>
<td>Faculty Lecturer</td>
</tr>
<tr>
<td>Sonya Zaor</td>
<td>Faculty Lecturer</td>
</tr>
</tbody>
</table>

Laurence Baret, Nora Wong, and Sonya Zaor, are genetic counsellors who are primarily responsible for cancer genetics. Maria Lalous is a full time genetic counsellor in the area of prenatal diagnosis. Heidi Rothenmund and Nassim Taherian are genetic counsellors who are responsible for cancer specialty clinics. Razia Chanda, Martah Urquilla and Samar Edriss provide administrative support for the department. Danielle Veyre has retired from her administrative post in September 2013.

Laurence Baret is a recent graduate from the University of Toronto Genetic Counselling Master Program. She is a full time genetic counsellor in cancer genetics and has been hired for a 1 year maternity leave replacement for Lynn Macrae. With her previous training in Bioethics (M.A. Bioethics), her research interests include the ethical issues in human genetics. Her previous work focused on the communication of genetic research results to research participants, which has been published in the *European Journal of Human Genetics*. Laurence Baret is involved in clinical projects aiming to improve the clinical model of delivering genetic services (*e.g.* developing an alternative model to deliver genetic counselling services for ‘low risk’ patients, helping to implement referral guidelines for breast and ovarian cancer, helping to implement blood send-outs from a core laboratory and implementing updated genetic testing approaches based on the current literature).

Laura Hayes has the responsibility to help patients who carry mutations in cancer susceptibility genes navigate the medical system and obtain the follow-up care they require. Her work is supported by the Hereditary Breast and Ovarian Cancer (HBOC) Foundation.

Heidi Rothenmund is a part-time genetic counsellor who has been working with Dr. Foulkes and Dr. George Chong to develop the Hereditary Colorectal Cancer Registry, which is based in the Cancer Prevention Centre at the Jewish General Hospital. The purpose of the Registry is to follow families affected with hereditary colorectal cancer that have been identified through the Cancer Genetics Clinics in the Department of Medical Genetics as well as other centres throughout the province. The clinical role of the registry is to provide regular contact with these high-risk individuals to update the
family history of cancer, coordinate clinical screening appointments, and to facilitate predictive genetic testing and clinical screening for at-risk relatives, where applicable. Patients and at-risk family members in the Registry may be referred to the GENG1 Clinic at the Jewish General Hospital where Heidi and Dr. Polymnia Galiatsatos in the Department of Gastroenterology will follow these high risk patients on an annual basis. For Registry participants who are not able to travel to the Jewish General Hospital, Heidi provides telephone appointments. On a research basis, the Registry recruits high-risk families who are eligible for gene-discovery efforts through the research laboratory of Dr. Foulkes. This involves various local and international collaborations that have been facilitated by both Dr. Foulkes and Dr. Chong. Heidi also works with a part-time research assistant (Angelica Randall) as the study coordinator Canadian Colorectal Cancer Consortium (C4): Clinical and Molecular Screening in Patients with Colorectal Cancer and their Family Members. This is a multi-centre study across Canada that is funded by the Terry Fox Research Institute and for which Dr. Foulkes is the Site Principal Investigator. Eligible patients with newly diagnosed colorectal cancer at the Jewish General Hospital and the Montreal General Hospital will be approached to participate in this study. The main goals of the study are to investigate the frequency of Lynch Syndrome in Canada; to determine the feasibility of implementing routine screening for Lynch Syndrome in newly diagnosed patients; to study quality of life factors for colorectal cancer patients; and to investigate the uptake of colonoscopy screening among at-risk relatives.

Nassim Taherian coordinates a specialty clinic for the evaluation of patients with familial renal cell carcinoma or prostate cancer. She works with the departments of Medical Genetics and Urology. Her clinics are held in the Cancer Prevention Centre with Dr. William Foulkes. She also coordinates various international research studies on prostate cancer genetics and is first author of a paper: Familial prostate cancer: the damage done and lessons learnt. Taherian, N., Hamel, N., Begin, L. R., Bismar, T. A., Goldgar, D. E., Feng, B. J. and Foulkes, W. D. January 2013 Nat. Rev. Urol. 10, 116-122 (2013).

Nora Wong continues her clinical focus on increasing the accessibility of genetic testing for cancer susceptibility and looking into alternative models for service delivery. Her two papers have been accepted for publication (see below). She also continues to devote time in various research projects, advisory boards and community programs. Experience of BRCA1/2 mutation-negative young women from families with hereditary breast and ovarian cancer: a qualitative study. Macrae L, de Souza AN, Loiselle CG, Wong N. Cultural aspects of healthy BRCA carriers from two ethnocultural groups in qualitative health research. Hered Cancer Clin Prac. October 2013. 16;11(1):14.

Sonya Zaor devotes her efforts in delivering and improving cancer genetic counselling services. The increase in referrals and recent implementation of new genetic testing technologies in the clinical setting, have synergistically impacted the current practice of genetic counseling. Sonya was instrumental in implementing referral criteria to increase accessibility to genetic testing for a greater number of families who are at high risk. Sonya is interested in other projects such as addressing the utility and acceptance of gene panels in the clinical setting for providers and their patients. Her efforts are also focused on addressing the needs for patients at lower genetic risk. She is looking at alternative models to delivery information for these individuals by developing alternative clinics as
well as improving access to information for patients and physicians using various media sources (McGill Website, educative pamphlet).

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, 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.

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 completed his term as Chair of the McGill Department of Human Genetics and his term at the end of June 2013. 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. 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.

SECTION II - GRANTS, PUBLICATIONS, AND SERVICE OUTSIDE OF McgILL

1. Grants and awards received

William Foulkes

CURRENT COMPETITIVE GRANTS

As Principal Investigator

- CCSRI: DICER1 and pituitary blastoma: Keys to understanding pituitary development and tumorigenesis

- Alex’s Lemonade Stand Foundation: DICER1, microRNAs and pediatric cancer: an emerging story

- Ride to Conquer Cancer: Establishing a Quebec Hereditary Colorectal Cancer Registry,
3. **Scholarly works published in the 2013 calendar year:**

**William Foulkes:**


**David Rosenblatt**


3. **Academic and community engagement service outside of McGill by individual members of the unit**

   **William Foulkes**

   February 5 2013
   **Title:** Triple negative breast cancer: germ-line mutations, classification and emerging targets
   Sírio Libanês Hospital
   São Paulo, Brazil

   March 13 2013
   **Title:** Triple Negative Disease: Molecular heterogeneity and emerging targets
   13th International St-Gallen Breast Cancer Conference
   St-Gallen, Switzerland

   April 6th 2013
   **Title:** Deconstructing the complexity of Triple Negative Breast Cancer
   2013 AACR Meeting
   Walter E. Washington Convention Center
   Waashington, DC

   October 4th 2013
   **Title:** Clinical aspect of digestive diseases
   10th International Symposium on the Physiology and Diseases of the Digestive Tract
   Estrimont Suites and Spa Orford
   Orford, Qc
David Rosenblatt

February 19, 2013
**Title:** Discovering New Genes in the One-carbon Pathway Using Exome Sequencing-A RaDiCAL Approach
Keystone Symposia on Molecular and Cellular Biology
Santa Fe, New Mexico

September 11, 2013
**Title:** Discovering new genes in the one-carbon pathway using exome sequencing: a RaDiCAL approach
9th International Conference on Homocysteine and One Carbon Metabolism
Dublin, Ireland

November 5, 2013
**Title:** Using next generation sequencing to discover novel steps in one-carbon Metabolism
Genomics in Metabolism
Copenhagen, Denmark

December 13, 2013
**Title:** Les maladies héréditaires du metabolisme de la vitamine B₁₂: contribution à la connaissance du metabolism cellulaire
Journées francophones de nutrition
Bordeaux, France

SECTION III - CONFIDENTIAL INFORMATION

1. **Consulting activities:**

Both Dr. Foulkes and Dr. Rosenblatt are involved in editorial reviews of manuscripts, and on various committees at the local, provincial and national levels.

Respectfully submitted,

David S. Rosenblatt, MD
Chief, Department of Medical Genetics, Jewish General Hospital
Head, Division of Medical Genetics, Department of Medicine, Jewish General Hospital and McGill University

January 26, 2014