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. 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

In the Hereditary Cancer Clinic, there were 520 new visits and 666 follow-up visits or telephone consultations in 2014. A low risk telephone consultation clinic was implemented where Lynn Macrae (Genetic Counsellor) counselled an additional 72 new consults, once the referred individuals were estimated as having a low probability of being affected by a hereditary cancer.

In the Urology-Genetics Clinic, led by Nassim Taherian (Genetic Counsellor), 17 new patients were seen with 2 follow-up visits.

In the Hereditary Colorectal Cancer Registry, led by Heidi Rothenmund, there were 34 new patients and 93 follow-up visits or telephone consults.

Nora Wong (Genetic Counsellor) continues her commitment in providing information about genetics risk for ovarian cancer risk at the annual Walk of Hope.

Carly Pouchet (Genetic Counsellor) returned from maternity leave in January 2014 and worked until August 2014 in both hereditary cancer and prenatal diagnosis. After she left the Department to pursue other personal and professional activities, Laurence Baret (Genetic Counsellor) and Lara Reichman (Genetic Counsellor) replaced her temporarily in the September to December 2014 period.

In the Prenatal Diagnosis Clinic there were 114 follow-up visits and 324 new visits in 2014, and 56 phone consultations were low-risk counselling and low-risk result sessions were
performed. The use of low-risk phone consultations has allowed for more open slots for higher-risk pregnancies in clinic.

The advent of next generation sequencing and the venue of new multiplex genes panel testing continue to have significant impact 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. Highlights of the year for Dr Foulkes were last author publications in Nature Genetics, Acta Neuropathol, and JCEM, as well as an editorial for Lancet Oncology and a major review for Nature Reviews Cancer. 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:

Nora Wong

HGEN 610, HGEN 611
Department: Human Genetics
Format: Independent research project for Rachel de Silva-Smith
Title: Intra-familial Communication about Hereditary Breast and Ovarian Cancer Syndrome in Orthodox Jewish Families
Role: Project advisor
Level: Graduate

Department: Psychiatry
Format: MSc Theses (completed and accepted)
Title: Psychosocial determinants of decision-making in women newly diagnosed with breast and ovarian cancer
Role: Co-supervisor for Sahar Sajadi
Level: Graduate

Maria Lalous

Research project supervisor:

Title: “The factors associated with elective termination of pregnancy of fetuses with congenital diaphragmatic hernia in Canada: A 9-year analysis of the CAPSNet data”

Abstract presentation at IFMSS Conference (International Fetal Medicine and Surgery Society) Sept. 7-11 2014:

Title: “The factors associated with elective termination of pregnancy of fetuses with congenital diaphragmatic hernia in Canada: A 9-year analysis of the CAPSNet data”

McGill Clinical Supervision by Genetic Counsellors

<table>
<thead>
<tr>
<th>Maria Lalous</th>
<th>Clinical Supervision</th>
<th>Length of Rotation (wks)</th>
<th>Number students</th>
<th>Total time (wks)</th>
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<tr>
<td>Second Year GC</td>
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<td>2</td>
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<td></td>
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<tr>
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<tr>
<td>MFM fellow</td>
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<td>1</td>
<td>4</td>
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</tr>
<tr>
<td>TOTAL</td>
<td></td>
<td></td>
<td>14</td>
<td>42</td>
</tr>
</tbody>
</table>

Carly Pouchet: - returned form Maternity leave in January 2014 and left the Departement in August 2014. She was involved part-time in PND clinics only.

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:** The Department of Medical Genetics has been working closely with the office of the Director of Professional Services to assess clinical needs and plan for a transition of leadership. This process has taken much longer than anticipated and it is hoped that 2015 will be the year in which decisions are finalized.

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 was elected to the Canadian Academy of Health Sciences in 2014 and was awarded the O. Harold Warwick Prize of the Canadian Cancer Society (Cancer Control) for 2013. Dr. Foulkes is a James McGill Professor of Medicine at McGill University. In 2013, Dr. Rosenblatt joined the Board of Directors of 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**

   - **William Foulkes**  
     Professor
   - **Laurence Baret**  
     Faculty Lecturer-returning to the JGH in January 2015
   - **Maria Lalous**  
     Faculty Lecturer
   - **Lynn Macrae**  
     Faculty Lecturer- on maternity leave as of December 2014
   - **Carly Pouchet**  
     Faculty Lecturer- left the JGH in August 2014
   - **Lara Reichman**  
     Genetic counsellor-Started December 2014 (replacing Sonya Zaor (see below))
   - **David Rosenblatt**  
     Professor
   - **Heidi Rothenmund**  
     Faculty Lecturer- will leave for Winnipeg in 2015
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 and Martah Urquilla provide administrative support for the department, with various other individuals providing help on a part time basis.

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.

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. 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 (most recently awarded first)

- Dept. of the Army USAMRAA, Military Medical Research and Development, Ovarian Cancer Research Program (OCRP): Systematic approaches to dissect driver pathways in small cell carcinoma of the ovary, hypercalcemic type

- Quebec Breast Cancer Foundation (QBCF): The genetics of breast cancer in Quebec populations: twenty years after BRCA1/2.

- CCSRI: Towards a biological understanding of small cell carcinoma of the ovary, hypercalcemic type

- 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,

- Prostate Cancer Canada: Funding for the Canadian BRCA1/2 Prostate Cancer Network,

- Susan G Komen for the Cure Komen Scholar: Hereditary breast cancer: cause and effect

- Cancer Research Society: Assessing the contribution of spindle assembly checkpoint genes to colorectal carcinoma

- Comité mixte Québec-Catalogne, du Ministère des Relations internationals: Identification de nouveaux gènes de susceptibilité à la néoplasie colorectale.

Co-investigator

- Dorval, M
Co-applicants: Foulkes WD et al.
CRS/QBCF: Cancer screening practices of non-carriers from BRCA1/2 mutation-positive families: Extent, determinants and psychosocial impact of over-screening

- Mes-Masson, A
Co-applicants: Foulkes WD et al.
FRQS: Budget alloué dans le cadre du projet FRQS-Réseau de recherche sur le cancer (Axe banque de tissus et de données)

- Tischkowitz, M
Co-applicants: Foulkes WD et al.
Cancer Research Society: A multimodal genomic analysis strategy to identify novel breast cancer genes

**David Rosenblatt**

**CURRENT COMPETITIVE GRANTS**

As Principal Investigator

CIHR, Operating Grant, PI – 2009-2014 (1 year extension to 2015)
Inborn Errors of Folate and Cobalamin Metabolism

3. **Scholarly works published in the 2014 calendar year:**

**William Foulkes**


David Rosenblatt


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

   **William Foulkes**

   January 30th 2014
   **Title:** DICER1: from ontogenesis to oncogenesis
   2014 AACR special conference on cancer genetics
   San Diego Westin Hotel
   San Diego, CA

   June 21st, 2014
   **Title:** Things I didn’t know – a (mainly) endocrine tale of deep voices, neck lumps and more
ICE-ENDO 2014
Mc Cormack Conference Centre
Chicago, IL.

September 8th, 2014
Title: Germ-line and somatic mutations characterize small cell carcinoma of the ovary, hypercalcemic type
Marsha Rivkin Centre-AACR ovarian cancer research symposium
Pacar Building, Seattle University
Seattle, WA

September 16th, 2014
Title: Inherited susceptibility to non-epithelial ovarian cancers
Centre of Cancer Research and Cell Biology,
Queen’s University, Belfast, Northern Ireland

September 17th, 2014
Title: Germ-line and somatic mutations characterize small cell carcinoma of the ovary, hypercalcemic type
HGV 2014 meeting,
Culloden Hotel and Spa, Belfast, Northern Ireland

September 18th, 2014
Title: DICER1: A review of the syndrome with a focus on endocrine aspects
European Society for Pediatric Endocrinology annual meeting,
Conference Centre Dublin, Dublin, Ireland

September 22, 2014

Sept 23rd, 2014
Title: Inherited susceptibility to non-epithelial ovarian cancers
UHN Department of Defense Ovarian cancer conference
Toronto General Hospital
Toronto, ON

November 6, 2014
Title: Beyond BRCA – PALB2, ATM, CHEK2 and other “new” breast cancer susceptibility genes
Dept Medicine Seminar
University of Ottawa
Ottawa, ON

December 5, 2014
Title: BRCA testing: a geneticist’s view
Gynecology-Oncology Canada, Communities of Practice, Fall meeting
SECTION III - CONFIDENTIAL INFORMATION

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. Dr. Rosenblatt serves on the committee of INESS that evaluated the introduction of new laboratory tests in Quebec. Dr Foulkes is Section Editor for Cancer Genetics, *Current Oncology*

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 30, 2015