Hôpital général juif
Jewish General Hospital

Hôpital d'enseignement de l'Université McGill
A McGill University Teaching Hospital

DÉPARTEMENT DE MÉDECINE GÉNÉTIQUE - DEPARTMENT OF MEDICAL GENETICS
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Annual Report
Division of Medical Genetics
Department of Medicine - Jewish General Hospital
January 1 - December 31, 2012

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. Those of Dr. William Foulkes can be found at www.mcgill.ca/cancergenetics/. 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. In 2012, Dr. Marc Tischkowitz completed his leave of absence and decided to remain at Cambridge where he holds a faculty position.

Clinical Activities

Our overall clinical activity continues to increase in prenatal and cancer activities. In the hereditary cancer clinic, there were 526 follow-up visits and 586 new visits for a total of 1,112 visits in 2012 compared to 436 follow-up visits and 553 new visits for a total of 989 visits in 2011. In the Prenatal Diagnosis clinic there were 371 follow-up visits and 204 new visits for a total of 575 visits in 2012 compared to 301 follow-up visits and 123 new visits for a total of 424 visits in 2011.

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. Perhaps the most exciting work in 2012 revolves around the recognition of the importance of *DICER1* mutations in cancer. This has become a major focus of Dr. Foulkes's research program.

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 involved in teaching in the M.Sc. program in Genetic Counselling and with residents.

McGill Genetic Counselling MSc Program

Lynn Macrae
Clinical Supervision: First Year Student, Clinical Rotation, 1 student, 4 weeks
Second Year Student, Clinical Rotation, 1 student, 3 weeks (6 weeks** rotation shared with Sonya Zaor) Total: 7 weeks
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

François Plourde
Intro to Research Ethics-HGEN 600-Genetic Counselling; Department: Human Genetics; Format: One Group Teaching; Role: Facilitator- 1 session; Titles: Consent Form Design, and the IRB Process; 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: Lecture and Small Group Teaching; Role: Lecturer; Titles: Cancer Genetics; Level: Medical Students; Time: 2 hour session
Intro to Research Ethics-HGEN 600-Genetic Counselling: Department: Human Genetics; Format: One Group Teaching; Role: Facilitator; Titles: Consent Form Design, and the IRB Process; Time: 1.5 hours

Summary of McGill Teaching Contributions by Genetic Counsellors

Maria Lalous
Clinical Supervision: Karen Canales - 1st year GC student - 4 wks ; Sonja Rummell - 1st year GC student - 4 wks

Lynn Macrae
Clinical Supervision: Alicia Schiavi - 1st year GC student - 4 wks,
Small Group Teaching Unit 8: Genetic Testing in Families and Populations (2 hrs)

François Plourde
HGEN 600 - Genetic Counselling 1st Year Practicum: Intro to Research Ethics, Consent Form Design, and the IRB Process - March 28, 2013 (1 ½ hrs)

Carly Pouchet
Clinical Supervision: Grace Kim - 2nd year GC student - 6 wks
Small Group Teaching Unit 8: Genetic Testing in Families and Populations (2 hrs)
HGEN 600 - Genetic Counselling 1st Year Practicum: Prenatal Diagnosis II - Maternal Serum Screening - Jan. 30, 2013 (1 ½ hrs)

Nassim Taherian
Small Group Teaching Unit 8: Genetic Testing in Families and Populations (2 hrs) Cancer Genetics (2 hrs), Intro to Research Ethics, Consent Form Design, and the IRB Process - March 28, 2013 (1 ½ hrs)

Nora Wong
Small Group Teaching Unit 8: Cancer Genetics (2 hrs), Genetics Rounds April 26, 2013 (1 hour)
HGEN 600 - Genetic Counselling 1st Year Practicum: Cancer Genetics Workshop III: Risk Assessment - Jan. 9, 2013 (1 ½ hrs)

Sonya Zaor
Clinical Supervision: Karen Canales - 1st year GC student - 4 wks; Grace Kim - 2nd yr GC student - 2 wks; Lara Reichman - 2nd yr GC student - 6 wks
HGEN 600 - Genetic Counselling 1st Year Practicum: Cancer Genetics Workshop III: Risk Assessment - Jan. 7, 2013 (1 ½ hrs)

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.

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:

As noted above, Dr. Marc Tischkowitz has retired from the Department to take up a faculty position at Cambridge, UK. There have been several additions to the genetic counseling staff in 2012. These include Carly Pouchet and Lynn Macrae as clinical genetic counselors as well as François Plourde and Alexandra Volenik as Research Genetic Counselors. Rachel Silva-Smith has joined our team in 2012 as a research assistant.

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.

7. Fundraising:

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

SECTION I - 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 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.

2. A nominative list of academic staff, their academic rank

William Foulkes	Professor
Maria Lalous	Faculty Lecturer
Lynn Macrae	Faculty Lecturer
Carly Pouchet	Faculty Lecturer
David Rosenblatt	Professor
Nora Wong	Faculty Lecturer
Sonya Zaor	Faculty Lecturer

Nora Wong, Sonya Zaor, and Lynn Macrae are genetic counsellors who are primarily responsible for cancer genetics. Maria Lalous is a full time genetic counsellor in the area of prenatal diagnosis. Carly Pouchet works clinically in both cancer genetics and in prenatal diagnosis. Razia Chanda, Martah Urquilla and Danielle Veyre provide administrative support for the Department. In addition, François Plourde and Nassim Taherian have contributed to the teaching of genetic counseling students (see summary table below).

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.

Nora Wong is a full-time genetic counselor whose clinical activities have focused on increasing the accessibility of genetic testing for mutations in BRCA1/2 in women newly diagnosed with breast and ovarian cancer. She has been working with members of the Department of Pathology, Oncology, Surgery and Gyneco-oncology to achieve this goal. She continues her research on psychosocial aspects in HBOC families. With the collaboration of Dr. Alicia Navarro de Souza, a psychiatrist from Brazil, Danielle Groleau PhD, a researcher in transcultural psychiatry, Carmen Loiselle RN, PhD, Chair in Psychosocial Oncology and Dr. Foulkes, she has focused on the cultural differences between French Canadians and Ashkenazi Jewish women living with a BRCA1 or BRCA2 mutation. With Lynn Macrae she has looked at the risk experience of young women who are non-carriers of a family-specific BRCA1/2-mutation. One project focuses on the attitudes and preferences for genetic testing in women newly diagnosed

with breast or ovarian cancer, and another multi-center study in Montreal and Quebec City looks at the screening practices of women found not to carrier their family-specific BRCA1/2 mutation. Michel Dorval, PhD, is the PI for this last project. Nora Wong is also a member of a Clinical Advisory Committee in Quebec looking at the development of tools for health professionals to help them best identify and support those women at risk of breast cancer.

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.

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 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. Please also refer to the annual report of the Program in Cancer Genetics for additional information: www.mcgill.ca/cancergenetics/.

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

1. Grants and awards received

William Foulkes

-Schutt Foundation, Operating, 2011-2012 (PI)
-Ride to Conquer Cancer, Operating, 2011-2012(PI)
- Prostate Cancer Canada, Operating, 2011-2012(PI)
- Canadian Cancer Society Bridge Funding, Operating, 2010-2011 no cost ext. 2012(PI)
- Susan G Komen for the Cure Komen Scholar, Operating, 2010-2014(PI)
-CBCRA, Idea, 2009-2011 no cost ext. 2012(PI)
-Cancer Research Society, Operating, 2011-2013(PI)

- CIHR, APOGEE, 2009-2014(CO-PI, no funds allocated to WDF)
- NIH (USA), Operating, renewed until 2012(CO-PI)
- CBCRA, Operating, 2009-2014 (CO-PI, no funds allocated to WDF)
- CRS, Operating, 2012-2014
-CRS/QBCF, Operating, 2012-2015(Co-PI)
-FRQS, Operating, renewed until 2014 (Co-PI)

David Rosenblatt
-CIHR, Operating Grant, PI – 2009-2014

2. Scholarly works published in the 2012 calendar year:

1. Cote S, Arcand SL, Royer R, Nolet S, Mes-Masson AM, Ghadirian P, **Foulkes WD**, Tischkowitz M, Narod SA, Provencher D, Tonin PN. The BRCA2 c.9004G>A (E2003K) variant is likely pathogenic and recurs in breast and/or ovarian cancer families of French Canadian descent. Breast Cancer Res Treat. 2012 Jan;131(1):333-40. Erratum in: Breast Cancer Res Treat. 2012 Jan;131(1):341.

2. Heravi-Moussavi A, Anglesio MS, Cheng S-WG, Senz J, Yang W, Prentice L, Fejes AP, Chow C, Tone A, Kalloger SE, Hamel N, Roth A, Ha G, Wan ANC, Maines-Bandiera S, Salamanca C, Pasini B, Clarke BA, Lee AF, Lee C-H, Zhao C, Young RH, Aparicio SA, Sorensen PHB, Woo MMM, Boyd N, Jones SJM, Hirst M, Marra MA, Gilks B, Shah SP, **Foulkes WD**, Morin GB, Huntsman DG. Recurrent somatic DICER1 mutations in non-epithelial ovarian cancers. *N Engl J Med.* 2012 Jan 19;366(3):234-42.

3. Natrajan R, Mackay A, Lambros MB, Weigelt B, Wilkerson PM, Manie E, Grigoriadis A, A'hern R, van der Groep P, Kozarewa I, Popova T, Mariani O, Turaljic S, Furney SJ, Marais R, Rodruigues DN, Flora AC, Wai P, Pawar V, McDade S, Carroll J, Stoppa-Lyonnet D, Green AR, Ellis IO, Swanton C, van Diest P, Delattre O, Lord CJ, **Foulkes WD**, Vincent-Salomon A, Ashworth A, Henri Stern M, Reis-Filho JS. A whole-genome massively parallel sequencing analysis of BRCA1 mutant oestrogen receptor-negative and -positive breast cancers. J Pathol. 2012 Feb 23.

4. Camp NJ, Cannon-Albright LA, Tammela TL, Schleutker J, Hoegel J, Herkommer K, Maier C, Vogel W, Wiklund F, Emanuelsson M, Grönberg H, Wiley KE, Isaacs SD, Walsh PC, Helfand BT, Kan D, Catalona WJ, Stanford JL, Fitzgerald LM, Johanneson B, Deutsch K, McIntosh L, Ostrander EA, Thibodeau SN, McDonnell SK, Hebbring S, Schaid DJ, Whittemore AS, Oakley-Girvan I, Hsieh CL, Powell I, Bailey-Wilson JE, Cropp CD, Simpson C, Carpten JD, Seminara D, Zheng SL, Xu J, Giles GG, Severi G, Hopper JL, English DR, **Foulkes WD**, Maehle L, Moller P, Badzioch MD, Edwards S, Guy M, Eeles R, Easton D, Isaacs WB; International Consortium for Prostate Cancer Genetics. Chromosomes 4 and 8 implicated in a genome wide SNP linkage scan of 762 prostate cancer families collected by the ICPCG. *Prostate*. 2012 Mar;72(4):410-23.

5. Kotsopoulos J, Lubisnki J, Salmena L, Lynch HT, Kim-Sing C, **Foulkes WD**, Ghadirian P, Neuhausen SL, Demsky R, Tung N, Ainsworth P, Senter L, Eisen A, Eng C, Singer C, Ginsburg O, Blum J, Huzarski T, Poll A, Sun P, Narod SA, Hbcc HB. Breastfeeding and

the Risk of Breast Cancer in BRCA1 and BRCA2 Mutation Carriers. Breast Cancer Res. 2012 Mar 9;14(2):R42.

6. Liu S, Lachapelle J, Leung S, Gao D, **Foulkes WD**, Nielsen TO. CD8+ lymphocyte infiltration is an independent favorable prognostic indicator in basal-like breast cancer.Breast Cancer Res. 2012 Mar 15;14(2):R48.

7. **Foulkes W**. Epigenetic modification and cancer: mark or stamp? Endocr Relat Cancer. 2012 Apr 10;19(2):C23-7.

8. Osher DJ, De Leeneer K, Michils G, Hamel N, Tomiak E, Poppe B, Leunen K, Legius E, Shuen A, Smith E, Arseneau J, Tonin P, Matthijs G, Claes K, Tischkowitz MD, **Foulkes WD**. Mutation analysis of RAD51D in non-BRCA1/2 ovarian and breast cancer families. Br J Cancer. 2012 Apr 10;106(8):1460-3.

9. Appleby-Tagoe JH, **Foulkes WD**, Palma L. Reading Between the Lines: A Comparison of Responders and Non-responders to a Family History Questionnaire and Implications for Cancer Genetic Counselling. *J Genet Couns*. 2012 Apr;21(2):273-91.

10. Tischkowitz M, Capanu M, Sabbaghian N, Li L, Liang X, Vallée MP, Tavtigian S, Concannon P, **Foulkes WD**; Leslie Bernstein, The WECARE Study Collaborative Group, Bernstein JL, Begg CB Rare germline mutations in PALB2 and breast cancer risk: A population-based study. Hum Mutat. 2012 Apr;33(4):674-80.

11. Arnes JB, Stefansson IM, Straume O, Baak JP, Lønning PE, **Foulkes WD**, Akslen LA. Vascular proliferation is a prognostic factor in breast cancer. Breast Cancer Res Treat. 2012 Jun;133(2):501-10.

12. McGee J, Kotsopoulos J, Lubinski J, Lynch HT, Rosen B, Tung N, Kim-Sing C, Karlan B, **Foulkes WD**, Ainsworth P, Ghadirian P, Senter L, Eisen A, Sun P, Narod SA. Anthropometric Measures and Risk of Ovarian Cancer Among BRCA1 and BRCA2 Mutation Carriers. Obesity (Silver Spring). 2012 Jun;20(6):1288-92.

13. Bailey-Wilson JE, Childs EJ, Cropp CD, Schaid DJ, Xu J, Camp NJ, Cannon-Albright LA, Farnham JM, George A, Powell I, Carpten JD, Giles GG, Hopper JL, Severi G, English DR, **Foulkes WD**, Mæhle L, Møller P, Eeles R, Easton D, Guy M, Edwards S, Badzioch MD, Whittemore AS, Oakley-Girvan I, Hsieh CL, Dimitrov L, Stanford JL, Karyadi DM, Deutsch K, McIntosh L, Ostrander EA, Wiley KE, Isaacs SD, Walsh PC, Thibodeau SN, McDonnell SK, Hebbring S, Lange EM, Cooney KA, Tammela TL, Schleutker J, Maier C, Borchum S, Hoegel J, Grönberg H, Wiklund F, Emanuelsson M, Cancel-Tassin G, Valeri A, Cussenot O, Isaacs WB. Analysis of Xq27-28 linkage in the international consortium for prostate cancer genetics (ICPCG) families. BMC Med Genet. 2012 Jun 19;13(1):46.

14. Lubinski J, Huzarski T, Byrski T, Lynch HT, Cybulski C, Ghadirian P, Stawicka M, **Foulkes WD**, Kilar E, Kim-Sing C, Neuhausen SL, Armel S, Gilchrist D, Sweet K, Gronwald J, Eisen A, Gorski B, Sun P, Narod SA; The Hereditary Breast Cancer Clinical

Study Group. The risk of breast cancer in women with a BRCA1 mutation from north america and poland. Int J Cancer. 2012 Jul 1;131(1):229-34.

15. Jin G, Lu L, Cooney KA, Ray AM, Zuhlke KA, Lange EM, Cannon-Albright LA, Camp NJ, Teerlink CC, Fitzgerald LM, Stanford JL, Wiley KE, Isaacs SD, Walsh PC, **Foulkes WD**, Giles GG, Hopper JL, Severi G, Eeles R, Easton D, Kote-Jarai Z, Guy M, Rinckleb A, Maier C, Vogel W, Cancel-Tassin G, Egrot C, Cussenot O, Thibodeau SN, McDonnell SK, Schaid DJ, Wiklund F, Grönberg H, Emanuelsson M, Whittemore AS, Oakley-Girvan I, Hsieh CL, Wahlfors T, Tammela T, Schleutker J, Catalona WJ, Zheng SL, Ostrander EA, Isaacs WB, Xu J; International Consortium for Prostate Cancer Genetics. Validation of prostate cancer risk-related loci identified from genome-wide association studies using family-based association analysis: evidence from the International Consortium for Prostate Cancer Genetics (ICPCG). Hum Genet. 2012 Jul;131(7):1095-103.

16. Kotsopoulos J, Lubinski J, Lynch HT, Kim-Sing C, Neuhausen S, Demsky R, **Foulkes WD**, Ghadirian P, Tung N, Ainsworth P, Senter L, Karlan B, Eisen A, Eng C, Weitzel J, Gilchrist DM, Blum JL, Zakalik D, Singer C, Fallen T, Ginsburg O, Huzarski T, Sun P, Narod SA. Oophorectomy after Menopause and the Risk of Breast Cancer in BRCA1 and BRCA2 Mutation Carriers.Cancer Epidemiol Biomarkers Prev. 2012 Jul;21(7):1089-96.

17. Sabbaghian N, Hamel N, Srivastava A, Albrecht S, Priest JR, **Foulkes WD**. Germline DICER1 mutation and associated loss of heterozygosity in a pineoblastoma. J Med Genet. 2012 Jul;49(7):417-9.

18. Wishart GC, Bajdik CD, Dicks E, Provenzano E, Schmidt MK, Sherman M, Greenberg DC, Green AR, Gelmon KA, Kosma VM, Olson JE, Beckmann MW, Winqvist R, Cross SS, Severi G, Huntsman D, Pylkäs K, Ellis I, Nielsen TO, Giles G, Blomqvist C, Fasching PA, Couch FJ, Rakha E, **Foulkes WD**, Blows FM, Bégin LR, Van't Veer LJ, Southey M, Nevanlinna H, Mannermaa A, Cox A, Cheang M, Baglietto L, Caldas C, Garcia-Closas M, Pharoah PD. PREDICT Plus: development and validation of a prognostic model for early breast cancer that includes HER2.Br J Cancer. 2012 Aug 21;107(5):800-7.

19. Choong CS, Priest JR, **Foulkes WD**. Exploring the endocrine manifestations of DICER1 mutations. Trends Mol Med. 2012 Sep;18(9):503-5.

20. **Foulkes WD**. Size Surprise? Tumour size, nodal status, and outcome after breast cancer. Curr Oncol. 2012 Oct;19(5):241-3.

21. Iqbal J, Ragone A, Lubinski J, Lynch HT, Moller P, Ghadirian P, **Foulkes WD**, Armel S, Eisen A, Neuhausen SL, Senter L, Singer CF, Ainsworth P, Kim-Sing C, Tung N, Friedman E, Llacuachaqui M, Ping S, Narod SA; the Hereditary Breast Cancer Study Group. The incidence of pancreatic cancer in BRCA1 and BRCA2 mutation carriers. Br J Cancer. 2012 Dec 4;107(12):2005-9.

22. Taherian N, Hamel N, Bégin LR, Bismar TA, Goldgar DE, Feng BJ, **Foulkes WD**. Familial prostate cancer: the damage done and lessons learnt. Nat Rev Urol. 2012 Dec 11;10(2):116-22.

23. Coelho D, Kim JC, Miouse IR, Fung S, du Moulin M, Buers I, Suormala T, Burda P, Frapolli M, Stucki M, Nürnberg P, Thiele H, Robenek H, Höhne W, Longo N, Pasquali M, Mengel E, Watkins D, Shoubridge EA, Majewski J, **Rosenblatt DS**, Fowler B, Rutsch F, Baumgartner MR. Mutations in *ABCD4* cause a new inborn error of vitamin B_{12} metabolism. *Nat. Genet.* 44:1152-1155, 2012.

24. Dempsey-Nunez L, Illson ML, Kent J, Huang Q, Brebner A, Watkins D, Gilfix BM, Wittwer C, **Rosenblatt DS**. High resolution melting analysis of the *MMAA* gene in patients with *cblA* and in those with undiagnosed methylmalonic aciduria. *Mol. Genet. Metab.* 107:363-367, 2012.

25. Deme JC, Miousse IR, Plesa M, Kim JC, Hancock MA, Mah W, **Rosenblatt DS**, Coulton JW. Structural features of recombinant MMADHC isoforms and their interactions with MMACHC, proteins of mammalian vitamin B_{12} metabolism. *Mol. Genet. Metab.* 107:352-362, 2012.

26. Moreno-Garcia M, **Rosenblatt DS**, Jerome-Majewska LA. The methylmalonic aciduria related genes, *Mmaa*, *Mmab* and *Mut*, are broadly expressed in placental and embryonic tissues during mouse organogenesis. *Mol. Genet. Metab.* 107:368-374, 2012.

27. Kim JC, Lee NC, Hwu WL, Chien YH, Fahiminiya S, Majewski J, Watkins D, **Rosenblatt DS**. Late onset symptoms in a patient with the *cblJ* inborn error of vitamin B_{12} metabolism: diagnosis and novel mutation revealed by exome sequencing. *Mol. Genet. Metab.* 107:664-668, 2012.

28. Mah W, Deme JC, Watkins D, Fung S, Janer A, **Rosenblatt DS**, Shoubridge EA, Coulton JW. Subcellular location of MMACHC and MMADHC, two human proteins central to intracellular vitamin B_{12} metabolism. *Mol. Genet. Metab.* 108:112-118, 2013.

29. Prasad C, Cairney AE, **Rosenblatt DS**, Rupar CA. Transcobalamin (TC) deficiency and newborn screening. *J Inher Metab Dis* 35:727, 2012.

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

William Foulkes

April 4 2012: *DICER1, microRNA and hereditary cancer: uncharted territories,* AACR Hereditary Cancers Symposium, McCormick West Convention Center, Chicago, Illinois

April 25-27, 2012: *Management and clinical Issues,* HBOC Symposium: From theory to practice, Montreal, Quebec

May 1, 2012: *Germline DICER1 mutations results in a surprising constellation of phenotypes,* Canadian Human and Statistical Genetics Meeting, White Oaks Conference Center, Niagara-on-the-Lake, Ontario

May 2, 2012: *Rare Diseases: Familial Ovarian Germ Cell Tumors,* CIHR and Canadian British Consulate Workshop, White Oaks Conference Center, Niagara-on-the-Lake, Ontario

May 7, 2012: *DICER1 mutations characterize a novel syndrome with endocrine features,* 15th International Congress of Endocrinology and 14th European Congress of Endocrinology, Fortezza da Basso, Florence, Italy

June 22, 2012: *Basal-like and triple negative breast cancer: same, different, who cares?* IUSCC seminar, Indianapolis, Indiana

June 29, 2012: *Basal-like and triple negative breast cancer: same, different, who cares?* Department of Medicine Grand Rounds, Memorial University, St-John's, Newfoundland

September 24, 2012: CIHR funded team in Familial Risks of Breast Cancer Workshop, Life *Insurance: Breast Cancer Research and Genetic Risk Prediction,* Loews Concorde Hotel, Quebec City, QC

David Rosenblatt

July 22-27, 2012: *Vitamin B_{12} Transport and Metabolism: lessons from Mendelian Disease,* Gordon Research Conference (GRC), Chemistry and Biology of Tetrapyrroles, Salve Regina University, Newport, Rhode Island

September 20-22, 2012: *Lessons in Biology from Patients with Inborn Errors of Vitamin B_{12} Metabolis,* Vitamin B_{12} Symposium, Nancy, France

September 24, 2012: *Lessons in Biology from Patients with Inborn Errors of Vitamin B_{12} Metabolis,* Medical Genetics Seminar, Hôpital Necker, Paris, France

October 24, 2012: *Lessons in Biology from Patients with Inborn Errors of Vitamin B_{12} Metabolism,* The Children's Hospital of Philadelphia, Grand Rounds-Annual Palmieri Lectureship, Philadelphia, Pennsylvania

November 15-16, 2012: *Disorders of Cobalamin Transport,* Neurometabolic disorders Related to B Vitamins, Orphan Europe Academy, Manchester, UK

SECTION III - CONFIDENTIAL INFORMATION

1. Consulting activities: None reported

Respectfully submitted,

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

June 2013