Hôpital général juif
Jewish General Hospital

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

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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 ful ime 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 rol in clinical and research aspects of hereditary cancer predisposition at the JGH. In his role as Director of the rogram 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

516-614B 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

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

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:

	Length Of Clinical Rotation (wks)	Number	Total time (wks)
FirstYearStudent	4	1	4
Second year Student	2	1	2
Total	-	2	6

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 d ?*
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:

	Length Of Clinical Rotation (wks)	Number	Total time (wks)
FirstYearStudent	4	1	4
Second Year Student	8	1	6

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:

	Length Of Clinical Rotation (wks)	Number	Total time (wks)
FirstYearStudent	4	1	4
Second Year Student	6	1	6

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 Me abolism: Lessons from the Clinic* Bethesda, MD

March 1, 2011
SIMD Annual Meeting
Title: *Clinical characterization of patients with various forms of homocystein uria* Asilomar, CA

Marc Tischkowitz

May 20th, 2010 *Investigating the PALB2 Hereditary Breast Cancer Gene n Quebec Families*, Québec— 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 diction of cancer risk and treatment response. Goodman Cancer Research Public Lecture Series, McGil 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 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)
–Marsha Rivkin Center for Ovarian Cancer Research, 2009-2010 (PI)
–US ARMY, Synergistic, 2008-2010 (PI)
–WEBC, 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)

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)

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.

Service Type	2005	2006	2007	2008	2009	2010
CANCER GENETICS						
new consultations	283	351	395	438	500	596
return visits	289	250	261	255	332	403
ward consultations	4	5	4	6	6	5
phone consultations	40	39	42	99	59	75
total (excluding phone consults)	576	606	660	693	828	1004
Total year-on-year increase	**616**	**645** **5%**	**702** **8%**	**798** **14%**	**887** **11%**	**1079** **22%**
PRENATAL DIAGNOSIS						
new consultations	245	281	260	203	304	283
return visits	75	162	112	306	156	97
Total year-on-year increase	**320**	**443** **38%**	**372*** **-16%**	**509** **37%**	**460** **-10%**	**380** **-12%**
GENERAL GENETICS						
new+ward consultations		49	34	71	66	69
return visits		11	23	29	30	27
Total		**62**	**57**	**100**	**96**	**96**
TOTAL FOR ALL AREAS year-on-	**936**	**1150** **23%**	**1131** **-2%**	**1407** **24%**	**1443** **3%**	**1555** **7%**

4) Academic Staff

Dr. David Rosenblatt continues to be Director of the U y Division of Medical Genetics in the McGill Department of Medicine; this enhances integrated servi livery 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 serv . 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 fo 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

1. **Foulkes WD**, Traffic Control for BRCA1. Foulkes WD. N Engl J Med. 010 Feb 25;362(8):755-6.

2. Greenwood CM, Sun S, Veenstra J, Hamel N, Niell B, Gruber S, **Foulkes WD**. How old is this mutation? - a study of three Ashkenazi Jewish founder mutations. *BMCGenet*. May 14;11:39, 2010.

3. Christensen GB, Baffoe-Bonnie AB, George A, Powell I, Bailey-Wilson JE, Carpten JD, Giles GG, Hopper JL, Severi G, English DR, **Foulkes WD**, Maehle L, Moller P, Eeles R, Easton D, Badzioch MD, Whittemore AS, Oakley-Girvan I, Hsieh CL, Dimitrov L, Xu J, Stanford JL, Johanneson B, Deutsch K, McIntosh L, Ostrander EA, Wiley KE, Isaacs SD, Walsh PC, Isaacs WB, Thibodeau SN, McDonnell SK, Hebbring S, Schaid DJ, Lange EM, Cooney KA, Tammela TL, Schleutker J, Paiss T, Maier C, Grönberg H, Wiklund F, Emanuelsson M, Farnham JM, Cannon-Albright LA, Camp NJ; International Consortium for Prostate Cancer Genetics. Genome wide linkage analysis of 1,233 prostate cancer pedigrees from the International Consortium for Prostate Cancer Genetics using novel sumLINK and sumLO analyses. *Prostate.* May 15;70(7):735-44, 2010.

4. Lachapelle J, Gilbert L, Artho G, Alcindor T, Amre R, Arseneau J, **Foulkes WD**. Colonic or coelomic? *Lancet*. May 22;375(9728):1844, 2010.

5. Blows FM, Driver KE, Schmidt MK, Broeks A, van Leeuwen FE, Wesseling J, Cheang MC, Gelmon K, Nielsen TO, Blomqvist C, Heikkilä P, Heikkinen T, Nevanlinna H, Akslen LA, Bégin LR, **Foulkes WD**, Couch FJ, Wang X, Cafourek V, Olson JE, Baglietto L, Giles GG, Severi G, McLean CA, Southey MC, Rakha E, Green AR, Ellis IO, Sherman ME, Lissowska J, Anderson WF, Cox A, Cross SS, Reed MW, Provenzano E, Dawson SJ, Dunning AM, Humphreys M, Easton DF, García-Closas M, Caldas C, Pharoah PD, Huntsman D. Subtyping of breast cancer by immunohistochemistry to investigate a relationship between subtype and short and long term survival: a collaborative analysis of data for 10,159 cases from 12 studies. *PLoS Med*. May 25; 7(5):e1000279, 2010.

6. **Foulkes WD**, Reis-Filho JS, Narod SA. Tumor size and survival in breast cancer--a reappraisal. *Nat Rev Clin Oncol.* Jun;7(6):348-53, 2010.

7. Brown LA, Johnson K, Leung S, Bismar TA, Benítez J, **Foulkes WD**, Huntsman DG. Co-amplification of CCND1 and EMSY is associated with an adverse outcome in ER-positive tamoxifen-treated breast cancers. *Breast Cancer Res Treat*. Jun; 121(2):347-54, 2010.

8. **Foulkes WD**, Tomlinson IP. Are we there yet?: journeying along th ancer genetic information superhighway. *Curr Opin Genet Dev*. Jun; 20(3):197-200, 2010.

9. Peixoto A, Santos C, Pinheiro M, Pinto P, Soares MJ, Rocha P, Gusmão L, Amorim A, van der Hout A, Gerdes AM, Thomassen M, Kruse TA, Cruger D, Sunde L, Bignon YJ, Uhrhammer N, Cornil L, Rouleau E, Lidereau R, Yannoukakos D, Pertesi M, Narod S, Royer R, Costa MM, Lazaro C, Feliubadaló L, Graña B, Blanco I, de la Hoya M, Caldés T, Maillet P, Benais-Pont G, Pardo B, Laitman Y, Friedman E, Velasco EA, Durán M, Miramar MD, Valle AR, Calvo MT, Vega A, Blanco A, Diez O, Gutiérrez-Enríquez S, Balmaña J, Ramon Y Cajal T, Alonso C, Baiget M, **Foulkes W**, **Tischkowitz M**, Kyle R, Sabbaghian N, Ashton-Prolla P, Ewald IP, Rajkumar T,Mota-Vieira L, Giannini G, Gulino A, Achatz MI, Carraro DM, de Paillerets BB, Remenieras A, Benson C, Casadei S, King MC, Teugels E, Teixeira MR. International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. *Breast Cancer Res Treat.* Jul 22, 2010. [Epub ahead of print]

10. Vicus D, Finch A, Rosen B, Fan I, Bradley L, Cass I, Sun P, Karlan B, McLaughlin J, Narod SA; Hereditary Ovarian Cancer Clinical Study Group. Risk factors for carcinoma of the

fallopian tube in women with and without a germline BRCA mutation. *Gynecol Oncol.* Aug 1;118(2):155-9, 2010.

11. Akbari MR, Tonin P, **Foulkes WD**, Ghadirian P, **Tischkowitz M**, Narod SA. RAD51C germline mutations in breast and ovarian cancer patients. *Breast Cancer Res.* Aug 19;12(4):404, 2010.

12. YilmazA, Hamel N,SchwartzCE, Houlston RS, HarperJI, **Foulkes WD**. A genome-wide analysis of loss of heterozygosity and chromosomal copy number variation in Proteus syndrome using high- density SNP microarrays. *J Hum Genet.* 2010 Sep; 55(9):627-30.

13. Rio Frio T, Haanpää M, Pouchet C, Pylkäs K, Vuorela M, **Tischkowitz M**, Winqvist R, **Foulkes WD**. Mutation analysis of the gene encoding the PALB2-binding protein MRG15 in BRCA1/2-negative breast cancer families. *J Hum Genet.* Sep 16, 2010. [Epub ahead of print]

14. Leong SP, Shen ZZ, Liu TJ, Agarwal G, Tajima T, Paik NS, Sandelin K, Derossis A, Cody H, **Foulkes WD**. Is breast cancer the same disease in Asian and Western countries? *WorldJ Surg.* Oct; 34(10):2308-24, 2010.

15. Shuen A, **Foulkes WD.** Clinical implications of next-generation sequencing for cancer medicine. *Curr Oncol.* 2010 Oct;17(5):39-42.

16. **Foulkes WD**, Smith IE, Reis-Filho JS. Triple-negative breast cancer. *N Engl J Med.* 2010 Nov 11;363(20) :1938-48.

17. Diabetes and breast cancer among women with BRCA1 and BRCA2 mutations. Bordeleau L, Lipscombe L, Lubinski J, Ghadirian P, **Foulkes WD**, Neuhausen S, Ainsworth P, Pollak M, Sun P, Narod SA; the Hereditary Breast Cancer Clinical Study roup. *Cancer.* 2010 Nov 10. [Epub ahead of print]

18. Metcalfe K, Lubinski J, Lynch HT, Ghadirian P, **Foulkes WD**, Kim-Sing C, Neuhausen S, Tung N, Rosen B, Gronwald J, Ainsworth P, Sweet K, Eisen A, Sun P, Narod SA; for the Hereditary Breast Cancer Clinical Study Group. Family History of Cancer and Cancer Risks in Women with BRCA1 or BRCA2 Mutations. *J Natl Cancer Inst.* 2010 Nov 23. [Epub ahead of print]

19. Dennis J, Ghadirian P, Little J, Lubinski J, Gronwald J, Kim-Sing C, **Foulkes W**, Moller P, Lynch HT, Neuhausen SL, Domchek S, Armel S, Isaacs C, Tung N, Sweet K, Ainsworth P, Sun P, Krewski D, Narod S; the Hereditary Breast Cancer Clinical Study Group. Alcohol consumption and the risk of breast cancer among BRCA1 and BRCA2 mutation carriers. *Breast.* 2010 Dec; 19(6):479-83.

20. Ginsburg OM, Kim-Sing C, **Foulkes WD**, Ghadirian P, Lynch HT, Sun P, Narod SA; Hereditary Breast Cancer Clinical Study Group. BRCA1 and BRCA2 families and the risk of skin cancer. *Fam Cancer. Fam Cancer.* 2010 Dec; 9(4):489-93.

21. Bahubeshi A, Bal N, Rio Frio T, Hamel N, Pouchet C, Yilmaz A, Bouron-Dal Soglio D, Williams GM, **Tischkowitz M**, Priest JR, **Foulkes WD**. Germline DICER1 mutations and familial cystic nephroma. *J Med Genet.* 2010 Dec;47(12):863-6.

22. Levesque S, Ahmed N, Nguyen VH, Nahal A, Blumenkrantz Puligandla P, Chong G, **Foulkes WD**. Pediatrics. Neonatal Gardner Fibroma: A Sentinel Prese tion of Severe Familial

Adenomatous Polyposis. *Pediatrics*. 2010 Dec;126(6):e1599-602.

23. Hamel N, Feng BJ, Foretova L, Stoppa-Lyonnet D, Narod SA, Imyanitov E, Sinilnikova O, Tihomirova L, Lubinski J, Gronwald J, Gorski B, Hansen TV, Nielsen FC, Thomassen M, Yannoukakos D, Konstantopoulou I, Zajac V, Ciernikova S, Couch FJ, Greenwood CM, Goldgar DE, **Foulkes WD**. On the origin and diffusion of BRCA1 c.5266dupC (538 insC) in European populations. *Eur J Hum Genet*. 2010 Dec 1. [Epub ahead of print]

24. Rio Frio T, Lavoie J, Hamel N, Geyer FC, Kushner YB, Novak DJ, Wark L, Capelli C, Reis-Filho JS, Mai S, Pastinen T, **Tischkowitz MD**, Marcus VA, **Foulkes WD**. Homozygous *BUB1B* mutation and susceptibility to gastrointestinal neoplasia. *N EnglJ Med* 2010 Dec: 363;27

25. Quadros EV, Lai S, Nakayama Y, Sequeira M, Hannibal L, Wang S, Jacobsen DW, Fedesov S, Wright E, Gallagher RC, Anastasio N, Watkins D, **Rosenblatt DS**. Positive Newborn Screen for Methylmalonic Aciduria identifies the First Mutation in TCblR/CD320, the Gene for Cellular Uptake of Transcobalamin-Bound Vitamin $_{B12}$. *Hum Mutat*31:924-929, 2010.

26. Anastasio N, Ben-Omran T, Teebi A, Ha KCH, Lalonde E ,Ali R, Almureikhi M,Der Kaloustian VM, Liu J, **Rosenblatt, DS**, Majewski J, Jerome-Majewska LA. Mutations in SCARF2 are Responsible for Van Den Ende-Gupta Syndrome. *Amer J Hum Genet*87: 553–559, 2010.

27. Plesa M, Kim J, Paquette SG, Gagnon H, Ng-Thow-Hing C, Gibbs BF, Hancock MA, **Rosenblatt DS,** Coulton JW. Interaction between MMACHC and MMADHC, two human proteins participating in intracellular vitamin $_{B12}$ metabolism. *Molec Genet Metab* 102: 139-148, 2010

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

29. Al Fares A, Millington GWM, **Tischkowitz MD** Dermatological features of inherited cancer syndromes in adults *Clinical and Experimental Dermatology* 2010 Jul;35(5):462-7

30. McQuirter M, Castiglia LL, Loiselle CG, **Wong N.** Decision-making process of women carrying a BRCA1 or BRCA2 mutation who have chosen pro ylactic mastectomy. Oncol Nurs Forum. 2010 May;37(3):313-20.

Book co-editorship

1. Cooney K and **Foulkes WD**. The genetics of male reproductive cancers. Springer, 2010.

Chapters

1. Watkins D and **Rosenblatt DS**. Vitamin B12 and folate metabolism. In: Mechanisms in Hematology, 4th Edition, Israels LG and Israels, E.D (eds), Core Health Services. http://wwwmechanismsinhematology.ca/chapter.aspx? Chapter =11,2010

2. Watkins D and **Rosenblatt DS.** Vitamin B12: disorders of absorption and metabolism. In: Encyclopedia of Life Sciences, John Wiley and Sons, Chichester. Doi:10.1002/9780470015902.a0002267.pub2, 2010.

3. **Rosenblatt DS**, Watkins D. Prenatal diagnosis of miscellaneous biochemical disorders. In: Genetics Disorders and the Fetus: Diagnosis, Preventio nd Treatment, 6th Edition, A. Milunsky (ed), Wiley-Blackwell, Oxford. 2010, Chapter 19 pp 614-627.

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