Annual Report
Division of Medical Genetics, Department of Medicine
Sir Mortimer B. Davis - Jewish General Hospital
April 1, 2006-May 31, 2007

1. Highlights

Medical Genetics has been functioning as a Department at the Jewish General Hospital in its own right for a full year. At the same time, it has maintained 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 practises as a full time medical geneticist. Nora Wong and Sonya Zaor are full time counsellors, primarily responsible for cancer genetics. Maria Lalous, genetic counsellor, who joined the department in August 2005 primarily in the area of prenatal diagnosis, went on maternity leave in December 2006. Jennifer Fitzpatrick, the director of the McGill M.Sc. program in Genetic Counselling returned as a temporary replacement. Razia Chanda and Danielle Veyre provide administrative support for the Department.

Marc Tischkowitz trained in both medical oncology and clinical genetics and his main clinical and research interest is hereditary cancer predisposition. Dr. William Foulkes also continues to play a major role in clinical and research aspects of hereditary cancer predisposition at the JGH.

On the eighth floor of the A Pavilion, the department has administrative space and a dedicated counselling room, primarily used for prenatal diagnosis cases. 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. The Program in Cancer Genetics, shared between the McGill Departments of Oncology and Human Genetics is now well established (www.mcgill.ca/cancergenetics/). This program is led by Dr. William Foulkes.

2. Evaluation of past academic year

a) Faculty

Dr. David Rosenblatt continues to be Director of the University Division of Medical Genetics in the Department of Medicine at both the JGH and MUHC sites; this enhances integrated service delivery. He is also Chair of the McGill Department of Human Genetics. The report of his academic activities for the past year can be obtained at www.mcgill.ca/finestone/.

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 has started a monthly multidisciplinary hereditary gastrointestinal cancer clinic with Dr. Polymnia Galiatsatos, a gastroenterologist at the JGH. In addition he has started 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/.

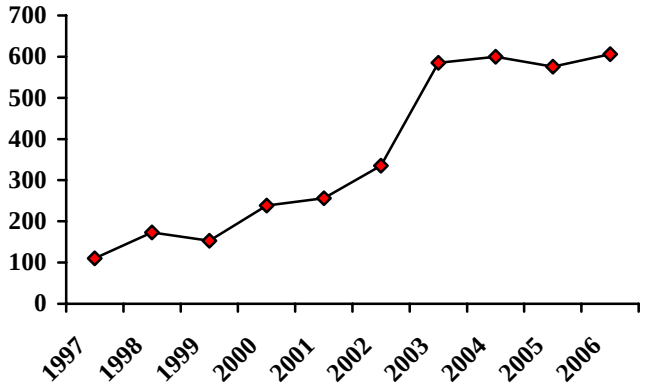
Dr. Eva Andermann, Dr. Serge Melancon, and Dr. Laura Russel, all of whom have active appointments at the MUHC, were appointed to the staff of the JGH as associate members. They join Dr. Vasken Der Kaloustian who also holds this appointment.

b) Clinical Activities

Cancer Genetics Clinical Activity

The following table and figure indicate the change in workload in cancer genetics from 1997 to 2006

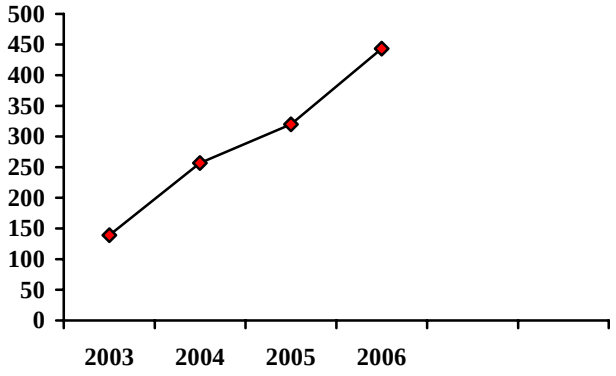
	1997	**1998**	**1999**	**2000**	**2001**	**2002**	**2003**	**2004**	**2005**	**2006**
Consultations	88	111	114	146	155	183	359	292	283	351
Visits	20	59	18	91	99	151	228	292	289	250
Ward	2	3	2	1	2	1	1	16	4	5
Phone	59	60	37	1	4	33	49		40	39
Total (excludes phone consults)	**110**	**173**	**153**	**238**	**256**	**335**	**585**	**600**	**576**	**606**



Prenatal Genetics Clinical Activity

The following table and figure indicate the change in workload in prenatal genetics from 2003-2006

	2003	2004	2005	2006
Consultations	101	213	245	281
Visits	38	44	75	162
Total	139	257	320	443



Other Clinics

Clinic Type		2006
General Genetics	**Consultations**	49
	Visits	11
	Total	**62**
Hereditary GI	**Consultations**	11
	Visits	2
	Total	**13**
Hereditary GYN	**Total**	**69**

c) Honours and Awards

As of 2007, Dr. William Foulkes is a Chercheur-National at the SMBD-Jewish General Hospital. This award was given to him by the Fonds de la recherche en Santé. He holds this award until 2011.

Since June 2003, he has been a William Dawson Scholar at McGill University. This is a major award equivalent to a Canada Research Char, Tier 2, and Dr. Foulkes holds this award until 2008.

In 2006, Dr. Marc Tischkowitz was awarded a Chercheur-Boursier Clinicien award from the FRSQ.

d) Teaching Activities – 2006-2007

GRADUATE AND UNDERGRADUATE COURSES

William Foulkes

516 – 614B	**Environmental Carcinogenesis**
Department:	Medicine (Div. Experimental Medicine)
Format:	Lecture
Title:	Cancer Genetics/Prevention
Role:	Lecturer
Level:	MSc program
Time (hr/yr):	One two-hour session

516 – 0635D	**Experimental and Clinical Oncology**
Format:	Lecture
Title:	***Cancer Genetics***
Role:	Lecturer
Level:	MSc program
Time (hr/yr):	1.5-hour seminar

521-690B	**Inherited Cancer Syndromes**
Department:	Department of Human Genetics
Format:	Lecture
Title:	***Cancer Genetics***
Role:	Lecturer
Level:	MSc program
Time (hr/yr):	Four two-hour sessions

Medical Genetics
Unit 8 small group teaching in medical genetics
Format: One 2 hour lecture and 4 small groups session, 3 hours each
Role: Lecturer
Level: Medical students

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-ordinator
Level: Undergraduate/Graduate
Time: 6 hours

Unit 8
Department: Human Genetics
Format: Lecture and Small Group Teaching
Role: Lecturer-2 sessions:
Titles: Introduction
Huntington Disease
Small Group Leader – 4 sessions
Level: Medical
Time: One hour lectures; 8 hours small groups

Marc Tischkowitz

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 (hr/yr): 1.5-hour seminar

Unit 8
Department: Human Genetics
Format: Lecture and Small Group Teaching
Role: Lecturer-2 sessions:
Titles: Cancer
Prenatal
Ethics
Small Group Leader – 2 sessions
Level: Medical
Time: One hour lectures; 8 hours small groups

Department: Human Genetics, Oncology
Format: Lecture and Small Group Teaching
Role: Lecturer
Titles: Using Pathology in Cancer Genetics
Level: Genetic Counselling MSc Course
Time: 1.5 hours

Maria Lalous (N.B. On maternity leave from January 2007)

McGill Genetic Counselling MSc Program

Clinical Supervision:

	Length Of Clinical Rotation (wks)	Number	Total time (wks)
First Year Student	4	2	8
Second year Student	8	2	16
Total	-	4	24

Medical Genetics
Unit 8 small group teaching in medical genetics
Format: One 2 hour lecture and 4 small group sessions, 3 hours each
Role: Lecturer
Level: Medical students

Nora Wong

HGEN600D2
Department: Human Genetics
Format: Lecture
Title: Risk Perception Factors Influencing and Behaviour
Role: Lecturer
Level: Graduate
Time: 1 X 3 hour session

HGEN601
Department: Human Genetics
Format: Lecture
Title: Decision-making under uncertainty
Role: Lecturer
Level: Graduate
Time: 1 X 3 hour session

McGill Genetic Counselling MSc Program

Clinical Supervision:

	Length Of Clinical Rotation (wks)	Number	Total time (wks)
First Year Student	4	2 (concurrent)	4

Workshop 2 - Cancer Risk Assessment
Methods used to evaluate cancer risk
Format: One small groups session, 3 hours
Role: Lecturer
Level: Genetic counseling students

Sonya Zaor

McGill Genetic Counselling MSc Program

Clinical Supervision:

	Length Of Clinical Rotation (wks)	Number	Total time (wks)
First Year Student	4	1	4

Workshop 1-Cancer Risk Assessment
Psychosocial Issues in Cancer Genetics: a patient testimony
Format: One small group session, 1.5 hours
Role: Lecturer
Level: Genetic counselling students

Medical Genetics
Unit 8 small group teaching in medical genetics
Format: One 2 hour lecture
Role: Lecturer
Level: Medical students

INVITED LECTURES, TALKS, PRESENTATIONS

William Foulkes

April 20, 2006
Title: ***Clinico-pathological features of basal-like/BRCA1 tumors***
"Basal-like and BRCA1-associated Breast Cancer" meeting
Harvard Club
Boston, MA, USA

June 15, 2006
Title: ***Genetics and Breast Cancer: An update***
Toronto Breast Cancer Symposium 2006
Metro Toronto Convention Center, Toronto, Ontario

August 16, 2006
Title: ***Recent advances in understanding of the inherited susceptibility to cancers of the prostate, pancreas, stomach and colorectum***
Australian Ovarian Cancer Study and the Family Cancer Clinics of Australia
Couran Cove Island Resort
Stradbroke Island, Australia

August 18, 2006
Title: ***Hereditary breast cancer: from pathology to treatment and beyond***
Australian Ovarian Cancer Study and the Family Cancer Clinics of Australia
Couran Cove Island Resort
Stradbroke Island, Australia

October 28, 2006
Title: ***Breakthrough treatments for BRCA1 and BRCA2 mutation carriers***
10th Annual Cincinnati Comprehensive Breast Cancer Conference
Cutting Edge Strategies in Breast Cancer: The next decade
Cincinnati, OH, USA

November 9, 2006
Title: ***Hereditary breast cancer: from pathology to treatment and beyond***
Cancer Colloquia IV: Cell and Molecular Biology of Breast Cancer
University of St-Andrews
St-Andrews, Scotland

January 5th, 2007
Title: ***DNA repair and cancer susceptibility***
Radiation Science Seminar
Montreal General Hospital

May 10th, 2007
Title: ***New developments in breast cancer genetics***
Manitoba Cancer Care workshop, ITMHRT
Winnipeg, Manitoba

David Rosenblatt

August 5-10, 2006
Chairman, FASEB Summer Research Conference
One Carbon Metabolism
Indian Wells, California

September 11, 2006
Moderator: ***Women and Health: Unravelling the Mysteries of Hormones***
National Council of Jewish Women of Canada
Gelber Conference Centre, Montreal, QC

September 26-29, 2006
Canadian Academy of Health Sciences
2nd Annual Meeting
Ottawa, Ontario

October 6-7, 2006
Genetic Testing
Center for Disease Control (CDC)
Atlanta, GA

April 19-22, 2007
Title: ***Inborn errors of vitamin B_{12}: From clinical phenotypic variability to novel metabolic steps***
Canadian Genetic Diseases Network
16th Annual Meeting
Saint-Sauveur, Quebec

Marc Tischkowitz

May 2, 2006
Title: ***BRCA1-associated "core basal phenotype" (ER-, HER2-, CK5/6+) of breast cancer is associated with a poor prognosis***
Réseau de Médicine Génétique Appliquées du FRSQ Journées Génétiques Meeting, Montréal

May 6, 2006
Title: ***Chromosome Breakage Syndromes and DNA repair***
Montreal Children's Hospital Friday Lecture and Seminar Series

May 8, 2006
Title:***HNPCC: Diagnostic Criteria and More…***
Polymnia Galiatsatos, Adrian Gologan, Marc Tischkowitz
Jewish General Hospital Medical Rounds

September 22, 2006
Title: ***An update on Hereditary Breast Cancer***
Hôpital Charles LeMoyne Oncology Rounds

October 26, 2006
Title: ***What's new in DNA Repair?***
Montreal Centre for Experimental Therapeutics in Cancer, 4th Annual Meeting

November 10, 2006
Title: ***Screening for genetic diseases in the Ashkenazi Jewish population***
Jewish General Hospital Obstetrics and Gynecology Rounds

December 14, 2006
Title: ***Genetic factors in breast and ovarian cancer: a primer for family doctors***
McGill Thursday Evening Lecture Series

February 15, 2007
Title: ***Pathology in Cancer Genetics***
Genetic Counselling Students Seminar

February 16, 2007
Title: ***Screening for genetic diseases in the Ashkenazi Jewish population***
Montreal Children's Hospital Friday Lecture and Seminar Series

February 26, 2007
Title: ***A stitch in time? - management issues in hereditary gastric cancer***
Jewish General Hospital Medical Rounds (also presented at Montreal General Hospital and Royal Victoria Hospital)

March 6, 2007
Title: ***It's a dangerous world out there: DNA repair and environmental toxins***
Environmental Carcinogenesis MSc course EXMD 614

March 23, 2007 (with Sonya Zaor)
Title: ***Dilemmas in Hereditary Gastric Cancer - a new case and an old case revisited.***
Montreal Children's Hospital Genetics rounds

April 13, 2007
Title: ***PALB2/FANCN – another link between Fanconi Anemia and breast cancer***
McGill University Hospitals Research Institute Genetics Axis and McGill Department of Human Genetics Seminar Series

April 13, 2007
Title: ***Fanconi anemia - an important cause of congenital malformations***
Montreal Children's Hospital neonatal rounds

May 16, 2007
Title: ***Genetics of Colorectal Cancer***
"Lets Talk about Colorectal Cancer" - Free Public Forum: Chevra Kadisha B'nai Jacob - Beit Hazikaron and Colorectal Association of Canada

May 27, 2007
Title: ***Hereditary Predisposition to Breast Cancer***
30 minute interview, Radio 940, Montreal, and
Title: ***Cancer- Are you at risk? Can you prevent it****?*
Public Seminar, Salon des Baby Boomers Plus, Palais des congrès, Montréal

Nora Wong

April 18, 2007
Title: ***Medical Genetics-Genetic risk and cancer***
Public Service Announcement
Minimed-SMBD-Jewish General Hospital
April 11, 18, 25 & May 2, 9, 16

May 31, 2007
Title: ***Cancer prevention Education: Insights from genetics and lifestyle changes***
Session A: Research in cancer patient education
Reasons to Hope, Knowledge to Cope: Innovations in Cancer Patient Education: Cancer Patient Education Network (CPEN)-Canada-5th Annual Conference Marriott Château Champlain, Montreal, Quebec May 31-June 2 2007

Sonya Zaor

March 23, 2007 (with Marc Tischkowitz)
Title: ***Dilemmas in Hereditary Gastric Cancer - a new case and an old case revisited.***
McGill Medical Genetic Grand Rounds

e) 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 the Past President of the Association of Medical Geneticists of Quebec. Dr. Foulkes is Director of the McGill Program in Cancer Genetics and plays a key role on the Board of Directors of the Hereditary Breast and Ovarian Cancer Foundation. 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. Nora Wong is the editor of the Hereditary Breast and Ovarian Cancer Foundation newsletter, an annual publication for individuals with BRCA1 or BRCA2 mutations.

f) Research Operating Funds – 2006-2007

William Foulkes

CBCRA – IDEA – 2006-2007 (PI)
CBCRA – Operating – 2006-2009 (PI)
CBCRA – Operating – 2005-2008 (PI)
CIHR – Operating – 2003-2006, extended 2007 (PI as of 12/18/06)
Susan G Komen BrCa Found. –2006-2009 (Co-I)
NIH – Group Grant – 2002-2006, extended 2007 (Co-I)
CIHR –Group Grant – 2003-2007 (Co-I)
PCRFC – Operating – 2005-2007 (Co-I)
CBCRI – Operating – 2004-2009 (Co-I)

David Rosenblatt

CIHR, Operating Grant, PI – 2006-2009
CIHR, Group Grant, Co-Investigator – 2007-2012

Marc Tischkowitz

Rethink Breast Cancer- Operating 2006-2008 (PI)

g) Publications for 2006-2007

William Foulkes

1. Soravia C, Delozier CD, Dobbie Z, Berthod CR, Arrigoni E, Brundler MA, Blouin JL, Foulkes WD, Hutter P. Double frameshift mutations in APC and MSH2 in the same individual. *Int J Colorectal Dis,* 21 (1): 79-83, 2006.

2. Barker KT, Foulkes WD, Schwartz CE, Labadie C, Monsell F, Houlston RS, Harper J. Is the E133K allele of VG5Q associated with Klippel-Trenaunay and other overgrowth syndromes? *J Med Genet*, 43(7): 613-614, 2006.

3. *Galiatsatos P, Foulkes WD. Familial adenomatous polyposis. *Am J Gastroenterol*, 101 (2): 385-98, 2006.

4. Nkondjock A, Ghadirian P, Kotsopoulos J, Lubinski J, Lynch H, Kim-Sing C, Horsman D, Rosen B, Isaacs C, Weber B, Foulkes WD, Ainsworth P, Tung N, Eisen A, Friedman E, Eng C, Sun P, Narod SA. Coffee consumption and breast cancer risk among BRCA1 and BRCA2 mutation carriers. *Int J Cancer,* 118 (1): 103-7, 2006.

5. *McVety S, Li L, Gordon PH, Chong G, Foulkes WD. Disruption of an exon splicing enhancer in exon 3 of MLH1 is the cause of HNPCC in a Quebec family. *J Med Genet*, 43 (2): 153-6, 2006.

6. Minoo P, Baker K, Goswami R, Chong G, Foulkes WD, Ruszkiewicz A, Barker M, Buchanan D, Young J, Jass JR. Extensive DNA methylation in normal colorectal mucosa in hyperplastic polyposis. *Gut*, 55(10): 1467-1474, 2006.

7. *Rudkin TM, Hamel N, Galvez M, Hogervorst F, Gille JJ, Moller P, Apold J, Foulkes WD. The frequent BRCA1 mutation 1135insA has multiple origins: a haplotype study in different populations. *BMC Med Genet*. 7 (1): 15, 2006.

8. *McVety S, Li L, Thiffault I, Gordon P.H, MacNamara E, Wong N, Australie K, Kasprzak L, Chong G, Foulkes WD. The value of multi-modal gene screening in HNPCC in Quebec: three mutations in mismatch repair genes that would have not been correctly identified by genomic DNA sequencing alone. *Familial Cancer*, 5: 21-8, 2006.

9. Oros KK, Leblanc G, Arcand SL, Shen Z, Perret C, Mes-Masson A-M, Foulkes WD, Ghadiran P, Provencher D, Tonin PN. Haplotype analysis suggest common founders in carriers of the recurrent BRCA2 mutation, 3398delAAAAG, in French Canadian hereditary breast and/ovarian cancer families. *BMC Medical Genetics*, 7: 23, 2006.

10. Li L, McVety S, Younan R, Du Sart D, Gordon PH, Hutter P, Hogervost FBL, Liang P, Chong G, Foulkes WD. Distinct patterns of Germ-Line Deletions in *MLH1* and *MLH2* : the Implication of Alu Repetitive Element in the genetic etiology of Lynch Syndrome (HNPCC). *Hum Mutations*, 27 (4): 388, 2006.

11. Friedman E, Kotsopoulos J, Lubinski J, Lynch HT, Ghadirian P, Neuhausen SL, Isaacs C, Weber B, Foulkes WD, Moller P, Rosen B, Kim-Sing C, Gershoni-Baruch R, Ainsworth P, Daly M, Tung N, Eisen A, Olopade OI, Karlan B, Saal HM, Garber JE, Rennert G, Gilchrist D, Eng C, Offit K, Osborne M, Sun P, Narod SA; the Hereditary Breast Cancer Clinical Study Group. Spontaneous and therapeutic abortions and the risk of breast cancer among BRCA mutation carriers. *Breast Cancer Res,* 8(2): R15, 2006.

12. *Morel CF, Thomas MA, Cao H, O'Neil CH, Pickering JG, Foulkes WD, Hegele RA. A *LMNA* splicing mutation in two sisters with severe Dunnigan-type familial partial lipodystrophy (FPLD2). *J.Clin.Endocrinol.Metab.* 91(7): 2689-2695, 2006.

13. Gronwald J, Tung N, Foulkes WD, Offit K, Gershoni R, Daly M, Kim-Sing C, Olsson H, Ainsworth P, Eisen A, Saal H, Friedman E, Olopade O, Osborne M, Weitzel J, Lynch H, Ghadirian P, Lubinski J, Sun P, Narod SA. Tamoxifen and contralateral breast cancer in BRCA1 and BRCA2 carriers: An update. *Int J Cancer*, 118 (9): 2281-4, 2006.

14. Narod SA, Lubinski J, Ghadirian P, Lynch HT, Moller P, Foulkes WD, Rosen B, Kim-Sing C, Isaacs C, Domcheck S, Sun P; for the Hereditary Breast Cancer Clinical Study Group. Screening mammography and risk of breast cancer in BRCA1 and BRCA2 mutation carriers: a case control study. *Lancet Oncol,* 7(5): 402-406, 2006.

15. Tischkowitz MD, Foulkes WD. The basal phenotype of BRCA1-Related Breast Cancer: Past, Present and Future. *Cell Cycle*, 5 (9): 963-7, 2006.

16. Goswami RS, Minoo P, Baker K, Chong G, Foulkes WD, Jass JR. Hyperplastic polyposis and cancer of the colon with gastrinoma of the duodenum. *Nat Clin Pract Oncol,* 3 (5): 281-4, 2006.

17. *Galiatsatos P, Kasprzak L, Chong G, Jass JR, Foulkes WD. Multiple primary malignancies in a patient with situs ambiguous. *Clin Genet,* 69 (6): 528-531, 2006.

18. Foulkes WD. BRCA1 and BRCA2: Chemosensitivity, Treatment Outcomes and Prognosis. *Fam Cancer*, 5 (2): 135-142, 2006.

19. Shinto E, Baker K, Tsuda H, Mochizuki H, Ueno H, Matsubara O, Foulkes WD, Jass JR. Tumor Buds Show Reduced Expression of Laminin-5 gamma 2 Chain in DNA Mismatch Repair Deficient Colorectal Cancer. *Dis. Colon and Rectum*., 49(8): 1193-1202, 2006.

20. Tischkowitz M, Gologan A, Srolovitz H, Khanna M, Foulkes WD. Muir Torre syndrome and MSH2 mutations: the importance of dermatological awareness. *Br J Cancer*, 95 (2): 243-244.

21. Finch A, Beiner M, Lubinski J, Lynch HT, Moller P, Rosen B, Murphy J, Ghadirian P, Friedman E, Foulkes WD, Kim-Sing C, Wagner T, Tung N, Couch F, Stoppa-Lyonnet D, Ainsworth P, Daly M, Pasini B, Gershoni-Baruch R, Eng C, Olopade OI, McLennan J, Karlan B, Weitzel J, Sun P, Narod SA, Hereditary Ovarian Cancer Clinical Study Group. Salpingo-oophorectomy and the risk of ovarian, fallopian tube and peritoneal cancers in women with BRCA1 or BRCA2 mutation. *JAMA*, 296 (2): 185-192, 2006.

22. Baker K, Chong G, Foulkes WD, Jass JR. Transforming growth factor-pathway disruption and infiltration of colorectal cancers by intraepithelial lymphocytes. *Histopathology*, 49(4): 371-380, 2006.

23. Oros, KK, Ghadirian P, Maugard CM, Perret C, Paredes Y, Mes-Masson AM, Foulkes WD, Provencher D, Tonin P. Application of BRCA1 and BRCA2 mutation carrier prediction models in breast and/or ovarian cancer families of French Canadian descent. *Clin Genet*, 70(4): 320-329, 2006.

24. Schaid DJ, McDonnell SK, Zarfas KE, Cunningham JM, Hebbring S, Thibodeau SN, Eeles RA, Easton DF, Foulkes WD, Simard J, Giles GG, Hopper JL, Mahle L, Moller P, Badzioch M, Bishop DT, Evans C, Edwards S, Meitz J, Bullock S, Hope Q, Guy M, Hsieh C-L, Halpern J, Balise RR, Oakley-Girvan I, Whittemore AS, Xu J, Dimitrov L, Chang B-L, Adams TS, Turner AR, Meyers DA, Friedrichsen DM, Deutsch K, Kolb S, Janer M, Hood L, Ostrander EA, Stanford JL, Ewing CM, Gielzak M, Isaacs SD, Walsh PC, Wiley KE, Isaacs WB, Lange EM, Ho LA, Beebe-Dimmer JL, Wood DP, Cooney KA, Seminara D, Ikonen T, Baffoe-Bonnie A, Fredriksson H, Matikainen MP, Tammela T LJ, Bailey-Wilson J, Schleutker J, Maier C, Herkommer K, Hoegel JJ, Vogel W, Paiss T, Wiklund F, Emanuelsson M, Stenman E, Jonsson B-A, Grönberg H, Camp NJ, Farnham J, Cannon-Albright LA, Catalona WJ, Suarez BK, Roehl KA. Pooled genome linkage scan of aggressive prostate cancer: results from the International Consortium for Prostate Cancer Genetics. *Human Genetics,* 120(4): 471-485, 2006.

25. Kotsopoulos J, Lubinski J, Lynch H.T, Klijn J, Ghadirian P, Neuhausen SL, Kim-Sing C, Foulkes WD, Moller P, Isaacs C, Domchek S, Randall S, Offit K, Tung N, Ainsworth P, Gershoni-Baruch R, Eisen A, Daly M, Karlan B, Saal HM, Couch F, Pasini B, Wagner T, Friedman E, Rennert G, Eng C, Weitzel J, Sun P, Narod SA, Hereditary Breast Cancer Clinical Study Group. Age at first birth and the risk of breast cancer in *BRCA1* and *BRCA2* mutation carriers. *Breast Cancer Res.* Jan 24 2007 [Epub ahead of print]

26. Refae MA, Wong N, Patenaude F, Bégin LR, Foulkes WD. Hereditary leiomyomatosis and renal cell cancer: an unusual and aggressive form of hereditary renal carcinoma. *Nature Clinical Practice Oncology*, 4(4): 256-61, 2007.

27. Hamel N, Wong N, Alpert L, Galvez M, Foulkes WD. Mixed ovarian germ cell tumor in a BRCA2 mutation carrier. *Int J Gybecol Pathol*, 26 (2): 160-4, 2007.

28. Tischkowitz M, Xia B, Sabbaghian N, Reis-Filho JS, Hamel N, Li G, Van Beers EH, Li, L, Khalil T, Quenneville L, Omeroglu A, Poll A, Wong N, Nederlof PM, Ashworth A, Tonin PN, Narod S, Livingston DM, Foulkes WD. Analysis of *PALB2/FANCN*–associated breast cancer families. *Proc Natl Acad Sci U S A*, 104(16): 6788-93, 2007.

David Rosenblatt

1. Worgan LC, Niles K, Tirone JC, Hofmann A, Verner A, Sammak A, Kucic T, Lepage P, Rosenblatt DS. The spectrum of mutations in *mut* methylmalonic academia and identification of a common Hispanic mutation and haplotype. *Hum Mutat* 27(1): 31-43, 2006.

2. Lerner-Ellis JP, Tirone JC, Pawelek PD, Dore C, Atkinson JL, Watkins D, Morel CF, Fujiwara TM, Moras E, Hosack AR, Dunbar GV, Antonicka H, Forgetta V, Fobson CM, Leclerc D, Gravel RA, Shoubridge EA, Coulton JW, Lepage P, Rommens JM, Morgan K, Rosenblatt DS. Identification of the gene responsible for methylmalonic aciduria and homocystinuria, *cblC* type. *Nat Genet* 38(1): 93-100, 2006. Erratum. *Nat Genet* 38(8): 957, 2006.

3. Zhang J, Dobson CM, Wu X, Lerner-Ellis JP, Rosenblatt DS, Gravel RA. Impact of cblB mutations on the function of ATP: Cob(I)alamin adenosyltransfrerase in disorders of vitamin B_{12} metabolism. *Mol Genet Metab* (87): 315-322, 2006.

4. Lerner-Ellis JP, Gradinger AB, Watkins D, Tirone JC, Villeneuve A, Lepage P, Dobson CM, Gravel RA, Rosenblatt DS. Mutation and biochemical analysis of patients belonging to the *cblB* complementation class of vitamin B_{-12} dependent methylmalonic aciduria. *Mol Genet Metab* (87): 219-225, 2006.

5. Dobson CM, Gradinger AB, Longo N, Wu X, Leclerc D, Lerner-Ellis JP, Lemieux M, Belair C, Watkins D, Rosenblatt DS, Gravel RA. Homozygous nonsense mutation in the *MCEE* gene and siRNA suppression of methylmalonyl-CoA epimerase expression: A novel cause of mild methylmalonic aciduria. *Mol Genet Metab* (88): 327-333, 2006.

6. Morel CF, Lerner-Ellis JP, Rosenblatt DS. Combined methylmalonic aciduria and homocystinuria (cblC): Phenotype-genotype correlations and ethnic-specific observations. *Mol Genet Metab* (88): 315-321, 2006.

7. Bikker H, Bakker HD, Abeling NGGM, Poll-The BT, Kleijer WJ, Rosenblatt DS, Waterham HR, Wanders RJA, Duran M. A homozygous nonsense mutation in the methylmalonyl-CoA epimerase gene results in mild methylmalonic aciduria. *Hum Mutat* 27(7): 640-643, 2006.

8. Moras E, Hosack A, Watkins D, Rosenblatt DS. Mitochondrial vitamin B_{12}-binding proteins in patients with inborn errors of cobalamin metabolism. *Mol Gen Metab* (90): 140-147, 2007.

9. Ernest S, Carter M, Shao H, Hosack A, Lerner N, Colmenares C, Rosenblatt DS, Yoh-Han Pao, Ross ME, Nadeau J. Parallel changes in metabolite and expression profiles in *Crooked-tail* mutant and folate-reduced wild-type mice. *Hum Mol Genet* 15(23): 3387- 93, 2006.

10. Gherasim C, Rosenblatt DS, Banerjee R. The polymorphic background of methionine synthase reductase modulates the phenotype of a disease-causing mutation. *Human Mutation* DOI 10.1002/humu.20563, 2007

11. Zhao R, Min SH, Qiu A, Sakaris A, Goldberg GL, Sandoval C, Malatack JJ, Rosenblatt DS, Goldman ID. The spectrum of mutations in the *PCFT* gene, coding for an intestinal folate transporter, that are the basis for Hereditary Folate Malabsorption. *Blood* DOI 10.1182/blood-2007-02-077099, 2007.

Marc Tischkowitz

1. Georgitsi M, Raitila A, Karhu A, van der Luijt RB, Aalfs CM, Sane T, Vierimaa O, Makinen MJ, Tuppurainen K, Paschke R, Gimm O, Koch CA, Gundogdu S, Lucassen A, Tischkowitz M, Izatt L, Aylwin S, Bano G, Hodgson S, De Menis E, Launonen V, Vahteristo P, Aaltonen LA Germline CDKN1B/p27Kip1 mutation in multiple endocrine neoplasia. *J Clin Endocrinol Metab*. 2007 May 22; [Epub ahead of print]

2. Tischkowitz M, Xia B, Sabbaghian N, Reis-Filho JS, Hamel N, Li G, Van Beers EH, Li L, Khalil T, Quenneville L, Omeroglu A, Poll A, Wong N, Nederlof PM, Ashworth A, Tonin PN, Narod S, Livingston DM and Foulkes WD. Analysis of *PALB2/FANCN*–associated breast cancer families. *Proc Natl Acad Sci U S A* 104 (16): 6788-6793, 2007.

3. Reid S, Schindler D, Hanenberg H, Barker K, Hanks S, Kalb R, Neveling K, Kelly P, Seal S, Freund M, Wurm M, Batish SD, Lach FP, Yetgin S, Neitzel H, Ariffin H, Tischkowitz M, Mathew CG, Auerbach AD, Rahman N. Biallelic mutations in *PALB2*, which encodes a BRCA2 interacting protein, cause Fanconi anemia subtype FA-N and predispose to childhood cancer. *Nat Gene* 39: 162-4, 2007.

4. Robin NH, Taylor CJ, McDonald-McGinn DM, Zackai EH, Bingham P, Collins KJ, Earl D, Gill D, Granata T, Guerrini R, Katz N, Kimonis V, Lin JP, Lynch DR, Mohammed SN, Massey RF, McDonald M, Rogers RC, Splitt M, Stevens CA, Tischkowitz MD, Stoodley N, Leventer RJ, Pilz DT, Dobyns WB. Polymicrogyria and deletion 22q11.2 syndrome: Window to the etiology of a common cortical malformation. *Am J Med Genet A* 140(22): 2416-25, 2006.

5. Tischkowitz M, Gologan A, Srolovitz H, Khanna M, Foulkes WD. Muir Torre syndrome and MSH2 mutations: the importance of dermatological awareness. *Br J Cancer* 95(2): 243-4, 2006.

6. Vogt J, Ryan E, Tischkowitz MD, Reardon W, Brueton LA. The tale of a nail sign in chromosome 4q34 deletion syndrome. *Clin Dysmorphol* 15(3): 127-132, 2006.

7. Tischkowitz MD, Foulkes WD. The basal phenotype of BRCA1-related breast cancer: past, present and future. *Cell Cycle* 5(9): 963-7, 2006.

Nora Wong

1. McVety S, Li L, Thiffault I, Gordon PH, Macnamara E, Wong N, Australie K, Kasprzak L, Chong G, Foulkes WD. The value of multi-modal gene screening in HNPCC in Quebec: three mutations in mismatch repair genes that would have not been correctly identified by genomic DNA sequencing alone. *Fam Cancer* 5(1): 21-8, 2006.

2. Tischkowitz M, Xia B, Sabbaghian N, Reis-Filho JS, Hamel N, Li G, Van Beers EH, Li L, Khalil T, Quenneville L, Omeroglu A, Poll A, Wong N, Nederlof PM, Ashworth A, Tonin PN, Narod S, Livingston DM and Foulkes WD. Analysis of *PALB2/FANCN*–associated breast cancer families. *Proc Natl Acad Sci U S A* 104 (16): 6788-6793, 2007.

3. Hamel N, Wong N, Alpert L, Galvez M, Foulkes WD. Mixed ovarian germ cell tumor in a BRCA2 mutation carrier. *Int J Gynecol Patho* 26(2): 160-4, 2007.

4. Refae MA, Wong N, Patenaude F, Begin LR, Foulkes WD. Hereditary leiomyomatosis and renal cell cancer: an unusual and aggressive form of hereditary renal carcinoma. *Nat Clin Pract Oncol* 4(4): 256-61, 2007.

3. 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 JGH. 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.

_____

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