Annual Report
Division of Medical Genetics
Department of Medicine
Jewish General Hospital
April 1, 2005-March 31, 2006

1. Highlights

The most important change over the last year is that Medical Genetics at the JGH has now been established as a department in its own right. This designation makes the JGH the first hospital in the McGill system to establish its own Department of Medical Genetics. There have also been a major expansion in personnel; Dr. Marc Tischkowitz, a medical geneticist trained in the United Kingdom, joined the Department in September 2005 and he is the first full time medical geneticist at the Jewish General Hospital since the departure of Dr. Leonard Pinsky. Maria Lalous, genetic counsellor, also joined the department in August 2005 and has taken over the prenatal diagnosis work from Jennifer Fitzpatrick who now works full time as director of the McGill M.Sc. program in Genetic Counselling. Razia Chanda also joined in August to take over the administrative work from Sabrina Notte who is now administrator to the McGill cancer genetics program.

The major clinical focus of the Department of Medical Genetics at the Jewish General Hospital continues to be in the areas of Hereditary Cancer and Prenatal Diagnosis. Nora Wong and Sonya Zaor are full time counsellors, primarily responsible for cancer genetics. 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. Maria Lalous works full-time in prenatal diagnosis - this post was previously a part-time post and the expansion of the post has brought additional capacity for seeing prenatal diagnosis cases and provides greater flexibility for patients in booking their appointments.

The department has undergone a major refurbishment and now has a dedicated counselling room, primarily used for prenatal diagnosis cases. Much of the departmental work continues to take place in the Cancer Prevention Centre, which has been incorporated into the new Segal Cancer Centre and provides state of the art facilities for managing individuals and families with a hereditary predisposition to cancer. This new space has also improved integration of the cancer genetics service with other oncology services.

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.

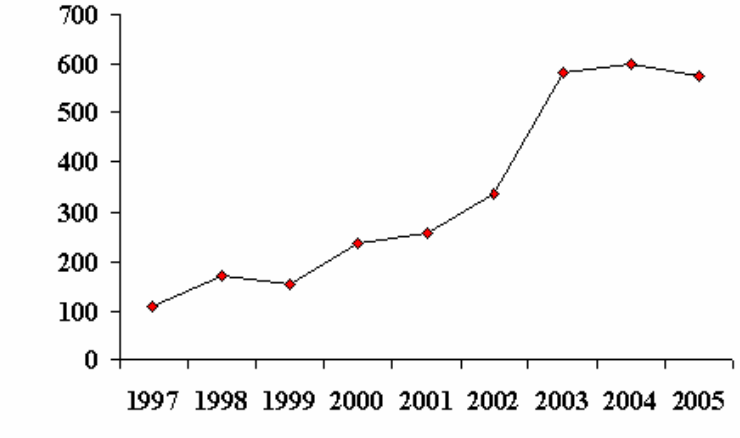
Dr. William Foulkes continues to run the cancer genetics service, dividing his time between the MUHC and the JGH. There are close links to the Departments of Oncology and Pathology, and to a number of surgical divisions, reflecting the multidisciplinary nature of the clinical service. Please also refer to the annual report of the Program in Cancer Genetics for additional information: www.mcgill.ca/cancergenetics/.

b) Clinical Activities

Cancer Genetics Clinical Activity

The following table and figures indicate the change in workload in cancer genetics over the years.

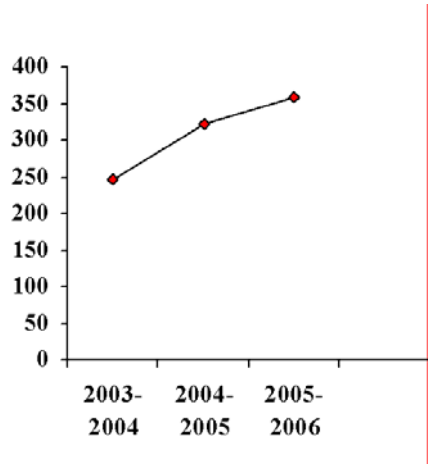
	1997	**1998**	**1999**	**2000**	**2001**	**2002**	**2003**	**2004**	**2005**
consultations	88	111	114	146	155	183	359	292	283
visits	20	59	18	91	99	151	228	292	289
ward	2	3	2	1	2	1	1	16	4
phone	59	60	37	1	4	33	49		40
total (excludes phone consults)	110	173	153	238	256	335	585	600	576



Prenatal Genetics Clinical Activity

During the past year, a total of 358 patients were seen for genetic counselling for prenatal diagnosis as compared to 322 patients last year. From the total 358 patients, 83 visits occurred during the period of April 1st 2005 to August 20, 2005, when the genetic counsellor, Jennifer Fitzpatrick, was working part-time in prenatal diagnosis. This averaged to 16.6 visits per period (~1month). Upon the hiring of a full-time prenatal diagnosis genetic counsellor, (August 21, 2005 to March 31, 2006) 273 visits occurred, averaging 34.12 visits per period (~1 month).

	2003-2004	**2004-2005**	**2005-2006**
Visits	247	322	358
Ward	-	-	3



c) Honours and Awards

Since 2002, Dr. William Foulkes has been a Chercheur-Boursier clinicien at a senior level 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 2007.

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 2005, Dr. David Rosenblatt was elected as an inauugural Fellow of the Canadian Academy of Health Sciences.

d) Teaching Activities-2005

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 group sessions, 3 hours each
Role: Lecturer
Level: Medical students

Unit 1 teaching
Format: Lecture
Role: Lecturer
Level: Medical Students
Time: One two-hour seminar

Back to Basics
Format: Lecture
Role: Lecturer
Level: Medical students

Time: One three-hour seminar, shared with Dr. Jeremy Jass

David Rosenblatt

Biology 568

Department: Biology/Human Genetics
Format: Lecture
Title: Somatic Cell Genetics
Role: Lecturer
Level: Undergraduate/Graduate
Time (hr/yr): 4 hours

Biology 575

Department: Biology/Human Genetics
Format: Lecture
Title: Inborn Errors of Folate and Cobalamin Transport and Metabolism
Role: Lecturer
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 Students
Time (hr/yr): 3 hours lectures; 8 hours small groups

Marc Tischkowitz

Advances in Human Genetics – Theme course

Department: Human Genetics
Format: Small Group Teaching
Title: ***Cancer Genetics***
Role: Lecturer
Level: MSc program
Time (hr/yr): Four 1.5 hour sessions

Maria Lalous

McGill Genetic Counselling MSc Program

Clinical Supervision:

	Length Of Clinical Rotation (wks)	Number	Total time (wks)
First Year Student	4	3	12
Second year Student	8	2	16
Total	-	5	28

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

Unit 1 teaching
Format: one two hour seminar
Role: Lecturer
Level: Medical students

Nora Wong

Superintegrated Small Group Session/Breast cancer
Format: one small group sessions, 3 hours each
Role: 1 of 3 tutors
Level: Medical students

Sonya Zaor

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

INVITED LECTURES, TALKS, PRESENTATIONS

William Foulkes

September 19, 2005
Title: *Role of genetic factors in cancer & familial diseases*
National Council of Jewish Women of Canada
Gelber Conference Centre
Montreal, Quebec

September 28, 2005
Title: *Genetics of colorectal cancer: What's new?*
CCMG 2005 Annual Meeting
Château Bromont
Bromont, Quebec

October 12, 2005
Title: *Genetic influence of breast and gynecological cancers in pre-menopausal women*
10th McGill International Symposium on Reproductive Endocrinology & Infertility and Women's Health
Centre Mont-Royal, Montreal, Quebec

October 20, 2005
Title: *Overview - 10 years of BRCA1 and BRCA2*
BRCA: Today & Tomorrow
First International Symposium on the Hereditary Breast and Ovarian Cancer Susceptibility Genes
Marriott Château Champlain, Montreal, Quebec

October 20, 2005
Title: *Outcome following BRCA1/2 related breast cancer*
BRCA: Today & Tomorrow
First International Symposium on the Hereditary Breast and Ovarian Cancer Susceptibility Genes
Marriott Château Champlain, Montreal, Quebec

February 2, 2006
Title: *Genetic Risk Assessment*
5th International "From Gene to Cure" Congress
Vrije Universiteit Amsterdam
Amsterdam, Netherlands

February 2, 2006
Title: *Prevention of Hereditary Breast Cancer*
5th International "From Gene to Cure" Congress
Vrije Universiteit Amsterdam
Amsterdam, Netherlands

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

Maria Lalous and Marc Tischkowitz

March 10th 2006
Title: *"Supermen" and "Superwomen" - Why are We Afraid of Them? Parental Decision-Making Following a Diagnosis of Sex chromosome abnormalities*.
JGH Obstetric and Gynaecology Rounds

David Rosenblatt

January 14, 2005
Title: *Mutation analysis in classical methylmalonic aciduria*
MUHC-RI Genetics Axis and McGill Department
of Human Genetics Seminar
Montreal Children's Hospital

March 6 – 9, 2005
Society for Inherited Metabolic Disorders
Asilomar, CA

March 16 – 17, 2005
Genetic Testing for Rare Diseases
Dallas, TX

Marc Tischkowitz

November 19th 2005
DNA repair syndromes – the clinical angle
McGill Oncology Department meeting, Lac Carling

February 27th 2006
Setting up remote clinics using telemedicine: Lessons from the UK experience with Medical Genetic Services
JGH medical rounds (also presented at MGH and RVH)

Nora Wong

September 30, 2005
Title: Workshop: *Challenges in Communicating Cancer Risk*
Concurrent Workshop Session I

Concurrent Workshop Session II
Canadian Association of Genetic Counsellors Annual Conference-
September 28-October 2, 2005

October 20 -21, 2005
Title: Workshop A: *Taking a History for Cancer Risk*
BRCA Today and Tomorrow; First International Symposium on the
Hereditary Breast and Ovarian Cancer Susceptibility Genes
Marriott Château Champlain, Montreal, Quebec

March 21, 2006
Title: *Cancer Genetics-Overview*
Biology 370 Applied Human Genetics

April 6, 2006
Title: Atelier: *Outils pour l'organisation des services de conseil génétique et pour le soutien au conseil génétique*: Letter writing
Association des conseillers et conseillères en génétique du Québec (ACCGQ) Annual Meeting-2006

April 28, 2006
Ti**t**le: *Extra, Extra Adrenal About it*
McGill Medical Genetics Case Presentation

Sonya Zaor

October 20 -21, 2005
Title: Workshop A: *Taking a History for Cancer Risk*
BRCA Today and Tomorrow; First International Symposium on the
Hereditary Breast and Ovarian Cancer Susceptibility Genes
Marriott Château Champlain, Montreal, Quebec

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.

f) Consulting Activities

William Foulkes

University Rank: Associate Professor
Period covered: June 1 2004-March 31 2006

g) Research Operating Funds – 2005-2006

William Foulkes

CBCRA – Operating – 2006-2009 (PI)
CBCRA – Operating – 2005-2008 (PI)
US Army –Concept Award – 2004, extended until 2006 (PI)
US Army –Concept Award –99-02, extended until 2006 (PI)
Susan G Komen BrCa Found. –2006-2009 (Co-I)
NIH – Group Grant – 2002-2006 (Co-I)
VRQ–Group Grant– 2002-2006 (Co-I)
FRSQ – Group Grant – 2000-2005(Co-I)
CIHR –Group Grant – 2003-2007 (Co-I)
PCRFC – Operating – 2005-2007 (Co-I)
CIHR – Operating – 2003-2006 (Co-I)
CBCRI – Operating – 2004-2009 (Co-I)

David Rosenblatt

CIHR – Operating – 2001-2006 (PI)
MOD – Operating – 2001-2005 (PI)
Garrod – Operating – 2004-2005 (PI)

h) Publications for 2005:

William Foulkes

1. Hope Q, Bullock S, Evans C, Meitz J, Hamel N, Edwards SM, Severi G, Dearnley D, Jhavar S, Southgate C, Falconer A, Dowe A, Muir K, Houlston RS, Engert JC, Roquis D, Sinnett D, Simard J, Heimdal K, Møller P, Maehle L, Badzioch M, The Cancer Research UK/British Association of Urological Surgeons' Section of Oncology Collaborators, Eeles RA, Easton DF, English DR, Southey M, Hopper JL, Foulkes WD, Giles GG. Macrophage Scavenger Receptor 1 (MSR1) 999C>T (R293X) mutation and risk of prostate cancer. *Cancer Epidemiol Biomark Prevent*, 14: 397-402, 2005 (WDF is corresponding author).

2. Metcalfe KA, Lynch HT, Ghadirian P, Tung N, Olivotto IA, Foulkes WD, Warner E, Olopade O, Eisen A, Weber B, McClennan J, Sun P, Narod SA. The risk of ovarian cancer after breast cancer in BRCA1 and BRCA2 carriers. *Obstet Gynecol Surg*, 60 (4): 235-6, 2005.

3. Metcalfe KA, Lynch HT, Ghadirian P, Tung N, Olivotto IA, Foulkes WD, Warner E, Olopade O, Eisen A, Weber B, McClennan J, Sun P, Narod SA. The risk of

ovarian cancer after breast cancer in BRCA1 and BRCA2 carriers corresponding. *Gynecol Oncol*, 96 (1): 222-6, 2005.

4. Chappuis PO, Donato E, Goffin JR, Wong N, Begin LR, Kapusta LR, Brunet JS,Porter P, Foulkes WD. Cyclin E expression in breast cancer: predicting germline BRCA1 mutations, prognosis and response to treatment. *Ann Oncol*, 16 (5): 735-742, 2005.

5. 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*, 20 (5): 466-70, 2005.

6. Collett K, Stefansson IM, Eide J, Braaten A, Wang H, Eide GE, Thoresen SO, Foulkes WD, Akslen LA. A Basal epithelial phenotype is more frequent in interval breast cancers compared with screen detected tumors. *Cancer Epidemiol Biomarkers Prev*, 14 (5): 1108-12, 2005.

7. Makriyianni I, Hamel N, Ward S, Foulkes WD, Graw S. BRCA1:185delAG found in the San Luis Valley probably originated in a Jewish founder. *J Med Genet*, 42 (5): e27, 2005 (WDF is corresponding author).

8. Arnes JB, Brunet JS, Stefansson I, Begin LR, Wong N, Chappuis PO, Akslen LA, Foulkes WD. Placental cadherin and the basal epithelial phenotype of BRCA1-related breast cancer. *Clin Cancer Res*, 11(11): 4003-11, 2005.

9. Weitzel JN, Robson M, Pasini B, Manoukian S, Stoppa-Lyonnet D, Lynch HT, McLennan J, Foulkes WD, Wagner T, Tung N, Ghadirian P, Olopade O, Isaacs C, Kim-Sing C, Moller P, Neuhausen SL, Metcalfe K, Sun P, Narod SA. A comparison of bilateral breast cancers in BRCA carriers. *Cancer Epidemiol Biomarkers Prev*, 14 (6): 1534-8, 2005.

10. Zauber NP, Sabbath-Solitare M, Marotta S, Zauber AG, Foulkes WD, Chan M, Turner F, Bishop DT. Clinical and genetic findings in an Ashkenazi Jewish population with colorectal neoplasms. *Cancer*, 15: 104 (4): 719-29, 2005.

11. Xu J, Dimitrov L, Chang BL, Adams TS, Turner AR, Meyers DA, Eeles RA, Easton DF, Foulkes WD, Simard J, Giles GG, Hopper JL, Mahle L, Moller P, Bishop T, Evans C, Edwards S, Meitz J, Bullock S, Hope Q, Hsieh CL, Halpern J, Balise RN, Oakley-Girvan I, Whittemore AS, Ewing CM, Gielzak M, Isaacs SD, Walsh PC, Wiley KE, Isaacs WB, Thibodeau SN, McDonnell SK, Cunningham JM, Zarfas KE, Hebbring S,Schaid DJ, Friedrichsen DM, Deutsch K, Kolb S, Badzioch M, Jarvik GP, Janer M, Hood L, Ostrander EA, Stanford JL, Lange EM, Beebe-Dimmer JL, Mohai CE, Cooney KA, Ikonen T, Baffoe-Bonnie A, Fredriksson H, Matikainen MP, Tammela TLj, Bailey-Wilson J, Schleutker J, Maier C, Herkommer K, Hoegel JJ, Vogel W, Paiss T, Wiklund F, Emanuelsson M, Stenman E, Jonsson BA, Gronberg H, Camp NJ, Farnham J, Cannon-Albright LA, Seminara D; ACTANE Consortium. A combined genomewide linkage scan of 1,233 families for prostate cancer-susceptibility genes conducted by the

international consortium for prostate cancer genetics. *Am J Hum Genet,* 77 (2): 219-29, 2005.

12. Kotsopoulos J, Lubinski J, Lynch HT, Neuhausen SL, Ghadirian P, Isaacs C, Weber B, Kim-Sing C, Foulkes WD, Gershoni-Baruch R, Ainsworth P, Friedman E, Daly M, Garber JE, Karlan B, Olopade OI, Tung N, Saal HM, Eisen A, Osborne M, Olsson H, Gilchrist D, Sun P, Narod SA. Age at menarche and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. *Cancer Causes Control,* 16 (6): 667-674, 2005.

13. McVety S, Younan R, Li L, Gordon PH, Wong N, Foulkes WD, Chong G. Novel genomic insertion-- deletion in MLH1: possible mechanistic role for non-homologous end-joining DNA repair. *Clin Genet,* 68 (3): 234-238, 2005.

14. Kotsopoulos J, Olopado OI, Ghadirian P, Lubinski J, Lynch HT, Isaacs C, Weber B, Kim-Sing C, Ainsworth P, Foulkes WD, Eisen A, Sun P, Narod SA. Changes in body weight and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Res,* 7 (5): R833-R843, 2005.

15. Sun S, Greenwood CM, Thiffault I, Hamel N, Chong G, Foulkes WD. The HNPCC associated MSH2*1906G-->C founder mutation probably originated between 1440 CE and 1715 CE in the Ashkenazi Jewish population. *J Med Genet,* 42 (10): 766-768, 2005.

16. Eisen A, Lubinski J, Klijn J, Moller P, Lynch HT, Offit K, Weber B, rebbeck T, Neuhausen SL, Ghadirian P, Foulkes WD, Gershoni-Baruch R, Friedman E, Rennert G, Wagner T, Isaacs C, Kim-Sing C, Ainsworth P, Sun P, Narod SA. Breast cancer risk following bilateral oophorectomy in BRCA1 and BRCA2 mutation carriers: an international case-control study. *J Clin Oncol,* 23 (30): 7491-6, 2005.

17. Cullinane CA, Lubinski J, Neuhausen SL, Ghadirian P, Lynch HT, Isaacs C, Weber B, Moller P, Offit K, Kim-Sing C, Friedman E, Randall S, Pasini B, Ainsworth P, Gershoni-Baruch R, Foulkes WD, Klijn J, Tung N, Rennert G, Olopade O, Couch F, Wagner T, Olsson H, Sun P, Weitzel JN, Narod SA. Effect of pregnancy as a risk factor for breast cancer in BRCA1/BRCA2 mutation carriers. *Int J Cancer,* 117 (6): 988-91, 2005.

18. Honrado E, Osorio A, Palacios J, Milne RL, Sanchez L, Diez O, Cazorla A, Syrjakoski K, Huntsman D, Heikkila P, Lerma E, Kallioniemi A, Rivas C, Foulkes WD, Nevanlinna H, Benitez J. Immunohistochemical expression of DNA repair proteins in familial breast cancer differentiate BRCA2-associated tumors. *J Clin Oncol,* 23 (30): 7503-11, 2005.

19. van der Klift H, Wijnen J, Wagner A, Verkuilen P, Tops C, Otway R, Kohonen-Corish M, Vasen H, Oliani C, Barana D, Moller P, Delozier-Blanchet C, Hutter P, Foulkes WD, Lynch H, Burn J, Moslein G, Fodde R. Molecular characterization of the spectrum of genomic deletions in the mismatch repair genes MSH2, MLH1,

MSH6, and PMS2 responsible for hereditary nonpolyposis colorectal cancer (HNPCC). *Genes Chromosomes Cancer*, 44 (2): 123-38, 2005.

David Rosenblatt

1. Guigonis V, Frémeaux-Bacchi V, Giraudier S, Favier R, Borderie D, Massy Z, Mougenot, Rosenblatt DS, and Deschênes G: Late-onset thrombocytic microangiopathy caused by *cblC* disease: Associated with a factor H mutation. *Am J Kidney Dis.* 45(3): 588-595, 2005.

2. Ernest S, Hosack A, O'Brien WE, Rosenblatt DS, and Nadeau JH: Homocysteine levels in A/J and C57BL/6J mice: genetic, diet, gender, and parental effects. *Physiol Genomics*. 21(3): 404-410, 2005.

3. Morel CF, Scott P, Christensen E, Rosenblatt DS, and Rozen R: Prenatal diagnosis for severe methylenetetrahydrofolate reductase deficiency by linkage analysis and enzymatic assay. *Mol Genet Metab*. 85(2): 115-120, 2005.

4. Morel CF, Watkins D, Scott P, Rinaldo P, and Rosenblatt DS: Prenatal diagnosis for methylmalonic acidemia and inborn errors of vitamin B_{12} metabolism and transport. *Mol Genet Metab.* 86:160-171, 2005.

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

6. Lerner-Ellis JP, Tirone JC, Pawelek PD, Dore C, Atkinson JL, Watkins D, Morel CF, Fujiwara TM, Moras E, Hossack 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. *Nature Genetics*, 38(1): 93-100, 2006.

7. 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. *Molecular Genetics and Metabolism* (In press).

8. 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 $B\text{-}_{12}$ dependent methylmalonic aciduria. *Molecular Genetics and Metabolism*, (87) 219-225, 2006.

9. 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. *Molecular Genetics and Metabolism* (In Press).

10. Morel CF, Lerner-Ellis JP, Rosenblatt DS: Combined methylmalonic aciduria and homocystinuria (cblC): Phenotype-genotype correlations and ethnic-specific observations. *Molecular Genetics and Metabolism* (In Press).

Marc Tischkowitz

1. Jordanna Joaquina Coelho, Angela Arnold, Jeremy Naylor, Marc Tischkowitz and James MacKay. An Assessment of the Efficacy of Cancer Genetic Counselling Using Real-time Videoconferencing Technology (Telemedicine) Compared to Face-to-Face Consultations. *European Journal of Cancer.* 2005; 41 2257-2261.

2. Neil V Morgan, Fahmida Essop, Ilja Demuth, Thomy de Ravel, Stander Jansen, Marc Tischkowitz, Cathryn M Lewis, Linda Wainwright, Janet Poole, Hans Joenje, Martin Digweed, Amanda Krause & Christopher G Mathew. A common Fanconi anaemia mutation in Black populations of Sub-Saharan Africa. *Blood.* 2005 1; 105(9): 3542-4.

3. H. V. New, C.M. Cale, M. Tischkowitz, B. Flores De Laurnaga, A. Jones, P.Telfer, P. Veys, A. D'Andrea, C.G. Mathew, and I. Hann. Nijmegen breakage syndrome diagnosed as Fanconi anaemia. *Pediatric Blood and Cancer* 2005; 44(5): 494-9

4. Elsa Callén, José A. Casado, Marc D. Tischkowitz, Juan A. Bueren, Amadeu Creus, Ricard Marcos, Angeles Dasí, Jesús M. Estella, Arturo Muñoz, Juan J. Ortega, Johan de Winter, Hans Joenje, Detlev Schindler, Helmut Hanenberg, Shirley V. Hodgson, Christopher G. Mathew, Jordi Surrallés. A common founder mutation in FANCA underlies the world highest prevalence of Fanconi anemia in Gypsy families from Spain. *Blood* 2005 Mar 1; 105(5**)**: 1946-9

Nora Wong

1. McVety S, Younan R, Li L, Gordon PH, Wong N, Foulkes WD, Chong G. Novel genomic insertion-- deletion in MLH1: possible mechanistic role for non-homologous end-joining DNA repair. Clin Genet. 2005 Sep; 68(3): 234-8.

2. Arnes JB, Brunet JS, Stefansson I, Begin LR, Wong N, Chappuis PO, Akslen LA, Foulkes WD. Placental cadherin and the basal epithelial phenotype of BRCA1-related breast cancer. Clin Cancer Res. 2005 Jun 1; 11(11): 4003-11.

3. Chappuis PO, Donato E, Goffin JR, Wong N, Begin LR, Kapusta LR, Brunet JS, Porter P, Foulkes WD. Cyclin E expression in breast cancer: predicting germline BRCA1 mutations, prognosis and response to treatment. Ann Oncol. 2005 May; 16(5): 735-42. Epub 2005 Mar 31.

4. Bayley JP, van Minderhout I, Weiss MM, Jansen JC, Oomen PH, Menko FH, Pasini B, Ferrando B, Wong N, Alpert LC, Williams R, Blair E, Devilee P, Taschner PE. Mutation analysis of SDHB and SDHC: novel germline mutations in sporadic head and neck paraganglioma and familial paraganglioma and/or pheochromocytoma. BMC Med Genet. 2006 Jan 11; 7:1

3. Objectives and Priorities

Our objective is 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. With the creation of a Department of Medical Genetics at the JGH in 2005, a mechanism is now in place to make the needs of both individuals and families in the area of medical genetics better known to the hospital and the community at large.

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David S. Rosenblatt, MD
Medical Geneticist