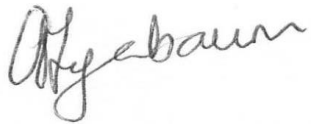

CURRICULUM VITAE

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EDUCATION

Degrees, Diplomas, Licensures and Certifications

1978	M.B., Ch.B., University of Cape Town Medical School Cape Town, South Africa
1979	ECFMG, USA
1979	Medical Practitioner, South African Medical and Dental Council
1980	Medical Practitioner, British Medical Council
1981	Diploma of Child Health, College of Medicine of South Africa
1981	VQE, USA
1985	F.C.P. (Paeds.), College of Medicine of South Africa Subject: Paediatrics
1986	MCCEE, Evaluating Examination Medical Council of Canada Ottawa, Ontario
1988	FRCP(C), Paediatrics Royal College of Physicians and Surgeons of Canada
1989	LMCC, Medical Council of Canada
1991	General License, College of Physicians and Surgeons of Ontario
2004	FCCMG, Fellow of the Canadian College of Medical Genetics (Biochemical Genetics)
2005	Certificate of Tutorial: Tricouncil Policy Statement Canada Subject: Ethical Conduct for research involving humans
2009	Diplomate of the American Board of medical genetics by Examination (Medical Biochemical genetics)
2011	General unrestricted medical license Medical Board of California

Specialty Training

1977	Boston Children's Hospital Boston, MA, USA 3 month elective Subject: Paediatric Pathology
1979	Groote Schuur Hospital Cape Town, South Africa Rotating Internship Subject: Obstetrics, Gynaecology, General Surgery, Cardiac Surgery, Urology
Jan 1980 - Jun 1980	Baragwanath Hospital Johannesburg, South Africa Senior house officer in Internal Medicine
Jul 1980 - Dec 1980	Baragwanath Hospital Johannesburg, South Africa Senior Paediatric House Officer
Jan-June 1981	Coronation Hospital Johannesburg, South Africa Senior Paediatric House Officer
1982 - 1985	Baragwanath, Coronation and Johannesburg Hospitals Johannesburg, South Africa Paediatric Registrar
1985	Johannesburg Hospital Johannesburg, South Africa Senior Registrar Subject: Paediatric Nephrology
1988	Jackson Laboratory Maine, USA Subject: Short Course in Human and Mammalian Experimental Genetics
1990	University of Toronto Toronto, Ontario Canada Subject: Course CH3113 in Clinical Epidemiology

Fellowships

1987 - 1989	The Hospital for Sick Children Toronto, Ontario Clinical Fellow Supervisor(s): Clarke J Subject: Genetics
1989	The Hospital for Sick Children Toronto, ON Research project during clinical fellowship Supervisor(s): Dr. Roy Gravel Subject: DNA Diagnostic Techniques in Tay-Sach's Disease

1989 - 1992	The Hospital for Sick Children Toronto. Research Institute MRC Canada Research Fellow Supervisor(s): Robinson B.H. Subject: Research Project: Molecular Biology Applications for Gene Characterization and Mutation Detection of Pyruvate Dehydrogenase E3

Other

1972	Rustenburg Girls High School Cape Town South Africa Matriculation certificate Subject: Distinction in Mathematics, Physical Sciences

APPOINTMENTS

University Appointments

1986-1987	University of Witwatersrand, Medical staff
1992 – 2007	Assistant Professor, Department of Paediatrics, Division of Clinical and Metabolic Genetics, The Hospital for Sick Children. University of Toronto, Toronto, Ontario
1992	Associate Member, Department of Molecular and Medical Genetics, University of Toronto, Toronto, Ontario
2007-	Associate Professor, Pediatrics, University of Toronto
2010-	Project Investigator, Division of Genetics, Metabolic Genetics Program, Department of Pediatrics,The Hospital for Sick and University of Toronto
2012 -	Associate Physician University of California San Diego.

Hospital Appointments

1979	Rotating Internship in Obstetrics, Gynaecology, General Surgery, Cardiac Surgery and Urology, Groote Schuur Hospital, Cape Town, South Africa
1980	Senior Paediatric House Officer, Baragwanath Hospital, Johannesburg, South Africa
1981	Senior Paediatric House Officer, Coronation Hospital, Johannesburg, South Africa
1982 - 1985	Paediatric Registrar, Baragwanath, Coronation and Johannesburg Hospitals, Johannesburg, South Africa
1985	Senior Registrar in Paediatric Nephrology, Johannesburg Hospital, Johannesburg, South Africa
1986	Attending Specialist in General Pediatrics and Paediatric Haematology/Oncology, Johannesburg Hospital, Johannesburg, South Africa
1987	Attending Consultant in Adult and Paediatric Endocrinology, Johannesburg Hospital, Johannesburg, South Africa
1989 - 1992	Examiner Paediatric OSCE, The Hospital for Sick Children,

	Toronto, ON
Jul 1990 - Jan 1991	Part time Paediatric Clinical Assistant/ Resident Clinical shift work Newborn Intensive Care Unit, The Hospital for Sick Children and Mount Sinai Hospital, Toronto, ON
1992 - 2010	Staff Pediatrician, Division of Clinical and Metabolic Genetics, Genetic Metabolics Disease program, The Hospital for Sick Children, Toronto.
2001 - 2010	Medical Director, Phenylketonuria Program, The Hospital for Sick Children, Toronto, ON
2005 - 2010	Project Director, Research Institute, Hospital for Sick Children, Toronto, Ontario
2007 - 2009	Section Head Genetic Metabolic Program Sickkids
2010 -	Project Investigator, Research Institute and Division of Clinical and Metabolic Genetics, Dept Pediatrics, Hospital for Sick Children, Toronto, Ontario
2012 -	Associate Physican, Rady Childrens Hospital, San Diego
2012-	Associate Physican, UCSD Genetics, San Diego
2014-	Associate Physician, Sharp Mary Birch Hospital, Pediatrics. San Diego.

Courtesy Cross Appointments under the appointment at Sickkids Hospital nonsalaried

1992 - 2001	The Hospital for Sick Children, Emergency Department, Part-time Staff Paediatrician
1996 - 2010	The Hospital for Sick Children, Department of Paediatrics, Medical Consultant
1999 - 2010	Mount Sinai Hospital, Department of Paediatrics, Courtesy
2003 - 2010	Associate staff, Toronto General Hospital Appointment/University Health Network, Department of Medicine
2007- 2010	Womens College Hospital, Department of Paediatrics, Courtesy

PUBLICATIONS

PA-denotes Principal Author; SRI-Senior Responsible Investigator; CPA-Co-Principal Author; C-Collaborator * indicates trainee supervised

Peer Reviewed - Journal Publications

Published

1. Mahuran DJ, Triggs-Raine BL, **Feigenbaum ASJ**, Gravel RA: The molecular basis of Tay-Sachs disease: Mutation identification and diagnosis. Clinical Biochemistry 1990: 23: pp 1-7. C
2. Triggs-Raine BL, **Feigenbaum ASJ,** Natowicz M, Skomorowski MA, Schuster SM, Clarke JTR, Mahuran DJ, Kolodny EH, Gravel R: Screening for carriers of Tay-Sachs disease among Ashkenazi Jews - A comparison of DNA-based and enzyme-

based tests. New England Journal of Medicine 1990: 323 (1): pp 6-12. CPA

3. Graham K, **Feigenbaum A,** Pastuszak A, Nulman I, Weksberg R, Einarson T, Goldberg S, Ashby S, Koren G: Pregnancy outcome and infant development following gestational cocaine use by social cocaine users in Toronto, Canada. Clinical and Investigative Medicine 1992: 15 (4): pp 384-394. CPA
4. *Tatuch Y, Christodoulou J, **Feigenbaum A,** Clarke JTR, Wherrett J, Smith C, Rudd N, Petrova-Benedict R, Robinson BH: Heteroplasmic mtDNA mutation (T - G) at 8993 can cause Leigh's disease when the percentage of abnormal mtDNA is high. American Journal of Human Genetics 1992: 50: pp 852-858. C
5. Van Allen MI, Siegel-Bartelt J, **Feigenbaum A,** Teshima IE: Craniosynostosis associated with partial duplication of 15q and deletion of 2q. American Journal of Medical Genetics 1992: 43: pp 688-692. C
6. **Feigenbaum A,** Robinson B: The structure of the human dihydrolipoamide dehydrogenase gene (DLD) and its upstream elements. Genomics 1993: 17: pp 376-381. PA
7. Koren G, Graham K, **Feigenbaum A,** Einarson T: Evaluation and counselling of teratogenic risk: The Motherisk approach. Journal of Clinical Pharmacology 1993: 33: pp 405-411. C
8. Hanley W, **Feigenbaum A,** Clarke JTR, Schoonheyt W, Austin V: Vitamin B12 deficiency in adolescent and young adults with phenylketonuria. Lancet 1993: 342(8877): pp 997. C
9. **Feigenbaum A,** Bergeron C, Richardson R, Wherrett J, Robinson B, Weksberg R: Premature atherosclerosis with photomyoclonic epilepsy, deafness, diabetes mellitus, nephropathy and neurodegenerative disease in two male siblings: A new syndrome? American Journal of Medical Genetics 1994: 49: pp 118-124. PA
10. Rovet J, Krekewich K, Perlman K, Weksberg R, Holland J, **Feigenbaum A:** Savant characteristics in a child with developmental delay and deletion in the short arm of chromosome 20. Developmental Medicine and Child Neurology 1995. C
11. **Feigenbaum A,** Chitayat D, Robinson B, MacGregor D, Myint T, Arbus G, Nowaczyk M: The expanding clinical phenotype of the tRNAleu(UUR)A->G mutation at 3243 of mitochondrial DNA: Diabetic embryopathy associated with mitochondrial cytopathy. American Journal of Medical Genetics 1996: 62: pp 404-409. PA
12. *Nowaczyk M, **Feigenbaum A,** Callahan J, Silver M, Levin A, Jay V: Bone marrow involvement and obstructive jaundice in Farber lipogranulomatosis: clinical and autopsy report of a new case. Journal of Inherited Metabolic Desease 1996: 19: pp 655-660. CPA
13. *Pitkanen S, **Feigenbaum A,** Lafromboise R, Robinson BH: NADH-coenzyme Q reductase (complex I) deficiency: heterogeneity in phenotype and biochemical findings. Journal of Inherited Metabolic Diseases 1996: 19: pp 675-686. CPA
14. Hanley WB, **Feigenbaum AS**, Clarke JT, Schoonheyt WE, Austin VJ: Vitamin B12 deficiency in adolescents and young adults with phenylketonuria. European Journal of Pediatrics 1996: 155: pp S145-7. CPA
15. Hanley WB, Demshar H, Preston MA, Borczyk A, Schoonheyt WE, Clarke JTR, **Feigenbaum A**: Newborn Phenylketonuturia (PKU) Guthrie (BIA) Screening and early hospital discharge. Early Human Development 1997: 47(1): pp 87-96. C
16. Gibson KM, **Feigenbaum A:** Ketothiolase deficiency. Journal of Inherited Metabolic Disease 1997: 20 (5): pp 712-713. CPA
17. Johnston J, Kelley RI, **Feigenbaum A,** Cox GF, Iyer GS, Funanage VL, Proujansky R: Mutation characterization and genotype-phenotype correlation in Barth syndrome.

American Journal of Human Genetics 1997: 61: pp 1053-1058. C

18. Strasberg P, Warren I, Skomorowski MA, **Feigenbaum A**: Homozygosity for the common Ashkenazi Jewish Tay-Sachs + IVS 12 splice - junction mutation: first report. Human Mutation 1997: 10: pp 82-83. CPA
19. Carbone MA, MacKay N, Ling M, Cole D, Douglas C, Rigat B, **Feigenbaum A,** Clarke JTR, Haworth JC, Greenberg CR, Seargent L, Robinson BH: Amerindian pyruvate carboxylase deficiency is associated with two distinct missense mutations. American Journal of Human Genetics 1998: 62 (6): pp 1312-1319. C
20. Garrett RM, Johnson JL, Graf TN, **Feigenbaum A**, Rajagopalan KV: Human sulfite oxidase R160Q. Identification of the mutation in a sulfite oxidase-deficient patient and expression and characterization of the mutant enzyme. Proceedings of the National Academy of Science 1998: 95 (11): pp 6394-6398. C
21. *Jankov RP, Boerkoel CF, Hellmann J, Sirkin WL, Tumer Z, Horn N, **Feigenbaum A:** Lethal neonatal Menkes disease with severe vasculopathy and fractures. Acta Paediatrica 1998: 87: pp 1297-1300. SRI
22. Shaag A, Saada AB, Berger I, Mandel H, Joseph A, **Feigenbaum A,** Elpeleg ON: Molecular basis of lipoamide dehydrogenase deficiency in Ashkenazi Jews. American Journal of Medical Genetics 1999: 82 (2): pp 177-182. C
23. Patel M, Callahan J, Zhang S, Unger S, Levin A, Skomorowski M-A, **Feigenbaum A,** O'Brien K, Hellmann J, Ryan G, Velsher L, Chitayat D: Early infantile galactosialidosis: prenatal presentation and postnatal follow-up. American Journal of Medical Genetics 1999: 85(1): pp 38-47. C
24. Smith ML, Saltzman J, Klim P, Hanley W, **Feigenbaum A**, Clarke JT: Neuropsychological function in mild hyperphenylalaninemia. Journal of Mental Retardation 2000: 105(2): pp 69-80. C
25. Seyda A, Newbold RF, Hudson TJ, Verner A, MacKay N, Winter S, **Feigenbaum A,** Malaney S, Gonzalez-Halphen D, Cuthbert AP, Robinson BH: A novel syndrome affecting multiple mitochondrial functions, located by microcell-mediated transfer to chromosome 2p14-2p13. American Journal of Human Genetics 2001: 68(2): pp 386-396. C
26. Nowaczyk MJ, Heshka T, Eng B, **Feigenbaum AJ**, Waye JS: DHCR7 genotypes of cousins with Smith-Lemli-Opitz syndrome. American Journal of Medical Genetics 2001: 100(2): pp 162-163. C
27. Brown NF, Mullur RS, Subramanian I, Esser V, Bennett MJ, Saudubray JM, **Feigenbaum AS**, Kobari JA, Macleod PM, McGarry JD, Cohen JC: Molecular characterization of L-CPT I deficiency in six patients: insights into function of the native enzyme. Journal of Lipid Research 2001: 42(7): pp 1134-1142. C
28. *Shah V, Friedman S, Moore AM, Platt BA, **Feigenbaum AS**: Selective screening for neonatal galactosemia: an alternative approach. Acta Paediatrica 2001: 90(8): pp 948-949. SRI
29. Salviati L, Scconi S, Mancuso M, Otaegui D, Camano P, Marina A, Rabinowitz S, Shiffman R, Thompson K, Wilson CM, **Feigenbaum A,** Naini A B, Hirano M, Bonilla E, DiMauro S and Vu TH: Mitochondrial DNA Depletion and dGK Gene Mutations. Annals of Neurology 2002: 52(3): pp 311-316. C
30. Van Der Knaap MS, Van Der Voorn P, Barkhof F, Van Coster R, Krageloh-Mann I, **Feigenbaum A,** Blaser S, Vles JSH, Rieckmann P, Pouwels PJW: A new leukoencephalopathy with brain stem and spinal cord involvement and high lactate. Annals of Neurology 2003: 53(2): pp 252-8. C
31. Schlame M, Kelley R, **Feigenbaum A,** Towbin J, Heerdt P, Schieble T, Wanders R,

Dimauro S, Blanck T: Phospholipid abnormalities in children with Barth syndrome. Journal of the American College of Cardiology 2003:42(11):pp 1994-9. C

32. **Feigenbaum A,** Moore R, Clarke J, Hewson S, Chitayat D, Ray PN, Stockley TL: Canavan Disease: Carrier-frequency determination in the Ashkenazi Jewish population and development of a novel molecular diagnostic assay. American Journal of Medical Genetics 2004: 142 (2): pp 142-7. SRI
33. *Stormon MO, Cutz E, Furuya K, Bedford M, Yerkes L, Tolan DR, **Feigenbaum A**: A six-month-old infant with liver steatosis. Journal of Pediatrics 2004: 144(2): pp 258-63. SRI
34. Mancuso M, Ferraris S, Pancrudo J, **Feigenbaum A,** Raiman J, Christodoulou J, Thorburn DR, DiMauro S: New DGK gene mutations in the hepatocerebral form of mitochondrial DNA depletion syndrome. Archives of Neurology 2005: 62(5): pp 745-7. C
35. Tadiboyina VT, Rupar A, Atkison P, **Feigenbaum A,** Kronick J, Wang J, Hegele RA: Novel mutation in DGUOK in hepatocerebral mitochondrial DNA depletion syndrome associated with cystasthioninuria. American Journal of Medical Genetics 2005: 135(3): pp 289-91. C
36. *Ben-Omran TI, Blaser S, Callahan J, Phillips H, **Feigenbaum A**: Atypical phenotype in a boy with maple syrup urine disease. Journal of Inherited Metabolic Disease 2006 Feb;29(1):195-200. SRI
37. *Glass H, **Feigenbaum A**, Clark JTR: A study on the nature of genetic metabolic practice at a major pediatric referral centre. Journal of Inherited Metabolic Disease 2006 Feb;29(1):175-8. C
38. Miller GL, Somani S, Nowaczyk MJ, **Feigenbaum A,** Davidson RG, Costa T, Levin AV: The ocular manifestations of Jacobsen syndrome: a report of four cases and review of the literature. Ophthalmic Genetics 2006: 27: pp 1. C
39. Cameron JM, Levandovskiy V, MacKay N, Raiman J, Renaud DL, Clarke JTR, **Feigenbaum A,** Elpeleg O, Robinson BH: Novel mutations in lipoamide dehydrogenase deficiency in two cousins with borderline-normal PDH complex activity. American Journal of Medical Genetics 2006: 140: pp 14. C
40. **Feigenbaum A,** Doherty E, Bai R, Kwon H, Tan D, Sloane A, Robinson B, Wong LJ: Novel mitochondrial DNA mutations associated with myopathy, cardiomyopathy, renal failure and deafness. American Journal of Medical Genetics A. 2006: Oct 15;140(20):2216-22. CPA
41. Blaser S, Propst E, James A, **Feigenbaum A**, Martin D, Shannon P, Papsin B: Inner ear dysplasia is common in children with down syndrome (trisomy 21). Laryngoscope. 2006 Dec;116(12):2113-9. C
42. Mahant S, **Feigenbaum A**: An infant with developmental delay. Canadian Medical Association Journal. 2006 Nov 21;175(11):1369. CPA
43. Scaglia F, Hsu CH, Kwon H, Bai RK, Perng CL, Dai P, Chang HM, O' Brian Smith E, Whiteman DAH, **Feigenbaum A,** Gropman A, Wong LJC: Molecular bases of hearing loss in multisystemic mitochondrial cytopathy. Genetics in Medicine. 2006 Oct;8(10):641-52 C
44. *Ben-Omran T, Wong H, **Feigenbaum A**: Late onset cobalamin-C disorder; a challenging diagnosis. American Journal of Medical Genetics Part A 143A:979–984 (2007). SRI
45. Levy HL, Milanowski A, Chakrapani A, Cleary M, Lee P, Trefz FK, Whitley CB, Feillet F, **Feigenbaum AS**, Bebchuk JD, Christ-Schmidt H, Dorenbaum A; Sapropterin Research Group. Efficacy of sapropterin dihydrochloride

(tetrahydrobiopterin, 6R-BH4) for reduction of phenylalanine concentration in patients with phenylketonuria: a phase III randomised placebo-controlled study. Lancet. 2007 Aug 11;370(9586):504-10. C

46. van der Knaap M.S., Linnankivi T, Paetau A., **Feigenbaum A**., Wakusawa K., Haginoya K., Kohler W., Henneke M., Dinopoulos A., Grattan-Smith P., Brockmann K., Schiffmann R., Blaser S., Hypomyelination with atrophy of the basal ganglia and cerebellum. Follow-up and pathology. Neurology 69 July 10, 2007 166-171. C
47. Arnold GL, Koeberl DD, Matern D, Barshop B, Braverman N, Burton B, Cederbaum S, **Fiegenbaum A**(misspelled on publication), Garganta C, Gibson J, Goodman SI, Harding C, Kahler S, Kronn D, Longo N. A Delphi-based consensus clinical practice protocol for the diagnosis and management of 3-methylcrotonyl CoA carboxylase deficiency. Mol Genet Metab. 2008 Apr;93(4):363-70. Epub 2007 Dec 21. C
48. Kroos M, Pomponio RJ, van Vliet L, Palmer RE, Phipps M, Van der Helm R, Halley D, Reuser A; **GAA Database Consortium (includes Feigenbaum A)** .Update of the Pompe disease mutation database with 107 sequence variants and a format for severity rating.Hum Mutat. 2008 Jun;29(6):E13-26. C
49. *Christina Gerth; Chantal F Morel, **Annette Feigenbaum**, Alex V. Levin. Ocular Phenotype in patients with methymalonic aciduria and homocystinuria, cobalamin C type. J AAPOS. 2008 Dec;12(6):591-6. Epub 2008 Oct 10. CPA
50. Dimmock DP, Dunn JK, **Feigenbaum A,** Rupar A, Horvath R, Freisinger P, Mousson de Camaret B, Wong LJ, Scaglia F Abnormal neurological features predict poor survival and should preclude liver transplantation in patients with deoxyguanosine kinase deficiency. Liver Transpl. 2008 Oct;14(10):1480-5. C
51. David P Dimmock, Pamela Trapane, **Annette Feigenbaum**, Catherine E. Keegan, Stephen Cederbaum, James Gibson, Michael J. Gambello, Keith Vaux, Patricia Ward, Gregory Rice, Jon Wolff, William E. O'Brien & Ping Fang. The Role of Molecular Testing and Enzyme Analysis in the Management of Hypomorphic Citrullinemia. On line October 2008 AJMG 2008 DOI: 10.1002/ajmg.a.32527. Am J Med Genet A. 2008 Nov 15;146A(22):2885-90. C
52. *Andrea Guerin[1], **Annette Feigenbaum**, Elizabeth Donner, Grace Yoon. Stepwise developmental regression associated with novel CACNA1A mutation. Pediatr Neurol. 2008 Nov;39(5):363-4. CPA
53. Lee P, Treacy EP, Crombez E, Wasserstein M, Waber L, Wolff J, Wendel U, Dorenbaum A, Bebchuk J, Christ-Schmidt H, Seashore M, Giovannini M, Burton BK, Morris AA; **Sapropterin Research Group (includes Feigenbaum A).** Safety and efficacy of 22 weeks of treatment with sapropterin dihydrochloride in patients with phenylketonuria. Am J Med Genet A. 2008 Nov 15;146A(22):2851-9. C
54. Feillet F, Clarke L, Meli C, Lipson M, Morris AA, Harmatz P, Mould DR, Green B, Dorenbaum A, Giovannini M, Foehr E; **Sapropterin Research Group (includes Feigenbaum A).** Pharmacokinetics of sapropterin in patients with phenylketonuria.Clin Pharmacokinet. 2008;47(12):817-25. C
55. Arnold GL, Vanhove J, Freedenberg D, Strauss A, Longo N, Burton B, Garganta C, Ficicioglu C, Cederbaum S, Harding C, Boles RG, Matern D, Chakraborty P, **Feigenbaum A.** A Delphi Clinical Practice Protocol for the Management of Very Long Chain Acyl-CoA Dehydrogenase Deficiency. Mol Genet Metab. 2009 Mar;96(3):85-90. C
56. Cameron JM, Maj M, Levandovskiy V, Barnett CP, Blaser S, Mackay N, Raiman J, **Feigenbaum A**, Schulze A, Robinson BH.Pyruvate dehydrogenase phosphatase 1

(PDP1) null mutation produces a lethal infantile phenotype. Hum Genet. 2009 Apr;125(3):319 -26. C

57. *Klaus G.E. Werner, Chantal F. Morel, MD, Adam Kirton, Susanne M. Benseler, John M. Shoffner, Jane B.L. Addis, Brian H. Robinson, Delilah M. Burrowes, Susan I. Blaser, Leon G. Epstein, **Annette S.J. Feigenbaum.** RoME: Rolandic Mitochondrial Encephalomyelopathy and ND3 mutations. Pediatr Neurol. 2009 Jul;41(1):27-33. SRI
58. * Emily W. Y. Tam, **Annette Feigenbaum,** Jane B.L. Addis, Susan Blaser, Nevi MacKay, Robert W. Taylor, Cameron Ackerley, Jessie M. Cameron, Brian H. Robinson. A Novel mitochondrial DNA mutation in COX1 Leads to Strokes, Seizures, and Lactic acidosis. Neuropediatrics 2009; 40:1-7 coSRI
59. Dimmock D., Trapane P, **Feigenbaum A,** Keegan CE, Cederbaum S, Gibson J, Gambello MJ, Vaux K, Ward P, Rice GM, Wolff JA, O'Brien WE, Fang P. Novel human pathological mutations. Gene symbol: ASS1. Disease: Citrullinaemia. Hum Genet. 2009 Aug;126(2):342 C
60. *Koifman, Arie; **Feigenbaum, Annette;** Bi, Weiman; Shaffer, Lisa; Chitayat, David. A homozygous deletion of 8q24.3 including the NIBP gene associated with severe delay, dysgenesis of the corpus callosum and dysmorphic facial features. Am J Med Genet A. 2010 May;152A(5):1268-72. CoSRI
61. Jennifer Seminara, Mendel Tuchman, Lauren Krivitzky, Jeffrey Krischer, Hye-Seung Lee, Cynthia LeMons, Matthias Baumgartner, Stephen Cederbaum, George A. Diaz, **Annette Feigenbaum**, Renata C. Gallagher, Cary O. Harding, Douglas S. Kerr, Brendan Lanpher, Brendan Lee, Uta Lichter-Konecki, Shawn E. McCandless, J. Lawrence Merritt, Mary Lou Oster-Granite, Margretta R. Seashore, Tamar Stricker, Marshall Summar, Susan Waisbren, Marc Yudkoff, Mark L. Batshaw. Establishing a consortium for the study of rare diseases: The Urea Cycle Disorders Consortium. Molecular Genetics and Metabolism 100 (2010) S97–S105. C
62. van der Lei HD, van Berkel CG, van Wieringen WN, Brenner C, **Feigenbaum A,** Mercimek-Mahmutoglu S, Philippart M, Tatli B, Wassmer E, Scheper GC, van der Knaap MS. Genotype - phenotype correlation in vanishing white matter disease. Neurology. 2010 Oct 26;75(17):1555-9.PMID: 20975056 C
63. Kranendijk M, Struys EA, van Schaftingen E, Gibson KM, Kanhai WA, van der Knaap MS, Amiel J, Buist NR, Das AM, de Klerk JB, **Feigenbaum AS,** Grange DK, Hofstede FC, Holme E, Kirk EP, Korman SH, Morava E, Morris A, Smeitink J, Sukhai RN, Vallance H, Jakobs C, Salomons GS. IDH2 Mutations in Patients with D-2-hydroxyglutaric aciduria. Science. 2010 Oct 15;330(6002):336. Epub 2010 Sep 16.PMID: 20847235 C
64. *Connolly BS, **Feigenbaum AS,** Robinson BH, Dipchand AI, Simon DK, Tarnopolsky MA. MELAS syndrome, cardiomyopathy, rhabdomyolysis, and autism associated with the A3260G mitochondrial DNA mutation. Biochem Biophys Res Commun. 2010 Nov 12;402(2):443-7. Epub 2010 Oct 20.PMID: 20965148. CoSRI
65. Uta Lichter Konecki; George A Diaz; John L Merritt II; **Annette Feigenbaum**; C Jomphe; J F Marier; M Beliveau; J Mauney; K Dickinson; Antonia Martinez; Masoud Mokhtarani; Bruce Scharschmidt; William Rhead. Ammonia Control in Children with Urea Cycle Disorders (UCDs): Phase 2 Comparison of Sodium Phenylbutyrate and Glycerol Phenylbutyrate. Mol Genet Metab. 2011 Aug;103(4):323-9. Epub 2011 May 5. C

66. * Shailly Jain Ghai; **Annette Feigenbaum**; Sandesh C Sreenath Nagamani; Susan Blaser; Komudi Siriwardena. Arginase I deficiency: Severe Infantile Presentation with hyperammonemia: more common than reported? Mol Genet Metab. 2011 Sep-Oct;104(1-2):107-11. Epub 2011 Jul 13. Erratum in: Mol Genet Metab. 2012 Jan;105(1):159. SRI
67. Urinary phenylacetylglutamine as dosing biomarker for patients with urea cycle disorders. Mokhtarani M, Diaz GA, Rhead W, Lichter-Konecki U, Bartley J, **Feigenbaum A,** Longo N, Berquist W, Berry SA, Gallagher R, Bartholomew D, Harding CO, Korson MS, McCandless SE, Smith W, Vockley J, Bart S, Kronn D, Zori R, Cederbaum S, Dorrani N, Merritt JL 2nd, Sreenath-Nagamani S, Summar M, Lemons C, Dickinson K, Coakley DF, Moors TL, Lee B, Scharschmidt BF. Mol Genet Metab. 2012 Aug 18. [Epub ahead of print]. Mol Genet Metab. 2012 Nov;107(3):308-14.
68. Ammonia Control And Neurocognitive Outcome Among Urea Cycle Disorder Patients Treated With Glycerol Phenylbutyrate. George A Diaz; Lauren S. Krivitsky; Masoud Mokhtarani; William Rhead; James Bartley; **Annette Feigenbaum;** Nicola Longo; William Berquist; Susan A Berry; Renata Gallagher; Uta Lichter-Konecki; Dennis Bartholemew; Cary O Harding; Stephen Cederbaum; Shawn E McCandless; Wendy Smith; Gerard Vockley; Stephan Bart; Mark S Korson; David Kronn; Roberto Zori; John L Merritt; Sandesh Sreenath-Nagamani; Joseph Mauney, Cynthia LeMons, Klara Dickinson; Tristen L Moors; Dion F Coakley; Bruce F Scharschmidt; Brendan Lee. HEP-12-1233.R1 Hepatology 2012 Sep 7. C
69. Design and Implementation of the First Randomized Controlled Trial of Coenzyme Q10 in Children with Genetic Mitochondrial Diseases.Peter W. Stacpoole; Ton. J. deGrauw; **Annette S. Feigenbaum**, Charles Hoppel, Douglas S. Kerr, Shawn E. McCandless; Michael V. Miles, Brian H. Robinson, and Peter H. Tang. Mitochondrion 12 (2012) 623–629. coPI.
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74. U Lichter-Konecki, GA Diaz, JL Merritt II, **A Feigenbaum**, C Jomphe, JF Marier, M Beliveau, J Mauney, K Dickinson, A Martinez, M Mokhtarani, B Scharschmidt, W Rhead. Ammonia Control in Children with Urea Cycle Disorders (UCDs): Phase 2 Comparison of Sodium Phenylbutyrate and Glycerol Phenylbutyrate **Col. A216.** Presented (oral) at the 60th Annual Meeting of The American Society of Human Genetics, November, 2010, Washington, DC.
75. ***Shailly Jain Ghai, Sandesh CS Nagamani, Brendan Lee, Susan Blaser, **Annette Feigenbaum**. Severe Infantile Presentation of Arginase I Deficiency. Poster SIMD Monterey USA February 2011. Abstract 51 MGM 102(3) page 291.
76. N Longo, **A Feigenbaum**, D Dimmock, S Stockler, K Workman, S Waisbren. Interim Report of Study PKU015: A Phase 3B study of Sapropterin Dihydrochloride (KUVAN®) in young children with PKU. ACMG March 2011.
77. Stacy Hewson, Elizabeth Kerr, **Annette Feigenbaum.** Long term follow-up of two siblings with Cobalamin E (MTRR) deficiency, one treated prenatally. SIMD Monterey USA February 2011. Abstract 48 MGM 102(3) page 290
78. GA Diaz, J Bartley, N Longo, W Berquist, **A Feigenbaum**, R Gallagher[6], W Rhead[7], D Bartholomew[8], CO Harding[9], MS Korson [10], U Lichter-Konecki[11], SA Berry[12], W Smith[13], SE McCandless[14], J Vockley[15], S Bart[16], D Kronn[17], R Zori[18], Sandesh Sreenath-Nagamani[22], M Summar[11], C Jomphe[19], M Beliveau[19], J Mauney[20], K Dickinson[21], M Mokhtarani[21], D Coakley[21], BF Scharschmidt[21], B Lee[22]. Phase 3 blinded, randomized, crossover comparison of sodium phenylbutyrate (NaPBA) and glycerol phenylbutyrate (GPB): Ammonia (NH3) control in adults with urea cycle disorders (UCDS). SIMD Monterey USA February 2011. Abstract 25 MGM 102(3) page 276

79. *Al- Hertani W, Siriwardena K, Cordeiro D, Blaser S, **Feigenbaum A**, Jakobs C. Clinical improvement after treatment of two siblings with L-2 hydroxyglutaric aciduria using riboflavin. Garrod society meeting Calgary, Canada. Poster June 2011.

80. *Almundher Al-Maawali, Grace Yoon,William Halliday, JTR Clarke, **Annette Feigenbaum**, Brenda Banwell, David Chitayat, Susan Blaser. Hypertrophy of the clava, a new MRI sign in patients with *PLA2G6* mutation. Poster ASHG Montreal 2011

81. ***Al-Hertani W, Blaser S, **Feigenbaum** A, Siriwardena K. Spongiform Leukonecephalopathy caused by NDUFV1 mutations. Poster SSIEM Geneva 2011

82. Prasad S, Burton B, **Feigenbaum A,** Grant M, Hendren R, Mardach R, Phillips J, Sanchez-Valle A, Singh R, Siriwardena K, Thomas J, Stahl S, Lang W, Kim S, Jurecki E. Baseline findings in the first 60 subjects in PKU ASCEND (016): A double-blind, placebo-controlled, randomized study to evaluate the safety and therapeutic effects of sapropterin dihydrochloride on neuropsychiatric symptoms in subjects with phenylketonuria (PKU). Poster ACMG 2012 Abstract# v89 and SIMD poster #92

83**.** Longo N, Burton B, Dimmock D, **Feigenbaum A ,** Potter M, Stockler S, Siriwardena K, Lang W, Kim S, Jurecki E, Prasad S**.** Safety and Efficacy of Sapropterin in Children aged 0 to 6 years with Phenylketonuria. Preliminary Findings from a Long Term Open Label Study. ACMG 2012 Abstract # 85

84. GA Diaz, L Krivitzky, M Mokhtarani W Rhead, J Bartley, **A Feigenbaum,** N Longo, W Berquist, SA Berry, R Gallagher, U Lichter-Konecki, D Bartholomew, CO Harding, S Cederbaum, SE McCandless, W Smith, J Vockley, S Bart, MS Korson, D Kronn, R Zori, JL Merritt, Sandesh Sreenath-Nagamani, J Mauney, K Dickinson, T Moors, D Coakley, BF Scharschmidt, B Lee. Ammonia (Nh3) Control And Improved Neurocognitive Outcome Among Urea Cycle Disorder (Ucd) Patients Treated With Glycerol Phenylbutyrate (Gpb)'. SIMD 2012 poster #24

85 GA Diaz, L Krivitzky, M Mokhtarani, W Rhead, J Bartley, **A Feigenbaum,** N Longo, W Berquist, SA Berry, R Gallagher, U Lichter-Konecki, D Bartholomew, CO Harding, S Cederbaum, SE McCandless, W Smith, J Vockley, S Bart, MS Korson, D Kronn, R Zori, JL Merritt, Sandesh Sreenath-Nagamani, J Mauney, K Dickinson, T Moors, D Coakley, BF Scharschmidt, B Lee. Ammonia (Nh_3) Control And Improved Neurocognitive Outcome Among Urea Cycle Disorder (Ucd) Patients Treated With Glycerol Phenylbutyrate (GPB). ACMG 2012 poster #301

86. M Mokhtarani, GA Diaz, W Rhead, J Bartley, **A Feigenbaum,** N Longo, W Berquist, SA Berry, R Gallagher, U Lichter-Konecki, D Bartholomew, CO Harding, S Cederbaum, SE McCandless, W Smith, J Vockley, S Bart, MS Korson, D Kronn, R Zori, JL Merritt, Sandesh Sreenath-Nagamani, J Mauney, K Dickinson, T Moors, D Coakley, B Lee, BF Scharschmidt. Elevated Phenylacetic Acid (Paa) Levels Appear Linked To Neurological Adverse Events In Healthy Adults But Not In Urea Cycle Disorder (Ucd) Patients. Poster # 79 SIMD 2012

87. JPR Monteleone, M Mokhtarani, GA Diaz, W Rhead, U Lichter-Konecki, J Bartley, **A Feigenbaum,** N Longo, W Berquist, SA Berry, R Gallagher, D Bartholomew, CO Harding, MS Korson, SE McCandless, W Smith, J Vockley, S Bart, D Kronn, R Zori, Sandesh Sreenath-Nagamani, M Summar, K Dickinson, D Coakley, B Lee, BF Scharschmidt, Population Pk Analysis Of Glycerol Phenylbutyrate (Gpb) And Sodium Phenylbutyrate (Napba) In Adult And Pediatric Patients With Urea Cycle Disorders (Ucd). Poster # 80 SIMD 2012

88. M Mokhtarani, GA Diaz, W Rhead, U Lichter-Konecki, J Bartley, **A Feigenbaum,** N Longo, W Berquist, SA Berry, R Gallagher, D Bartholomew, CO Harding, MS Korson, SE McCandless, W Smith, J Vockley, S Bart, D Kronn, R Zori, Sandesh Sreenath-Nagamani, M Summar, K Dickinson, D Coakley, B Lee, BF Scharschmidt. Urinary Phenylacetylglutamine Appears To Be A More Useful Marker Than Metabolite Blood Levels For Therapeutic Monitoring Of Phenylacetic Acid (Paa) Prodrugs Simd 2012 Poster 78
89. Burton B, Longo, Dimmock D, **Feigenbaum A,** Potter M, Stockler S, Siriwardena K, Lang W, Kim S, Jurecki E, Prasad S. Safety and Effectiveness of Sapropterin in Children aged 0 to 6 years with Phenylketonuria. Early Findings from a Seven Year Study. National PKU Alliance meeting July 2012
90. F. Rutsch, M. MacDougall, C. Lu, Y. Nitschke, I. Buers, O. Mamaeva, D.K. Crossman, J. Dong, C. Müller, H.G. Kehl, J. Kleinheinz, P. Barth,K. Barczyk, D. Bazin, J. Altmüller, H. Thiele, P. Nürnberg, W. Höhne, **A.S. Feigenbaum**, R. Hennekam: Exome Sequencing Identifies an IFIH1 Mutation Causing Singleton-Merten Syndrome by Dysregulation of the Innate Immune Response. ASHG Meeting San Francisco November 2012 Poster # 2759T.
91. Nicola Longo, Komudi Siriwardena, **Annette Feigenbaum**, David Dimmock, Barbara Burton, Sylvia Stockler, Susan Waisbren, William Lang, Elaina Jurecki, Suyash Prasad. Long-Term Developmental Progression in Young Children Taking Sapropterin for Phenylketonuria: A Two-Year Analysis of Safety and Efficacy. ACMG annual Meeting March 2013, Phoenix USA poster coI. Abstract Number: 169.
92. Pranesh Chakraborty, Jonathan Kronick, Beth Potter, Hilary Vallance, Maria Karaceper, Alicia Chan, Doug Coyle, Sarah Dyack, **Annette Feigenbaum**, Michael Geraghty, Monica Hernandez. A Framework for Developing Case Definitions and Clinical Measures to Support Longitudinal Research on Outcomes for Inborn Errors of Metabolism. ACMG Annual Meeting March 2013, Phoenix USA poster coI. Abstract Number: 56
93. Beth Potter, Pranesh Chakraborty, Doug Coyle, Sarah Dyack, **Annette** F**eigenbaum**, Monica Hernandez, Maria Karaceper, Sara Khangura, Jonathan Kronick, Jennifer MacKenzie, John Mitchell, Chitra Prasad, Sylvia Stockler, Yannis Trakadis, Brenda Wilson, Kumanan Wilson. The Canadian Inherited Metabolic Diseases Research Network: Development of a Pan-Canadian Practice-Based Research Network for Inborn Errors of Metabolism ACMG annual Meeting March 2013, Phoenix USA poster coi. Abstract Number: 188
94. Maria Karaceper, Robin Casey, Meranda Nakhla, Marni Brownell, Pranesh Chakraborty, Doug Coyle, Linda Dodds, Cheryl Greenberg, Astrid Guttmann, Jonathan Kronick, Anne-Marie Laberge, Kumanan Wilson, Beth Potter, **Annette Feigenbaum**, Aizeddin Mhanni, Fiona Miller, Chitra Prasad, Hilary Vallance, Brenda Wilson. The Epidemiology and Health System Impact of Medium-Chain acyl-coa Dehydrogenase Deficiency Among Affected Children and Those with False Positive Newborn Screening Results in Ontario, Canada ACMG annual Meeting March 2013, Phoenix USA poster coi. Abstract Number: 193.
95. Optimizing ammonia (NH_3) control in urea cycle disorder (UCD) patients: short and long-term implications. B Lee, M Mokhtarani, GA Diaz, W Rhead, U Lichter-Konecki, **A Feigenbaum**, SA Berry, J Bartley, N Longo, W Berquist, W Smith, R Gallagher, CO Harding, S McCandless A Schulze, S Nagamani, C Lemons, K Dickinson, DF Coakley, TL Moors, D Millikien, M Marino, BF Scharschmidt. Garrod Association , Sherbrooke Canada May 2013. Poster

96. Ammonia (NH_3) amino acid and hyperammonemic crises (HACS) in pediatric and adult patients with urea cycle disorders (UCDS) during dosing with sodium phenylbutyrate (NaPBA) vs. Glycerol phenylbutyrate (GPB). George A. Diaz, Masoud Mokhtarani, William Rhead, James Bartley, **Annette Feigenbaum,** Nicola Longo, William Berquist, Susan A. Berry, Renata Gallagher, Uta Lichter-Konecki, Cary O. Harding, Shawn E. Mccandless, Wendy Smith, Andrea Schulze, Sandesh Sreenath-Nagamani, Joseph Mauney, Cynthia lemons, Klara Dickinson, Tristen L. Moors, Eun Lee, Dion F. Coakley, Bruce F. Scharschmidt, Brendan Lee· Garrod Association , Sherbrooke Canada May 2013. Poster
97. Ammonia (NH_3) amino acid and hyperammonemic crises (HACS) in pediatric and adult patients with urea cycle disorders (UCDS) during dosing with sodium phenylbutyrate (NaPBA) vs. Glycerol phenylbutyrate (GPB). George A. Diaz, Masoud Mokhtarani, William Rhead, James Bartley, **Annette Feigenbaum,** Nicola Longo, William Berquist, Susan A. Berry, Renata Gallagher, Uta Lichter-Konecki, Cary O. Harding, Shawn E. Mccandless, Wendy Smith, Andrea Schulze, Sandesh Sreenath-Nagamani, Joseph Mauney, Cynthia lemons, Klara Dickinson, Tristen L. Moors, Eun Lee, Dion F. Coakley, Bruce F. Scharschmidt, Brendan Lee· SERGG: Southeastern Regional Genetics Group™, Inc Annual meeting Nashville, North Carolina, July, 2013 Platform presented by M Mokhtarani.
98. P-026 ICIEM September 2013 Barcelona Spain Neuropsychiatric outcomes in PKU patients treated with sapropterin:results from the randomized, controlled PKU ascend (PKU 016) trial. Prasad S, Burton B , **Feigenbaum A**, GrantM, HendrenR, SinghR, Stahl S, Zhang C: J Inherit Metab Dis (2013) 36 (Suppl 2):S91–S342
99. P-089 ICIEM September 2013 Barcelona Spain. Optimizing ammonia (NH3) control in Urea Cycle Disorder (UCD) patients: short and long-term implications. Lee B, Mokhtarani M, Diaz GA, Rhead W, Lichter-Konecki U,**Feigenbaum A**, Berry SA, Bartley J, Longo N, BerquistW, Smith W, Gallagher R, Harding CO, McCandless S, Schulze A,Nagamani S, Le MonsC, Dickinson K, Coakley DF, Moors TL ,Millikien D, Marino M, Scharschmidt BF: J Inherit Metab Dis (2013) 36 (Suppl 2):S91–S342
100. P-248 ICIEM September 2013 Barcelona Spain. Cervical spondylosis and myelopathy: a complication of glutaric aciduria type 1. Cordeiro D, Blaser S, Clarke J, Drake J, **Feigenbaum A,** Raiman J,Siriwardena K, Mecija M. 1 J Inherit Metab Dis (2013) 36 (Suppl 2):S91–S342
101. P-382 ICIEM September 2013 Barcelona Spain Exome sequencing identifies complex I NDUFV2 mutations as a novel cause of Leigh syndrome. **Feigenbaum A,** Cameron JM, MacKay N, Tarnopolsky M, Blaser S,Robinson BH, Schulze A: J Inherit Metab Dis (2013) 36 (Suppl 2):S91–S342
102. P-389 ICIEM September 2013 Barcelona Spain Outcome for DGUOK associated liver failure 16.5 years postorthotopic Transplantation. **Feigenbaum A**, Raiman J, Ling S, NgV, RobinsonB, Siriwardena K. J Inherit Metab Dis (2013) 36 (Suppl 2):S91–S342
103. P-420 ICIEM September 2013 Barcelona Spain Retrospective review of all GSD type VI and IX patients at the hospital for sick children. Roscher A, Hewson S, **Feigenbaum A,** Kronick J, Raiman J,Schulze A, Siriwardena K, Mercimek-Mahmutoglu S: J Inherit Metab Dis (2013) 36 (Suppl 2):S91–S342
104. P-538 ICIEM September 2013 Barcelona Spain Two new patients with pyridoxine dependent epiliepsy caused by ALDH7A1 genetic defect: long-term follow-up and normal neurodevelopmental outcome. Cordeiro D, Mamak E, Donner E,

Feigenbaum A, Siriwardena K,Mercimek-Mahmutoglu S J Inherit Metab Dis (2013) 36 (Suppl 2):S91–S342

105. P-855 ICIEM September 2013 Barcelona Spain. The Canadian Inherited Metabolic Diseases Research Network (CIMDRN): a national, practice-based research network for inborn errors of metabolism. Kronick JB, Chakraborty P, Coyle D, Wilson K, Dyack S ,**Feigenbaum A,** Hernandez M, Khan A, Khangura SD, Mitchell JJ, Potter M, Prasad C, Siriwardena K, Sparkes R, Speechley KN, Potter BK, CIMDRN, Ottawa, Canada: J Inherit Metab Dis (2013) 36 (Suppl 2):S91–S342
106. P-866 ICIEM September 2013 Barcelona Spain. Prevalence of inherited metabolic disorders in epilepsy patients: a large, single center study. Imhof E, Zak M, Hewson S, **Feigenbaum A,** Kobayashi J, Minassian B, Raiman J, Siriwardena K1, Tein I, Clarke J, Mercimek-Mahmutoglu S: J Inherit Metab Dis (2013) 36 (Suppl 2):S91–S342
107. Poster #98 SIMD March 2014, Monterey, California. Treatment-Related Signs and Symptoms among Patients with Urea Cycle Disorders (UCDs) during Treatment with Sodium Phenylbutrate (NaPBA) and Glycerol Phenylbutyrate (GPB) SC Nagamani, GA Diaz, W Rhead, SA Berry, C Le Mons, U Lichter-Konecki, J Bartley, A Feigenbaum, N Longo, W Berquist, , R Gallagher, D Bartholomew, CO Harding, MS Korson, SE McCandless, W Smith, J Vockley, D Kronn, R Zori, D Coakley, BF Scharschmidt, M Mokhtarani, B Lee. MGM 111(3):287.
108. Poster 2298M 64th Annual Meeting of The American Society of Human Genetics, October, 2014 San Diego.Predictive Value of Blood Ammonia and Glutamine to HyperammonemicCrises in Patients with Urea Cycle Disorders. B. Lee, G.A. Diaz, W. Rhead, U. Lichter-Konecki, A. Feigenbaum, S.A. Berry, C. Le Mons, J. Bartley, N. Longo, S.C. Nagamani, W. Berquist10, R.C. Gallagher, D. Bartholomew, C.O. Harding, M.S. Korson, S.E McCandless, W. Smith, S. Cederbaum, D. Wong, J.L. Merritt II, A. Schulze, J.Vockley, D. Kronn, R. Zori, M. Summar, D.A. Milikien, M. Marino, D.F. Coakley, M. Mokhtarani, B.F Scharschmidt. AJHG

PRESENTATIONS

Invited Visits to Other Hospitals/Universities/Etc.

1. Miami Children's Research Institute. 5th Symposium, Miami, FL, Dec 1994. Neurological, neurophysiological and MRI findings in adult PKU.
2. Society for the Study of Inborn Errors of Metabolism, Vienna, Austria, May 1997. Local Experience in the use of Dichloracetate.
3. Hamilton University and MacMaster Medical Centre. Department of Genetics, Hamilton, Ontario, 1998. Mitochondrial disease.
4. Markham Stouffville Hospital network CME. Department of Paediatrics, Markham, Ontario, Mar 2000. Genetic metabolic diseases in presenting infancy.
5. Mitochondria 2001: Combine Meeting of Mitochondrial Medicine Society and The United Mitochondrial Disease Foundation, San Diego, California, Mar 2001. Long-term managment of mitochondrial disease - putting your medical team together.
6. National Council of Jewish Women of Canada, Toronto, Ontario, Oct 2001. Canavan and Tay Sachs population screening and clinics.
7. Garrod Society Meeting, Monterey, California, USA, Mar 2002. Talk: Jewish genetic screening in Canada.

8. National Council of Jewish Women of Canada and Montreal Children's Hospital, Montreal, Quebec - Montreal Children's Hospital, May 2002. Jewish genetic diseases - population carrier screening.
9. Chinese TV, Toronto, Ontario, 2002. Neurological and mitochondrial disease.
10. Barth Syndrome Society, Baltimore, USA, Oct 2002. Clinical management of your child with Barth syndrome.
11. United Mitochondrial Diseases Foundation Meeting - Invited Lecture, San Diego, USA, Jun 2003. Diagnostic dilemmas in mitochondrial disease.
12. Health on the line; City TV One, Toronto, Ontario, 2003. Neurodegenerative diseases.
13. Toronto, Ontario, Mar 2004. Kolel Education Workshop: Jewish genetic diseases Kolel Centre.
14. Weizmann Institute of Canada Public Symposium, Toronto, Ontario - Ontario Science Centre, Apr 2004. Screening for Jewish genetic diseases.
15. Medicine Grand Rounds, Ajax Pickering Hospital, May 2004. Mitochondrial disease.
16. Barth Syndrome Foundation, Orlando, USA, Jul 2004. Ask the doctor.
17. Barth Syndrome Foundation, Orlando, USA, Jul 2004. Clinical standards panel.
18. Toronto Board of Jewish Education, Toronto, Ontario, Sep 2004. Ashkenazi Jewish genetic screening.
19. Global TV News, Toronto, Ontario, Oct 2004. Ashkenazi Jewish disease.
20. Latin America Society of Inborn Errors of Metabolism and Neonatal Screening (SLEIMPN), Costa Rico, Nov 2005. Inborn errors of metabolism and the central nervous system.
21. Weizmann Institute of Canada Public Symposium, Montreal October 2006. Screening for Jewish and French Canadian genetic diseases.
22. Mitochondrial diseases in Endocrinology. Endocrinology Grand Rounds. May 2007 Womens college hospital Host : Dr C Kelly.
23. SIMD/ACMG: Newborn screening dilemmas: Tyrosinemia March 2007, Nashville USA.
24. Magen David Adom Canada: Ashkenazi Jewish Genetic Diseases: the power of prevention. Toronto May 16 2007.
25. Treatment of Genetic Diseases. University of California San Diego. April 2008. Host: Dr Robert Naviaux
26. Everything you wanted to know about Metabolic disease. City wide Pediatric rounds Royal Victoria Hospital Barrie Ontario October 16 2008. Host: Dr Chee Chen
27. Autism and metabolic disease. Bloorview Childrens Autism research meeting 2008.
28. Canadian PKU Physician advisory meeting (Biomarin) Montreal November 2009.
29. Canadian PKU Physician advisory meeting (Biomarin) Toronto December 2-3, 2010: Invited lecture: PKU Outcomes and therapy.
30. Invited speaker ACMG 2012 Annual meeting North Carolina March 31 2012: Prenatal and maternal issues in mitochondrial disease
31. Safety and Efficacy of Sapropterin in Children aged 0 to 6 years with Phenylketonuria: Study Design and Preliminary Findings from a Long Term Open Label Study PKU 015. To Biomarin and Investigators Charlotte North Carolina March 31 2012.
32. Maternal, fetal and neonatal presentations of Mitochondrial Disease. Childrens Hospital of Orange County. Pedatrics Grand Rounds. September 12, 2012.
33. A Pot Pourri of Metabolic and Mitochondrial Diseases: Lessons learned from real cases. Neurology, Genetics, Neuroradiology Rounds Invited Speaker Johannesburg

General Hospital Gauteng South Africa October 16, 2012.

34. Safety and Efficacy of Sapropterin in Children aged 0 to 6 years with Phenylketonuria: Study Design and Preliminary Findings from a Long Term Open Label Study PKU 015. 2013 North America Commercial Team Meeting, Biomarin Pharmaceuticals Inc., Marina del rey January 2013.
35. Nutritional Management of Mitochondrial diseases. Abbott Nutrition RD Training June 2013, Nashville USA.
35. Update on PKU. Network PKU. Boston USA. October 2013.
36. How High Phe affects the Brain. Medaccess/Biomarin February 2014 San Diego.
37. Update on PKU research Atlanta USA Network PKU March 2014
38. Challenges in PKU. ACMG Annual meeting. Satellite meeting March 28 2014, Nashville USA.
39. PKU Update. Seattle WA. Network PKU. May 2015.

Peer Reviewed Papers Read at Scientific Meetings (First author presenting)

1. Kalk WJ, **Feigenbaum A** (C): Glomerular hyperfiltration, albuminuria and the duration of Type I (insulin-dependent) diabetes. European Association for the Study of Diabetes, Leipzig, East Germany, Sep 1987.
2. Triggs-Raine B, **Feigenbaum A** (CPA), Gravel R, Mahuran D: Development of DNA based diagnostics for Jewish Tay-Sachs Disease. International Congress of Clinical Enzymology, Toronto, Ontario, Jun 1989.
3. Holland J, Perlman K, Walker D, Teshima I, **Feigenbaum A** (CA ; C), Weksberg R, McInnes R: Short Arm Chromosome 20 Deletion and Growth Hormone Deficiency. 3rd Joint Meeting of the ESPE/LWPES, Jerusalem, Israel, Oct 1989.
4. Triggs-Raine BL, **Feigenbaum A** (CPA), Skomorowski MA, Natowicz M, Kolodny E, Clarke JTR, Mahuran D, Gravel RA: Ashkenazi Tay-Sachs Disease: Comparison of DNA and enzyme-based methods for carrier testing. American Society of Human Genetics, Baltimore, MD, Nov 1989.
5. Blaser SI, Smith AM, **Feigenbaum ASJ** (CPA), Clarke JTR, Becker LE, Roessman U, Carollo C, Hochhauser L, Chuang SH: Magnetic Resonance Imaging of dilated Virchow-Robin spaces in the mucopolysaccharidoses. XIV Symposium Neuroradiologicum, London, UK, Jun 1, 1990.
6. Blaser SI, Smith AM, **Feigenbaum ASJ** (CPA), Clarke JTR, Becker LE, Roessman U, Carollo C, Hochhauser L, Chuang SH: Magnetic Resonance Imaging of dilated Virchow-Robin spaces in the mucopolysaccharidoses. RSNA, Chicago, IL, Nov 1990.
7. Blaser SI, Smith AM, **Feigenbaum ASJ** (CPA), Clarke JTR, Becker LE, Roessman U, Carollo C, Hochhauser L, Chuang SH: Magnetic Resonance Imaging of dilated Virchow-Robin spaces in the mucopolysaccharidoses. International Paediatric Radiology Society, 2nd Meeting, Stockholm, Sweden, May 1991.
8. Blaser SI, Feigenbaum ASJ (CPA), Becker LE, Whyte HJ, Harwood-Nash DC: Lethal neonatal mitochondrial disorder, radiographic mimic of perinatal asphyxia. American Society of NeuroRadiology. 31st Annual Meeting, Vancouver, BC, 1993.
9. Tsai CH, Costa MT, Chen WC, **Feigenbaum A** (C), Teshima I: Terminal chromosome 11q deletion (Jacobsen syndrome) - report of three cases. Chinese Taipei Pediatric Association. 136th Scientific Meeting, Acta Pediatrica Sinica, 1993.
10. **Feigenbaum A** (CPA), Toone J, Kooh S, Vallance H, Jabbar M, Applegarth D: Mitochondrial DNA deletion/duplication syndrome: From neonatal sideroblastic

anaemia to Kearns-Sayre at age 11 years. Garrod Association, Edmonton, AB, Apr 21, 1994.

11. **Feigenbaum A** (PA), Ben-Zeev B, Blaser S, Taylor M, Schoonheyt W, Hanley W. (R): Neurological, Neurophysiological and MRI findings in Adult PKU (Platform Presentation). Miami Childrens Research Institute, 5th Symposium, Miami, Fl., Dec 1994.
12. Hanley WB, Demshar H, Preston MA, Borczyk A, Schoonheyt W, Clarke JTR, **Feigenbaum A** (C): Newborn PKU screening and early hospital discharge. 11th Annual Neonatal Screening Symposium, Corpus Christie, TX, Sep 1995.
13. Hanley WB, Demshar H, Preston MA, Borczyk A, Schoonheyt W, Clarke JTR, **Feigenbaum A** (C): Newborn PKU screening and early hospital discharge. Annual Meeting Irish and American Pediatric Society, Vermont, Sep 1995.
14. **Feigenbaum ASJ** (PA): Local experience in the use of Dichloroacetate. Society for the Study of Inborn Errors and Metabolism, Vienna, Austria, May 1997.
15. Laughlin S, Blaser SI, Becker LE, **Feigenbaum A** (CPA): Menkes disease simulating non-accidental injury. European Society of Pediatric Radiology, Lugano, Switzerland, May 1997.
16. Herman G, **Feigenbaum A (CPA),** Zhao W, Finegold M, deGouyon B, Laporte J, Mandel JL, Metzenberg A: Medical complications in long-term survivors with X-linked myotubular myopathy (MTM1) (Platform Presentation). 47th Annual ASHG Meeting, Baltimore, MD, Oct 1997.
17. **Feigenbaum A (PA),** Heon J, Wherrett J, Bilbao J, Stone ME: Charcot Marie Tooth disease and Stargardt disease in a consanguinous family. World Federation of Neurogenetics Research Group Meeting, Toronto, Ontario, Apr 1999.
18. Farnzahen-Far A, Patel M, **Feigenbaum A (C),** Weissberg PL, Proudfoot D: The Osteopoprotogerin and Matrix gla protein genes in human vascular calcification: Several polymorphisms but no pathogenic mutation in a family with Singleton Merton Syndrome. American Heart Association Meeting, Jul 1999.
19. **Feigenbaum A (E):** Canavan and tay sachs population screening and clinics. National Council of Jewish Women of Canada, Toronto, Ontario, Oct 2001.
20. H. Levy, A. Milanowski, A. Chakrapani, M. Cleary, F. Trefz, C. Whitley, F. Feillet, **A. Feigenbaum**, J. Bebchuk, H. Christ-Schmidt, A. Dorenbaum. A Phase 3 study of the efficacy of sapropterin dihydrochloride (tetrahydrobiopterin, 6R-BH_4) in reducing phe levels in subjects with phenylketonuria. SSIEM 2006 Japan. **CoI, site director.**
21. H. Levy, A. Milanowski, A. Chakrapani, M. Cleary, F. Trefz, C. Whitley, F. Feillet, **A. Feigenbaum**, J. Bebchuk, H. Christ-Schmidt, A. Dorenbaum. A Phase 3 study of the efficacy of sapropterin dihydrochloride (tetrahydrobiopterin, 6R-BH_4) in reducing phe levels in subjects with phenylketonuria AJHG 2006 Platform presentation by Dr. Levy; New Orleans USA Abstract #57. **CoI, site director**.
22. G. L. Arnold, D. D. Koeberl, B. A. Barshop, B. K. Burton, S. Cederbaum, **A. Feigenbaum**, C. O. Harding, D. Kronn, D. Matern, J. B. Gibson, C. L. Garganta, N. Braverman, N. Longo, S. G. Kahler, the 3-MCC working group Clinical practice protocols for 3-methylcrotonyl CoA carboxylase (3-MCC) deficiency.(273) (09:30AM-09:45AM on Sat) ASHG San Diego October 2007 **Co-I**
23. Hussain, M (CRA**), Feigenbaum A.** Efficacy of Sapropterin in subjects with Phenylketonuria, Garrod Society Annual Meeting May 2008. **PI**
24. **Feigenbaum A,** Jain S. Arginase deficiency-severe early presentation with hyperammonemia. Platform presentation. Garrod Society June 2010. **PI**
25. U Lichter-Konecki, GA Diaz, JL Merritt II, **A Feigenbaum**, C Jomphe, JF Marier, M

Beliveau, J Mauney, K Dickinson, A Martinez, M Mokhtarani, B Scharschmidt, W Rhead. Ammonia Control in Children with Urea Cycle Disorders (UCDs): Phase 2Comparison of Sodium Phenylbutyrate and Glycerol Phenylbutyrate **CoI.** Presented platform by U. Lichter ASHG meeting October 2010. AJHG A.

Poster Presentations

1. **Feigenbaum A,** Kumar V, Weksberg R: The Singleton-Merten Syndrome: Autosomal Dominant Inheritance with Variable Expression. (Poster) American Society of Human Genetics Meetings, New Orleans, LA, Oct 1988.
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99. P-855 ICIEM September 2013 Barcelona Spain. The Canadian Inherited Metabolic Diseases Research Network (CIMDRN): a national, practice-based research network for inborn errors of metabolism. Kronick JB, Chakraborty P, Coyle D, Wilson K, Dyack S ,**Feigenbaum A,** Hernandez M, Khan A, Khangura SD, Mitchell JJ, Potter M, Prasad C, Siriwardena K, Sparkes R, Speechley KN, Potter BK, CIMDRN, Ottawa, Canada: J Inherit Metab Dis (2013) 36 (Suppl 2):S91–S342
100. P-866 ICIEM September 2013 Barcelona Spain. Prevalence of inherited metabolic disorders in epilepsy patients: a large, single center study. Imhof E, Zak M, Hewson S, **Feigenbaum A,** Kobayashi J, Minassian B, Raiman J, Siriwardena K1, Tein I, Clarke J, Mercimek-Mahmutoglu S: J Inherit Metab Dis (2013) 36 (Suppl 2):S91–S342
101. Poster #98 SIMD March 2014, Monterey, California. Treatment-Related Signs and Symptoms among Patients with Urea Cycle Disorders (UCDs) during Treatment with Sodium Phenylbutrate (NaPBA) and Glycerol Phenylbutyrate (GPB) SC Nagamani, GA Diaz, W Rhead, SA Berry, C Le Mons, U Lichter-Konecki, J Bartley, A Feigenbaum, N Longo, W Berquist, , R Gallagher, D Bartholomew, CO Harding, MS Korson, SE McCandless, W Smith, J Vockley, D Kronn, R Zori, D Coakley, BF Scharschmidt, M Mokhtarani, B Lee. MGM 111(3):287.
102. 2014 Joint Garrod and Canadian Newborn & Child Screening Symposium, May 29-31, 2014, Ottawa Ontario Canada. Treatment-Related Signs and Symptoms among Patients with Urea Cycle Disorders (UCDs) during Treatment with Sodium Phenylbutrate (NaPBA) and Glycerol Phenylbutyrate (GPB) SC Nagamani, GA Diaz, W Rhead, SA Berry, C Le Mons, U Lichter-Konecki, J Bartley, **A Feigenbaum,** N Longo, W Berquist, , R Gallagher, D Bartholomew, CO Harding, MS Korson, SE McCandless, W Smith, J Vockley, D Kronn, R Zori, D Coakley, BF Scharschmidt, M Mokhtarani, B Lee.

103. 2014 Joint Garrod and Canadian Newborn & Child Screening Symposium, May 29-31, 2014, Ottawa Ontario Canada. The epidemiology and health services impact of medium-chain acyl-CoA dehydrogenase deficiency among affected children and those with false positive newborn screening results in Ontario. Maria D Karaceper, Marni Brownell, Robin Casey, Pranesh Chakraborty, Doug Coyle, Linda Dodds, **Annette Feigenbaum,** Deshayne Fell, Scott Grosse, Astrid Guttmann, Anne-Marie Laberge, Aizeddin Mhanni, Fiona Miller, Meranda Nakhla, Cheryl Rockman-Greenberg, Rebecca Sparkes, Hilary Vallance, Brenda Wilson, Kumanan Wilson, Beth K Potter, on behalf of the Canadian Inherited Metabolic Diseases Research Network (CIMDRN).

104. 2014 Joint Garrod and Canadian Newborn & Child Screening Symposium, May 29-31, 2014, Ottawa Ontario Canada. A REDCap Database to Support Longitudinal Follow-up of Pediatric Patients with Inborn Errors of Metabolism. Hernandez M, Chakraborty P, Kronick JB, Potter BK 2, Chan AKJ, Coyle D, Dyack S, **Feigenbaum A,** Geraghty M, Karaceper M, Khan A, Little J, MacKenzie J, Maranda B, Mhanni A, Mitchell G, Mitchell JJ, Potter M, Prasad C, Siriwardena K, Stockler S, Tingley K, Trakadis Y, Turner L, Van Karnebeek C, Vallance H, Wilson B, Wilson K, on behalf of the Canadian Inherited Metabolic Diseases Research Network (CIMDRN)

RESEARCH AND INVESTIGATION

Senior Investigator's Name Appears in Bold

Research Funding – Active
Toronto

PKU 015: Biomarin: 2009. A Phase 3b Open-Label Study to Evaluate the Effect of Kuvan on Neurocognitive Function, Maintenance of Blood Phenylalanine Concentrations, Safety, and Population Pharmacokinetics in Young children with Phenylketonuria. REB# 1000013704 **PI at Sickkids A Feigenbaum 2009-2011, 2014-**

Neuropsychological and Quality of Life outcomes in untreated adults with mild hyperphenylalaninemia with phenylalanine levels between 360 and 600 µmol/L. Investigator (Dr. Siriwardena) initiated study sponsored by Biomarin Pharmaceuticals. 2013. REB # 1000039726**. Feigenbaum PI 2014-**

CIHR (Canadian Institutes of Health Research), IG (Institute of Genetics) and IHSPR (Institute of Health Services and Policy Research). 2012-2017. Emerging Team In rare diseases: achieving the 'triple aim' for inborn errors of metabolism. PI Elizabeth Potter PhD; Pranesh Chakraborty, Douglas Coyle, Jonathan Kronick, Kumanan Wilson, L. Dodds, A. Feigenbaum, D. Fell, M. Geraghty, S. Grosse, A. Guttmann, J. Little, A. Mhanni, F. Miller, J. Mitchell, M. Nakhla, K. Siriwardena, K. Speechley, H. Vallance, B. Wilson. Total: $298,000/year. **Coinvestigator.** Rate 4.38.

Urea Cycle Diseases Consortium: Longitudinal study of urea cycle disorders. Feigenbaum A. Site PI 2007-to 2011 then coPI with Dr A Schulze.
Multicentre consortium PI Mendel Tuchman/Mark Batshaw USA. **NIH ORD** funded. 2007/8 $ 7000 start up to A. Feigenbaum for Sickkids Site.
Grant Number: 9U54HD061221
Project Title: Rare Disease Clinical Research Consortia (RDCRC) for the RDCR Network
Project Period 09/30/2003 – 7/31/2014
PI: Mark L. Batshaw, MD
$ 350,000 private philanthropic donation to Sickkids centre.
Refunded 2014-2019.

HPN100-011: Hyperion 2010- . Long-Term Use of HPN-100 in Urea Cycle Disorders**.** Industry initiated**. Site PI A Feigenbaum to 2011; then coI with Dr A Schulze**

Insulin status in Glycogen Storage Disease: PI (Feigenbaum 2009-2011); Dr A Schulze Ochshorn Fund for GSD. **Coinvestigator**. Sickkids Foundation Private donations ($43, 000): 2011- .

Ongoing Unfunded studies at Sickkids:

Are nutrient intakes of children and adolescents with PKU meeting requirements? (Bone Health in PKU) - REB 1000004798. **Austin V,** Kuperberg K, Feigenbaum A, Antle B, Clarke JTR.

Evaluating Unknown Leukodystrophies. REB File No. 1000004802 2005- date PI Dr. **Susan Blaser;** Coinvestigators: Feigenbaum A, vanderknaap M.

Retrospective Survey of Medical Management and Outcomes for Patients with Phenylketonuria (PKU) treated at the Hospital for Sick Children REB# 1000041534. **Feigenbaum A.,** Siriwardena K., Mitchell J.

Essential fatty acid status in young children and women of chld bearing age with Phenylketonuria. **PI Feigenbaum A,** coI, Nagy L, Austin V.

Active Funded studies Rady Childrens Hospital/UCSD San Diego:

BioMarin Pharmaceutical, Inc. 2012- . A Phase 1/2 Open-label Study of the Safety, Tolerability, Pharmacokinetics, Pharmacodynamic and Preliminary Efficacy of BMN 701 (GILT-tagged Recombinant human GAA) in Patients with Late-onset Pompe Disease, Protocol Number: POM-001. **Site PI: B. Barshop**, IRB approval # 110520 approx $330,000 total. **CoInvestigator**

BioMarin Pharmaceutical, Inc. 2012- . A Long-Term Study for Extended BMN 701Treatment of Patients with Pompe Disease who have Completed BMN 701 Studies; POM-002. **Site PI: B. Barshop** IRB approval # 111326. $38,000/pt/6 month cycle; now ~$2,300/visit Q 3 months. **CoInvestigator**

BioMarin Pharmaceutical, Inc. 2012- . PKUDOS—PKU Demographics, Outcomes and Safety Registry. **Site PI: W. Nyhan.** IRB approval # 081356. $250 per quarter for entry made within 30 days of when info was collected. **CoInvestigator**

BioMarin Pharmaceutical, Inc. 2012- . A Phase 3b Open-Label Study to Evaluate the Effect of Kuvan® on Neurocognitive Function, Maintenance of Blood Phenylalanine Concentrations, Safety, and Population Pharmacokinetics in Young Children with Phenylketonuria; Protocol Number: **PKU-015**. **Site PI: W. Nyhan.** IRB approval # 090582. Approx $2,000/visit Q 6 months. **CoInvestigator**

BioMarin Pharmaceutical, Inc. 2014- . A Phase 3, Open-Label, Randomized, Multi-Center Study to Assess the Safety and Tolerability of an Induction, Titration, and Maintenance Dose Regimen of BMN 165 Self-Administered by Adults With Phenylketonuria Not Previously Treated with BMN 165; HRPP# 130921 **(PKU 165-301 PRISM). Site PI: A. Feigenbaum.** Protocol $26,000/pt completing study; $9100 start up. **Site PI.**

BioMarin Pharmaceutical, Inc. 2014- . A Three-Part, Phase 3, Randomized, Double-Blind, Placebo-Controlled, Four-Arm, Discontinuation Study to Evaluate the Efficacy and Safety of Subcutaneous Injections of BMN 165 Self Administered by Adults With Phenylketonuria. 165; HRPP# 140493 **(PKU 165-302 PRISM). Site PI: A. Feigenbaum. Site PI.**

Hyperion /Horizon Pharma: 2014- Observational Study That Will Collect Information on Patients With Urea Cycle Disorders (UCDs) (THRIVE). **Site PI: A. Feigenbaum. Site PI.**

Research Funding - Peer Reviewed-Completed
Neurological outcome in adult PKU. **Feigenbaum A:** *Paediatric Consultants. The Hospital for Sick Children* ($5,000 1995)

Magnetic Resonance Spectroscopy in PKU. **Feigenbaum A:** *Garrod Association of Canada* ($14,500 1998)

Canavan disease screening in Toronto. **Feigenbaum A,** Stockley T, Ray P: *Canadian Foundation of Control for Neurodegenerative Disease* ($104,000 1999 - 2001)

Canavan disease screening in Toronto. **Stockley T, Feigenbaum A:** *Ontario Mental Health Foundation* ($25,000 1999)

More on my own: A pilot transition planning/education project. **Feigenbaum A, Antle B,** Anderson D: *Garrod Association of Canada* ($16,850 2000 - 2001).

ALID-006-01: A multicenter, multinational open-label extension study of the safety and efficacy of recombinant human alpha-L-iduronidase in patients with mucopolysaccharidosis I (MPS I). **Clarke JTR,** Mahuran JJ, Feigenbaum A (co-investigator): *Biomarin/Genzyme* LLC (private company) ($207,578 2001 - 2002) ($291,616 2003-4).

Supporting "Diet for Life": A group program for young people with PKU and their parents. **Antle BJ,** Anderson D, Austin V, Bulthuis W, Kerr E, Feigenbaum A: *HSC Internal Seed Grant Competition* ($204,800 2002 - 2004)

Why is she having so much trouble, she seems so smart?: A PKU retrospective analysis of non-verbal learning disabilities in young people with PKU. **Kerr E,** Antle BJ, Feigenbaum A: *Garrod Association* ($23,800 2002 - 2003)

All you ever wanted to know about PKU: Information outreach program for clients with PKU. **Antle B, Feigenbaum A,** Anderson D, Austin V, Bulthuis W, Kerr E: *Garrod Association of Canada* ($12,200 2003 - 2004)

Molecular basis of Leigh syndrome. **Robinson, B,** Feigenbaum A, Cameron J: *Jacobs Ladder* : *Canadian Foundation of Control for Neurodegenerative Disease* ($125,000 2004 - 2006)

PKU 001: Phenoptin in PKU - Phase II multi-center Open label clinical trial. **Feigenbaum A at HSC site,** Multicentre trial international: Biomarin Pharmaceuticals ($60,000 2005 - 2006)

An open label extension of study TKT 024 - evaluating long term safety and clinical outcomes in MPS II patients receiving iduronate 2 sulfatase enzyme replacement therapy. **Clarke JTR,** Feigenbaum A, Friedman J: TKT ($111,498 2005).

PKU 003: Phenoptin in PKU - Phase II Muliti-center Double Blind Placebo Controlled Clinical Trial. **Feigenbaum A at HSC site,** Multicentre international: Biomarin Pharmaceuticals ($45,000 2005 - 2006)

2005 CIHR Canada International Opportunities Program Development/Planning Grant Competition. Rare diseases clinical research centre for urea cycle disorders. **Clarke JTR,** Feigenbaum A: (25,000) and private donation (40,000) ($65,000 2005 - 2006) 200509OPD

PKU 004: Phenoptin in PKU - Phase III multi-center open label variable dose clinical trial. **Feigenbaum A at HSC site,** Multicentre international: Biomarin Pharmaceuticals ($41,000 2006 - 2007)

PKU 006: multidose trial of Phenoptin in PKU. Study approved and funded but closed by sponsor before enrolment. **Feigenbaum A at HSC site,** Multicentre study international: Biomarin Pharmaceuticals ($16,485 2006)

Autism Treatment Network:Cooperative Multi-Center Program for Research and Treatment of Autism: Canadian Centre. PI: Wendy Roberts, Alvin Loh, **Co investigator: A Feigenbaum**, M. Marcon, S. Weiss, C. Goldfarb, G. Berall, E. Bradley. $150,000 one year. Site visit September 2007.

HPN100-006: Hyperion 2009-2010. A Phase 3, Randomized, Double-Blind, Cross-Over, Active-Controlled Study of the Efficacy and Safety of HPN-100, Glyceryl tri-(4-

phenylbutyrate), for the Treatment of Adults with Urea Cycle Disorders (Help UCD). **PI at Sickkids A Feigenbaum.**

PKU 008: Biomarin. Phenoptin in PKU - Open Label Extension Study. **Feigenbaum A PI at HSC site,** Multicentre international: Biomarin Pharmaceutical ($25,500 per year 2006 - 2009) In analysis.

Autism Treatment Network: Cooperative Multi-Center Program for Research and Treatment of Autism. Sickkids site: **Roberts SW,** Loh A, Feigenbaum A, Weiss S, Marcon C, Goldfarb C, Berall G, Bradley E, Brian J, Freeman N. Atkinson A, Taylor M, Stinson J, Mitchell S, Green P: Collaborator ; chair of Metabolic Group
Autism Speaks ($408,000 2008 - 2010)

Funding of an autism intervention research network for physical health (AIR-P). Roberts SW, Loh A, Feigenbaum A, Weiss S, Marcon C, Goldfarb C, Berall G, Bradley E, Brian J, Freeman N, Atkinson A, Taylor M, Stinson J, Mitchell S, Green P:
Health Resources and Services Administration (HRSA) ($400,882 2009 - 2011).

PKU 016: Biomarin: 2010. A double-blind, placebo-controlled, randomized study to evaluate the safety and therapeutic effects of sapropterin dihydrochloride on neuropsychiatric symptoms in subjects with phenylketonuria (PKU ASCEND) **PI at Sickkids A Feigenbaum CoPI K. Siriwardena Study active Feigenbaum resigned**

Phase III clinical trial of Coenzyme Q10 in Mitochondrial Disease. Multi-center double blind placebo controlled randomized crossover trial. PI: **Stacpoole P. Site Director Sickkids: Feigenbaum A**: NIH, Department of Health and Human Services, Office of Orphan Products Development ($1,000,000 2006 - 2009) $ 37 500 to Sickkids/year (2006-2010), Prime Award No. R01 FD003032 Subaward No. UF06129

R34 planning grant NICHD awarded to **PI: Dr Peter Stacpoole** U Florida. Multicentre group. A Feigenbaum, et al. Collaborator, Group leader. 2010-2011. Planning for a Phase 3 RCT of Dichloracetate in Pyruvate dehydrogenase deficiency.

HPN100-005: Hyperion 2009-2011. A Phase 2, Fixed-Sequence, Open-Label, Switch-Over Study of the Safety and Tolerability of HPN-100 Compared to Sodium Phenylbutyrate in Children 6–17 Years of Age with Urea Cycle Disorders, with a Long-Term Safety Extension **PI at Sickkids A Feigenbaum**

HPN100-006 and 007: Hyperion 2009-2012. A Phase 3, Open-Label Study of the Safety of **H**PN-100 for the Long-Term **Treat**ment of **U**rea **C**ycle **D**isorders (**Treat UCD). Industry initiated. Site PI at Sickkids A Feigenbaum to 2011 then coI with Dr A Schulze.**

Health Resources and Services Administration (HRSA award UA3MC11054), Autism Intervention Research Network for Physical Disorders (AIR-P) "The Study of Toddlers with Autism and Regression (STAR) Protocol: Biomarkers of Inflammation And Immune activation in ASD". PI: Loh, A. ; James, D. Menon ,E. Anagnostou, C. Pardo, S. Benseler, A. Schulze, A. Feigenbaum, W. Logan, J. Brian, K. Burns, S. Asghar, B. Banwell, S. Spence, M. Bauman, A. Zimmerman, D. Beversdorf, K. Sohl. ($213,854 2011 - 2012): **Coinvestigator**

Autism Speaks Autism Treatment Network Registry. **A. Loh. W. Roberts, E. Anagnostou,** M. Marcon, S. Weiss, J. Brian, A. Feigenbaum, A. Schulze, W. Logan, D. Chitiyat, R. Weksberg, P. Green, J. Weiss, S. Mitchell, G. Berall, E. Bradley, N. Freeman, J. Stinson, M. Ornstein, A. Atkinson, C. Goldfarb, J. Flanagan, D. Superina, E. Jiminez, N. Jones Stokreef, A. Orsino, B. Isaacs, M. Handley Derry: ($420,000 2011 - 2013) **Coinvestigator.**

BioMarin Pharmaceutical, Inc. 2012-2014 . A double-blind, placebo-controlled, randomized study to evaluate the safety and therapeutic effects of sapropterin dihydrochloride on neuropsychiatric symptoms in subjects with phenylketonuria **(PKU ASCEND). Site PI UCSD: B. Barshop.** Protocol Number: PKU-016 IRB approval # 110709. $16,000/pt completing study; $9,600 start up. **CoInvestigator**

Research Funding - Non Peer Reviewed

Mitochondrial disease research. **Robinson B,** Feigenbaum A: Kirkland Kilbride Mito March ($33,992 2004 - 2005)

Population screening clinic operating fund. **Feigenbaum, A:** National council of Jewish Women/Uger Estate ($26,000 2004 - 2005)

Mitochondrial disease research. **Robinson B,** Feigenbaum A: Kirkland Kilbride Mito March ($32,000 2005 - 2006)

Halee Ochshorn Fund: Glycogen storage disease. HSC Foundation. ($25,000 2006-7) for GSD Days May 2007 and research into GSD.

Genetic Metabolic diseases and Phenylketonuria Transition Day 2007 Sickkids $1500

Urea Cycle Travel clinic 2008: Urea Cycle Diseases Consortium. A Feigenbaum. $7500

Mitochondrial disease research. **Robinson B,** Feigenbaum A: Kirkland Kilbride Mito March ($40,000/year – 2007-2012)

Philanthropic support for Urea Cycle Diseases research: The Canadian Urea cycle diseases Network $375,000 **Feigenbaum A**

Phenylketonuria Education Day 2008; The Odessa Sloane Fund Sickkids Foundation

Phenylketonuria Transition Day 2010 The Odessa Sloane Fund Sickkids Foundation

Research Endeavours - Not Funded

Completed

Frontal Lobe Function in Type III PKU. **Smith ML,** Hanley WB, Clarke JT, Feigenbaum A.

Psychological/developmental outcome in Glycogen Storage disease. **Feigenbaum A.**

The use of Dichloroacetate in Chronic congenital lactic acidemia. A multicentre trial, Base centre: UCSD. Sickkids PI **Feigenbaum A**

Vitamin B12 status in phenylketonuria. **Feigenbaum A,** Antle B, Hanley W.

Natural History of Familial Dysautonomia. **Feigenbaum A.**

Can visual evoked potentials differentiate optic neuritis vs Lebers hereditary optic neuropathy in children? **Banwell B,** Feigenbaum A, Buncic R.

MECP2 variants in children with developmental delay/autism. PI **John Vincent PhD**.

Rett syndrome: PI **Rosanna Weksberg**

Mitochondrial disease research 2000-2012. **Robinson B,** Feigenbaum A: partially funded by Kirkland Kilbride Mito March

Barth Syndrome in Canada: more common than we think? **PI Annette Feigenbaum** Yigal Dror, Paul Kantor, Richard Kelley, Aneal Khan, Jonathan B. Kronick, Aziz Mhanni, Michael Geraghty.

HONOURS AND AWARDS RECEIVED

Linda Stevens Travel Award. 1987

MRC Fellowship Award by Medical Research Council of Canada. 1989-1991

Scientific Exhibit (certificate of merit). RSNA. Chicago, IL. 1990

Scientific Exhibit (Geurbet prize). International Paediatric Radiology Society. 2nd Meeting. Stockholm, Sweeden. 1991

3rd Poster Prize. Miami Children's Research Institute, 6th Annual Symposium. Miami, FL. 1995

Humanitarian Award nomination Sickkids Family Advisory Committee 2007.

Inter- professional Practice Award in Paediatric Palliative Care. December 2008 The Hospital for Sick Children. Palliative care service award to the Metabolic Genetics team at Sickkids.

March 2011: Tapestry Team Award to the Clinical and Metabolic Genetics team at Sickkids for work done while Section Head:
to *celebrate SickKids staff and volunteers who promote cultural competence values at the Hospital by:*
Working with colleagues, families and patients in a respectful manner that takes into consideration the diversity of their social, cultural and linguistic backgrounds and beliefs.

Recognizing that patients and families may face challenges that impact family and child health, such as language barriers, poverty and other socioeconomic issues, and limited health literacy, and making all efforts to addressing these issues.
To recognize a team that As a group, seeks out opportunities to enhance their own cultural competence and teaches others the benefits of culturally competent care and practice.
Is respectful and understanding of others' viewpoints.
Looks for areas in which cultural competence seeds can be planted and cared for until they grow.

COMMITTEE INVOLVEMENT

Active:

Genetic testing committee 2013-
Rady Childrens Hospital San Diego

HIPAC 2013-
Rady Childrens Hospital San Diego

(completed) :
University of Toronto, Faculty of Medicine Committees

- Undergraduate Medical Electives Committee, Division of Clinical and Metabolic Genetics, Member
  1997 - 2010

- Advisory Committee for Elective Tutors, Member
  1999 - 2005

- Problem Based Learning: Case Review Committee, Member
  2001 - 2007

- Toronto Complementary and Alternative Medicine Research Network's CAM Research Methods Subcommittee, Member
  2001 - 2003

Hospital Committees

- HSC Foundation External Grant Applications, The Hospital for Sick Children, Toronto, Ontario, Scientific Reviewer
  1994 - 2010

- Elective Undergraduate Student Supervisor, Genetics, The Hospital for Sick Children, Toronto, ON, Supervisor
  1995 - 2009

- Genetics Grand Rounds (weekly), The Hospital for Sick Children, Toronto, ON, Organizer/Coordinator
  1995 - 2002

- Medical Research Council of Canada Grant Application, The Hospital for Sick Children, Toronto, Ontario, ad hoc Reviewer
  1995 – 2011

- Research Ethics Board, The Hospital for Sick Children, Toronto, ON, Active Panel Member
  1995 - 2001

- Research Ethics Board, The Hospital for Sick Children, Toronto, Ontario, Ethics Reviewer and Scientific Reviewer
  1995 - 2010

- Search Committee for Chief, Division of Clinical Genetics, The Hospital for Sick Children, Toronto, Ontario, Division Representative
  1998

- Cardiomyopathy Working Group, The Hospital for Sick Children, Toronto, ON, Active Panel Member
  2000 - 2010

- Focus Group re Triannual Review, The Hospital for Sick Children, Toronto, ON, Active Panel Member
  2000

- Complementary and Alternative Medicine/HSC Foundation Collaboration, The Hospital for Sick Children, Toronto, ON, Member
  2001 - 2002

- HSC Foundation External Grants Review Committee, The Hospital for Sick Children, Toronto, Ontario, Active Panel Member
  2001 - 2010

- Mitochondrial Working Group, The Hospital for Sick Children, Toronto, ON, Member
  2001 - 2012

- Clinical and Metabolic Genetic Division Retreat. Overview of the Metabolic Genetics Program, The Hospital for Sick Children, Toronto, ON, Metabolic Program Presenter
  2002

- Genetic Screening Center Organizing Committee, The Hospital for Sick Children, Toronto, ON, Active Member
  2002 - 2005

- Safety Monitoring Committee for Zavesca in San Fillipo Study - Dr. N. Polowski, Dr. J. Clarke, The Hospital for Sick Children, Toronto, ON, Active Member
  2002 - 2005

- Canadian College of Medical Genetics Accreditation - site visit, The Hospital for Sick Children, Toronto, ON, Panel Representative for Metabolic section of HSC Training Program
  2003

- Clinical/Admission Chiefs Committee, The Hospital for Sick Children - Pediatrics, Toronto, ON, attendee
  2003

- Quality Management Committee, The Hospital for Sick Children (Division of Clinical and Metabolic Genetics), Toronto, ON, Active Member
  2003 - 2004

- Jewish Genetic Screening Program, The Hospital for Sick Children, Toronto, Ontario,
  2005 - 2010

- Muscle Biopsy Task Force (Chair - Dr. Ben Alman), The Hospital for Sick Children, Toronto, ON,
  2005 - 2008

- Safety Monitoring Committee for Zavesca in GM2 Gangliosidosis (Dr. G. Maegawa, Dr. J. Clarke), The Hospital for Sick Children, Toronto, ON,
  2005

- Search Committee, Genetic Metabolic Disease, The Hospital for Sick Children, Toronto, Ontario,
  2005 - 2006

- Supervisory Committee - Dr. C. Morel (fellow), Biochemical Genetics, The Hospital for Sick Children, Toronto, Ontario,
  2005 - 2009

- Joe Clarke Symposium, The Hospital for Sick Children, Toronto, Ontario, Moderator and Organizer (June 12, 2006)
  2006

- Morbidity and Mortality, Division of Clinical and Metabolic Genetics, Sickkids, Toronto, Ontario, Chair
  2006 - 2010

- Residents Research Day Commitee, The Hospital for Sick Children, Toronto, Ontario, Poster Judge at Research Day - May 10, 2006
  2006

- Genetic Metabolic Diseases Program, Sickkids, Toronto, Ontario, Chair - Program

Meetings
July 2006 – January 2009

- PKU Program Sickkids. Program Meetings: Chair
  2004- 2010

- Newborn screening program Sickkids : member, Division of Clinical and Metabolic diseases representative.
  2006 – 2009

- Training Committee Division of Clinical and Metabolic genetics
  2006- 2009

- Ambulatory Functional Plan – Metabolics representative. Sickkids
  2007

- Autism Treatment Network Ontario. Chairs: Wendy Roberts Alvin Loh; member.
  2007 - 2010

- Search Committee for Metabolic Genetics program : Chair
  2007 – 2008

- Patent review Committee for Dr Brian Robinson October 2007

- Trainee Supervisory Committee
- Dr. Julian Raiman(fellow), Biochemical Genetics, The Hospital for Sick Children, Toronto, Ontario,2003-2005
- Dr. F. Al-Mushedi (fellow), Biochemical Genetics, The Hospital for Sick Children, Toronto, Ontario, 2006 – 2009
- Dr. E. Crushell (fellow), Biochemical Genetics, The Hospital for Sick Children, Toronto, Ontario,2007-2008
- Dr. H. Faghfoury (fellow), Biochemical Genetics, The Hospital for Sick Children, Toronto, Ontario2007 – 2010
- Dr. M. Maj (fellow), Biochemical Genetics, The Hospital for Sick Children, Toronto, Ontario 2007 – 2010
-Dr Rawda Sunbul, (fellow), Biochemical Genetics, The Hospital for Sick Children, Toronto, Ontario2008-2009

- Pediatric Resident interviews 2007, 2008, 2010

- Division of Neonatology Mortality Review meetings: Metabolic representative Sickkids
  2009 – 2010

- Clinical Task Force, Genetic Metabolic Diseases , Sickkids Co-chair
  2009- 2010

- Billing Champions: Metabolic rep: 2009-2010

Medical/Scientific Organizations-Committees/Affiliations

Active:

American College of Medical Geneticists
2009-

National Urea Cycle Foundation

2007 – present Scientific and Medical Advisory Board member
Occasional:

American Journal of Medical Genetics

2000,2,3,4,6: Ad Hoc Reviewer

American Journal of Pathology

1998 Ad Hoc Reviewer

Annals of Neurology

2002 Ad Hoc Reviewer

2007 Ad Hoc Reviewer

Autism Treatment Network

2010- 2012 Chair Metabolic Genetic subgroup

Canadian Medical Association Journal

2005 Ad Hoc Reviewer

Canadian College of Medical Genetics

2005 - 2009 Examinations committee, Biochemical Genetics Examiner
2005- 2013 Biochemical Genetics committee

Canadian Institutes of Health Research

2009 Ad hoc grant reviewer

Canadian Organization for Rare Diseases

2006 Medical Advisor

Canadian Pediatric Society

1999 - 2001 Canadian Pediatric Society Surveillance Program, Progressive Intellectual and Neurological Deterioration Panel Member

Cardiovascular Pathology

2002 Ad Hoc Reviewer
2007 Ad Hoc Reviewer

CDG Network North America

2008 - 2009 Invited member Canadian representative

Diabetes

1999 Ad Hoc Reviewer

Disease Models and Mechanisms

2013 Ad Hoc Reviewer

Epilepsy Research

2012 Ad Hoc Reviewer

European Journal of Paediatrics

2011 Ad Hoc Reviewer

FEBS Letters

2006 Ad Hoc Reviewer

Garrod Society of Canada

2008 Annual meeting: Convener and Chair Organizing and Scientific Committee

2014: Moderater *Session 3* May 30, 2014 Ottawa Ontario

Irish Medical Research Charities Group / Health Research Board Joint funding Scheme

2012 Grant application reviewer

Jacobs Ladder Foundation for Neurodegenerative Disease

1999 - 2009 Medical Advisor

Journal of Inherited Metabolic Diseases

2000 Ad Hoc Reviewer
2004
2005
2008
2009
2012

Journal of Intensive Care

1996 Ad Hoc Reviewer

Journal of Paediatrics

2001 Ad Hoc Reviewer
2006 Ad Hoc Reviewer

Journal of Rheumatology

2001 Ad Hoc Reviewer

Lancet

1995 Ad Hoc Reviewer

Medical Research Council of Canada

1994 - 2000 Ad Hoc Reviewer

Medical Science Monitor

2004 Ad Hoc Reviewer

Metabolic Information Network USA

1997 Board, Member

Ministry of Community Safety and Correctional Services

2005 Laison with Project Leader Performance Outcome System

Ministry of Health and Long-Term Care Ontario

2002 - 2011 Inherited Metabolic Diseases Task Force, Member

Ministry of Health of Ontario

1999 Drug Program Branch, Ad Hoc Reviewer

Mitochondrial and Molecular Genetics

2004 Ad Hoc Reviewer

Mitochondrial Medicine Society

2000 Clinical Trials Task Force Committee, Member with UMDF

2001 - 2005 Scientific Abstract Reviewer for Annual Meeting

2001 - 2002 Convening/Nominating/Search Committee

2006 Nominated as President of the Society

Molecular Genetics and Metabolism

2006 Ad Hoc Reviewer
2008
2009
2013

National Council of Jewish Women - Canada

2001 - 2010 Genetic Screening Program, Medical Advisor: liaison with Jacob's Ladder, CORD, CTSAD, FD Foundation

NIH

2008- 2010 Grant reviewer Therapeutic Approaches to Genetic Diseases section: Genes genome and Genetics

North American Metabolic Academy

2007	Content provider

Paediatric Research

2000, 2003,4,5	Ad Hoc Reviewer

Paediatrics and Child Health

2002,3,4,5	Ad Hoc Reviewer

Reproductive Toxicology

2004,5	Ad Hoc Reviewer

Society of Inherited Metabolic Diseases

2002 - 2008	Elected Executive Board Member at large, Canadian Representative
2005	2005 Asilomar Meeting, Organizing Committee for 2005 Asilomar meeting, moderator 1/2 day session
2006 - 2008	Organising Program Committee for the annual SIMD meeting ½ day session Moderator and Ask the Expert Panel

The Barth Syndrome Foundation Inc.

2002 - 2008	Scientific Grant Reviewer
2002 - 2008	Clinical Advisory Panel
2002 - 2005	Scientific and Medical Advisory Board invited Member

United Mitochondrial Disease Foundation UMDF

2001 - 2011	Scientific Grant Reviewer, Active Panel Member
2001 - Present	Ask the Mitodoc Active Panel Member
2003 – 2008	Scientific and Medical Advisory Board Active Member - 6 monthly meetings

Urea Cycle Disease Rare Diseases Consortium USA

2005 - 2006	Observer
2007	Toronto Site PI and organizer for travel clinics
2007- Present	Member
2008- 2012	site PI for Longitudinal study-Toronto/Canada
2012-	coI for Longitudinal study-Toronto/Canada

Weitzmann Institute of Canada

2004 - 2009	Jewish Genetic Screening, Medical Advisor

MEDICAL/SCIENTIFIC ORGANIZATIONS-MEMBERSHIPS

- American Society of Human Genetics (and FASEB). 1990 -
- Canadian College of Medical Genetics. 2004-
- Canadian Paediatric Society. 1992 -
- College of Physicians and Surgeons of Ontario. 1991 -
- Federation of American Societies for Experimental Biology. 1999-
- Garrod Association of Canada. 1997 -
- Medical Practitioner, British Medical Council. 1980
- Medical Practitioner, South African Medical and Dental Council. 1979
- Mitochondrial Medicine Society. 2000 -
- Mitochondrial Research Society. 2000 -
- Ontario Medical Association. 1992-
- Paediatrician, Royal College of Physicians and Surgeons, Canada. 1988
- Paediatrician, South African Medical and Dental Council. 1986
- Society for Inherited Metabolic Diseases. 1999 -
- Society for the Study of Inborn Errors of Metabolism. 1993 -

Conferences/Workshops Attended - CME

1) American Society of Human Genetics. 1988, 1989, 1990, 1992
2) March of Dimes Clinical Genetics Conference. 1991
3) Society for Inherited Metabolic Diseases. 1990, 1991, 1992, 1993, 1996, 1998
4) Miami Children's Hospital Research Institute Annual Symposium. 1993, 1994, 1995
5) Society for the Study of Inborn Errors of Metabolism. 1994, 1995, 1997, 1999
6) UMDF International Conference on Mitochondrial Diseases. 1997, 1999
7) Mitochondrial Medicine. San Diego, CA. 1997
8) Human Ethics Workshop. University of Toronto. 1998
9) UCSD Mitochondrial Medicine Conference. 1998
10) Association of Genetic Colleagues of Ontario. 1998
11) Prenatal and Genetics Update. The Hospital for Sick Children. 1998
12) Mini symposium NIH. Mitochondrial Genetics, Health and Disease. 1998, 1999
13) Workshop on Inborn Errors of Metabolism. Genoa, Italy. September 1999
14) Society fo the Study of Inborn Errors of Metabolism. Genoa, Italy. SSIEM. 1999
15) World Federation of Neurology (WFN) Neurogenetics Research Group Meeting. 1999
16) World Neurology Conference and Neurogenetics meeting. 1999
17) Canadian Paediatric Society. 1999
18) European Mitochondrial Meeting. 1999
19) Teaching Dossier Workshop PMH. 1999
20) CMA Leadership Workshop for Medical Women. 1999
21) REB Retreat. 1997, 1999
22) Giving Feedback - Workshop. Undergraduate Faculty Development. University of Toronto. Toronto General Hospital. February. 2000
23) NIH Mitochondrial Symposium. March 2000
24) Small Group Teaching. Undergraduate faculty development. University of Toronto. Toronto General Hospital. February 200
25) CAM. Toronto Network Workshop. February 9, 2000
26) Planning for a Multidisciplinary Complementary/Alternative Medicine (CSM) Research Network. 2000
27) Prenatal and Genetics Update. The Hospital for Sick Children. 1998, 2000, 2001
28) UMDF Meeting. Cleveland, OH. June 2000
29) Planning for a Multidisciplinary Complementary/Alternative Medicine (CAM). University of Toronto. June 9, 2000
30) 8th Annual Symposium: New Developments in Prenatal Diagnosis and Medical Genetics
31) CAM. University of Toronto. June 2000
32) Phenylketonuria Consensus Conference, NIH. October 2000
33) Medical Education Day. The 2nd Annual. 2000
34) Mitochondria 2001. San Diego 2001
35) ACMG. Miami, FL. March 2001
36) SIMD. Miami, FL. March 2001
37) CAM Toronto Network. February 9, 2001, June 2001
38)Prenatal and Genetics Update. The Hospital for Sick Children. 2001
39) SSIEM, Prague . September 2001
40) CAM HSC Foundation Network. November 2001
41) SIMD. Monterey, California. March 2002

42) Garrod Association Meeting. Monterey, California 2002
43) Pediatric Update. The Hospital for Sick Children. April 2002
44) National Council of Jewish Women. National Board Meeting Launch of Genetic Screening Project. Montreal, Quebec. May 2002
45) United Mitochondrial Diseases Meeting. Dallas, TX. June 2002
46) American Society of Human Genetics. Baltimore, MD. October 2002
47) Barth Syndrome Society. Baltimore, MD. October 2002
48) UMDF Mito - Dallas 2002 Symposium. 2002
49) UMDF Brainstorming Workshop. Chicago, Il. November 11, 2002
50) Clinical Dossier Seminar. Dr. E. Harvey. The Hospital for Sick Children. February 2003
51) Teaching Dossier Seminar. Dr. S. Tallet/Dr. K. Leslie. The Hospital for Sick Children. March 2003
52)Garrod Society Meeting. Winnipeg, Canada. May 2, 2003
53) Mitochondrial Medicine Society Meeting. San Diego, USA. June 11 -14, 2003
54) United Mitochondrial Diseases Foundation: Grant review. Invited. San Diego, USA. June 2003
55) United Mitochondrial Diseases Foundation: Meeting. Invited lecture. San Diego, USA. June 2003
56) Nyhan Centre Brainstorming. Invited panel member. Palo Alto, USA. January 2004
57) Society of Inherited Metabolic Diseases. Orlando, Florida. March 2004
58) United Mitochondrial Diseases Foundation. Grant review. Chicago, USA. 2004
59) Newborn Screening Symposium. Ministry of Health. Toronto, Ontario. June 2004
60) Barth Syndrome Foundation. Panel meeting. Invited lecture. Orlando, Florida. July 2004
61) Euromit. Nijmegen, Netherlands. July 2004
62) SHS Metabolic Conference - PKU. Teleconference. October 1, 2004
63) American Society of Human Genetics. Poster presentation. Toronto, Ontario. October 2004
64) PKU BH4 Conference. Miami, Florida. February 2005
65) SIMD. Board member. Session moderator. Monterey, California. March 2005
66) Latin American Society of Inborn Errors of Metabolism and Neonatal Screening (SLEIMPN). Invited plenary speaker. Costo Rica. November 2005
67) CDCP. Workshop. The Hospital for Sick Children. February 2006
68) ACMG.SIMD. San Diego, USA. March 2006
69) UMDF. Scientific Meeting. Atlanta, USA. June 14 - 17, 2006
70) Joe Clarke's Symposium. Organizing committee and moderator. June 2006
71) ACMG/SIMD annual meeting. Nashville USA March 2007.
72) Newborn screening Ontario meeting Ottawa April 2007.
73) Urea cycle ½ day at Sickkids: visitor: Dr Mark Batshaw Urea Cycle Consortium. May 2007: Organiser and moderator.
74) North American Metabolic Academy: Course provider, Inaugural committee. Meeting Washington August 2007
75) North American Metabolic Academy Monterey March 2008
76) Urea cycle ½ day at Sickkids: visitor: Dr Mark Batshaw Urea Cycle Consortium. May 2007: Organiser and moderator.
77) GSD days at Sickkids: Dr David Weinstein and University of Florida Gainesville Team: 2 day clinics and workshops with patients and parents. Grand rounds Host. Organiser. May 30, 31 2007.

78) Genetic Metabolic Diseases Program: Transition Day for families and professionals October 20 2007. Organising committee.
79) Tissue Culture DPLM QM issues Committee member 2006-7. Chair: Dr PhilGordon.
80)Organiser monthly Neuroradiology rounds for the Division of Clinical and Metabolic genetics. 2007- 2010
81)Urea Cycle Diseases Travel Clinics: Organiser and Chair. January 9, 10 2008.
82)SIMD Monterey USA March 2008
83)Cystinosis Foundation Meeting Newport Beach USA April 2008
84)Garrod Society Annual meeting:Toronto. Organiser and Chair, scientific reviewer May 2008.
85)United Mitochondrial Disease Foundation Indianapolis June 2008
86)Urea Cycle Foundation Orlando June 2008.
87)Autism Treatment Network: Medical Aspects of Autism Care. Toronto March 2009
88)Newborn Screening. CHEO Ottawa. April 2009.
89) Prenatal Symposium. University of Toronto, Toronto, Ontario. May 13, 2009
90)United Mitochondrial Disease Foundation Washington June 2009
91)ICIEM San Diego September 2009
92)PKU update San Diego September 2009
93)Urea Cycle Update (UCDC/NUCDF) San Diego September 2009
94)Urea Cycle disease- research update (Hyperion) San Francisco October 2009
95)Canadian PKU Physician advisory meeting (Biomarin) Montreal November 2009.
96)Biomarin speakers meeting Scottsdale Arizona February 2010.
97) Garrod Asssociation Annual Meeting St Johns Newfoundland June 2010: 10 hours.
98) Urea Cycle Diseases Foundation Meeting Boston July 2010
99) Urea Cycle Disease Consortium Investigators meeting Boston July 2010
100) Teleconference: Whats new in PKU. Biomarin . July 2010 1 hour.
101) France Foundation Webinar: October 26 2010. Dr Vockley New and Emerging Treatment Options for PKU. 1hour.
102) France Foundation Webinar: November 18 2010 1 hour. : Current Management of PKU – Dietary and Nutritional Considerations.
103) Good Clinical Practices (GCP) Information Session. Health Canada Toronto November 23 2010.
104) Canadian PKU Physician advisory meeting (Biomarin) Toronto December 2-3, 2010: 11 hours.
105) SIMD Monterey USA February 27-March2, 2011
106) Garrod society Calgary Canada June 9-10, 2011
107) SSIEM Geneva Switzerland August 29–September 2, 2011
108) Region 4 genetics collaborative meeting: Newborn Screening by MS/MS Quality Improvement Project.. San diego USA November 6 2011.
109) 2011 Newborn Screening & Genetic Testing Symposium APHL - San Diego, CA November 7-10 2011
110) Autism Spekas: Autism Treatment network: Fall program meeting November 17-18 2011 Washington DC: Genetic Metabolic sub-committee co chair
111) ATN/AIR-P Works in Progress Webinar-Recruitment Panel. November 21 2011
112) NIH Consensus conference on Phenylketonuria: by Webinar February 22- 23rd 2012
113) Rare diseases Day at Burnham Sanford Institute February 24 2012
114)) ATN/AIR-P Works in Progress Webinar-Update. February 27 2012
115) Speakers training: Symbiotix for Biomarin Dallas March 9-10 2012
116) ACMG Annual Meeting Charlotte NC March 29-31 2012

117) SIMD Annual Meeting Charlotte NC April 1-4 2012
118) UCDC Meeting Charlotte NC April 12012
119)Rady Childrens Hospital: Giving feedback worskhop June 2 2012
120) Annual NUCDF/UCDC meeting July 2-4 2012 Washington DC
121) PALS course Rady Childrens Sept 13-14 2012
122) Neuromuscular meeting Rady Childrens Hospital December 5 2012
123) Sanford Burnham Rare Disease day February 28 2013
124) UCLA lysosomal meeting March 1 2013
125) ACMG Phoenix March 19-23 2013
126) UMDF Newport Beach June 12-15 2013- parent session speaker
127) Abbott Nutrition Metabolic Conference June 27-29 2013: invited speaker
128) UCDC –EIMD Meeting Barcelona Spain September 2013
129) SSIEM Barcelona Spain September , 2013
130) Sanford Burnham Rare Disease day March 2014
131) SIMD Annual Meeting Monterey CA March 2014
132) ACMG Annual Meeting Nashville TN March 2014
133) Garrod Society Annual meeting May 2014 Ottawa
134) SIMD Salt Lake City March 2015
134) Network PKU Seattle May 2015
135) Garrod Society Annual meeting May 2015 Vancouver

CREATIVE PROFESSIONAL ACTIVITY

Outside Agencies
1) **Ashkenazi Jewish Screening Clinic**: National Council of Jewish Women, Jacobs Ladder Foundation, CTSAD, Familial Dysautonomia Foundation and The Hospital for Sick Children: Coordinator for Ashkenazi Jewish Screening Clinic; Toronto; October 2001; reporting out of all results with information package; n=312
2) **Ashkenazi Jewish Diseases**: Pamphlet for referring physicians developed 2006
3) **Barth Syndrome Foundation:** Internet "Ask The Doctor" Invited Participant; 2000 - 2008
4) **Barth Syndrome Foundation of Canada**: Barth Syndrome multi-disciplinary clinic at HSC 2005
5) **Barth Syndrome Foundation**: Medical and Scientific advisor, grant reviewer, clinical panel. October 2002, February 2003, November 2003, May 2004, January 2005
6) **Canadian Society for the Weizmann Institute of Science**: Organizer and Speaker : Jewish genetic disease; April 28, 2004; Symposium at Ontario Science Center: Toronto.
7) **Hospital for Sick Children:**
HSC Foundation External Grants Committee and Reviewer: Bi-annual grants review meetings; 2001 - present
Research Ethics Board: Ethics reviewer and scientific reviewer; 1995 - present
CIHR: Internal peer review; Dr. Eve Roberts - HSC; Mitochondrial liver disease
HSC: Internal peer scientific review, Dr. Brian Robinson, 2003 - Leigh Disease;
Internal peer scientific review, Dr. Joe Clarke, 2003 - Fabry Disease;
Internal peer scientific review, Dr. Joe Clarke, 2003 - GM2 gangliosidosis;
Internal peer scientific review, Valerie Austin, January 2004 - PKU;
Internal peer scientific review, Dr. Brian Robinson, 2003 - Leigh Disease;
Internal peer scientific review, Dr. Joe Clarke, 2003 - GM2 gangliosidosis;
Internal peer scientific review, Dr. Paul Kantor, Cardiology - HSC, 2005 - Pediatric

Cardiomyopathy Registry;
Internal peer scientific review, Dr. Brian Robinson, 2005 - Leigh Disease;
Internal peer scientific review, Dr. Brian Robinson, 2006 - Leigh Disease;
8) Inquest: Urea cycle defect in detention Milton Ontario Expert witness
9) **Jacobs Ladder**: The Canadian Foundation for Control of Neurodegenerative Disease: Medical advisor; Scientific advisor to 2008
10) **Medical Research Council of Canada:** Ad hoc scientific reviewer; 1995 – 2000, 2009.
11) **Mitochondrial Medicine Society:** Scientific abstract reviewer; Search committee. Nominated for President 2006.
13) **National Council for Jewish Women**: Toronto Chapter: Jewish Genetic Screening and Awareness Program: Arranging community based access for DNA testing for Canavan and Familial Dysautonomia testing; 2003 - 2011
14) **National Council for Jewish Women Canada**: Medical advisor for A Trans-Canada Jewish Genetic Screening Program; 2002 - 2011
15) Other reviews:
Caring for metabolic patients; Marija Bojic RN - HSC, 2004;
Metabolic diseases chapter, Dr. Andrew Argent - South Africa, 2004
16) **Society of Inherited Metabolic Diseases USA:** Moderator and coordinator; 1/2 day session at annual meeting; Monterey, California; March 2005; Moderator And Panel member ½ day each- newborn screening and 'ask the expert" : Nashville USA March 2007.
17) **United Mitochondrial Diseases Foundation**: Clinical and Scientific Advisor; Ad Hoc Scientific Reviewer; Internet Chat Line "Ask The Doctor" Participation; 2000 -2009
18) **United Mitochondrial Diseases Foundaton**: Invited participation; Family Symposium 2006; Atlanta; Panel member: "Ask the mito doc"; One on one 3 hour session 5 families: "the doctor is in".
19) United Mitochondrial Diseases Foundation: Grant review 2001, 2002 (Dallas, USA), 2003 (San Diego, USA), 2004 (Chicago, USA), 2004 (LOI), 2005 (Letters of intent, grant reviews), 2006 (Letters of intent, grant reviews)
20) **Canadian College of Medical Geneticists:** Biochemical Genetics examiner/Examinations committee/Biochemical training committee 2006-
21) National Council of Jewish Women Canada: Organiser for Jewish genetic screening day and send out of reports. Toronto 2007 May 7. n=279.
22) **Magen David Adom Canada:** Jewish Genetic diseases: The power of Prevention. Toronto May 17 2007.
23) Festschrift for Dr Eve Roberts. Sickkids and Sunnybrooke Estates. May 26 2007. Attendee.
24) Mitomarch for the Cure: Mitochondrial fundraiser (Kirkland Kilbride Fund) 2007, 2008.
25) **Glycogen Storage Disease** Days Sickkids. May 30th and 31st 2007. Chair and Organiser.
26) Delhi method to establish evidence based guidelines for management of 3MCCD. Chair: Dr G Arnold Rochester New York. Panel member. Published- coauthor
27) **Urea Cycle Diseases Consortium USA**. Toronto Site PI and Organiser. 2007. Meeting Washington November 2007
28) Organiser: PKU and GMD **transition** day – Sickkids 2007.
28) Delhi method to establish evidence based guidelines for management of VLCADD. Chair: Dr. G. Arnold Rochester New York. Panel member
29) Organiser and chair: **Garrod Society** annual meeting Toronto Canada May 2008.
30) Organiser, Presenter and Chair: **PKU education day** for Patients and Lay persons. April 4 2009. Toronto. Diet for Life in PKU- presentation to Lay persons. April 24 2009.

31) **Ashkenazi Jewish Genetic Screening day with National Council of Jewish Women Toronto Canada**: Medical Organiser: Toronto. November 2009
32**) Urea Cycle** disease- research update (Hyperion) Presenter of Toronto data. San Francisco October 2009
33) Canadian **PKU** Physician advisory meeting (Biomarin) Montreal. Presenter: Kuvan in PKU. November 2009.
34) **Biomarin** speakers meeting. Scottsdale Arizona February 2010.
35) **Glycogen Storage Disease** Fundraiser. Toronto. Sickkds Rep and thank families. March 2010.
36) PKU Educational Day April 24 2010. Toronto. Organiser and presenter.
37) **Autism Treatment Network:** Genetic Metabolic subgroup tele-meetings: monthly. Chair.
38) **PKU** camp doctor and educator. Sponsor: Specilaty Food Shop Sickkids. October 16-18, 2010. Parry Sound Ontario
39) Lecture to parents of children with **urea cycle defects**: Metabolic Family Day Sickkids Toronto November 6 2010. Update on urea cycle disorder research
40) **Health Canada:** Scientific Advisory Panel on Neurological (neurodegenerative) Diseases: Therapeutic Products Directorate. Nominated member; voluntary position. October 2011
41)**National Urea Cycle Disease Foundation**: Guidelines for families with UCD. 2012-
42) **Barth Syndrome**: 2012: worked with Barth Syndrome Foundation Canada to apply to **Canadian Pediatric Society Surveillance Program** for Project: Barth Syndrome in Canada: more common than we think?: PI, lit review, wrote application, chosen to present as Webinar to CPS. Not funded.
43) **Irish Medical Research** Charities Group / Health Research Board Joint funding Scheme ad hoc grant reviewer
44) **UMDF 2013:** Ask the Mitodoc; panel speaker
45) **Network PKU:** Familty education re Phenylketonuria 2013-
46) Biomarin: Speaker Bureau 2009-
47) Biomarin: PKU 016 Steering committee 2012-
48) Hyperion DSMB member for HPN100-0009 study 2014-

Pharmaceutical/Industry affiliations:
1) Tishcon Corporation:
- Medical lead Canada/Health Canada liason for CoQ study to 2012

2)Biomarin Pharmaceuticals:
- Site PI for Studies funded as listed
- 2010-2014: PKU016 ascend study: Steering committee member;
- 2009-2014: Educational Speakers member
- 2012 PKU Speaker Training - March 9-10, 2012 - Dallas, Texas.
- November 1-2, 2013 PKU Speaker's Bureau - Scottsdale, Arizona.
- Site CoI for PKU 015, PKU 016, PKUDOS, POM1 and POM2 studies
- 2013: Site PI for Prism 301, 302 studies (PegPal)

3) Hyperion Therapeutics
- Site PI for Studies funded as listed to 2012
- Medical lead investigator Health Canada liason 2008-2012
- Coinvestigator

- DSMB for HPN 009 study 2015
- Site PI Thrive registry UCSD 2014

Lectures given at UCSD 2012-:
Urea Cycle diseases and aminoacidopathies: Genetics trainees
December 17 2012 1.5 hours;
March 2014.1.5 hours

Teaching UCSD 2012-:
GIM small group teaching 2013, 2014 2hours

Rady Childrens Hospital 2012-:
Urea Cycle defects: CPC February 2015