CURRICULUM VITAE

Bruce A. Barshop, M.D., Ph.D.

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EDUCATION/TRAINING

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	YEAR(s)	FIELD OF STUDY
Brandeis University, Waltham, MA	A.B.	1976	Biochemistry
Washington University, St. Louis, MO	M.D.	1984	Medicine
Washington University, St. Louis, MO	Ph.D.	1984	Molecular Biology

A. Personal Statement

A major focus of my career has been in chemometrics and numerical modeling, and my laboratory has focused on large-scale quantitative clinical metabolomic measurements. My background is in metabolic regulation and control, and I direct the William L. Nyhan Biochemical Genetics and Metabolomics Laboratory at UCSD, a CLIA-compliant, CAP-certified facility which processes large numbers of clinical samples. I have served on the CAP-ACMG Genetics Resource Committee charged with designing and evaluating proficiency testing in Biochemical Genetics. I am clinically active in the practice of Biochemical Genetics and am Clinical Chief of Genetics at Rady Children's Hospital San Diego. I have experience in clinical trials and have been involved in many investigator-initiated and sponsored studies.

B. Positions and Honors.

PROFESSIONAL EXPERIENCE

1984-1986: Intern and Resident in Pediatrics (PL-1,-2), University of California San Diego
1986-1988: Clinical Fellow, Human Genetics, Department of Pediatrics, UCSD: Dr. William Nyhan.
1988-1990: Research Fellow, Department of Medicine, UCSD: Dr. J. Seegmiller.
1990-1991: Assistant Research Scientist, Department of Pediatrics, UCSD
1991-1992: Senior Resident in Pediatrics (PL-3), UCSD
1992-1998: Assistant Professor in Residence, Department of Pediatrics, UCSD
1998-2004: Associate Clinical Professor, Department of Pediatrics, UCSD
2004-2019: Professor of Clinical Pediatrics, Department of Pediatrics, UCSD
2019-: Professor Emeritus (Recalled/Active), Department of Pediatrics, UCSD

HONORS AND AWARDS

High honors in Biochemistry, Brandeis University (1976); Aaron B. Chausmer Prize in Biomedical Computing, National Student Research Forum (1982); Outstanding Resident Teaching Award, UCSD Department of Pediatrics (1992); Best Doctors in America (2003-present). Benard L. Maas Chair in Inherited Metabolic Disease, UCSD (2007-2019). Co-chair, session on Metabolic Diseases, 2014 meeting of the American Society of Human Genetics. Opening talk at 2014 SIMD meeting on "Metabolomic Approaches to Metabolic Testing." Opening plenary talk at 2016 SSIEM meeting, "Untargeted Metabolomics in Inborn Errors of Metabolism."

SERVICE APPOINTMENTS

Director, Biochemical Genetics Laboratory, Department of Pediatrics, UCSD (1994-); Assistant Director, UCSD Pediatric Pharmacology Research Unit (1996-2003); Chair, UCSD Clinical Research Center Advisory Committee (1997-2001); Research Safety Advisor, UCSD General Clinical Research Center (2001-2010); Chief Medical Consultant, California Newborn Screening, Region V (2002-2018)

REGIONAL, NATIONAL AND INTERNATIONAL COMMITTEE SERVICE

Board of Directors, Society for Inherited Metabolic Diseases (2000-); Steering Committee, Mitochondrial Medicine Society (2001-3); Advisory Committee, Expanded Newborn Screening Project, California Department of Health Services, Genetic Disease Branch (2002-); Item Writer, American Board of Medical

Genetics, Section on Biochemical Genetics (2001-2004); American College of Medical Genetics, Web Site Oversight Committee (2006-2009), College of American Pathologists, Biochemical and Molecular Genetics Resource Committee (2007-2013); Data Safety Monitoring Boards, StemCells, Inc. Phase I trial of neuronal stem cell therapy in a) neuronal ceroid lipofuscinosis (2007-2013), b) Pelizaeus-Merzbacher disease (2010-2013); Communicating Editor, Journal of Inherited Metabolic Disease (2005-present). Subcommittee on Biochemical Genetics Testing, CDC/CLIA (2009-2010).

C. Publications

A. Published Primary Work

a) Research Articles

1 Margossian SS, Lowey S, Barshop B. Effect of DTNB light chain on the interaction of vertebrate skeletal myosin with actin. Nature. 13;258(5531):163-6, 1975. PMID: 1186897

2 Barshop BA, Wrenn RF, Frieden C. Analysis of numerical methods in computer simulation of chemical kinetic processes: Development of KINSIM, a flexible, portable system. Analytical Biochemistry 130:134-145, 1982. PMID: 6688159

3 Barshop BA, Frieden C. Analysis of the interaction of rabbit skeletal muscle adenylate deaminase with myosin subfragments: A kinetically regulated system. Journal of Biological Chemistry 259:60-66, 1983.

4 Nyhan WL, Mascarello J, Barshop BA, Doroski D, Hirshhorn K. Duplication of 16q and deletion of 15q. American Journal of Medical Genetics 34:183-186, 1989.

5 Barshop BA, Alberts AS, Gruber HE. Kinetic studies of mutant human adenylosuccinase. Bioch Biophys Acta 999:19-23, 1989.

6 Wolff JA, Barshop BA, Nyhan WL, Leslie J, Seegmiller JE, Gruber HE, Garst M, Winter S, Michals K, Matalon R. Effects of ascorbic acid in alkaptonuria: Alterations in benzoquinone acetic acid and an ontogenic effect in infancy. Pediatric Research 26:140-144, 1989.

-- 7 Barshop BA, Breuer J, Holm J, Leslie J, Nyhan WL. Excretion of hippuric acid during sodium benzoate therapy in patients with hyperglycinaemia or hyperammonaemia. Journal of Inherited Metabolic Disease 12:72-79, 1989.

8 Press GA, Barshop BA, Haas R, Nyhan WL, Glass RF, Hesselink JR. Abnormalities of the brain in nonketotic hyperglycinemia: Magnetic resonance manifestations. American J. of Neuroradiology 10:315-321, 1989.

9 Barshop BA, Wolff J, Nyhan WL, Yu A, Prodanos C, Jones GC, Sweetman L, Leslie J, Holm J, Green R, Jacobsen DW, Cooper B, Rosenblatt D. Transcobalamin II deficiency presenting with methylmalonic aciduria, homocystinuria and abnormal cobalamin absorption. American Journal of Medical Genetics 35:222-228, 1990.

10 Barshop BA, Adamson DT, Vellom DC, Rosen F, Epstein BL, Seegmiller JE. Luminescent immobilized enzyme test systems for pyrophosphate: Assays using firefly luciferase and nicotinamide-mononucleotide adenylyl transferase or adenosine-5'-triphosphate sulfurylase. Analytical Biochemistry, 197:266-272, 1991.

11 Ko F-J, Nyhan WL, Wolff J, Barshop BA, Sweetman L. 3-Hydroxyisobutyric aciduria: An inborn error of valine metabolism. Pediatric Research, 30:322-326, 1991.

12 Barshop BA, Yoshida I, Ajami A, Sweetman L, Wolff J, Prodanos C, Sweetman FR, Smith M, Nyhan WL. Metabolism of 1-13C-propionate in patients with disorders of propionate metabolism. Ped Research, 30:15-22, 1991.

13 Stone RL, Aimi J, Barshop BA, Jaeken J, Van den Berghe G, Zalkin H, Dixon JE. A mutation in adenylosuccinate lyase associated with mental retardation and autistic features. Nature Genetics, 1:59-63, 1992.

14 Marsden D, Barshop BA, Capistrano-Estrada S, Rice M, Prodanos C, Sartoris D, Wolff J, Jones KL, Spector S, Nyhan WL. Anabolic effect of human growth hormone: Management of inherited disorders of catabolic pathways. Biochemical Medicine and Metabolic Biology 52:145-54, 1994.

15 Page T, Barshop BA, Yu A, Nyhan WL. Treatment of Lesch-Nyhan syndrome with AICAR. Adv Exp Med Biol. 370:353-6, 1994.

16 Haas RH, Light M; Rice M; Barshop BA. Oxidative metabolism in Rett syndrome .1.Clinical studies. Neuropediat 26:90-4, 1995.

17 Nyhan WL, Rice-Kelts M, Klein J, Barshop BA. Treatment of the acute crisis in maple syrup urine disease. Arch Pediatr Adolesc Med 152:593-8, 1998.

18 Haas RH, Barshop BA. Diet change in the management of metabolic encephalomyopathies. BioFactors 7:259-62, 1998.

19 Faunt KK, O'Brien DP, Barshop BA, Thorburn DR, Shelton GD. Malonic aciduria in Maltese dogs. J Vet Int Med 12:236, 1998.

20 Shelton GD, Nyhan WL, Kass PH, Barshop BA, Haas RH. Analysis of organic acids, amino acids, and carnitine in dogs with lipid storage myopathy. Muscle and Nerve 21:1202-1205, 1998.

21 Naviaux RK, Nyhan WL, Barshop BA, Poulton J, Markusic D, Karpinski NC, Haas RH. Mitochondrial DNA polymerase gamma deficiency and mtDNA depletion in a child with Alpers' syndrome. Ann Neurol, 45:54-58, 1999.

22 Naviaux RK, Markusic D, Barshop BA, Nyhan WL, Haas RH. Sensitive assay for mitochondrial DNA polymerase gamma. Clin Chem 45(10):1725-33, 1999.

23 O'Brien DPO, Barshop BA, Faunt KK, Johnson GC, Gibson KM, Shelton GD. Malonic aciduria in Maltese dogs: Normal methylmalonic acid concentrations and malonyl-CoA decarboxylase activity in fibroblasts. J Inher Metab Dis 22:883-890, 1999.

24 Barshop BA, Nyhan WL, Naviaux RK, McGowan KA, Friedlander M, Haas RH. Kearns-Sayre syndrome presenting as 2-oxoadipic aciduria. Molecular Genetics and Metabolism 69(1):64-8, 2000.

25 Graf WD, Marin-Garcia J, Gao HG, Pizzo S, Naviaux RK, Markusic D, Barshop BA, Courchesne E, Haas RH. Autism associated with the mitochondrial DNA G8363A transfer RNA(Lys) mutation. J Child Neurol, 15(6):357-361, 2000.

26 Spruijt L, Naviaux RK, McGowan KA, Nyhan WL, Sheean G, Haas RH, Barshop BA. Nerve conduction changes in mitochondrial patients treated with dichloroacetate. Muscle and Nerve, 24(7):916-924, 2001.

27 Lee YS, Yap HK, Barshop BA, Lee YS, Rajalingam S, Loke KY. Mitochondrial tubulopathy: the many faces of mitochondrial disorders. Pediatr Nephrol, 16:710-2, 2001.

28 Barshop BA, Nyhan WL, Climent C, Rubio V. Pitfalls in the detection of heterozygosity by allopurinol in a variant form of ornithine carbamoyltransferase deficiency. J Inherit Metab Dis, 24: 513-4, 2001.

29 Marsden D, Nyhan WL, Barshop BA. Creatine kinase and uric acid: early warning for metabolic imbalance resulting from disorders of fatty acid oxidation. Eur J Pediatr. 160(10):599-602, 2001.

30 O'Donnell J, Finer NN, Rich W, Barshop BA, Barrington KJ. Role of L-carnitine in apnea of prematurity: a randomized, controlled trial. Pediatrics. 109(4):622-626, 2002.

31 Nyhan W, Khanna A, Barshop B, Naviaux R, Precht A, Lavine J, Hart M, Hainline B, Wappner R, Nichols S, Haas R. Pyruvate carboxylase deficiency- Insights from liver transplantation. Mol Genet Metab. 77(1-2):143-149, 2002.

32 Farag NH, Barshop BA, Mills PJ. Effects of estrogen and psychological stress on plasma homocysteine levels. Fertil Steril. 79(2):256-260, 2003.

33 Gao HZ, Kobayashi K, Tabata A, Tsuge H, Iijima M, Yasuda T, Kalkanoglu HS, Dursun A, Tokatli A, Coskun T, Trefz FK, Skladal D, Mandel H, Seidel J, Kodama S, Shirane S, Ichida T, Makino S, Yoshino M, Kang JH, Mizuguchi M, Barshop BA, Fuchinoue S, Seneca S, Zeesman S, Knerr I, Rodes M, Wasant P, Yoshida I, De Meirleir L, Abdul Jalil M, Begum L, Horiuchi M, Katunuma N, Nakagawa S, Saheki T. Identification of 16 novel mutations in the argininosuccinate synthetase gene and genotype-phenotype correlation in 38 classical citrullinemia patients. Hum Mutat. 22(1):24-34, 2003.

34 Barshop BA, Nyhan WL, Steenhout PH, Endres W, Tolan TR, Clemens RA. Fructo-oligosaccharide tolerance in patients with hereditary fructose intolerance. A preliminary nonrandomized open challenge short-term study. Nutrition Research, 23:1003-1011, 2003.

35 Wang G, Medeiros FA, Barshop BA, Weinreb RN. Total plasma homocysteine and primary open-angle glaucoma. Am J Ophthalmol. 137(3):401-6, 2004.

36 McGowan KA, Nyhan WL, Barshop BA, Naviaux RK, Yu A, Haas RH, Townsend JJ. The role of methionine in ethylmalonic encephalopathy with petechiae. Arch Neurol. 61(4):570-4, 2004.

37 Barshop BA, Naviaux RK, McGowan KA, Levine F, Nyhan WL, Loupis-Geller A, Haas RH. Chronic Treatment of mitochondrial disease patients with dichloroacetate. Molec Genet Metab, 83(1-2):138-49, 2004.

38 Moore DF, Ye F, Brennan ML, Gupta S, Barshop BA, Steiner RD, Rhead WJ, Brady RO, Hazen SL, Schiffmann R. Ascorbate Decreases Fabry Cerebral Hyperperfusion Suggesting a Reactive Oxygen Species Abnormality – An arterial spin tagging study. J Mag Res Imag 20(4):674-83, 2004.

39 Barshop BA. Metabolomic approaches to mitochondrial disease: correlation of urine organic acids. Mitochondrion, 4/5-6:521-7, 2004, 2004.

40 Wolf B, Jensen KP, Barshop B, Blitzer M, Carlson M, Goudie DR, Gokcay GH, Demirkol M, Baykal T, Demir F, Quary S, Shih LY, Pedro HF, Chen TH, Slonim AE. Biotinidase deficiency: novel mutations and their biochemical and clinical correlates. Hum Mutat 25(4):413, 2005.

41 Dohil R, Fidler M, Barshop B, Newbury R, Sellers Z, Deutsch R, Schneider J. Esomeprazole therapy for gastric acid hypersecretion in children with cystinosis. Pediatr Nephrol 20(12): 1786-93, 2005.

42 Barshop BA, Khanna A. Domino hepatic transplant in maple syrup urine disease. New Engl J Med 353(22): 2410-1, 2005.

43 Eraly SA, Vallon V, Vaughn DA, Gangoiti JA, Richter K, Nagle M, Monte JC, Rieg T, Truong DM, Long JM, Barshop BA, Kaler G, Nigam SK. Decreased renal organic anion secretion and plasma accumulation of endogenous organic anions in OAT1 knockout mice. J Biol Chem 281(8): 5072-83, 2006.

44 Khanna A, Hart, M, Nyhan WL, Hassanein T, Panyard-Davis J, Barshop BA. Domino liver transplantation in maple syrup urine disease. Liver Transplantation 12(5):876-82, 2006.

45 Dohil R, Fidler M, Barshop BA, Gangoiti J, Deutsch R, Martin M, Schneider JA. Understanding intestinal cysteamine bitartrate absorption. J Pediatr. 148(6):764-9, 2006.

46 Fidler MC, Barshop BA, Gangoiti J, Deutsch R, Martin M, Schneider J, Dohil, R. Pharmacokinetics of cysteamine bitartrate following gastrointestinal infusion. Br J Clin Pharm, 63(1):36-40, 2006.

47 Barshop BA, Gangoiti JA. Analysis of coenzyme Q in human blood and tissues. Mitochondrion 7S:S89-93, 2007.

48 Sethi R, Barshop B, Stucky ER. Vomiting-again? J Hosp Med. 2(3):189-93, 2007.

49 Wikoff WR, Gangoiti JA, Barshop BA, Siuzdak G. Metabolomics identifies perturbations in human disorders of propionate metabolism. Clin Chem 53(12):2169-76, 2007.

50 Lien J, Nyhan WL, Barshop BA. Fatal initial adult-onset presentation of urea cycle defect. Arch Neur 64:1777-9, 2007.

51 Wolfe LA, Finegold DN, Vockley J, Walters N, Chambaz C, Suormala T, Koch HG, Matern D, Barshop BA, Cropcho LJ, Baumgartner MR, Gibson KM. Potential misdiagnosis of 3-methylcrotonyl-coenzyme A carboxylase deficiency associated with absent or trace urinary 3-methylcrotonylglycine. Pediatrics 120(5):e1335-40, 2007.

52 Eraly SA, Vallon V, Rieg T, Gangoiti JA, Wikoff WR, Siuzdak G, Barshop BA, Nigam SK. Multiple organic anion transporters contribute to net renal excretion of uric acid. Physiol Genomics. 33(2):180-92, 2008.

53 Vallon V, Eraly SA, Wikoff WR, Rieg T, Kaler G, Truong DM, Ahn SY, Mahapatra NR, Mahata SK, Gangoiti JA, Wu W, Barshop BA, Siuzdak G, Nigam SK. Organic anion transporter 3 contributes to the regulation of blood pressure. J Am Soc Nephrol. 19(9):1732-40, 2008.

54 Arnold GL, Koeberl DD, Matern D, Barshop B, Braverman N, Burton B, Cederbaum S, Fiegenbaum A, 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 93(4):363-70, 2008.

55 Barshop BA, Summar ML. Attitudes regarding vaccination among practitioners of clinical biochemical genetics. Mol Genet Metab 95(1-2):1-2, 2008.

56 Van Hove JL, Josefsberg S, Freehauf C, Thomas JA, Thuy le P, Barshop BA, Woontner M, Mock DM, Chiang PW, Spector E, Meneses-Morales I, Cervantes-Roldán R, León-Del-Río A. Management of a patient with holocarboxylase synthetase deficiency. Mol Genet Metab. 95(4):201-5, 2008.

57 Dhar SU, Scaglia F, Li FY, Smith L, Barshop BA, Eng CM, Haas RH, Hunter JV, Lotze T, Maranda B, Willis M, Abdenur JE, Chen E, O'Brien W, Wong LJ. Expanded clinical and molecular spectrum of guanidinoacetate methyltransferase (GAMT) deficiency. Mol Genet Metab. 96(1):38-43, 2009.

58 Lee TM, Addonizio LJ, Barshop BA, Chung WK. Unusual presentation of propionic acidaemia as isolated cardiomyopathy. J Inherit Metab Dis. 32 Suppl 1:S97-101, 2009.

59 West M, Nicholls K, Mehta A, Clarke JT, Steiner R, Beck M, Barshop BA, Rhead W, Mensah R, Ries M, Schiffmann R. Agalsidase alfa and kidney dysfunction in Fabry disease. J Am Soc Nephrol. 20(5):1132-9, 2009.

60 Nyhan WL, Willis M, Barshop BA, Gangoiti J. Positive newborn screen in the biochemically normal infant of a mother with treated holocarboxylase synthetase deficiency. J Inherit Metab Dis. Apr 11. [Epub ahead of print], 2009.

61 Fidler MC, Gangoiti JA, Schneider JA, Barshop BA. Time before isolating cystinotic leukocytes affects reliability of cystine determination. Pediatr Nephrol. Apr 25. [Epub], 2009.

62 Barshop BA, Greene CL. Laboratory Referral Practices in Biochemical Genetics in the United States. Mol Genet Metab. 2009 Sep-Oct;98(1-2):149-51.

63 Vivatrat N, Barshop BA, Jones KL. Severe hypertriglyceridemia and recurrent pancreatitis in a girl with type Ia glycogen storage disease and type III hyperlipoproteinemia. Am J Med Genet A. 2009 149A(11):2557-2559.

64 Dohil R, Fidler M, Gangoiti JA, Kaskel F, Schneider JA, Barshop BA. Twice-daily cysteamine bitartrate therapy for children with cystinosis. J Pediatr. 2010 Jan;156(1):71-75.

65 Dohil R, Gangoiti JA, Cabrera BL, Fidler M, Schneider JA, Barshop BA. Long-term treatment of cystinosis in children with twice-daily cysteamine. J Pediatr. 2010 May;156(5):823-7. Epub 2010 Feb 6.

66 Gangoiti JA, Fidler M, Cabrera BL, Schneider JA, Barshop BA, Dohil R. Pharmacokinetics of enteric-coated cysteamine bitartrate in healthy adults: a pilot study. Br J Clin Pharmacol. 2010 Sep;70(3):376-82. PMID: 20716238

67 Nguyen KV, Naviaux RK, Patra S, Barshop BA, Nyhan WL. Novel mutations in the human MCCA and MCCB gene causing methylcrotonylglycinuria. Mol Genet Metab. 2011 Feb;102(2):218-21. PMID: 21071250

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