

BIOGRAPHICAL SKETCH

NAME	POSITION TITLE		
Vivian E. Shih, MD	Professor of Neurology Associate Neurologist, Pediatrician		
EDUCATION/TRAINING <i>(Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)</i>			
INSTITUTION AND LOCATION	DEGREE <i>(if applicable)</i>	YEAR(s)	FIELD OF STUDY
National Taiwan University, Taiwan	M.D.	1958	Premedical; medical
Edgewater Hospital, Chicago, Illinois	Internship	1959-1960	Rotating
Philadelphia General Hospital, Philadelphia	Residency	1960-1962	Pediatrics
Children's Hospital of Philadelphia	Fellowship	1962-1965	Pediatric Neurology
Massachusetts General Hospital, Boston	Res. Fellow	1965-1967	Neurology

A. Positions and Honors.

Positions and Employment

1967/1976/1982–	Assistant in Neurology/Assistant Neurologist/Associate Neurologist; Massachusetts General Hospital
1967/1970/1976–	Instructor/Assistant Professor/Associate Professor in Neurology; Harvard Medical School
1967–1980	Consultant & Co-principal Investigator, Mass. Metabolic Disorder Program, Mass. Dept. Public Health
1967–	Director, Amino Acid Disorders Laboratory, Neurology Service, Massachusetts General Hospital
1979/1984/1988–	Assistant Pediatrician/Associate Pediatrician/Pediatrician; Massachusetts General Hospital
1998–	Professor of Neurology, Harvard Medical School
1999–2001	Mentor, Clinical Investigator Training Program, Harvard-M.I.T. Div. of Health Sciences & Technology
1999–	Investigator, Clinical Nutrition Research Center at Harvard
1999–	Consultant, New England Newborn Screening Program, University of Massachusetts Medical School
2003-2005	Mentor, Clinical Investigator Training Program, Harvard-M.I.T. Div. of Health Sciences & Technology.

Honors

1985; 1992	Javits Neuroscience Investigator Award
1998	Teacher of the Year Award, Pediatric Neurology Service, Massachusetts General Hospital

Memberships

1969–	The Massachusetts Medical Society
1971–	The Society for Pediatric Research
1974	Ad Hoc Member, Metabolism Study Section, NIH
1976–	The American Society of Human Genetics
1978–	The Society for Inherited Metabolic Disorders
1979–	The American Pediatric Society
1980–	Society for the Study of Inborn Errors of Metabolism
1981–1983	Genetic Services Committee, The American Society of Human Genetics
1982–1988	Scientific Program Advisory Committee, NINCDS
1982	Ad Hoc Member, Special Study Section, NIH
1985	Site Visit Team, Biochemistry Study Section, NIH
1987	Ad Hoc Member, Metabolic Study Section, NIH
1988/1991/1996	Site Visit Team, NICHD
1988	Nominating Committee, American Board of Medical Genetics
1988–1990	Board of Directors, Society for Inherited Metabolic Disorders
1990	Special Reviewer, Medical Biochemistry Study Section, NIH
1992–1993	President, Society for Inherited Metabolic Disorders
1992–	Founding member, American College of Medical Genetics
1999	Special Reviewer, Mental Retardation Research Subcommittee, NICHD

B. Selected peer-reviewed publications (in chronological order)

(Publications selected from 146 peer-reviewed publications)

- Shih VE, Efron ML, Moser HW. Hyperornithinemia, hyperammonemia, and homocitrullinuria. A new disorder of amino acid metabolism associated with myoclonic seizures and mental retardation. *Amer J Dis Child* 1969; 117: 83-92.
- Shih VE, Berson EL, Gargiulo M. Reduction of hyperornithinemia with a low protein, low arginine diet and pyridoxine in patients with a deficiency of ornithine-ketoacid transaminase (OKT) activity and gyrate atrophy of the choroid and retina. *Clin Chim Acta* 1981;113:243-251.
- Shih VE, Mandell R, Herzfeld A. Defective ornithine metabolism in cultured skin fibroblasts from patients with the syndrome of hyperornithinemia, hyperammonemia and homocitrullinuria. *Clin Chim Acta.* 1982; 118: 149-157.
- Ramesh V, Shaffer MM, Allaire JM, Berson EL, Shih VE, Gusella JF. Investigation of gyrate atrophy using a cDNA clone for human ornithine aminotransferase. *DNA.* 1986; 5: 493-501.

- Johnson JL, Wuebbens MM, Mandell R, Shih VE. Molybdenum cofactor biosynthesis in humans: Identification of two complementation groups of cofactor-deficient patients and preliminary characterization of a diffusible molybdopterin precursor. *J Clin Invest*. 1989; 83:897-903.
- Shih VE, Axel SM, Tewksbury JC, Watkins D, Cooper BA, Rosenblatt DS. Defective lysosomal release of vitamin B₁₂ (cblF). A hereditary cobalamin metabolic disorder associated with sudden death. *Am J Med Genet* 1989; 33:555-563.
- Tuchman M, Knopman DS, Shih VE. Episodic hyperammonemia in adult siblings with hyperornithinemia, hyperammonemia, and homocitrullinuria syndrome. *Arch Neurol* 1990;47:1132-1137.
- Kvedar JC, Baden HP, Baden LA, Shih VE, Kolodny EH. Dietary Management Reverses Grooving and Abnormal Polarization of Hair Shafts in Argininosuccinase Deficiency. *Am J Med Gen* 1991;40:211-213.
- Ramesh V, Gusella J, Shih VE. Molecular pathology of gyrate atrophy of the choroid and retina due to ornithine aminotransferase deficiency. *Mol Biol Med*, 1991, 8:81-93.
- Shih VE, Laframboise R, Mandell R, Pichette J. Neonatal form of the Hyperornithinemia, Hyperammonemia and Homocitrullinuria (HHH) Syndrome and Prenatal Diagnosis. *Prenatal Diag* 1992;12:717-723.
- Hu F, Gu Z, Kozich V, Kraus JP, Ramesh V, Shih VE. Molecular basis of cystathionine β -synthase deficiency in B₆ responsive and nonresponsive homocystinuria. *Human Molecular Genetics* 1993;2:1857-1860.
- Shih VE, Fringer JM, Mandell R, Kraus JP, Heidenreich RA, Korson MS, Levy HL, Ramesh V. A missense mutation (I278T) in the cystathionine β -synthase gene prevalent in pyridoxine responsive homocystinuria and associated with mild clinical phenotype. *Am J Hum Genet* 1995;57:34-39.
- Zammarchi E, Ciani F, Pasquini E, Bonocore G, Shih VE, Donati MA. Neonatal Onset of Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome with Favourable Outcome. *J Pediatr*, 1997;131:440-443.
- Coughlin EM, Christensen E, Kunz PL, Krishnamoorthy KS, Walker V, Dennis NR, Chalmers RA, Elpeleg ON, Whelan D, Pollitt RJ, Ramesh V, Mandell R, Shih VE. Molecular analysis and prenatal diagnosis of human fumarase deficiency. *Molecular Genetics and Metabolism*, 1998;63:254-262.
- Shih VE, Safran AP, Ropper AH, Tuchman M. Ornithine Transcarbamylase Deficiency: Unusual Clinical Findings and Novel Mutation. *J Inher Metab Dis*. 1999 22:672-673.
- Huang TS, Yang W., Pereira AC, Craigen WJ, Shih VE. Cloning and characterization of Putative Human D-2-Hydroxyacid Dehydrogenase in Chromosome 9q. *Biochem and Biophys Res Commun*, 2000;268:298-301.
- Takeoka M, Soman TB, Shih VE, Caviness VS, Krishnamoorthy KS. Carbamyl Phosphate Synthetase 1 Deficiency: A Destructive Encephalopathy. *Pediatr Neurol*. 2001 24:193-9.
- Zytkovicz TH, Fitzgerald EF, Marsden D, Larson CA, Shih VE, Johnson DM, Strauss AW, Comeau AM, Eaton RB, Grady GF. Tandem mass spectrometric analysis for amino, organic, and fatty acid disorders in newborn dried blood spots: a two-year summary from the New England Newborn Screening Program. *Clin Chem*. 2001 47:1945-55.
- Kelly PJ, Rosand J, Kistler JP, Shih VE, Silveira S, Plomaritoglou A, Furie KL. Homocysteine, MTHFR 677C->T polymorphism, and risk of ischemic stroke: Results of a meta-analysis. *Neurology*, 2002; 59:529-36
- Waisbren SE, Read CY, Ampola M, Brewster TG, Demmer L, Greenstein R, Ingham CL, Korson M, Msall M, Pueschel S, Seashore M, Shih VE, Levy HL; New England Consortium of Metabolic Programs. Newborn screening compared to clinical identification of biochemical genetic disorders. *J Inher Metab Dis* 2002 Nov;25(7):599-600.
- Kelly PJ, Furie KL, Kistler JP, Barron M, Picard EH, Mandell R, Shih VE. Stroke in young patients with hyperhomocysteinemia due to cystathionine beta-synthase deficiency. *Neurology* 2003 60:275-9.
- Picker JD, Puga AC, Levy HL, Marsden D, Shih VE, Degirolami U, Ligon KL, Cederbaum SD, Kern RM, Cox GF. Arginase deficiency with lethal neonatal expression: evidence for the glutamine hypothesis of cerebral edema. *J Pediatr* 2003 142:349-52.
- Kelly PJ, Shih VE, Kistler JP, Barron M, Lee H, Mandell R, Furie KL. Low vitamin B6 but not homocyst(e)ine is associated with increased risk of stroke and transient ischemic attack in the era of folic acid grain fortification. *Stroke*. 2003 Jun;34(6):e51-4. Epub 2003 May 08.
- Waisbren SE, Albers S, Amato S, Ampola M, Brewster TG, Demmer L, Eaton RB, Greenstein R, Korson M, Larson C, Marsden D, Msall M, Naylor EW, Pueschel S, Seashore M, Shih VE, Levy HL. Effect of expanded newborn screening for biochemical genetic disorders on child outcomes and parental stress. *JAMA*. 2003;290:2564-72.
- Stoler JM, Sabry MA, Hanley C, Hoppel CL, Shih VE. Successful long-term treatment of hepatic carnitine palmitoyltransferase I deficiency and a novel mutation. *J Inher Metab Dis*. 2004 27(5):679-684.
- Kelly PJ, Kistler JP, Shih VE, Mandell R, Atassi N, Barron M, Lee H, Silveira S, Furie KL. Inflammation, homocysteine, and vitamin B6 status after ischemic stroke. *Stroke*. 2004 Jan;35(1):12-5. Epub 2003 Dec 04.
- Tan WH, Eichler FS, Hoda S, Lee MS, Baris H, Hanley CA, Grant PE, Krishnamoorthy KS, Shih VE. Isolated Sulite Oxidase Deficiency: A Case Report with a Novel Mutation and Review of the Literature. *Pediatrics* 2005;116:757-766.
- Cleary MA, Dorland L, de Koning TJ, Poll-The BT, Duran M, Mandell R, Shih VE, Berger R, Olpin SE, Besley GT. Ornithine aminotransferase deficiency: Diagnostic difficulties in neonatal presentation. *J Inher Metab Dis*. 2005;28:673-679.
- Browning MF, Levy HL, Wilkins-Haug LE, Larson C, Shih VE. Fetal Fatty Acid oxidation defects and maternal liver disease in pregnancy. *Obstet Gynecol*. 2006;107:115-20.
- Zeng WQ, Gao H, Brueton L, Hutchin T, Gray G, Chakrapani A, Olpin S, Shih VE. Fumarase deficiency caused by homozygous P131R mutation and paternal partial isodisomy of chromosome 1. *Am J Med Genet A*. 2006 May 1;140:1004-9
- Shih VE, Mandell R. Fumarase Deficiency. In: *GeneReviews at GeneTests: Medical Genetics Information Resource* [database online]. Copyright, University of Washington, Seattle, 1997-2006. Available at <http://www.genetests.org>. July 2006.
- Wattanasirichaigoon D, Khowsathit P, Visudtibhan A, Suthutvoravut U, Charoenpipop D, Kim SZ, Levy HL, Shih VE. Pericardial effusion in primary systemic carnitine deficiency. *J Inher Metab Dis*. 2006;29:589.
- Eichler F, Tan WH, Shih VE, Grant PE, Krishnamoorthy K. Proton magnetic resonance spectroscopy and diffusion-weighted imaging in isolated sulfite oxidase deficiency. *J Child Neurol*. 2006;21:801-5.