CURRICULUM VITAE

**University of Pittsburgh**

**School of Medicine**

**BIOGRAPHICAL**

**Name: Jerry Vockley, M.D., Ph.D.** **Birth Date:** 5/24/1956

**Business Address** University of Pittsburgh School of Medicine **E-Mail Address:** Jerry.vockley@chp.edu

 Children’s Hospital of Pittsburgh of UPMC

 Department of Medical Genetics

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 Pittsburgh, PA 15224

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# EDUCATION AND TRAINING

**UNDERGRADUATE:**

9/1974 – 5/1978 Carnegie Mellon University B.S. Biology

 Pittsburgh, PA

**GRADUATE:**

7/1978 – 5/1984 University of Pennsylvania M.D., Ph.D. Harry Harris, M.D. (Thesis advisor)

 School of Medicine Medicine, Genetics

**POST GRADUATE:**

6/1984 – 6/1987 University of ColoradoInternship and ResidenceEva Sujansky, M.D**.**

 Health Science Center Pediatrics

7/1987 – 6/1991 Yale University Fellowship Kay Tanaka, M.D. (Research advisor)

School of Medicine Pediatrics and Human Genetics

**APPOINTMENTS AND POSITIONS**

**ACADEMIC:**

6/1990 – 6/1991Yale University School of MedicineInstructor of Pediatrics and Human Genetics

7/1991 – 12/1995 Mayo Clinic Assistant Professor of Medical Genetics

1/1996 – 12/1999 Mayo Clinic Associate Professor of Medical Genetics

1/2000 – 12/2003 Mayo Clinic Professor of Medical Genetics

1/2000 – 12/2003 Mayo Clinic Chair, Department of Medical Genetics

1/2004 ­ present UPMC Pittsburgh Children’s Hospital Chief of Genetic and Genomic Medicine

 University of Pittsburgh School of Medicine Professor of Pediatrics

 University of Pittsburgh Graduate School Professor of Human Genetics

of Public Health

6/2015-present UPMC Children’s Hospital of Pittsburgh Director, Center for Rare Disease Therapies

11/2015-presemt University of Pittsburgh Cleveland Family Endowed Chair in Pediatric

 Research

## CERTIFICATION AND LICENSURE

**SPECIALTY CERTIFICATION:**

**Certifying Board Certificate Number Month/Year**

American Board of Pediatrics 041677 6/1989

American Board of Medical Genetics 870745 1/1990

 Clinical Genetic

 Biochemical/molecular

**MEDICAL OR OTHER PROFESSIONAL LICENSURE:**

**Licensing Board/State** PA MD423265 11/2005

**DEA** Federal **Certificate Number** BV10054935/31/2005

## MEMBERSHIPS IN PROFESSIONAL AND SCIENTIFIC SOCIETIES

**Organization**

American College of Medical Genetics and Genomics: Board of Directors 2021-2027

American College of Pediatrics

American Society for Clinical Investigation

Society for Inherited Metabolic Disorders: Board of Directors (1999-current); President Elect (2003-2004); President (2005- 2007)

American Society of Human Genetics

American Academy of Pediatrics

American Association for the Advancement of Science

Society for the Study of Inborn Errors of Metabolism

International Network for Fatty Acid Oxidation and Research: Founder and Chair

Association for Clinical Translational Science

**HONORS**

**Title of Award/Honor Month/Year**

T. Denny Sanford Visiting Lecturer 4/2023

Elected Fellow of the American Association for the Advancement of Science 6/2021

Organic Acidemia Association Lifetime Service Award 3/2021

Keynote Speaker, National PKU Alliance Annual Meeting 6/2020

Keynote Speaker, International Conference on Genetic and Metabolic Diseases (Cairo) 3/2020

Emmanuel Shapira Award. Molecular Genetics and Metabolism, Top Publication 2019 2/2020

Plenary Speaker, International Inborn Errors or Metabolism and Nutrition Congress (Istanbul) 4/2019

University of Pittsburgh Faculty Innovator 10/2019

Keynote Speaker, Jinan University Grand Rounds, China 11/2018

Invited Faculty, Recordati Course on IEMs, Rio de Janeiro 10/2018

Garrod Award (SSIEM, Best article published in JIMD for 2017) 10/2018

Invited Faculty, SSIEM Academy 4/2016-Present

Award for Excellence in Patient Experience, Wolff Center at UPMC 11/2017

Plenary Lecture. International Paediatric Medical Congress. Dubai, UAE. 11/2017

Mansen Visiting Professor. Boston Children’s Hospital. (Pediatric Grand Rounds) 10/2017

Plenary Lecture (2). International Congress of Inborn Errors of Metabolism, Rio de Janeiro. 9/2017

Haworth Visiting Professor, University of Manitoba and Children’s Hospital of Manitoba 4/2017

Pittsburgh Magazine “Top Doc” Award 2007-2020

Keynote speaker, 4th Annual Plain Population Translational Medicine Meeting 8/2016

Chair, International Network for Fatty Acid Oxidation Research and Management (INFORM) Annual Meeting 6/2016-Present

Aaron Michael Graham Memorial Lectureship, Children’s Hospital of Los Angeles 2/2016

Distinguished Editor, NIH Director’s New Innovator Award Editorial Board 1/2016

Postdoctoral trainee Emir Tas, 2016 Fellow Basic Research Award, Society for Pediatric Research 3/2016

March of Dimes Champion for Babies Award 10/2015

Founder and Chair, International Network for Fatty Acid Oxidation Disorders Research and Management 2014- Present

Ron Scott Memorial Lecturer, University of Washington, Seattle 2014

Program Chair, United Mitochondrial Disease Foundation Annual Meeting. 2014

Faculty Honors University of Pittsburgh, Pittsburgh 2014

Founding Faculty Member Asia Pacific Inborn Errors of Metabolism Academy 2014

Pediatric Grand Rounds Lecture, Children’s Hospital, University of Edmonton 2013

Chair, Sterol and Isoprenoid Research Collaborative Annual Meeting 2013

Chair, American College of Medical Genetics PKU Guideline Workgroup 2012-13

Chair, American College of Medical Genetics Therapeutics Committee 2012- Current

Postdoctoral Trainee Dr. Kristen Suhrie winner of Neil Buist outstanding trainee Award at SIMD annual meeting 2011

Pediatric Grand Rounds Lecture, Children’s Hospital, Halifax, Canada 2011

Pediatric Grand Rounds Lecture, University of California at San Diego 2010

Session chair SSIEM annual meeting (Istanbul) 9/2010

Michael Palmerie Memorial Lecturer, Children’s Hospital of Pittsburgh 7/2010

Plenary Session Chair Xth International Congress on IEMs, San Diego 9/2009

Keynote speaker, SSIEM annual meeting Garrod Day Celebration (Lisbon) 9/2008

Member HRSA Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children 2007-2011

Chair, HRSA Advisory Committee Technical Subcommittee 2007-2011

Chair International Organizing Committee Xth International Congress on IEMs, San Diego 2007- 2009

Member ACMG National Clinical Trial Collaborative Working Group 2006-Current

Member, PA State Newborn Screening Advisory Committee 2006-2010

Postdoctoral Trainee Dr. Miao He winner of Neil Buist outstanding trainee Award at SIMD annual meeting 2007

Past-President SIMD 2007-2009

Donough O’Brien Presidential Lecturer, SIMD Annual Meeting, Nashville, TN 2007

Founder and Managing Editor North American Metabolic Academy 2006- Current

Facilitator, HRSA evidence process working group 2006

Invited speaker and plenary session chair, IXth International Congress on Inborn Errors of Metabolism, Tokyo 2006

Member International Organizing Committee IXth International Congress on Inborn Errors of Metabolism, Tokyo 2006

Invited Speaker, Genetic Metabolic Dietician International Conference, Atlanta 2006

Invited Speaker, International Workshop on Fatty Acid Oxidation, Amsterdam 2005

President Society for Inherited Metabolic Disorders (SIMD) 2005-2007

Nominated to Board of Directors, American Board of Medical Genetics 2005

Elected to Pediatric Academic Society 2004

Research Educator of the Year, Mayo Clinic 2001

Elected to American Society for Clinical Investigation 1999

Young Investigator Award, Society for Inherited Metabolic Disorders 1989, 1990

Selected James Hudson Brown-Alexander B. Coxe Fellow, Yale School of Med. 1989

PHS Individual National Research Service Award, Yale School of Med. 1989

Medical Scientist Training Program Award, Univ. Penn. 1978-1984

University Honors, Carnegie-Mellon University 1974-1978

United States Steel Scholar, Carnegie-Mellon University 1974-1978

**PUBLICATIONS**

1. **Refereed articles**

1. Ray K, **Vockley J**, Harris H. (1984). Epitopes of human intestinal alkaline phosphatases, defined by monoclonal antibodies. *FEBS Letters.* **174:** 294-9.

2. **Vockley J**, Bednarz K, Harris H. (1984). Purification of monoclonal antibodies to human alkaline phosphatases by antigen-immunoaffinity chromatography: comparisons of their molar binding values. *J Immunol Methods.* **74:** 23-30.

3. **Vockley J**, D'Souza MP, Foster CJ, Harris H. (1984). Structural analysis of human adult and fetal alkaline phosphatases by cyanogen bromide peptide mapping. *Proc Natl Acad Sci U S A.* **81:** 6120-3. PMC391871.

4. **Vockley J**, Harris H. (1984). Purification of human adult and foetal intestinal alkaline phosphatases by monoclonal antibody immunoaffinity chromatography. *Biochemical Journal.* **217:** 535-41. PMC1153246.

5. **Vockley J**, Meyer LJ, Harris H. (1984). Differentiation of human adult and fetal intestinal alkaline phosphatases with monoclonal antibodies. *American Journal of Human Genetics.* **36:** 987-1000. PMC1684528.

6. Matsubara Y, Indo Y, Naito E, Ozasa H, Glassberg R, **Vockley J**, Ikeda Y, Kraus J, Tanaka K. (1989). Molecular cloning and nucleotide sequence of cDNAs encoding the precursors of rat long chain acyl-coenzyme A, short chain acyl-coenzyme A, and isovaleryl-coenzyme A dehydrogenases. Sequence homology of four enzymes of the acyl-CoA dehydrogenase family. *Journal of Biological Chemistry.* **264:** 16321-31.

7. **Vockley J**, Inserra JA, Breg WR, Yang-Feng TL. (1991). "Pseudomosaicism" for 4p- in amniotic fluid cell culture proven to be true mosaicism after birth. *American Journal of Medical Genetics.* **39:** 81-3.

8. **Vockley J**, Parimoo B, Tanaka K. (1991). Molecular characterization of four different classes of mutations in the isovaleryl-CoA dehydrogenase gene responsible for isovaleric acidemia. *American Journal of Human Genetics.* **49:** 147-57. PMC1683224.

9. Nagao M, **Vockley J**, Parimoo B, Tanaka K. (1992). Processing error of type-2 variant isovaleryl-CoA dehydrogenase precursor is caused by inefficient binding to the mitochondrial import receptor. *Progress in clinical and biological research.* **375:** 541-51.

10. **Vockley J**, Nagao M, Parimoo B, Tanaka K. (1992). The variant human isovaleryl-CoA dehydrogenase gene responsible for type II isovaleric acidemia determines an RNA splicing error, leading to the deletion of the entire second coding exon and the production of a truncated precursor protein that interacts poorly with mitochondrial import receptors. *Journal of Biological Chemistry.* **267:** 2494-501.

11. **Vockley J**, Parimoo B, Nagao M, Tanaka K. (1992). Identification of the molecular defects responsible for the various genotypes of isovaleric acidemia. *Progress in clinical and biological research.* **375:** 533-40.

12. **Vockley J**, Vockley CM, Lin SP, Tuchman M, Wu TC, Lin CY, Seashore MR. (1992). Normal N-acetylglutamate concentration measured in liver from a new patient with N-acetylglutamate synthetase deficiency: physiologic and biochemical implications. *Biochemical Medicine and Metabolic Biology.* **47:** 38-46.

13. Yokota I, Saijo T, **Vockley J**, Tanaka K. (1992). Impaired tetramer assembly of variant medium-chain acyl-coenzyme A dehydrogenase with a glutamate or aspartate substitution for lysine 304 causing instability of the protein. *Journal of Biological Chemistry.* **267:** 26004-10.

14. Kogekar N, Teebi AS, **Vockley J**. (1993). Sandrow syndrome of mirror hands and feet and facial abnormalities. *American Journal of Medical Genetics.* **46:** 126-8.

15. Wolfe L, Jethva R, Oglesbee D, **Vockley J**. Short-Chain Acyl-CoA Dehydrogenase Deficiency. in GeneReviews((R)) Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, Eds. Seattle (WA), 1993), .

16. Kim J-JP, Wang M, Paschke R, Djordjevic S, Bennett DW, **Vockley J**. Three dimensional structures of acyl-CoA dehydrogenases: structural basis of substrate specificity. in Flavins and Flavoproteins 1993 Yagi K, Eds. (Walter deGruyter, New York, 1994), pp. 273-82.

17. Rozen R, **Vockley J**, Zhou L, Milos R, Willard J, Fu K, Vicanek C, Low-Nang L, Torban E, Fournier B. (1994). Isolation and expression of a cDNA encoding the precursor for a novel member (ACADSB) of the acyl-CoA dehydrogenase gene family. *Genomics.* **24:** 280-7.

18. **Vockley J**. (1994). The changing face of disorders of fatty acid oxidation. *Mayo Clin Proc.* **69:** 249-57.

19. Battaile KP, Kim J-J, **Vockley J**. (1995). Identification of the catalytic residue and characterization of the substrate binding pocket of rat SCAD by *in vitro* mutagenesis and x-ray crystallography. *Am J Hum Genet.* **57:** A175.

20. Lindor NM, Karnes PS, Michels VV, Dewald GW, Goerss J, Jalal S, Jenkins RB, **Vockley J**, Thibodeau SN. (1995). Uniparental disomy in congenital disorders: a prospective study. *American Journal of Medical Genetics.* **58:** 143-6.

21. Mattson LR, Lindor NM, Goldman DH, Goodwin JT, Groover RV, **Vockley J**. (1995). Central Pontine Myelinolysis As a Complication Of Partial Ornithine Carbamoyl Transferase Deficiency. *American Journal of Medical Genetics.* **60:** 210-3.

22. Mohsen AW, **Vockley J**. (1995). Identification of the active site catalytic residue in human isovaleryl-CoA dehydrogenase. *Biochemistry.* **34:** 10146-52.

23. Battaile KP, Mohsen AW, **Vockley J**. (1996). Functional role of the active site glutamate-368 in rat short chain acyl-CoA dehydrogenase. *Biochemistry.* **35:** 15356-63.

24. Ushikubo S, Aoyama T, Kamijo T, Wanders RJA, Rinaldo P, **Vockley J**, Hashimoto T. (1996). Molecular characterization of mitochondrial trifunctional protein deficiency - formation of the enzyme complex is important for stabilization of both alpha- and beta-subunits. *American Journal of Human Genetics.* **58:** 979-88.

25. Willard J, Vicanek C, Battaile KP, Van Veldhoven PP, Fauq AH, Rozen R, **Vockley J**. (1996). Cloning of a cDNA for short/branched chain acyl-Coenzyme A dehydrogenase from rat and characterization of its tissue expression and substrate specificity. *Archives of Biochemistry and Biophysics.* **331:** 127-33.

26. Dakoji S, Shin I, Battaile KP, **Vockley J**, Liu HW. (1997). Redesigning the active-site of an acyl-CoA dehydrogenase: new evidence supporting a one-base mechanism. *Bioorg Med Chem.* **5:** 2157-64.

27. Schaller RA, Mohsen AW, **Vockley J**, Thorpe C. (1997). Mechanism-based inhibitor discrimination in the acyl-CoA dehydrogenases. *Biochemistry.* **36:** 7761-8.

28. Thorpe C, Schaller RA, Mohsen A-W, **Vockley J** The acyl-CoA dehydrogenases: Some mechanistic aspects (University of Calgary Press,, Calgary, Canada, 1997).

29. Tiffany KA, Roberts DL, Wang M, Paschke R, Mohsen AWA, **Vockley J**, Kim JJP. (1997). Structure of human isovaleryl-coA dehydrogenase at 2.6 angstrom resolution - basis for substrate specificity. *Biochemistry.* **36:** 8455-64.

30. Tiffany KA, Wang M, Paschke R, Mohsen A-W, **Vockley J**, Kim JJ. (1997). Structural basis for substrate specificity in acyl-CoA dehydrogenases: What makes isovaleryl-CoA dehydrogenase specific for a branched chain substrate? *Flavins and Flavoproteins 1996.* 649-52.

31. Battaile KP, McBurney M, Van Veldhoven PP, **Vockley J**. (1998). Human long chain, very long chain and medium chain acyl-CoA dehydrogenases are specific for the S-enantiomer of 2- methylpentadecanoyl-CoA. *Biochimica et Biophysica Acta.* **1390:** 333-8.

32. Binzak B, Willard J, **Vockley J**. (1998). Identification of the catalytic residue of human short/branched chain acyl-CoA dehydrogenase by in vitro mutagenesis. *Biochimica et Biophysica Acta.* **1382:** 137-42.

33. Kurtz DM, Rinaldo P, Rhead WJ, Tian L, Millington DS, **Vockley J**, Hamm DA, Brix AE, Lindsey JR, Pinkert CA, O'Brien WE, Wood PA. (1998). Targeted disruption of mouse long-chain acyl-CoA dehydrogenase gene reveals crucial roles for fatty acid oxidation. *Proc Natl Acad Sci U S A.* **95:** 15592-7. PMC28088.

34. Mohsen AW, Anderson BD, Volchenboum SL, Battaile KP, Tiffany K, Roberts D, Kim JJ, **Vockley J**. (1998). Characterization of molecular defects in isovaleryl-CoA dehydrogenase in patients with isovaleric acidemia. *Biochemistry.* **37:** 10325-35.

35. O'Sullivan DA, Torres VE, de Groen PC, Batts KP, King BF, **Vockley J**. (1998). Hepatic lymphangiomatosis mimicking polycystic liver disease. *Mayo Clin Proc.* **73:** 1188-92.

36. Schievink WI, Wijdicks EF, Michels VV, **Vockley J**, Godfrey M. (1998). Heritable connective tissue disorders in cervical artery dissections: a prospective study. *Neurology.* **50:** 1166-9.

37. Babovic-Vuksanovic D, Patterson MC, Schwenk WF, O'Brien JF, **Vockley J**, Freeze HH, Mehta DP, Michels VV. (1999). Severe hypoglycemia as a presenting symptom of carbohydrate-deficient glycoprotein syndrome. *Journal of Pediatrics.* **135:** 775-81.

38. Mathur A, Sims HF, Gopalakrishnan D, Gibson B, Rinaldo P, **Vockley J**, Hug G, Strauss AW. (1999). Molecular heterogeneity in very-long-chain acyl-CoA dehydrogenase deficiency causing pediatric cardiomyopathy and sudden death. *Circulation.* **99:** 1337-43.

39. Mohsen A-W, **Vockley J**. Biochemical characteristics of recombinant human isovaleryl-CoA dehydrogenase pre-treated with ethylenediaminetetraacetate. in Flavins and Flavoproteins 1999 Ghisla S, Kroneck P, Macheroux P, Sund H, Eds. (Rudolf Weber, New York, 1999), pp. 515-8.

40. Moolenaar SH, Poggi-Bach J, Engelke UF, Corstiaensen JM, Heerschap A, de Jong JG, Binzak BA, **Vockley J**, Wevers RA. (1999). Defect in dimethylglycine dehydrogenase, a new inborn error of metabolism: NMR spectroscopy study. *Clinical Chemistry.* **45:** 459-64.

41. Tein I, Haslam RH, Rhead WJ, Bennett MJ, Becker LE, **Vockley J**. (1999). Short-chain acyl-CoA dehydrogenase deficiency: a cause of ophthalmoplegia and multicore myopathy. *Neurology.* **52:** 366-72.

42. Binzak BA, **Vockley J**G, Jenkins RB, **Vockley J**. (2000). Structure and analysis of the human dimethylglycine dehydrogenase gene. *Mol Genet Metab.* **69:** 181-7.

43. Gibson KM, Burlingame TG, Hogema B, Jakobs C, Schutgens RB, Millington D, Roe CR, Roe DS, Sweetman L, Steiner RD, Linck L, Pohowalla P, Sacks M, Kiss D, Rinaldo P, **Vockley J**. (2000). 2-Methylbutyryl-coenzyme A dehydrogenase deficiency: a new inborn error of L-isoleucine metabolism. *Pediatric Research.* **47:** 830-3.

44. Lea W, Abbas AS, Sprecher H, **Vockley J**, Schulz H. (2000). Long-chain acyl-CoA dehydrogenase is a key enzyme in the mitochondrial β-oxidation of unsaturated fatty acids. *Bba-Mol Cell Biol L.* **1485:** 121-8.

45. Reinard T, Janke V, Willard J, Buck F, Jacobsen H-J, **Vockley J**. (2000). Cloning of a gene for an acyl-CoA dehydrogenase from Pisum sativum L. and purification and characterization of its product as an isovaleryl-CoA dehydrogenase. *J Biol Chem.* **275:** 33738-43.

46. Treacy EP, Lambert DM, Barnes R, Boriack RL, **Vockley J**, O'Brien LK, Jones PM, Bennett MJ. (2000). Short-chain hydroxyacyl-coenzyme A dehydrogenase deficiency presenting as unexpected infant death: A family study. *Journal of Pediatrics.* **137:** 257-9.

47. **Vockley J**, Mohsen al WA, Binzak B, Willard J, Fauq A. (2000). Mammalian branched-chain acyl-CoA dehydrogenases: molecular cloning and characterization of recombinant enzymes. *Methods Enzymol.* **324:** 241-58.

48. **Vockley J**, Rinaldo P, Bennett MJ, Matern D, Vladutiu GD. (2000). Synergistic heterozygosity: disease resulting from multiple partial defects in one or more metabolic pathways. *Mol Genet Metab.* **71:** 10-8.

49. **Vockley J**, Rogan PK, Anderson BD, Willard J, Seelan RS, Smith DI, Liu W. (2000). Exon skipping in IVD RNA processing in isovaleric acidemia caused by point mutations in the coding region of the IVD gene. *American Journal of Human Genetics.* **66:** 356-67. PMC1288088.

50. Volchenboum SL, **Vockley J**. (2000). Mitochondrial import and processing of wild type and type III mutant isovaleryl-CoA dehydrogenase. *Journal of Biological Chemistry.* **275:** 7958-63.

51. Binzak BA, Wevers RA, Moolenaar SH, Lee YM, Hwu WL, Poggi-Bach J, Engelke UF, Hoard HM, **Vockley J**G, **Vockley J**. (2001). Cloning of dimethylglycine dehydrogenase and a new human inborn error of metabolism, dimethylglycine dehydrogenase deficiency. *American Journal of Human Genetics.* **68:** 839-47. PMC1275637.

52. Corydon MJ, **Vockley J**, Rinaldo P, Rhead WJ, Kjeldsen M, Winter V, Riggs C, Babovic-Vuksanovic D, Smeitink J, De Jong J, Levy H, Sewell AC, Roe C, Matern D, Dasouki M, Gregersen N. (2001). Role of common gene variations in the molecular pathogenesis of short-chain acyl-CoA dehydrogenase deficiency. *Pediatric Research.* **49:** 18-23.

53. Cox KB, Hamm DA, Millington DS, Matern D, **Vockley J**, Rinaldo P, Pinkert CA, Rhead WJ, Lindsey JR, Wood PA. (2001). Gestational, pathologic and biochemical differences between very long-chain acyl-CoA dehydrogenase deficiency and long-chain acyl-CoA dehydrogenase deficiency in the mouse. *Human Molecular Genetics.* **10:** 2069-77.

54. Hoard HM, Benson LM, **Vockley J**, Naylor S. (2001). Microelectrospray ionization analysis of noncovalent interactions within the electron transferring flavoprotein. *Biochemical and Biophysical Research Communications.* **282:** 297-305.

55. Matern D, Hart P, Murtha AP, **Vockley J**, Gregersen N, Millington DS, Treem WR. (2001). Acute fatty liver of pregnancy associated with short-chain acyl-coenzyme A dehydrogenase deficiency. *Journal of Pediatrics.* **138:** 585-8.

56. Mohsen AW, Navarette B, **Vockley J**. (2001). Identification of Caenorhabditis elegans isovaleryl-CoA dehydrogenase and structural comparison with other acyl-CoA dehydrogenases. *Mol Genet Metab.* **73:** 126-37.

57. Volchenboum SL, Mohsen AW, Kim JJ, **Vockley J**. (2001). Arginine 387 of human isovaleryl-CoA dehydrogenase plays a crucial role in substrate/product binding. *Mol Genet Metab.* **74:** 226-37.

58. Willard JM, Reinard T, Mohsen A, **Vockley J**. (2001). Cloning of genomic and cDNA for mouse isovaleryl-CoA dehydrogenase (IVD) and evolutionary comparison to other known IVDs. *Gene.* **270:** 253-7.

59. Battaile KP, Molin-Case J, Paschke R, Wang M, Bennett D, **Vockley J**, Kim JJ. (2002). Crystal structure of rat short chain acyl-CoA dehydrogenase complexed with acetoacetyl-CoA: comparison with other acyl-CoA dehydrogenases. *Journal of Biological Chemistry.* **277:** 12200-7.

60. Nguyen TV, Andresen BS, Corydon TJ, Ghisla S, Abd-El Razik N, Mohsen AW, Cederbaum SD, Roe DS, Roe CR, Lench NJ, **Vockley J**. (2002). Identification of isobutyryl-CoA dehydrogenase and its deficiency in humans. *Mol Genet Metab.* **77:** 68-79.

61. Nguyen TV, Riggs C, Babovic-Vuksanovic D, Kim YS, Carpenter JF, Burghardt TP, Gregersen N, **Vockley J**. (2002). Purification and characterization of two polymorphic variants of short chain acyl-CoA dehydrogenase reveal reduction of catalytic activity and stability of the Gly185Ser enzyme. *Biochemistry.* **41:** 11126-33.

62. **Vockley J**, Singh RH, Whiteman DA. (2002). Diagnosis and management of defects of mitochondrial beta-oxidation. *Current opinion in clinical nutrition and metabolic care.* **5:** 601-9.

63. **Vockley J**, Whiteman DA. (2002). Defects of mitochondrial beta-oxidation: a growing group of disorders. *Neuromuscular disorders : NMD.* **12:** 235-46.

64. He M, Burghardt TP, **Vockley J**. (2003). A novel approach to the characterization of substrate specificity in short/branched chain Acyl-CoA dehydrogenase. *Journal of Biological Chemistry.* **278:** 37974-86.

65. Koeberl DD, Young SP, Gregersen NS, **Vockley J**, Smith WE, Benjamin DK, Jr., An Y, Weavil SD, Chaing SH, Bali D, McDonald MT, Kishnani PS, Chen YT, Millington DS. (2003). Rare disorders of metabolism with elevated butyryl- and isobutyryl-carnitine detected by tandem mass spectrometry newborn screening. *Pediatric Research.* **54:** 219-23.

66. Matern D, He M, Berry SA, Rinaldo P, Whitley CB, Madsen PP, van Calcar SC, Lussky RC, Andresen BS, Wolff JA, **Vockley J**. (2003). Prospective diagnosis of 2-methylbutyryl-CoA dehydrogenase deficiency in the Hmong population by newborn screening using tandem mass spectrometry. *Pediatrics.* **112:** 74-8.

67. Nishigaki Y, Tadesse S, Bonilla E, Shungu D, Hersh S, Keats BJ, Berlin CI, Goldberg MF, **Vockley J**, DiMauro S, Hirano M. (2003). A novel mitochondrial tRNA(Leu(UUR)) mutation in a patient with features of MERRF and Kearns-Sayre syndrome. *Neuromuscular disorders : NMD.* **13:** 334-40.

68. Pedersen CB, Bross P, Winter VS, Corydon TJ, Bolund L, Bartlett K, **Vockley J**, Gregersen N. (2003). Misfolding, degradation, and aggregation of variant proteins. The molecular pathogenesis of short chain acyl-CoA dehydrogenase (SCAD) deficiency. *Journal of Biological Chemistry.* **278:** 47449-58.

69. Rao KS, Albro M, **Vockley J**, Frerman FE. (2003). Mechanism-based inactivation of human glutaryl-CoA dehydrogenase by 2-pentynoyl-CoA: rationale for enhanced reactivity. *Journal of Biological Chemistry.* **278:** 26342-50.

70. Seidel J, Streck S, Bellstedt K, Vianey-Saban C, Pedersen CB, **Vockley J**, Korall H, Roskos M, Deufel T, Trefz KF, Sewell AC, Kauf E, Zintl F, Lehnert W, Gregersen N. (2003). Recurrent vomiting and ethylmalonic aciduria associated with rare mutations of the short-chain acyl-CoA dehydrogenase gene. *J Inherit Metab Dis.* **26:** 37-42.

71. Battaile KP, Nguyen TV, **Vockley J**, Kim JJ. (2004). Structures of isobutyryl-CoA dehydrogenase and enzyme-product complex: comparison with isovaleryl- and short-chain acyl-CoA dehydrogenases. *Journal of Biological Chemistry.* **279:** 16526-34.

72. Chu G, Kerr JP, Mitton B, Egnaczyk GF, Vazquez JA, Shen M, Kilby GW, Stevenson TI, Maggio JE, **Vockley J**, Rapundalo ST, Kranias EG. (2004). Proteomic analysis of hyperdynamic mouse hearts with enhanced sarcoplasmic reticulum calcium cycling. *FASEB J.* **18:** 1725-7.

73. Ensenauer R, **Vockley J**, Willard JM, Huey JC, Sass JO, Edland SD, Burton BK, Berry SA, Santer R, Grunert S, Koch HG, Marquardt I, Rinaldo P, Hahn S, Matern D. (2004). A common mutation is associated with a mild, potentially asymptomatic phenotype in patients with isovaleric acidemia diagnosed by newborn screening. *American Journal of Human Genetics.* **75:** 1136-42. PMC1182150.

74. Hoard-Fruchey HM, Goetzman E, Benson L, Naylor S, **Vockley J**. (2004). Mammalian electron transferring flavoprotein.flavoprotein dehydrogenase complexes observed by microelectrospray ionization-mass spectrometry and surface plasmon resonance. *Journal of Biological Chemistry.* **279:** 13786-91. PMID: 14744856.

75. Nasser I, Mohsen AW, Jelesarov I, **Vockley J**, Macheroux P, Ghisla S. (2004). Thermal unfolding of medium-chain acyl-CoA dehydrogenase and iso(3)valeryl-CoA dehydrogenase: study of the effect of genetic defects on enzyme stability. *Biochimica et Biophysica Acta.* **1690:** 22-32.

76. Yu W, Liang X, Ensenauer RE, **Vockley J**, Sweetman L, Schulz H. (2004). Leaky beta-oxidation of a trans-fatty acid: incomplete beta-oxidation of elaidic acid is due to the accumulation of 5-trans-tetradecenoyl-CoA and its hydrolysis and conversion to 5-trans-tetradecenoylcarnitine in the matrix of rat mitochondria. *Journal of Biological Chemistry.* **279:** 52160-7.

77. Ensenauer R, He M, Willard JM, Goetzman ES, Corydon TJ, Vandahl BB, Mohsen AW, Isaya G, **Vockley J**. (2005). Human acyl-CoA dehydrogenase-9 plays a novel role in the mitochondrial beta-oxidation of unsaturated fatty acids. *Journal of Biological Chemistry.* **280:** 32309-16.

78. Goetzman ES, Mohsen AW, Prasad K, **Vockley J**. (2005). Convergent evolution of a 2-methylbutyryl-CoA dehydrogenase from isovaleryl-CoA dehydrogenase in Solanum tuberosum. *Journal of Biological Chemistry.* **280:** 4873-9.

79. Saenger AK, Nguyen TV, **Vockley J**, Stankovich MT. (2005). Thermodynamic regulation of human short-chain acyl-CoA dehydrogenase by substrate and product binding. *Biochemistry.* **44:** 16043-53.

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84. Subgroup Analysis of Pegvaliase in Adults with Phenylketonuria in Phase 3 PRISM Studies: Evaluating Efficacy and Safety Based on Previous Sapropterin Response or Dietary Phenylalanine Intake. SIMD Annual Meeting. 2019
85. IVD deficient fibroblasts show improvement in phenotype with epigallocatechin gallate treatment. SIMD Annual Meeting 2019
86. Treatment of bioenergetic dysfunction in propionic acidemia. SIMD Annual Meeting. 2019
87. A Phase 1/2a, First-in-human, Oral Single and Multiple Dose-Escalation, Randomized, Double-blind, Placebo-controlled Study of SYNB1618 in Healthy Adult Volunteers and Adult Subjects with Phenylketonuria to Evaluate Safety, Tolerability, Kinetics, and Pharmacodynamics. SSIEM. Annual Meeting. 2019.
88. A comparative multi-omic study to characterize VLCAD patient response to triheptanoin vs. medium chain triglyceride management. SSIEM Annual Meeting. 2019
89. Successful Orthotopic Heart Transplantation in CPTII deficiency. SSIEM Annual Meeting. 2019
90. Interim results from an open-label, long-term extension study to evaluate the safety and efficacy of triheptanoin (UX007) in LC-FAOD. SSIEM Annual Meeting. 2019
91. Fatty acid oxidation-respiratory chain proteins and their interactions are abnormal in end stage cardiomyopathy. SSIEM Annual Meeting. 2019
92. Assessing Referral of Plain Community Members for Genetic Services at UPMC Children’s Hospital of Pittsburgh: A Quality Improvement Study. ACMG Annual Meeting. 2020
93. Long-term follow-up (up to 24 months) of glycerol phenylbutyrate for patients 2 months to 2 years with urea cycle disorders. SIMD Annual Meeting. 2020
94. Phase 3 prism studies: efficacy and safety of pegvaliase 60 mg dose in adult patients with phenylketonuria. SIMD Annual Meeting. 2020
95. Rare metabolic disorders of energy dysfunction in patients with treatment-resistant depression. SIMD Annual Meeting. 2020
96. Interim results from an open-label, long-term extension study to evaluate the safety and efficacy of triheptanoin (UX007) in long-chain fatty acid oxidation disorders (LC-FAODs). SIMD Annual Meeting. 2020
97. Comparison of distal and proximal inhibition of the leucine pathway in IVD deficient cells. SIMD Annual Meeting. 2020
98. Compassionate use of triheptanoin in patients with pyruvate carboxylase deficiency. SIMD Annual Meeting. 2020
99. Potential Therapies for Mitochondrial Bioenergetics Dysfunction in Fibroblasts from a 2-Hydroxyglutaric Aciduria Patient. SIMD Annual Meeting. 2020
100. Metabolic analysis reveals evidence for branched chain amino acid catabolism crosstalk and the potential for improved treatment of organic aciduria. SIMD Annual Meeting. 2020
101. Outcomes and genotype-phenotype correlations in 45 individuals with *HADHA* and *HADHB* mutations diagnosed by NBS and enrolled in the Inborn Errors of Metabolism – Information System database. ACMG 2021 Meeting. Virtual fomrmat
102. **C**linical Outcomes of Major Clinical Events and Emergency Triheptanoin in Critically Ill Patients With Long-chain Fatty Acid Oxidation Disorders. ACMG 2021 Annual Meeting. Virtual format.
103. Phenom: A prospective clinical study on the clinical impact of phenylketonuria in adults. ACMG Annual 2021 Meeting. Virtual format.
104. Comparison of triheptanoin and succinate as anaplerotic treatment in a very long-chain acyl-CoA dehydrogenase deficiency mouse model. SSIEM 2021 Annual Meeting, Sydney Australia
105. Comparison of triheptanoin and succinate as anaplerotic treatment in a very long-chain acyl-CoA dehydrogenase deficiency mouse model. SSIEM 2021 Annual Meeting, Sydney Australia
106. Mitochondrial-targeted reactive oxygen species scavenger JP4-039 prevents oxidative stress in cerebral cortex and striatum of glutaryl-CoA dehydrogenase-deficient mice. SSIEM 2021 Annual Meeting, Sydney Australia
107. Characterization of variants of uncertain significance in isovaleryl-CoA dehydrogenase identified through newborn screening: An approach for faster analysis. SSIEM 2021 Annual Meeting, Sydney Australia.
108. Reduction in Plasma Phenylalanine Levels in Patients with Phenylketonuria with Live Bacterial Therapeutic SYNB1618. SSIEM 2021 Annual Meeting, Sydney Australia.
109. Restoration of interaction between FAO and ETC proteins in VLCAD and VLCAD-deficient mice mitochondria by addition of recombinant VLCAD. SSIEM 2021 Annual Meeting, Sydney Australia.
110. Defining Therapeutic Options for Combined D,L-2 Hydroxyglutaric Aciduria. SSIEM 2021 Annual Meeting, Sydney Austrailia.
111. Synthetic Messenger RNA Rescues Very Long-Chain Acyl-CoA Dehydrogenase Deficiency in Fibroblasts from Patients and a Murine Model. UMDF 2021 Annual Meeting. Virtual format.
112. Correlation of genotype and molecular phenotype in mitochondrial trifunctional protein deficiency. ACMG 2022 Annual Meeting. Nashville, TN.
113. An open-label, phase 1/2 trial of gene therapy 4D-310 in adult males with Fabry disease. WORLD 2022 Annual Meeting. San Diego, CA.
114. Cardiolipin Remodeling Deregulation and Mitochondrial Bioenergetics Alterations in Trifunctional Protein (TFP) Deficiency. SIMD 2022 Annual Meeting. Orlando, FL
115. Characterization of variants of uncertain significance in very long chain acyl-CoA dehydrogenase identified through newborn screening. SIMD 2022 Annual Meeting. Orlando, FL
116. Improvement in major clinical events (MCEs) in children treated with triheptanoin for long-chain fatty acid oxidation disorders (LC-FAOD). SIMD 2022 Annual Meeting. Orlando, FL
117. Oxidative stress and mitochondrial respiration impairment induced by sulfite in rat brain are prevented by the mitochondria-targeted antioxidant XJB-5-131. SSIEM 2022 Annual Meeting. Freyburg, Germany.
118. Restoring succinyllysine antigenic signal and improving O2 consumption of CPT II deficient cells treated with anaplerotic compounds. SSIEM 2022 Annual Meeting. Freyburg, Germany.
119. A new class of anaplerotic compounds restores cellular lysine succinylation and antigenic signal. SSIEM 2022 Annual Meeting. Freyburg, Germany.
120. An Open-Label Study To Determine The Safety And Tolerability Of 12 Weeks’ Treatment With Oral REN001 In Subjects With Fatty Acid Oxidation Disorders(FAOD). SSIEM 2022 Annual Meeting. Freyburg, Germany.
121. A CRISPR/Cas9 genome-edited PAH-deficient cell line for studying PKU. SSIEM 2022 Annual Meeting. Freyburg, Germany.
122. **Patents Awarded and Pending**
123. Antioxidant Therapy of Inborn Errors of Fatty Acid Oxidation and Oxidative Phosphorylation (5/5/2016)
124. Metabolomics of Treatment Refractory Depression and Suicidal Behavior (2/10/2016)
125. Use of a knock-out Yucatan mini-pig model for characterization and treatment of phenylalanine hydroxylase (PAH) deficiency (phenylketonuria; PKU) (1/8/2016)
126. Novel anaplerotic agents for treatment of disorders of propionate and long chain fat metabolism (12/14/2015)
127. Use of gene transcription modifiers for treatment of rhabdomyolyis due to fatty acid oxidation and of unknown cause (10/8/2015)
128. Treatment of Rhabdomyolysis due to Fatty Acid Oxidation Disorders and Other Causes with Bendavia (10/8/2015)
129. Development of acyl-CoA dehydrogenases micro/nano enzyme assay for clinical diagnosis and newborn screening (6/29/2015)
130. Triheptanoin IND (Awarded 1/19/2011)
131. Treatment of medium chain acyl-CoA dehydrogenase deficiency (Awarded 10/12/2011)

# PROFESSIONAL ACTIVITIES

**TEACHING:**

University of Pittsburgh Graduate School of Public Health, Department of Human Genetics

Lecturer in first year Health Human Genetics course (2005-current)

University of Pittsburgh School of Medicine

 Director, UPMC Medical Biochemistry Fellowship (2010-2020)

 Lecturer in first year Biochemistry course (2005-current)

 Lecturer in first year Medical Genetics course (2007-current)

 Lecturer in Molecular Medicine course (2007-2019)

 Clinical supervisor Medical Genetics rotation for pediatric residents and genetic counseling students (2004-current)

Mayo Medical and Graduate School Teaching:

Medical Genetics (Medical School 1992)

Cell and Molecular Biology (Medical School 1993)

Biochemistry 8001: Enzyme Kinetics, Mechanism and Structure: Graduate School (1994–97)

Molecular Biology 8102: Regulation of Protein Synthesis: Graduate School (1994–98)

Molecular Biology 8600: Human Genetics: Co-course director, Graduate School (1994, 1995)

Graduate School Core Curriculum: Course director, Molecular Genetics (1999-2003)

Molecular Genetics Journal Club: Course director (1998-2003)

Mayo Clinical Faculty Genome Education Initiative (1999-2002) Program founder.

Doctoral and Postdoctoral (non-clinical) Trainees

Al-Walid A. Mohsen. Postdoctoral Trainee. Mayo Clinic. 1992-1995.

Kevin Pruitt Battaile. Ph.D. Trainee. Mayo Graduate School. 1992-1996.

Samuel L. Volchenboum. M.D., Ph.D. Trainee. Mayo Graduate School. 1993-1998.

Barbara A. Binzak. Ph.D. Trainee. Mayo Graduate School 1994-1999. Selected as outstanding predoctoral trainee at the 1998 annual meeting of the American Society of Human Genetics.

Heidi Hoard. Ph.D. Trainee. Mayo Graduate School 1998-2002. Mayo Clinic Medical School

Tien Nguyen, Ph.D. Postdoctoral Trainee. Mayo Clinic. 1999-2004. Mayo Clinic Medical School

Dusica Babovic-Vuksanovic, M.D. Postdoctoral Trainee. Mayo Clinic 1997-2000.

Miao He, Ph.D. Postdoctoral Trainee. Mayo Clinic. 2000-2006. Selected as outstanding postdoctoral trainee at the 2011 annual meeting of the Society for Metabolic Disorders.

Eric Goetzman, Ph.D., Postdoctoral Trainee, Mayo Clinic. 2001- 2007.

Zuzana Swigonova, Ph.D. Postdoctoral Trainee, University of Plattsburgh, 2005-2007

Kristen Harris Suhrie, M.D., Postdoctoral Fellow in Pediatrics, 2009-2011.

Manuel Schiff, M.D., Postdoctoral Fellow, Fullbright Scholar, 2011-2013.

Kristen Skvorak Postdoctoral Fellow, 2012-current

Heejung Kang. Ph.D. Trainee. University of Pittsburgh Graduate School of Public Health. 2010-2014

Wei Wang, Ph.D. Trainee. University of Pittsburgh Graduate School of Public Health. 2010-2014

Kaitlyn Kormanik Bloom. Ph.D. Trainee. University of Pittsburgh Graduate School of Public Health. 2010-2014

Chikara Otsubo, M.D., Ph.D., Postdoctoral Trainee, University of Pittsburgh, 2013-2016

Lina Ghaloul-Gonzalez, M.D., Postdoctoral Trainee, University of Pittsburgh, 2013-2016

Steve McCalley, Ph.D. Trainee. University of Pittsburgh Graduate School of Public Health. 2014-2018

Andrew Sinsheimer, Ph.D. Trainee. University of Pittsburgh Graduate School of Public Health. 2014-2019

Guilhian Leipnitz, Ph.D., Visiting Scientist, Universidade Federal do Rio Grande do Sul, 2015-2016

Bianca Seminotti, Ph.D., Postdoctoral Trainee, University of Pittsburgh, 2015-2017

Olivia Dannibale, Ph.D. Trainee, University of Pittsburgh Graduate School of Public Health, 2018-2022

Yuleng Phua, Pd.D. Trainee. University of Pittsburgh School of Medicine. 20

Eduardo Vieira Neto, Ph.D., Clinical Biochemical Fellow, University of Pittsburgh School of Medicine, 2019-2022

Frances Johnson, Ph.D. Trainee, University of Pittsburgh Graduate School of Public Health, 2021- current

Justin Dutta, Ph.D. Trainee, University of Pittsburgh Graduate School of Public Health, 2022- current

Suja Somanadhan, Ph.D., Postdoctoral Trainee, Fullbright Scholar, University of Pittsburgh School of Medicine, 2022

Doctoral Candidate Thesis Committees

Kevin Battaile (Biochemistry and Molecular Biology, Mayo Clinic Medical School)

Samuel Volchenboum (Biochemistry and Molecular Biology, Mayo Clinic Medical School)

Barbara Binzak (Biochemistry and Molecular Biology, Mayo Clinic Medical School)

Monique Purdock (Biochemistry and Molecular Biology, Mayo Clinic Medical School)

Brian Johnson (Biochemistry and Molecular Biology, Mayo Clinic Medical School)

Marcie Mikesell (Biochemistry and Molecular Biology, Mayo Clinic Medical School)

Scott Kuhns (Biochemistry and Molecular Biology, Mayo Clinic Medical School)

Lori Lebruska (Biochemistry and Molecular Biology, Mayo Clinic Medical School)

Kurt Krummel (Biochemistry and Molecular Biology, Mayo Clinic Medical School)

Heidi Hoard (Biochemistry and Molecular Biology, Mayo Clinic Medical School)

Heather O’Neil (Biochemistry and Molecular Biology, Mayo Clinic Medical School)

Rachaneekorn Punyashthiti (Molecular Biology, Mayo Clinic Medical School)

 Sarah Van Driest (Molecular Biology, Mayo Clinic Medical School)

Kristen Skvorik (Biochemistry, University of Pittsburgh School of Medicine)

Wei Wang (Human Genetics, University of Pittsburgh Graduate School of Public Health)

Heejung Kang (Human Genetics, University of Pittsburgh Graduate School of Public Health)

Kaitlyn Kormanik (Human Genetics, University of Pittsburgh Graduate School of Public Health)

Ryan McAndrew (Biochemistry, Medical College of Wisconsin)

Megan Beck (Human Genetics, University of Pittsburgh Graduate School of Public Health)

Steve McCalley (Human Genetics, University of Pittsburgh Graduate School of Public Health)

Andrew Sinsheimer (Human Genetics, University of Pittsburgh Graduate School of Public Health)

Olivia Dannibale (Human Genetics, University of Pittsburgh Graduate School of Public Health)

Keaton Solo (Human Genetics, University of Pittsburgh Graduate School of Public Health)

National and International Teaching

 North American Metabolic Academy (2006- current; Founder and Director)

 SSIEM (European) Academy (2012- current; Course faculty)

 Asia Pacific Inborn Errors of Metabolism Course (2014-2015; Course faculty)

 Recordati Academy (2013-current; Intermittent course faculty)

 South American Inborn Errors of Metabolism Course (2014-current; Intermittent course faculty)

**RESEARCH:**

**RESEARCH FUNDING**

**Vockley, Gerard**

**ACTIVE**

**ACTIVE**

\*Title: Characterization of Branched Chain Amino Acid Metabolism and Its Deficiency

Major Goals: The long-range goal of this project is to characterize the metabolism of branched chain acyl-CoAs and to identify the consequences of its failure in humans.

\*Status of Support: Active

Project Number: 5R01DK109907-06

Name of PD/PI: Gerard Vockley (PI)

\*Source of Support: NIDDK

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 07/01/2016 – 02/28/2025

\* Total Award Amount (including Indirect Costs): $4,123,041

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 2.10 calendar |
| 2. 2022 | 2.10 calendar |
| 3. 2023 | 2.10 calendar |
| 4. 2024 | 2.10 calendar |

\*Title: Phase 3 Trial of DCA in PDC Deficiency: (IND 028,625, 02/04/2015)

Major Goals: The primary goal will be based on an improved clinical status, as determined by a novel FDA-approved, Observer Reported Outcome (ObsRO) measure. Our secondary endpoints will include 1) confirmation of better clinical outcome by the Karnofsky/Lansky performance scale of home functionality in children with life- threatening disease; and 2) decreased lactatemia.

\*Status of Support: Active

Project Number: 5R01FD005407-03

Name of PD/PI: Peter Stacpoole (PI)

\*Source of Support: FDA

\*Primary Place of Performance: University of Florida

Project/Proposal Start and End Date: 06/01/2016 – 06/30/2023

\* Total Award Amount (including Indirect Costs): $39,098

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.12 calendar |
| 2. 2022 | 0.12 calendar |

\*Title: Use of a home phenylalanine meter to help manage PKU

Major Goals: Our long-term goal is to create low-cost, effective tools that empower people to take charge of their healthcare though field diagnosis and therapy monitoring, and thereby reduce death, disability, and healthcare costs.

\*Status of Support: Active

Project Number: 5R01NR016991-03

Name of PD/PI: Gerard Vockley (PI)

\*Source of Support: NINR

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 09/01/2017 – 06/30/2022

\* Total Award Amount (including Indirect Costs): 2,376,288.00

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 1.81 calendar |

\*Title: Creating a paper-based phenylalanine test for personalized therapy monitoring by patients with phenylketonuria

Major Goals: The long-range goal of this project is to characterize the metabolism of branched chain acyl-CoAs and to identify the consequences of its failure in humans.

\*Status of Support: Active

Project Number: 1R01HD091175

Name of PD/PI: Elaine Fu (PI)

\*Source of Support: NIDDK

\*Primary Place of Performance: Oregon State University

Project/Proposal Start and End Date: 09/01/2017 – 05/31/2021

\* Total Award Amount (including Indirect Costs): $86,127

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.12 calendar |

\*Title: North American Mitochondrial Disease Consortium

Major Goals: Patient Enrollment in to the NAMDC Patient Registry and Bio-Repository.

\*Status of Support: Active

Project Number: 5U54NS078059-10

Name of PD/PI: Michio Hirano (PI)

\*Source of Support: NINDS

\*Primary Place of Performance: Columbia University

Project/Proposal Start and End Date: 09/30/2011 – 08/31/2024

\* Total Award Amount (including Indirect Costs): $48,035

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.12 calendar |
| 2. 2022 | 0.12 calendar |
| 3. 2023 | 0.12 calendar |

\*Title: Hepatocyte Transplantation for Liver-Based Metabolic Disease

Major Goals: In this study, we will treat patients with liver-based metabolic disease by hepatocyte transplantation with the goal of opening the door for broader use of hepatocyte transplants in the treatment of metabolic liver disease.

\*Status of Support: Active

Project Number: 5R01DK117916-03

Name of PD/PI: Ira Fox (PI), Gerard Vockley (MPI)

\*Source of Support: NIDDK

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 08/10/2018 – 06/30/2024

\* Total Award Amount (including Indirect Costs): $1,268,334

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2020 | 0.91 calendar |
| 2. 2022 | 1.20 calendar |
| 3. 2023 | 1.20 calendar |

\*Title: PKUDOS – PKU Demographics, Outcomes, and Safety Registry

Major Goals: The objective of this study is to evaluate the safety of long-term treatment with Kuvan.

\*Status of Support: Active

Project Number: PKUDOS-01/17869

Name of PD/PI: Gerard Vockley (PI)

\*Source of Support: BioMarin

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 09/26/2008 – 96/26/2023

\* Total Award Amount (including Indirect Costs): $55,700

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.12 calendar |
| 2. 2022 | 0.12 calendar |

\*Title: An Observational, Longitudinal, Prospective, Long-Term Registry of Patients with Hypophosphatasia

Major Goals: This multinational, multicenter, observational, prospective, long-term registry is designed to collect data on epidemiology, HPP history, clinical course, symptoms (including multi-systematic aspects of disease), and burden of disease from patients of all ages who have a diagnosis of HPP.

\*Status of Support: Active

Project Number: ALX-HPP-501/18581

Name of PD/PI: Gerard Vockley (PI)

\*Source of Support: Alexion

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 10/19/2015 – 10/18/2024

\* Total Award Amount (including Indirect Costs): $62,250

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.12 calendar |
| 2. 2022 | 0.12 calendar |
| 3. 2023 | 0.12 calendar |

\*Title: An Open Label Study to Determine the Safety and Tolerability of 12 Weeks Treatment with Oral REN001 in Subjects with Fatty Acid Oxidation Disorders (FAOD)

Major Goals: The objective of this study is to evaluate the safety and tolerability of 12 weeks treatment with REN001 in subjects with FAOD.

\*Status of Support: Active

Project Number: REN001-102/23225

Name of PD/PI: Gerard Vockley (PI)

\*Source of Support: Reneo

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 02/19/19 – 02/18/22

\* Total Award Amount (including Indirect Costs): $157,025

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.12 calendar |

\*Title: Precision Medicine Implementation and Commercialization

Major Goals: Our goal is to build upon previous successes in administering pilot awards to stimulate precision medicine research, and our experience in implementation science (pharmacogenomics) and education (Test2Learn) to focus this proposal on the implementation of genomic sequencing in the neonatal intensive care unit (NICU), the development of healthcare provider genomics education through novel solutions and driving precision medicine initiatives across Pitt and UPMC towards commercialization.

\*Status of Support: Active

Project Number: Not Applicable

Name of PD/PI: Adrian Lee (PI)

\*Source of Support: The Richard King Mellon Foundation

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 05/01/2019 – 04/30/2022

\* Total Award Amount (including Indirect Costs): $449,859

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.12 calendar |

\*Title: A Phase 1/2 Open-Label, Randomized, Concurrently-Controlled, Dose Escalation Study to Evaluate the Safety and Efficacy of HMI-102 in Adult PKU Subjects with PAH Deficiency

Major Goals: The primary objective of the study is to evaluate the safety, tolerability, and efficacy of a single dose of HMI-102 when administered to subjects with phenylalanine hydroxylase (PAH) deficiency.

\*Status of Support: Active

Project Number: Not Applicable

Name of PD/PI: Gerard Vockley (PI)

\*Source of Support: Homology Medicines

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 07/03/2019 – 07/02/2022

\* Total Award Amount (including Indirect Costs): $646,739

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.12 calendar |

\*Title: Rare disease research network: Congenital disorders of glycosylation

Major Goals: The goal of this project is to establish a nation-wide network of regional centers for diagnosis, follow up, treatment and clinical research in CDG in order to be able to improve quality of life and life expectancy in congenital disorders of glycosylation.

\*Status of Support: Active

Project Number: 5U54NS115198-03

Name of PD/PI: Eva Morava-Kozicz (PI)

\*Source of Support: NINDS

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 09/15/2019 - 07/31/2024

\* Total Award Amount (including Indirect Costs): $149,435

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.46 calendar |
| 2. 2022 | 0.60 calendar |
| 3. 2023 | 0.60 calendar |

\*Title: The Natural History of LCHAD Retinopathy

Major Goals: The goal for this study is conducting a prospective deep phenotyping study of LCHAD/TFP deficient retinopathy among 44 patients diagnosed with LCHAD/TFP deficiencies followed over time.

\*Status of Support: Active

Project Number: 5R01HD095968‐02

Name of PD/PI: Melanie Gillingham

\*Source of Support: NICHHD

\*Primary Place of Performance: Oregon Health & Science University

Project/Proposal Start and End Date: 08/01/2019 - 11/31/2023

\* Total Award Amount (including Indirect Costs): $859,285

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.60 calendar |
| 2. 2022 | 0.60 calendar |

\*Title: Precision Medicine in the Diagnosis of Genetic Disorders in Neonates

Major Goals: We hypothesize that TNGS will perform comparably to WGS while providing enhanced detection of copy number variants and genes with high degrees of homology. The impact of TNGS will determined on targeted therapeutics (e.g. medications, diet, surgery, other interventions) enrollment in clinical trials, reduction in diagnostic studies/therapeutics, or initiation of palliative care.

\*Status of Support: Active

Project Number: 5U01TR002271-04

Name of PD/PI: Jonathan Davis

\*Source of Support: NCATS

\*Primary Place of Performance: Tufts Medical Center

Project/Proposal Start and End Date: 08/15/2018 - 07/31/2023

\* Total Award Amount (including Indirect Costs): $326,371

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.60 calendar |
| 2. 2022 | 0.60 calendar |

\*Title: A Prospective Clinical Study of Phenylketonuria (PKU)

Major Goals: The purpose of this study is to learn more about health problems in people with PKU and how we measure these problems over time. This knowledge will help us design future studies to understand whether health problems get better when we test new drugs for PKU.

\*Status of Support: Active

Project Number: Not Applicable

Name of PD/PI: Gerard Vockley

\*Source of Support: BioMarin

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 07/24/2020 - 07/23/2022

\* Total Award Amount (including Indirect Costs): $384,421

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.12 calendar |

\*Title: A Longitudinal, Prospective, Observational Study in Adult Males with Fabry Disease

Major Goals: Characterize FD-specific biomarkers, symptoms and outcomes for use as baseline/external control data in interventional clinical trials and Assess patient-specific variance of cardiac biomarkers (serum, non-invasive imaging, and ECG) associated with cardiac involvement in FD.

\*Status of Support: Active

Project Number: Not Applicable

Name of PD/PI: Gerard Vockley

\*Source of Support: 4D Molecular Therapeutics

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 07/13/2020 - 07/12/2022

\* Total Award Amount (including Indirect Costs):$17,716

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.12 calendar |

\*Title: An Open-label, Phase 1/2 Trial of Gene Therapy 4D-310 in Adult Males with Classic Fabry Disease

Major Goals: Evaluate the safety and tolerability of 4D-310 following a single intravenous (IV) dose.

\*Status of Support: Active

Project Number: Not Applicable

Name of PD/PI: Gerard Vockley

\*Source of Support: 4D Molecular Therapeutics

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 11/13/2020 - 11/12/2022

\* Total Award Amount (including Indirect Costs): $247,396

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.12 calendar |

\*Title: An Open-label Study of the Efficacy and Safety of SYNB1618 in Patients with Phenylketonuria (SynPheny-1)

Major Goals: The purpose of this study is to determine the efficacy of SYNB1619 in reducing area under the curve (AUC) for plasma D5-phenylalanine (Phe).

\*Status of Support: Active

Project Number: Not Applicable

Name of PD/PI: Gerard Vockley

\*Source of Support: Synlogic

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 11/20/2020 – 11/19/2022

\* Total Award Amount (including Indirect Costs): $44,607

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.12 calendar |

\*Title: AAV Gene therapy of VLCAD Deficiency

Major Goals: The purpose of this study is to determine the efficacy of SYNB1619 in reducing area under This project analyzes VLCAD deficiency in mice.

\*Status of Support: Active

Project Number: Not Applicable

Name of PD/PI: Gerard Vockley

\*Source of Support: Kriya Pharmaceuticals

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 01/14/2021 – 01/13/2023

\* Total Award Amount (including Indirect Costs): $344,684

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2022 | 0.50 calendar |

\*Title: Novel Mouse Models to Assess AAV Gene Therapy of VLCAD Deficiency

Major Goals: This project assesses AAV Gene Therapy in mice.

\*Status of Support: Active

Project Number: Not Applicable

Name of PD/PI: Gerard Vockley

\*Source of Support: Kriya Pharmaceuticals

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 01/14/2021 – 01/13/2023

\* Total Award Amount (including Indirect Costs): $344,684

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2022 | 0.50 calendar |

\*Title: Enzyme Substitution Therapy of a Porcine Model of PKU

Major Goals: To examine the effectiveness of Nestlé’s novel therapeutic in the minipig disease model of phenylketonuria.

\*Status of Support: Active

Project Number: Not Applicable

Name of PD/PI: Gerard Vockley

\*Source of Support: Nestle

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 11/04/2021 - 11/03/2023

\* Total Award Amount (including Indirect Costs): $800,000

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 1.00 calendar |
| 2. 2022 | 1.00 calendar |

\*Title: A Phase 1/2 Open-label Clinical Study of Hlb-001 Gene Therapy in Pediatric Patients with Methylmalonic Acidemia Characterized by MMUT Mutations

Major Goals: To assess the safety and tolerability of hLB-001 in pediatric patients with methylmalonic acidemia (MMA)

\*Status of Support: Active

Project Number: Not Applicable

Name of PD/PI: Gerard Vockley

\*Source of Support: Logic Bio

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 11/09/2020 – 11/08/2022

\* Total Award Amount (including Indirect Costs): $105,678

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2021 | 0.12 calendar |

**PENDING**

\*Title: Metabolic Engineering for Management of PAH Deficient PKU Osteopenia

Major Goals: We will identify analytes informing bone restoration and therapeutic response that will be assayable within patient collected dried blood spots adding a novel element to this long-established aspect of PKU management.

\*Status of Support: Pending

Project Number: R01 HD105994

Name of PD/PI: Steven Dobrowolski / Harry Blair

\*Source of Support: NIAMSD / NIH

\*Primary Place of Performance: University of Pittsburgh

Project/Proposal Start and End Date: 09/2022 – 08/2027

\* Total Award Amount (including Indirect Costs): $3,258,091

\* Person Months (Calendar/Academic/Summer) per budget period.

| Year (YYYY) | Person Months (##.##) |
| --- | --- |
| 1. 2022 | 0.30 calendar |
| 2. 2023 | 0.30 calendar |
| 3. 2024 | 0.30 calendar |
| 4. 2025 | 0.30 calendar |
| 5. 2026 | 0.30 Calendar |

**PRIOR FUNDING**

P01 HL59412 (Terry Flotte) 09/01/06 – 08/31/07

NIH (University of Florida)

**Project #2 Gene Therapy using Viral Vector for Lung and Cardiovascular Disease**

The purpose of the study is to provide tandem MS analysis and other biochemical assays crucial for determining the correction of the FAO disorders in cell culture and animal models.

R01 DK054936 (Gerard Vockley) 02/15/99 – 02/28/14

NIH

**Metabolism of Short Chain Acyl-CoA’s and its Deficiency**

The purpose of the study is to identify molecular defects responsible for causing SCAD, SBCAD, and IBD deficiencies, and to characterize the effects of mutations on enzyme function.

R01 DK045482 (Gerard Vockley) 08/01/93 – 04/30/06

NIH

**Molecular Characterization of Acyl-CoA Dehydrogenases**

The purpose of the study is to Investigate important structure/function relationships in the ACD gene family.

BMRN PKU-006 (Gerard Vockley) 01/01/05 – 12/31/07

BioMarin Pharmaceutical Inc.

**A Phase 3, Multicenter, Randomized, Double-blind. Placebo-controlled Study to Evaluate the Safety and Efficacy of Phenoptin 20mg/kg/day to increase Phenylalanine Tolerance in Phenylketonuric Children on a Phenylalanine-restricted diet.**

The purpose of this diet study is to evaluate the effect of Phenoptin on Phenylalanine tolerance.

BMRN PKU-001 (Vockley) 01/01/05 – 12/31/07

BioMarin Pharmaceutical Inc.

**A Phase 2, Multicenter, Open-Label Study to Evaluate the Response to and Safety of an 8-day Course of Phenoptin Treatment in Subjects with Phenylketonuria Who Have Elevated Phenylalanine Levels.**

The purpose of the study is in support of a Phenoptin’s regulatory approval application submitted to the U.S. Food and Drug Administration.

BMRN PKU-003 (Gerard Vockley) 01/01/05 – 12/31/07

BioMarin Pharmaceutical Inc.

**A Phase 3, Randomized, Double-Blind, Placebo-Controlled Study to Evaluate the Safety and Efficacy of Phenoptin in Subjects with Phenylketonuria Who Have Elevated Phenylalanine Levels.**

The purpose of the study is in support of a Phenoptin’s regulatory approval application submitted to the U.S. Food and Drug Administration.

U54 RR019455 (Ronald Sokol) 08/01/05 – 07/31/07

NIH

**ACD9 Deficiency: A New Genetic Cause of Acute Liver Failure**

The purpose of the study is to test the hypothesis that ACD9 Deficiency represents a previously unrecognized cause of liver disease, including sudden liver failure in otherwise healthy individuals.

R01 NS051813 (Maureen Durkin) 10/01/06 – 09/30/09

NIH

**Epidemiology of 2-Methylbutyryl-CoA Dehydrogenase Deficiency (SBCADD) in the Hmong-American population of Wisconsin**

My role in this project is to provide molecular and biochemical diagnosis of SBCAD deficiency in patients.

0655512U (Gerard Vockley) 07/01/06 - 06/30/08

American Heart Association

**Cardiomyopathy in β-oxidation defects**

The purpose of this study is to characterize the molecular determinants predictive of the development of cardiomyopathy in very long chain acyl-CoA dehydrogenase deficiency

R01 DK049200 (Vladimir Ritov) 06/01/07 – 05/31/09

NIH

**Mitochondrial Impairment in Muscle Insulin Resistance**

Our first specific aim will be to measure the functional capacity of mitochondria in human skeletal muscle in obesity and type 2 DM. The second specific aim will be to examine the morphology of mitochondria in human skeletal muscle in obesity and type 2 DM. The third specific aim will be to examine the pathogenesis of mitochondrial damage in obesity and type 2 DM. The fourth specific aim will be to assess whether exercise and diet can heal mitochondria in obesity and type 2 DM.

DK078775 (Gerard Vockley) 04/01/08 - 03/31/21

NIH

**Inborn Errors of Long Chain Fat Metabolism**

The goal of this study is to characterize the physiological roles of LCAD, VLCAD, and ACD9 and explore the ramifications of genetic deficiencies of these enzymes in humans and mouse models.

U54 HD061939 (William Rizzo) 09/29/09 – 08/31/20

NIH

**Sterol and Isoprenoid Disease Research Consortium (STAIR)**

The STAIR Consortium was created to study a group of inherited diseases bound by common biochemistry. The consortium will conduct four major clinical trials and a therapeutic trial to evaluate the efficacy of cholesterol therapy in Smith-Lemli-Opitz Syndrome (SLOS). The consortium will support a full-scale training program in the field of sterol and isoprenoid diseases and will share resources with the NIH Rare Diseases Clinical Research Network (RDCRN).

PAL-003(Gerard Vockley) 11/19/09 – 11/18/15

BioMarin

**Phase 2 Ext. Open-Label, Dose-Finding Study to evaluate the Safety, Efficacy and Tolerability of Multiple sub-Q Doses of rAvPAL-PEG in Subjects with PKU.**

The primary objective of the study is: to evaluate the effect of long-term administration of Sc injections of rAvPAL-PEG on blood Phe concentrations in subjects with PKU.

HPN-100-005 (Gerard Vockley) 01/01/10 – 12/31/11

Hyperion Therapeutics

**A Phase 2, Fixed Sequence, 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**

The primary objective of the study is: to evaluate the safety and pharmacokinetic (PK) characteristics of HPN-100 as compared to sodium phenylbutyrate (NaPBA) in pediatric subjects with urea cycle disorders (UCDs).

HPN-100-006 (Gerard Vockley) 01/01/10 – 12/31/11

Hyperion Therapeutics

**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)**

The primary objective of the study is: to establish the non-inferiority of HPN-100 as compared to sodium phenylbutyrate (NaPBA) as assessed by venous ammonia.

HPN-100-007 (Gerard Vockley) 01/01/10 – 12/31/11

Hyperion Therapeutics

**A Phase 3, Open-Label Study of the Safety of HPN-100 for the Long-Term Treatment of Urea Cycle Disorders (Treat UCD)**

The primary objective of the study is: to evaluate the long-term safety and it’s control of venous ammonia in the management of urea cycle disorders (UCDs).

N/A (Gerard Vockley) 09/01/10 – 08/31/15

Enobia Pharma

**Safety and Efficacy of ENB-0040 in Infants & Children < 5 with Hypophosphosphatasia**

The primary objective is to study the effect of ENB-0040 treatment on skeletal manifestations of HPP as measured by radiographs.

N/A (Gerard Vockley) 09/26/08 – 09/26/21

BioMarin

**PKUDOS – PKU Demographics, Outcomes, and Safety Registry**

The objective of this study is to evaluate the safety of long-term treatment with Kuvan.

SAP4100033126 (Bret Goodpaster) 09/01/06 – 05/31/10

PA Department of Health

**Determinants and Treatment of Obesity.**

My subsection of this grant focuses on identification of variations in energy metabolism in morbid obesity.

N/A (Gerard Vockley) 03/17/10 – 03/16/12

SHS International

**Safety and Efficacy of Lanaflex II in Improving Plasma Phenylalanine Levels in PKU Subjects who are Non-Compliant with a Phe-Restricted Diet**

The primary objective is to investigate the effects of Lanaflex II in reducing plasma Phe levels.

HPN-100-011 (Gerard Vockley) 02/10/11 – 02/09/13

Hyperion Therapeutics

**Long-Term Use of HPN-100 in Urea Cycle Disorders**

The primary objective of the study is: to follow the subjects’ transition to the study and provide a six month follow up for mental testing, ammonia levels and routine safety screens.

R01 FD003895 (Melanie Gillingham) 05/20/11 – 04/30/15

Oregon Health & Science University

**Triheptanoate Therapy of Long-chain Fatty Acid Oxidation Disorders**

The overall goal of the project is to examine the relationship between Fatty Acid oxidation (FAO) and the Citric Acid Cycle (CAC) in patients with FAO defects and develop novel, improved therapies for their treatment.

PAL-004 (Gerard Vockley) 08/18/11 – 08/17/13 BioMarin

**Phase 2 study to Evaluate the Safety, Efficacy and Tolerability of Sub-Q Doses of rAvPAL-PEG administered Daily in Subjects with PKU.**

The primary objective of the study is to evaluate the effect of daily administration of rAvPAL-PEG on the reduction of blood Phe concentrations in subjects with PKU.

PRISM 165-301 (Gerard Vockley) 05/07/13 – 05/06/15

BioMarin

**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 in Adults Not Previously Treated with BMN-165**

The primary objective of the study is to evaluate the effect of treatment with BMN-165 on an adult population.

PRISM 165-302 (Gerard Vockley) 06/25/13 – 09/24/16

BioMarin $1,322,850

**Three-Part, Phase 3, Double-Blind, Placebo-Controlled, Four-arm, Discontinuation Study to Evaluate the Efficiency and Safety of Sub-Cutaneous Injections of BMN-165 Self-Administered**

This is the follow-up study for PRISM 165-302.

N/A (Gerard Vockley) 07/12/13 – 07/11/15

Ultragenyx Pharmaceuticals

**A Retrospective Medical Record Review Study to Assess the Clinical Outcome of Triheptanoin Treatment on Patients Affected by Long-Chain Fatty Oxidation Disorders**

This study is a retrospective chart review of FAOD-affected subjects who participate in the triheptanoin compassionate use program. Based on eligibility criterion data from the medical records will be abstracted to collect the important clinical events, safety events and laboratory results that characterize FAOD as defined in the protocol.

N/A (Gerard Vockley) 08/12/13 – 06/30/20

Ultragenyx Pharmaceuticals

**Dietary Therapy for Inherited Disorders of Energy Metabolism**

The primary hypothesis of this study is that triheptanoin is an effective dietary supplement for the treatment of inherited disorders of energy metabolism in substitute for standard medium chain triglycerides. Efficacy will be evaluated through standard of care clinical and laboratory parameters used to follow patients as part of their metabolic care. This includes monitoring of the known associated complications of these diseases including hypoglycemia, cardiomyopathy and episodes of rhabdomyolysis. Data obtained through this protocol will lead to development of a formal clinical trial for FDA approval of use triheptanoin.

U54 NS078059 (Michio Hirano) 09/30/12 - 08/31/24

NIH/Columbia Univeristy

**North American Mitochondrial Disease Consortium**

Patient Enrollment in to the NAMDC Patient Registry and Bio-Repository.

N/A (Gerard Vockley) 11/12/13 – 05/11/15

Ultragenyx Pharmaceuticals

**An Open-Label Phase 2 Study to Assess the safety and Clinical Effects of UX007 on Long-Chain Fatty Oxidation Disorders (LC-FAOD)**

This study will assess whether UX007 is safe and effective in subjects with LC-FAOD. FAOD is an inherited disorder in which the body is unable to break down fats present in normal foods, which would produce energy. People with FAOD are usually treated by limiting the amount of fat in their diets and then placed on a diet which includes a supplement of medium chain triglycerides (MCT). MCT are shorter length fats that can be normally absorbed by people with LC-FAOD and used to make energy.

N/A (Gerard Vockley) 03/01/14 – 02/28/14

Children’s Research Institute

**N-carbamlyglutamate in the treatment of hyperammonenmia**

The overall objective of this project is to determine whether treatment of acute hyperammonemia with N-carbamyl-L-glutamate (NCG) in propionic academia (PA), methylmalonic academia (MMA), carbamyl phosphate synthetase 1 deficiency (CPSD) and ornithine transcarbalmylsae deficiency (OTCD) changes in the clinical outcome of diseases.

N/A (Gerard Vockley) 03/18/14 – 03/17/16

Shire Human Genetic Therapies, Inc.

**Mucopolysaccharidosis (MPS) I, II, and VI Screening in a High-Risk Population With Previous Surgical Repair or Presence of Inguinal and/or Umbilical Hernia in Combination With Pediatric Ear, Nose and Throat (ENT) Surgery (Adenoidectomy and/or Tonsillectomy and/or Tympanostomy) (The HATT Project)**

The primary objective of this study is to evaluate the positive screening rate of MPS II subjects by screening a high-risk male pediatric population who have had or are scheduled for 1 or more specific ENT surgical procedures (adenoidectomy and/or tonsillectomy and/or tympanostomy) and who have a previously repaired or present evidence of an inguinal and/or umbilical hernia. Also, to evaluate the positive screening rate of MPS I and VI in the same high-risk population.

N/A (Gerard Vockley) 04/17/14 – 04/16/16

Wellstat Therapeutics

**Open-Label Study of Uridine Triacetate in Pediatric Patients with Hereditary Orotic Aciduria**

The primary objective is to document the continued clinical benefit of exogenous uridine when patients are

switched from oral administration of uridine to oral administration of uridine triacetate.

N/A (Gerard Vockley) 11/17/14 – 10/31/20

Ultragenyx Pharmaceutical Inc.

**An Open-label Long-Term Safety and Efficacy Extension Study in Subjects with Long-Chain Fatty Acid Oxidation Disorders (LC-FAOD) Previously Enrolled in UX007 or Triheptanoin Studies**

The primary objective of the study is to evaluate the long-term safety and efficacy of UX007 in LC-FAOD subjects. The secondary objectives of the study are to evaluate the effect of UX007 on energy metabolism in LC-FAOD and to evaluate impact of UX007 on clinical events associated with LC-FAOD.

N/A (Gerard Vockley) 01/01/15 – 12/31/16

Ultragenyx Pharmaceutical, Inc.

**ACAD9 and its role in energy metabolism**

The overall aim of this project is to better understand the role of ACAD9 in normal mitochondrial energy metabolism and the physiologic derangements induced by its deficiency.

N/A (Gerard Vockley) 02/01/15 - 01/31/16

National PKU Alliance

**Hepatocyte and Human Amnion Epithelial Cell Transplantation**

Objective: To improve transplant methods, optimize liver environment to maximize cell engraftment, and stabilize PAH to attain the highest enzyme efficiency. When successful, these studies will generate preclinical data relevant to help move AE cell transplant for this disorder from bench to the clinic.

R01 GM108073 (Michael John Palladino) 04/01/15 - 01/31/19

NIH

**Pre-clinical studies of novel mitochondrial gene therapy**

Mitochondrial diseases are devastating untreatable diseases that affect ~1 in 3-5000 humans. We propose preclinical research to develop and demonstrate efficacy of a novel mitochondrial-targeted RNA approach as a viable gene therapy. This approach will be developed and tested in vitro in human cells and in vivo using an established invertebrate genetic model of mitochondrial disease.

AT1001-042 (Gerard Vockley) 05/15/15 – 05/14/19

Amicus Therapeutics

**An Open-Label Extension Study to Evaluate the Long Term Safety and Efficacy of Migalastat Hydrochloride Monotherapy in Subjects with Fabry Disease**

The purpose of this study is to investigate the long-term safety and explore the efficacy/pharmacodynamics of migalastat HCl administered 150 mg every other day (QOD).

N/A (Gerard Vockley) 06/25/15 – 06/24/17

ArmaGen

**A Phase 1 Safety and Dose-Finding Study of a Human Insulin Receptor Monoclonal Antibody-Human Iduronate 1-Sulfatese (IDS) Fusion Protein, AGT-182 in Adult Patients with MPS II**

The primary objective of this study is to determine a safe and tolerated dose of AGT-182 in adults with MPS II.

N/A (Gerard Vockley) 06/17/15 – 06/16/17

ArmaGen

**A Phase I Safety and Dose-Finding Study of a Human Insulin Receptor Monoclonal Antibody-Human**

**Iduronate 2-Sulfatase (IDS) Fusion Protein, AGT-181 in Adult Patients with Mucopolysaccharadisosis (MPS 1)**

The objective of this study is to determine a safe and well tolerated dose of AGT-181 in adults with MPS I.

N/A (Gerard Vockley) 07/28/15 – 07/27/18

Reata Pharmaceuticals

**A phase 2 study of the safety, efficacy, and pharmacodynamics of RTA 408 in the treatment of Mitochondrial Myopathy**

This two-part study will evaluate the efficacy, safety, and pharmacodynamics of RTA 408 in the treatment of patients with mitochondrial myopathy.

N/A (Gerard Vockley) 10/07/15 – 10/06/17

Alexion Pharma

**An open-label, multicenter, expanded access program for asfotase alfa treatment for patients with infantile- or juvenile-onset Hypophosphatasia (HPP)**

The primary objective is to assess the safety and tolerability of subcutaneous (SC) injections of asfotase alfa administered 6 times weekly (at a dose of 1 mg/kg) or 3 times weekly (at a dose of 2 mg/kg).

N/A (Gerard Vockley) 10/19/15 – 10/18/20

Alexion

**An Observational, Longitudinal, Prospective, Long-Term Registry of Patients with Hypophosphatasia**

This multinational, multicenter, observational, prospective, long-term registry is designed to collect data on epidemiology, HPP history, clinical course, symptoms (including multi-systematic aspects of disease), and burden of disease from patients of all ages who have a diagnosis of HPP.

N/A (Gerard Vockley) 11/12/15 – 11/11/16

Mitobridge, Inc.

**Evaluation of PPARd modulators in cellular models of FAOD**

We will test Mitobridge-provided PPAR delta modulators in various fatty acid oxidation deficiency (FAOD) patient cell lines. This study will be carried out in two phases; phase 1 will test compounds in four cell lines per disease in three FAOD mutations. Phase 2 will test compounds in ten or more cell lines to achieve sufficient statistical power in focused FAOD mutation(s).

R44 HD088211 (Scott Norton) 09/01/16 - 07/31/20

Baebies, Inc.

**Point-of-Birth Newborn Screening for MCAD/VLCAD and Galactosemia to Eliminate Deadly Delays for Time Critical Conditions**

The project objective is to demonstrate the technical feasibility of using digital microfluidics to detect medium-chain acyl-CoA dehydrogenase (MCAD) deficiency, very-long-chain acyl-CoA dehydrogenase (VLCAD) deficiency and galactosemia in whole blood samples for improved newborn screening at point-of-birth.

N/A (Gerard Vockley) 12/20/17 - 12/19/20

Stealth BioTherapeutics

**A Phase 3 Randomized, Double-Blind, Parallel-Group, Placebo-Controlled Trial to Evaluate the Efficacy and Safety of Daily Subcutaneous Injections of Elamipretide in Subjects with Primary Mitochondrial Myopathy Followed by an Open-Label Treatment Extension**

The objectives of this study are divided into two parts. PART 1 is a 24-week, randomized, double-blind, parallel-group, placebo-controlled assessment of the efficacy and safety of single daily subcutaneous (SC) doses of 40 mg elamipretide (vs placebo) administered through the elamipretide delivery system as a

treatment for subjects with primary mitochondrial myopathy (PMM). PART 2 is an up to 144-week, open-label assessment of the long-term safety and tolerability of single daily SC doses of 40 mg elamipretide administered through the elamipretide delivery system in subjects with PMM.

N/A (Gerard Vockley) 03/23/18 – 06/30/18

Homology

**A 5-year retrospective analysis of two cohorts (BCH and Pitt) of Individuals 10-40 with a diagnosis of Hyperphenylalaninemia from two specialized tertiary care clinics**

The goal of this study is to evaluate individuals 10-40 with a diagnosis of Hyperphenylalaninemia.

N/A (Gerard Vockley) 04/19/18 – 04/18/19

Reneo Pharmaceuticals, Inc.

**Reneo Pharmaceuticals Research Collaboration for Laboratory Experiments to Test a Potential Therapy for Patients with Fatty Acid Oxidation Disorders**

This study proposes to test PPAR activators provided by Reneo Pharmaceuticals in a number of vitro assays.

N/A (Gerard Vockley) 04/17/18 – 04/16/19

Reneo Pharmaceuticals, Inc.

**Reneo FAOD Patient Survey**

This study proposes to better understand the FAOD population and to pre-identify patients who might be eligible to participate in a clinical trial of REN001.

N/A (Gerard Vockley) 12/17/18 – 12/31/19

Carnot, LLC

**Treatment of Fatty Acid Oxidation Disorder by cardiolipin binding peptides**

The goal of this project is to examine the status of the TCA cycle in patients with PA and MMA before and after treatment with succinate/citrate, and to examine its effect in cardiomyopathy in patients with PA.

N/A (Gerard Vockley) 12/20/18 – 12/19/19

Synlogic

**A Phase 1/2a, First-in0human, Oral Single and Multiple Dose Escalation, Randomized, Double-blinded, Placebo-controlled Study of SYNB1618 in Healthy Adult Volunteers and Adult Subjects with Phenylketonuria to Evaluate Safety, Tolerability, Kinetics, and Pharmacodynamics**

The primary objective is to evaluate the safety and tolerability of SYNB1618 following single and multiple doses in health volunteers and subjects diagnosed with PKU.

N/A (Gerard Vockley) 07/01/18 – 12/31/19

Imbria Pharmaceuticals, Inc.

**Anapleurotic therapy for Disorders of Propionate Metabolism**

The goal of this project is to examine the status of the TCA cycle in patients with PA and MMA before and after treatment with succinate/citrate, and to examine its effect in cardiomyopathy in patients with PA.

1. **Seminars and invited lectureships related to your research.**

**Visiting Professor/Named Lecturer**

Rady Children’s Hospital Grand Rounds, San Diego, CA, (2010)

Michael Palmeri Memorial Lectureship, Children’s Hospital of Philadelphia (2011)

Grand Rounds, Halifax Children’s Hospital (2011)

Grand Rounds, University of Edmonton Children’s Hospital (2013)

Grand Rounds, University of British Columbia, Vancouver Children’s Hospital (2014)

Ron Scott Memorial Lecture, University of Washington Children’s Hospital, Seattle (2014)

Aaron Michael Graham Memorial Lectureship, Children’s Hospital of Los Angeles (2016)

Haworth Visiting Professor, University of Manitoba and Children’s Hospital of Manitoba (2017)

Boston Children’s Hospital Grand Rounds (Manton Lecture), Boston, MA (2017)

Franklin and Marshall College. Leadership in Biomedical Research and Medicine Lecture3 (2018)

Brown University Department of Pediatrics Grand Rounds. (2021)

General Hospital Medical Genetics Grand Rounds. (2022)

University of South Florida Department of Genetics Grand Rounds. (2022)

**Invited Presentations at National Meetings (Selected from several hundred)**

1. Society for Inherited Metabolic Disorders Annual Meeting. (1989). Orlando, FL. Molecular Characterization of Type III Isovaleric Acidemia.

2. Society for Inherited Metabolic Disorders Annual Meeting. (1991). Santa Fe, NM. Central Pontine Myelinolysis in Ornithine Transcarbamylase Deficiency.

3. Second International Congress on Disorders of β-Oxidation. (1991). Philadelphia, PA. Molecular Defects in Isovaleric Acidemia.

4. Society for Inherited Metabolic Defects Annual Meeting. (1993). Asilomar, CA. Molecular and Biochemical Characterization of Mutations Responsible for Isovaleric Acidemia.

5. Society for Inherited Metabolic Disorders Annual Meeting. (1995). Perdido Beach, AL. Cloning and Characterization of Short/Branched Chain Acyl-CoA Dehydrogenase.

6. Society for Inherited Metabolic Disorders Annual Meeting. (1996). Cocoyoc, Mexico. Molecular Modeling as an Aid to Study Point Mutations in Isovaleric Acidemia.

7. NIH Workshop on Structural Biology in Inborn Errors of Metabolism. (1996). Bethesda, MD. Structure/Function Relationships in Acyl-CoA Dehydrogenases.

1. March of Dimes and American College of Medical Genetics Annual Meeting. (1998). Los Angeles, CA. Short chain acyl-CoA dehydrogenase deficiency.
2. American Society of Human Genetics Annual Meeting. (1998). San Francisco, CA. Splicing mutations in the IVD gene in isovaleric acidemia.
3. Society for Inherited Metabolic Disorders Annual Meeting. (1999). Lake Lanier, Georgia. New advances in inborn errors of metabolism. Workshop organizer and speaker.
4. American Society of Human Genetics Annual Meeting. (2001).Philadelphia, PA. Short/branched chain acyl-CoA dehydrogenase deficiency.
5. Society for Inherited Metabolic Disorders Annual Meeting (2002). Asilomar, CA. Beyond the human genome project. Workshop chair and speaker.
6. American Society of Human Genetics Annual Meeting. (2002). Baltimore, MD. Isobutyryl-CoA dehydrogenase deficiency.
7. Pediatric Grand Rounds. University of California, Davis. (2003). Davis, CA. Expanded newborn screening for inborn errors of metabolism.
8. Society for Inherited Metabolic Disorders Annual Meeting. Asilomar CA (2005). Redefining long chain fat metabolism in humans.
9. Donough O’Brien Presidential Lecturer, Society for Inherited Metabolic Disorders Annual Meeting. Nashville, TN (2007). Newborn Screening: After the Thrill is Gone.
10. American College of Medical Genetics Annual Meeting, Phoenix AZ. (2007) Inborn errors of metabolism and complex patterns of disease.
11. Society for Inherited Metabolic Disorders Annual Meeting. Asilomar. CA (2008). A new inborn error of sterol metabolism.
12. New developments in the treatment of fatty acid oxidation disorders. (2010). Presented at the annual meeting of the American College of Medical Genetics, Albuquerque, NM
13. Clinical trials for mitochondrial myopathies. **Vockley J** (2011). Invited Plenary lecture. Presented at the United Mitochondrial Disease Foundation 2011 Annual Meeting.
14. Long chain fatty acid oxidation disorders. **Vockley J** (2012). Invited Plenary lecture. Presented at the American College of Medical Genetics 2012 Annual Meeting.
15. Personalized medicine and inborn errors of metabolism. **Vockley J** (2012). Invited Plenary lecture. Presented at the Society of Inborn Errors of Metabolism 2012 Annual Meeting.
16. Development of a New PKU Guideline. **Vockley J.** (2013) Invited lecture, American College of Medical Genetics 2013Annual Meeting
17. Infantile hypophosphatasia. Presented at the American College of Medical Genetics 2014 Annual Meeting. Memphis TN
18. The mitochondrial architecture of energy metabolism. Keynote Address at the United Mitochondrial Disease Foundation Annual Meeting, 2014. Pittsburgh, PA.
19. Results from a randomized trial of triheptanoin compared to MCT oil in patients with long chain fatty acid oxidation defects. (2015). Presented at the Annual Meeting of the Society for Inherited Metabolic Disorders, Salt Lake City, UT
20. Mitochondrial respiratory chain disorders in the Old Order Amish Population. (2015). Presented at the Annual Meeting of the Society for Inherited Metabolic Disorders, Salt Lake City, UT
21. Keynote Speaker, A Plain Population Genome Project, Amish Translational Medicine Meeting. (2016) South Bend, IN
22. Plenary Speaker, Update on the International Network for Fatty Acid Oxidation Research and Management. (2016). Presented at the Annual Meeting of the Society for Inherited Metabolic Disorders, Jacksonville, FL
23. Positive response to Infliximab in a patient with very long-chain acyl-CoA dehydrogenase deficiency. (2017). Presented at the annual meeting of the American College of Medical Genetics and Genomics. Phoenix, AZ
24. Newborn screening for heavy metals. Annual Meeting of the Newborn Screening Translational Research Network. Baltimore, MD. 2017
25. Results from a 78-week single-arm, open-label phase 2 study to evaluate UX007 in pediatric and adult patients with moderate to severe long-chain fatty acid oxidation disorders (LC-FAOD). SIMD Annual Meeting. San Diego. 2018
26. Mitochondrial targeted antioxidants improve mitochondrial function in very long-chain acyl-coA dehydrogenase deficient fibroblasts. SIMD Annual Meeting. San Diego. 2018
27. Identifying rare metabolic disorders of energy dysfunction in patients with treatment-resistant depression. ACMG Annual Meeting. 2019
28. Treatment of bioenergetic dysfunction in propionic acidemia. SIMD Annual Meeting. Seattle. 2019
29. Metabolic analysis reveals evidence for branched chain amino acid catabolism crosstalk and the potential for improved treatment of organic aciduria. SIMD Annual Meeting. Virtual presentation. 2020
30. A long day’s journey: new therapies for rare disorders. Brown University Department of Pediatrics Grand Rounds.Virtual presentation. 2021
31. Metabolic imbalance in chronic depression: implications for diagnosis and treatment. Massachusetts General Hospital Medical Genetics Grand Rounds. Virtual presentation. 2022
32. Long chain fatty acid oxidation disorders. University of South Florida Department of Pediatrics Grand Rounds. Virtual presentation. 2022
33. A New Era in Therapies for Genetic Disorders: FAODs. Texas Newborn Screening Program Annual Meeting. Virtual presentation. 2022

**Invited Presentations at International Meetings (Selected from several hundred)**

1. Vth International Congress on Inborn Errors of Metabolism. (1990). Asilomar, CA. Molecular Characterization of Mutations Responsible for Isovaleric Acidemia.

2. VIth International Congress on Inborn Errors of Metabolism. (1994). Milan, Italy. Mutations in Isovaleric Acidemia. Molecular and Biochemical Characterization of Mutations Responsible for Isovaleric Acidemia.

3. International Working Group on Disorders of β-Oxidation. (1995). Aarhus, Denmark. Chair of Session on Structure and Substrate Specificity of Acyl-CoA Dehydrogenases.

4. VII International Congress on Inherited Metabolic Disorders. (1997). Vienna, Austria. The use of molecular modeling in inborn errors of metabolism.

1. SCAD collaborative group workshop. (1999). Aarhus, Denmark. SCAD deficiency.
2. VII International Congress on Inborn Errors of Metabolism. (2000). Cambridge, England. Plenary Session Chair. Workshop chair. Invited lecture on Disorders of ß-Oxidation
3. Annual meeting of the Society for Study of Inherited Metabolic Disorders. (2002). Dublin, Ireland. Workshop chair. Invited presentation on Synergistic Heterozygosity
4. International Working Group on Disorders of ß-Oxidation. (2003). Session chair. Invited presentation on New Disorders of ß-Oxidation.
5. IXth International Congress on Inborn Errors of Metabolism. (2003). Brisbane, Australia. Plenary Session Chair, Disorders of Mitochondrial Energy Metabolism.
6. International Symposium on -oxidation. Amsterdam. (2005). Acyl-CoA dehydrogenases: new enzymes and new disorders.
7. Mitochondrial -oxidation: new developments. (2006). Presented at the International Congress on Inherited Metabolic Disorders. Chiba. Japan
8. Keynote address. Annual meeting of the Society for Study of Inherited Metabolic Disorders. Lisbon. Spain, (2008). Energy metabolism as a complex genetic trait.
9. Tissue specificity of mitochondrial -oxidation. (2008). Fulda. Germany.
10. Xth International Congress on Inborn Errors of Metabolism. San Diego, CA. (2009). Characterization of a multi-functional protein complex containing the mitochondrial respiratory chain and fatty acid oxidation.
11. Presented at the annual meeting of the Society for Inherited Metabolic Disorders Annual Meeting. Istanbul, Turkey. (2010). Changes in the mitochondrial proteome in fatty acid oxidation deficient mice.
12. Presented at the XIth Annual Congress on Inborn Errors of Metabolism, Barcelona, Spain. (2013) The molecular Architecture of Mitochondrial Energy Metabolism
13. Moonlighting in mitochondria; ACAD9 plays a duel role in energy metabolism in mitochondria. (2013). Presented at XI th International Congress on Inborn Errors of Metabolism. Barcelona, Spain
14. Transplant for metabolic disease. Presented at the Garrod Society Annual Meeting, Ottowa, CA
15. Novel therapy of long chain fatty acid oxidation disorders using triheptanoin. **Vockley J.** (2013) XI th International Congress on Inborn Errors of Metabolism. Barcelona, Spain.
16. Emerging clinical trials for fatty acid oxidation disorders. (2015). Presented at the annual meeting of the International Fatty Acid Oxidation Research and Management Network. Lyon. France.
17. Molecular Architecture of mitochondrial energy metabolism. (2015). Presented at the annual meeting of the Society for the Study of Inherited Metabolic Disorders. Lyon. France.
18. Molecular Architecture of mitochondrial energy metabolism. (2015). Presented at the annual meeting of the Society for the Study of Inherited Metabolic Disorders. Lyon. France.
19. Advances in therapy for PKU. (2016). Presented at the Annual Meeting of the Garrod Society, Halifax, Canada
20. Advances in fatty acid oxidation research. (2016). Presented at the annual meeting of the International Network for Fatty Acid Oxidation Research and Management (INFORM). Boston, MA
21. Novel therapies for fatty acid oxidation disorders. (2016). Presented at the annual meeting of the Society for the Study of Inherited Metabolic Disorders. Rome, Italy
22. Triheptanoin in the treatment of long chain fatty acid oxidation disorders: Report of two FDA Phase 2 trial results. (2017). Presented at the annual meeting of the Portuguese Society of Metabolic Disorders. Lisbon, Portugal.
23. Fatty acid oxidation update. (2017). Plenary Lecture. International Congress on Inherited Metabolic Disorders. Rio De Janeiro, Brazil
24. Results from a 78-week Single-arm, Open-label Phase 2 Study to Evaluate UX007 in Pediatric and Adult Patients with Moderate to Severe Long-Chain Fatty Acid Oxidation Disorders (LC-FAOD). (2017). International Congress on Inherited Metabolic Disorders. Rio De Janeiro, Brazil
25. Phase 2 Long-term Pegvaliase Treatment for Adults with Phenylketonuria: Updated Year 5 Safety and Efficacy Data from the PAL-003 Extension. (2017). International Congress on Inherited Metabolic Disorders. Rio De Janeiro, Brazil
26. Endoplasmic reticulum-mitochondria crosstalk and redox homeostasis disruption in very long-chain acyl-CoA dehydrogenase deficient fibroblasts. (2017). International Congress on Inherited Metabolic Disorders. Rio De Janeiro, Brazil
27. Antioxidant therapy as an adjunct for treatment of long chain fatty acid oxidation disorders. (2017). International Congress on Inherited Metabolic Disorders. Rio De Janeiro, Brazil
28. Novel mechanisms of pathogenesis in mitochondrial trifunctional protein deficiency: implications for clinical outcome and treatment. (2017). International Congress on Inherited Metabolic Disorders. Rio De Janeiro, Brazil
29. Elucidating the mitochondrial architecture of branched-chain amino acid metabolism enzymes. (2017). International Congress on Inherited Metabolic Disorders. Rio De Janeiro, Brazil
30. PKU Research Update. (2017). Asia-Pacific PKU Summit. Osaka. Japan.
31. Targeting cardiolipin: a new therapeutic approach to treat long chain hydroxy acyl CoA dehydrogenase and mitochondrial trifunctional protein deficiency. SSIEM Annual Meeting. 2018. Athens, Greece
32. A Phase 1/2a, First-in-human, Oral Single and Multiple Dose-Escalation, Randomized, Double-blind, Placebo-controlled Study of SYNB1618 in Healthy Adult Volunteers and Adult Subjects with Phenylketonuria to Evaluate Safety, Tolerability, Kinetics, and Pharmacodynamics. SSIEM. Annual Meeting. 2019. Amsterdam, The Netherlands
33. Interim results from an open-label, long-term extension study to evaluate the safety and efficacy of triheptanoin (UX007) in LC-FAOD. SSIEM Annual Meeting. 2019. Amsterdam, The Netherlands
34. Assessing Referral of Plain Community Members for Genetic Services at UPMC Children’s Hospital of Pittsburgh: A Quality Improvement Study. ACMG Annual Meeting. 2020
35. Rare metabolic disorders of energy dysfunction in patients with treatment-resistant depression. SIMD Annual Meeting. 2020
36. Metabolic analysis reveals evidence for branched chain amino acid catabolism crosstalk and the potential for improved treatment of organic aciduria. SIMD Annual Meeting. 2020
37. **C**linical Outcomes of Major Clinical Events and Emergency Triheptanoin in Critically Ill Patients With Long-chain Fatty Acid Oxidation Disorders. ACMG 2021 Annual Meeting. Virtual format.
38. Characterization of variants of uncertain significance in isovaleryl-CoA dehydrogenase identified through newborn screening: An approach for faster analysis. SSIEM 2021 Annual Meeting, Sydney Austrailia.
39. Restoration of interaction between FAO and ETC proteins in VLCAD and VLCAD-deficient mice mitochondria by addition of recombinant VLCAD. SSIEM 2021 Annual Meeting, Sydney Austrailia.
40. Defining Therapeutic Options for Combined D,L-2 Hydroxyglutaric Aciduria. SSIEM 2021 Annual Meeting, Sydney Austrailia.
41. Gene therapy for inborn errors of metabolism: current challenges and opportunities. Middle East Genetic and Metabolic Society. Virtual presentation. 2021
42. Whole genome sequencing for genetic diseases across the age spectrum. Arab Medicine. Virtual presentation. 2021
43. Targeting treatments of rare diseases: Biology comes to life! Arab Pediatrics. Virtual presentation. 2021
44. Advances and Treatments of LCFAODs. Brazilian Society for Inborn Errors of Metabolism. Virtual presentation. 2021
45. Inborn errors of long chain fatty acid oxidation. Taiwan Neurology Society. Virtual presentation. 2021
46. A long day’s journey: new therapies for rare disorders. South American Society for Inborn Errors of Metabolism. Virtual presentation. 2022
47. Moving forward one disease at a time. Irish North/South Rare Disease Day. Virtual presentation. 2022
48. Cardiolipin Remodeling Deregulation and Mitochondrial Bioenergetics Alterations in Trifunctional Protein (TFP) Deficiency. SIMD 2022 Annual Meeting. Orlando, FL
49. Restoring succinyllysine antigenic signal and improving O2 consumption of CPT II deficient cells treated with anaplerotic compounds. SSIEM 2022 Annual Meeting. Freyburg, Germany.
50. A new class of anaplerotic compounds restores cellular lysine succinylation and antigenic signal. SSIEM 2022 Annual Meeting. Freyburg, Germany.
51. A CRISPR/Cas9 genome-edited PAH-deficient cell line for studying PKU. SSIEM 2022 Annual Meeting. Freyburg, Germany.

 **3. Other research related activities.** Include editorships, journal refereeing, study section memberships, extra-mural grant reviewing, national course directorships and specialty board memberships.

Meeting Chair. Annual meeting of the International Network for Fatty Acid Oxidation Research and Management. (2020)

Chair DMSB, NIH NIDDK Radiant Project 6/2019-current

Meeting Chair. Annual meeting of the International Network for Fatty Acid Oxidation Research and Management. (2019)

Meeting Chair. Annual meeting of the International Network for Fatty Acid Oxidation Research and Management. (2018)

Meeting Chair. Annual meeting of the International Network for Fatty Acid Oxidation Research and Management. (2017)

Chair, Scientific Advisory Board. Newborn Screening Translational Research Network. (2016-Current)

Member, NIH Director’s Young Investigator Award Second Tier Review Group (NIH). (2016-current)

Member, Undiagnosed Disease Network Metabolomics Core Scientific review committee (NIH). (2015)

Member, Big Data Award ad hoc review committee (NIH) (2014)

Member, NIH Director’s Award review committee (NIH) (2012-current)

Founder and Chair International Network for Fatty Acid Oxidation Disorders Research and Management (INFORM). 2013-current

Program Chair, United Mitochondrial Disease Foundation Annual Meeting (2013)

Chair, Sterol and Isoprenoid Research Collaborative Annual Meeting (2012)

Member, Pioneer Award ad hoc review committee (NIH (2012-2016)

Chair, Metabolomics K Award Study Section (NIH) (2012-Current)

Founder and Managing Editor, North American Metabolic Academy (2006-Current)

Communicating editor, Journal of Inherited Metabolic Disorders (2010-Current)

Assistant editor, Molecular Genetics and Metabolism (2008-Current)

Chair, International Congress of Inherited Metabolic Disorders, (2007-2009)

President, Society for Inherited Metabolic Disorders, 2005-2007

Chair, NIH National Institute of General Medical Sciences Human Mutant Cell Repository Advisory Panel (2003-2004)

Program Committee Member, American College of Medical Genetics, 2002-2005

Program Director, Society for Inherited Metabolic Disorders, 2002-2003

Chair, NIH National Institute of General Medical Sciences Human Mutant Cell Repository Advisory Panel (2005)

Member, NIH National Institute of General Medical Sciences Human Mutant Cell Repository Advisory Panel (2001 - 2005)

Medical Biochemistry Study Section, NIH, permanent member, 1997-2001

Board of Directors, The Metabolic Information Network (1998-2006)

Board of Directors, The Society for Inherited Metabolic Diseases (1998- 2007)

Member, *Ad Hoc* Site Visit Committee Maternal and Child Health Mental Retardation Center (1995)

Member, NIH Medical Biochemistry Study Section, Center for Scientific Review (1997-2001)

Program Chair, Society for Inherited Metabolic Disorders, North American Metabolic Academic Annual Meeting (2011)

**LIST OF CURRENT RESEARCH INTERESTS:**

1. Molecular architecture of mitochondrial energy metabolism
2. Molecular basis of defects of ß-oxidation and energy metabolism
3. Systems biology of energy metabolism
4. Structure/function relationships in acyl-CoA dehydrogenases
5. Gene modifying effects in disorders of energy metabolism
6. Gene therapy of acyl-CoA dehydrogenases
7. Novel therapies for disorders of energy metabolism
8. Novel genetic diagnostic techniques
9. Development of novel therapeutics for inborn errors of metabolism
10. Hepatocyte transplant for inborn errors of metabolism

**SERVICE:**

**1. University and Medical School.** Include committee service and chairmanships, administrative appointments and assignments.

Chair, Pennsylvania Rare Disease Advisory Council, Clinical Research Subcommittee, 2019-Current

Member, Pennsylvania Rare Disease Advisory Council, 2019-Current

Chair, NIH Ad Hoc Review Panel for RFA Center for Identification and Study of Individuals with Atypical Diabetes (3/2018)

Advisory Committee on Lead Exposure and the Hazards of Lead Poisoning in Pennsylvania (2017-Current)

Pennsylvania Birth Defects Registry Committee (2016-Current)

Member, Steering Committee, New York, Mid-Atlantic Regional Newborn Screening Consortium (2016- Current)

Chair, Newborn Screening Translational Research Network Steering Committee (2016-2018)

Member, United Mitochondrial Disease Foundation Scientific and Medical Advisory Board, 2015- Current

Member University of Pittsburgh School of Medicine Tenure Committee (2015- 2017)

Director, Center for Rare Disease Therapeutics, Children’s Hospital of Pittsburgh, (2015-Current)

Member of the Precision Medicine Steering Committee, University of Pittsburgh School of Medicine (2012-Current)

Chief of Medical Genetics, Children’s Hospital of Pittsburgh, (2004-Current)

Chair, Research Committee, Department of Medical Genetics, Mayo Clinic (1992-1998)

Chair, Department of Medical Genetics Staff Search Committee (1998-2002)

Member, Rochester Research Committee (1998-1999)

Chair, Interdepartmental Genetics Society, Mayo Clinic (1993-1997)

Educational Director, Biochemical Disease Detection Laboratory, Mayo Clinic (1994-1998)

Member, M.D, Ph.D. Committee (1997-Present)

Member, Institutional Animal Care and Use Committee, Mayo Clinic (1994-1999)

Vice-Chair, Institutional Animal Care and Use Committee, Mayo Clinic (1997-1998)

Chair, Institutional Animal Care and Use Committee, Mayo Clinic (1998-1999)

Member, Research Web Task Force (1998-1999)

Member, Molecular Medicine Task Force and Search Committee (1994-1998)

Member, Molecular Medicine Executive Committee (1995-1998)

Member, Research Committee, Department of Pediatrics (1994-1996)

Member, Balfour/Kendall Research Award Selection Committee (1994 and 1995)

Member, Eagles Grant Review Committee (1996)

Member, Department of Biochemistry and Molecular Biology Graduate Student Qualifying Examination Committee (1994-1997)

Member, Department of Lab Medicine, Biochemical Genetics Testing Task Force (1995-2004)

Member, Division of Lab Genetics Search and Planning Committee (1998-2004)

Member, Pediatric Council Task Force of Pediatric Research at Mayo (1997)

Member, Medical Chairs Forum, Mayo Clinic (2000-2004)

1. **Community Activities.** Include hospital appointments and consultantships.

Chair, HRSA Secretary’s Advisory Committee on Heritable Diseases in Childhood, Technology Subcommittee. (2010-2014)

Chair, American College of Medical Genetics Committee on Therapeutics (2010-2014)

Member Advisory Board Rare Disease Forum (2018-Current)

Member, Pennsylvania Rare Disease Advisory Council (2018-Current)

Member, Pennsylvania State Birth Defects Surveillance Advisory Board (2017-Current)

Member, Pennsylvania State Birth Defects Lead Advisory Board (2016-2019)

Member, HRSA Secretary’s Advisory Committee on Heritable Diseases in Childhood (2008-2014)

Chair, Pennsylvania State Newborn Screening Advisory Committee (2010-2019)

Member, Pennsylvania State Newborn Screening Advisory Committee (2006-Current)

Member, State of Minnesota Newborn Screening Ad Hoc Advisory Committee (1994-2004)

Medical advisor, Organic Acidemia Family Foundation (1991-Current)

Medical advisor, Fatty Acid Oxidation Family Support Group (1991-Current)

Medical advisor, Saving Lives through Screening Foundation (2004-Current)

Musician, St. Joseph Church Contemporary Music Group (2004-Current)

Assistant Scoutmaster, Boy Scouts of America (1996-2012)

Member, Rochester Catholic Schools Council (2002-2004)

Member, St. John the Evangelist Catholic Church Pastoral Council (2000-2004)

Co-director, St. Joseph Parish, contemporary Choir (2004-current)

Boy Scouts of America Troop 380 Scoutmaster (2012-2016)

Boy Scouts of America, Japeechen District Chair (2019- Current)

Boy Scouts of America, Laurel Highlands Council, Leadership Board (2019-current)

3. **Continuing Medical Education (CME) activities.**

Regular presenter for Children’s Hospital of Pittsburgh outreach CME programs

Course Director, Challenging the Paradigms: Liver Transplantation for Metabolic Disease, May 4-5, 2012, Pittsburgh, PA.

Many others!

Medscape 2020. MMA and PA, Is liver transplant always the best treatment option.

Medscape 2020. Long chain fatty acid oxidation disorders

Touch IME. Long chain fatty acid oxidation disorders: pathophysiology, diagnosis and management

Rockpoint CME. Breaking down long chain fatty acid oxidation disorder treatment. Review of current care and the latest advances

Total CME. 2020. Long chain fatty acid oxidation disorders

TEDx Talk:. 2/2017. A matter of balance: changing the rules in depression treatment