Boston Children's Hospital
CURRICULUM VITAE

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Place of Birth: Philadelphia, PA

Education:

1967-1971	B.S. University of Notre Dame
1971-1975	M.D. Jefferson Medical College

Postdoctoral Training:

1975-1976	Intern in Internal Medicine (PGY1), Mercy Catholic Medical Center, Philadelphia, PA
1976-1978	Resident in Pediatrics (PGY1 & PGY2), Thomas Jefferson University Hospital, Philadelphia, PA
1978-1979	Clinical Fellow, The Division of Biochemical Development and Molecular Diseases and the Division of Endocrinology/Diabetes, The Children's Hospital of Philadelphia
1979-1981	Research Fellow, The Division of Biochemical Development and Molecular Diseases, The Children's Hospital of Philadelphia

Academic Appointments:

1981-1989	Assistant Professor of Pediatrics, Department of Pediatrics, University of Pennsylvania School of Medicine
1989-1995	Associate Professor of Pediatrics, Department of Pediatrics, University of Pennsylvania School of Medicine
1995-2001	Professor of Pediatrics, Department of Pediatrics, University of Pennsylvania School of Medicine
2001-2003	Professor of Pediatrics (Tenure Track), Department of Pediatrics,

	George Washington University School of Medicine and Health Sciences
2003-2006	Professor of Pediatrics (Tenure Track), Department of Pediatrics, Jefferson Medical College of Thomas Jefferson University
2004-2006	Professor of Biochemistry, Thomas Jefferson University
2011-	Professor of Pediatrics, Harvard Medical School

Hospital or Affiliated Institution Appointments:

1981-2001	Staff Member, The Children's Hospital of Philadelphia
1995-2001	Staff Member, Hospital of the University of Pennsylvania
1995-2001	Member, Institute for Human Gene Therapy, University of Pennsylvania
1996-2001	Member, Cell and Molecular Biology Graduate Group, University of Pennsylvania
1997-2001	Medical Staff Member, Pennsylvania Hospital, Philadelphia, PA
2001-2003	Medical Staff Member, Children's National Medical Center, Washington, D.C
2003	Medical Staff Member, Georgetown University Hospital, Washington, D.C.
2003-2006	Medical Staff Member, Thomas Jefferson University Hospital, Philadelphia, PA
2006-	Medical Staff Member, Boston Children's Hospital
2007-	Associate Staff Member, Brigham and Women's Hospital, Boston, MA
2008- 2022	Harvey Levy Chair in Metabolism, Boston Children's Hospital
2009-	Associate Member, Broad Institute of Harvard and MIT
2011-	Consulting Staff Member, Beth Israel Deaconess Medical Center, Boston, MA

Hospital and Health Care Organization Service Responsibilities:

1981-1985	Assistant Physician, Division of Metabolism, The Children's Hospital of Philadelphia
1985-1989	Associate Physician, Division of Metabolism, The Children's Hospital of Philadelphia
1989-1998	Senior Physician, Division of Metabolism, The Children's Hospital of Philadelphia
1998-2001	Senior Physician, Division of Endocrinology/Diabetes and the Division of Human Genetics and Molecular Diseases, The Children's Hospital of Philadelphia
1995-2001	Member, Division of Medical Genetics, Department of Medicine, Hospital of the University of Pennsylvania
1997-2002	Medical Staff Member, Pennsylvania Hospital, Philadelphia, PA
2001-2003	Medical Staff Member, Children's National Medical Center, Washington, D.C
2001-2003	Director, Biochemical Genetics and Molecular Diagnostic Laboratory, Children's National Medical Center, Washington, D.C.
2003	Medical Staff Member, Georgetown University Hospital, Washington, D.C.
2003-2006	Medical Staff Member, Thomas Jefferson University Hospital,

	Philadelphia, PA
2006- 2022	Director, Metabolism Program, Boston Children's Hospital
2007-	Associate Staff Member, Brigham and Women's Hospital, Boston, MA
2011-	Consulting Staff Member, Beth Israel Deaconess Medical Center, Boston, MA

Major Administrative Leadership Positions:

2001-2003	Chief, Division of Metabolism, Children's National Medical Center, Washington, D.C.
2001-2003	Director, Biochemical Genetics and Molecular Diagnostic Laboratory, Children's National Medical Center, Washington, D.C.
2003-2005	Vice Dean for Research, Jefferson Medical College
2006-2022	Director, Metabolism Program, Boston Children's Hospital
2006-2022 Program	Director, Harvard Medical School Medical Biochemical Genetics Training
2006-2023	Director, Harvard Medical School Clinical Biochemical Genetics Training Program

Committee Service:

2006	Chair, Committee to Evaluate "Genetics in Medicine" Education in the Jefferson Medical School curriculum
2006	Member, 2007 LCME Self Study, Educational Program Committee, Jefferson Medical College
2010	Member, Drexel University, Biomedical Sciences Ph.D. program, Thesis defense committee (Robert Buccafusca)

Academic Committees at the University of Pennsylvania

1984 -2001	Member, Fellowship Committee, The Children's Hospital of Philadelphia
1990 -2001	Member, University of Pennsylvania Diabetes Research Center
1994 -2001	Member, Committee for Protection of Human Subjects (Institutional Review Board), The Children's Hospital of Philadelphia
1997 -2001	Member, General Clinical Research Center Advisory Committee, The Children's Hospital of Philadelphia
1997 -2001	Member, Intern Selection Committee, The Children's Hospital of Philadelphia
1999 -2001	Chair, General Clinical Research Center Budget Subcommittee, The Children's Hospital of Philadelphia

Academic Committees at the Harvard Medical School (HMS)

2007-	Member, HMS Genetics Training Program Admissions Committee
2012-2015	Member, HMS Promotions and Appointments Committee
2013	Member, HMS Ad Hoc Committee for Dr. Paola Dal Cin's promotion to Professor
2013	Member, HMS, Systems Biology Ph.D. program, Thesis defense committee (Steve Hershman)

2013	Member, HMS, Chemical Biology Ph.D. program, Thesis defense committee (Laura Strittmatter)

Local Committees

1994-2001	Consultant, Newborn Screening Advisory Panel, New Jersey State Department of Health and Senior Services, Newborn Biochemical Screening Program, State of New Jersey
2006	Member, Technical Advisory Committee, Division of Newborn Screening and Genetics, Department of Health, State of Pennsylvania
2007-	Consultant, Newborn Screening Advisory Panel, Department of Health, State of Massachusetts
2007-2014	Member, Board of Directors, Business Committee, New England Regional Genetics Group (NERGG), State of Massachusetts

National Committees

2000-2004	Member, National Newborn Screening and Genetics Resource Center (NNSGRC), Genetics Advisory Committee
2000-2005	Member, American Board Medical Genetics Item-Writing Committee, Clinical Biochemical Genetics Examination Committee
2001	Ad Hoc Reviewer, Orphan Products Development Grant Program, Food and Drug Administration (FDA)
2001-2008	Chair, Newborn Screening Committee, Society for Inherited Metabolic Diseases (SIMD)
2005	Member, Clinical Biochemical Genetics Standard Setting Webcast Study, National Board of Medical Examiners (NBME)
2005-2006	Member, New York-Mid Atlantic Consortium for Genetic and Newborn Screening Services (NYMAC) and the Middle Atlantic Regional Human Genetic Network (MARHGN)
2005-2006	Member, Panel on Newborn Screening ACT Sheets, American College of Medical Genetics (ACMG) and the Department of Health and Human Services (HHS)
2006	Ucyclyd Pharma, Inc. Physician Advisory Board
2015-	Co-Chair, Metabolomics Working Group, Undiagnosed Diseases Network (UDN) (NIH/NHGRI U01HG007690)
2021-	Member, CAP/ACMG BCMG Committee

International Committees

1985-	Member, The Society for the Study of Inborn Errors of Metabolism (SSIEM)
2000-2006	Corresponding Member for the USA, The Society for the Study of Inborn Errors of Metabolism (SSIEM)
1997-2007	Member, Scientific Advisory Board, United Mitochondrial Disease Foundation (UMDF)
2001-	Medical Advisory Board, Propionic Acidemia Foundation

2004-2005	Member of the International Scientific Organizing Committee, on The International Symposium on Galactosemia: Facts and unresolved issues, Fulda, Germany. November 16-18, 2005.
2009	Co-Chair, Organizing Committee, Stanton Segal Symposium at the 11th International Congress of Inborn Errors of Metabolism (ICIEM), San Diego, CA. September 1st, 2009.
2021-	Member, CLSI DDC NBS (Galactosemia) Committee
2021-	Member, IOC Committee, *ICIEM 2021* International Meeting

Professional Societies

International Societies:

1985-	Member, The Society for the Study of Inborn Errors of Metabolism (SSIEM)
2000-2006	Corresponding Member for the USA, The Society for the Study of Inborn Errors of Metabolism (SSIEM)
1997	Member, International Society for Neonatal Screening

National Societies:

1993-	Founding Fellow, American College of Medical Genetics (ACMG)
1987-	Member, American Society of Human Genetics (ASHG)
2003-	Member, The American Society for Biochemistry and Molecular Biology (ASBMB)
2000-	Member, American Society of Gene Therapy
1996-	Member, American Pediatric Society (APS)
1983-	Member, Society for Pediatric Research (SPR)
1986-	Member, American Diabetes Association (ADA)
1986-	Member, Juvenile Diabetes Foundation (JDF)
1987-	Member, The Lawson Wilkins Pediatric Endocrine Society
1999-	Member, The Endocrine Society
2003-	Member, Sigma Xi Scientific Research Society
1987-	Member, Society for Inherited Metabolic Disorders (SIMD)
2001-2008	Member, Board of Directors, Society for Inherited Metabolic Disorders (SIMD)
2002-2008	Chair, Newborn Screening Committee, Society for Inherited Metabolic Disorders (SIMD)
2009-	Member, Board of Directors, Society for Inherited Metabolic Disorders (SIMD)
2015-2017	Membership Chair and Officer, Board of Directors, The Society for Inherited Metabolic Disorders (SIMD)
2017-2019	Program Chair and Officer, Board of Directors, The Society for Inherited Metabolic Disorders (SIMD)
2003-	Member, Medical Advisory Board, Propionic Acidemia Foundation

Local Societies:

Member, Philadelphia Pediatric Society
Member, Philadelphia Endocrine Society
Member, Philadelphia County Medical Society
Member, Pennsylvania Medical Society

Editorial Boards:

2001-2006	Associate Editor, Diabetes
2005-	Member, Editorial Advisory Board, Current Diabetes Reviews
2004-	Communicating Editor, The Journal of Inherited Metabolic Diseases
2012-	Member, Editorial Board, Metabolism- Clinical and Experimental
2019-2021	Co-Editor (Edward R.B. McCabe, Co-editor), Molecular Genetics and Metabolism-Reports

Editorial Review:

Ad hoc reviewer for:

American Journal of Clinical Nutrition
American Journal of Human Genetics
American Journal of Medical Genetics-Part A
American Journal of Physiology
Annals of Internal Medicine
Annals of Neurology
Annals of Nutrition and Metabolism
Archives of Biochemistry and Biophysics
Archives of General Psychiatry
Archives of Pediatrics and Adolescent Medicine
Biochem. Biophys. Res. Commun.
Biochimica et Biophysica Acta
Biochimica et Biophysica Acta- Molecular Basis of Disease
Biochimie
BioMed Central Pediatrics
Biomedicine & Pharmacotherapy
Brain Research
British Medical Journal
Canadian Journal of Neurological Sciences
Cell Biology and Toxicology
Chemistry and Physics of Lipids
Clinical Biochemistry
Clinical Chemistry
Clinical Chemistry and Laboratory Medicine
Clinical Genetics
Diabetes
Disability and Rehabilitation
Early Human Development

Epilepsia
European Journal of Human Genetics
European Journal of Paediatric Neurology
European Neuropsychopharmacology
Expert Opinion on Orphan Drugs
FEBS Letters
Fertility and Sterility
Frontiers in Genetics
Future Medicinal Chemistry
Gene Reports
Genetic Testing
Genetics in Medicine
Hepatic Medicine: Evidence and Research
Human Molecular Genetics
Human Mutation
Institute for Laboratory Animal Research Journal
IUBMB Life
Journal of Biological Chemistry
Journal of Cell Physiology
Journal of Child Neurology
Journal of Clinical Investigation
Journal of Inherited Metabolic Disease
Journal of Membrane Biology
Journal of Neurochemistry
Journal of Pediatrics
Journal of Personalized Medicine
Journal of the American Medical Association
Life Sciences
Metabolism- Clinical and Experimental
Molecular Genetics and Genomic Medicine
Molecular Genetics and Metabolism
Molecular Psychiatry
Molecular Therapy
Nature Reviews Endocrinology
NeuroMolecular Medicine
Neuropsychopharmacology
Neurotherapeutics
New England Journal of Medicine
Orphanet Journal of Rare Diseases
Pediatric Research
Pediatrics
Plos One
Proceedings of the National Academy of Sciences
Reproduction
Science
Science Signaling

Science Translational Medicine
World Journal of Pediatrics

Review Panel Service:

2003-2009	Member, Gene Therapy and Inborn Errors Special Emphasis Panel, ZRG1 GTIE-A (01) (S), National Institutes of Health (NIH)
12/2005	Ad hoc Member, General Clinical Research Center (GCRC) Review Panel, National Center for Research Resources (NCRR), National Institutes of Health (NIH)
3/27/06	Ad hoc Member, Drug Discovery Special Emphasis Panel, ZRGI MDCN-C (91), National Institutes of Health (NIH)
6/9/06	Ad hoc Member, "Innovative Therapies and Clinical Studies for Screenable Disorders" Review Panel, ZHD1 MRG-C (07), National Institutes of Health (NIH)
3/28/08	Ad hoc Member, "Innovative Therapies and Clinical Studies for Screenable Disorders" Review Panel, ZHD1 MRG-C (07), National Institutes of Health (NIH)
8/12/08	Ad hoc Member, "Innovative Therapies and Clinical Studies for Screenable Disorders" Review Panel, ZHD1 MRG-C (PA), National Institutes of Health (NIH)
7/8/09	Ad hoc Member, "Innovative Therapies and Clinical Studies for Screenable Disorders" Review Panel, ZHD1 MRG-C (07), National Institutes of Health (NIH)
11/3/09	Ad hoc Member, "Screening for Inherited Disorders" Review Panel, ZHD1 DSR-N (08), National Institutes of Health (NIH)
2006-2009	Chairman, Rare Diseases Clinical Research Network (RDCRN) Data and Safety Monitoring Board/Observational Study Monitoring Board (DSMB/OSMB)2, National Center for Research Resources (NCRR), National Institutes of Health (NIH)
8/21/08	Committee member, Working Group for Gene Therapy and Inborn Errors Study Section, National Institutes of Health (NIH)
2/12/09	Committee member, Working Group for Gene Therapy and Inborn Errors Study Section, National Institutes of Health (NIH)
2/27/09	Committee member, Developmental Biology Subcommittee, National Institutes of Health (NIH)
2009-2013	Member, Therapeutic Approaches to Genetic Diseases (TAG) Study Section, National Institutes of Health (NIH)
2016	Member, Genetics, Genomes and Genes Working Group to Access Peer Review, National Institutes of Health (NIH)

Research Grant Reviewer

Canadian MRC
Welcome Trust
The Israel Science Foundation
Barth Syndrome Foundation

Propionic Acidemia Foundation

Honors and Prizes:

1981	NICHD Clinical Investigator Award - Pediatrics
1993	Ethel Brown Foerderer Fund Award for Excellence in Clinical Research
1999	Ethel Brown Foerderer Fund Award for Excellence in Clinical Research
2005	The 2004 Emmanuel Shapiro Society for Inherited Metabolic Disorders Award
2010	Journal of Inherited Metabolic Diseases Communicating Editor Award
2010	Harvard University, Honorary Masters Degree in Medicine
2012	Journal of Inherited Metabolic Diseases Guest Editor Award
2016	Jefferson Medical College Alumni Achievement Award
2019-2021	President-elect, The Society for Inherited Metabolic Disorders (SIMD)
2021-2023	President, The Society for Inherited Metabolic Disorders (SIMD)

Report of Funded and Unfunded Projects

Past Funded Projects:

07/01/81-06/30/84 PI, NIH, $146,100
NIH – KO8 HD000427 NICHD Clinical Investigator Award- Pediatrics
The purpose of this proposal was to use rat brain synaptosomes as a model to study galactose toxicity-induced abnormalities and phosphoinositide metabolism.

07/01/86-07/28/87 PI, NIH, $21,750
NIH-University of Pennsylvania, Diabetes Center-Pilot Project
The purpose of this proposal was to generate a macrovascular endothelial cell culture system to study abnormalities in polyol metabolism that are relevant to diabetic complications.

09/01/87-08/31/89 PI, Juvenile Diabetes Foundation, $66,100
Juvenile Diabetes Foundation
The purpose of this proposal was to utilize macrovascular endothelial cells in culture to study glucose and galactose induced alterations in myo-inositol and phosphatidylinositol turnover.

07/01/87-06/30/89 PI, WW Smith Charitable Trust, $103,543
WW Smith Charitable Trust
The purpose of this proposal was to study diabetic complications that involve the vascular endothelium using a cell culture model system.

07/01/88-06/30/93 PI, NIH, $520,000
NIH-RO1 DK040382, The Biochemical Basis of Diabetic Angiopathy
The purpose of this proposal was to study glucose and galactose induced abnormalities in endothelial transport of myo-inositol, turnover of phosphatidylinositol and bradykinin related signal abnormalities.

07/01/93-06/30/96 PI for Project 1, NIH, $262,000
NIH-PO1 HD29847, Enigma of Galactosemia - Searching for Answers, Project 1: Galactose Pathways and Their Regulation in Galactosemic Patients, Program PI: Stanton Segal
The purpose of project 1 of this proposal was to study a whole body galactose metabolism including breath testing in patients in the general clinical research setting.

03/01/94-08/31/94 PI, University of Pennsylvania Diabetes Center, $12,305
University of Pennsylvania Diabetes Center, Myo-Inositol Transporter Gene
The purpose of this proposal was to clone the sodium myo-inositol cotransporter gene and delineate its organization.

09/01/94-08/31/96 PI, Juvenile Diabetes Foundation, $100,000
Juvenile Diabetes Foundation, Osmoadaptive Myo-Inositol Transporter Gene and Diabetes
The purpose of this proposal was to determine whether signal nucleotide polymorphisms or mutations in the sodium cotransporter gene are linked to an increased risk for development of diabetic renal complications.

09/01/95-08/31/98 PI, NIH, $363,000
NIH - RO1 HD033922, Transport-Mediated Regulation of Cellular Myo-Inositol Levels
The purpose of this proposal was to determine whether the increased expression of the sodium myo-inositol cotransporter gene due to three copies in trisomy 21 state is linked to phenotype abnormalities in down syndrome.

07/01/96-06/30/00 PI for Project 1, NIH, $270,427
NIH-PO1 HD 29847, Project 1: Galactose Pathways and their Regulation in Galactosemic Patients, Program PI: Stanton Segal
The purpose of this proposal was to continue our work on whole body galactose metabolism modeling in the patient with hereditary galactosemia and to establish important genotype-phenotype relationships.

09/01/96-08/31/98 PI, University of Pennsylvania Institute for Human Gene Therapy, $75,801
University of Pennsylvania Institute for Human Gene Therapy, The Na+/myo-inositol cotransporter (SLC5A3) gene defect in the murine species
The purpose of this work was to generate a homozygous ablation of the murine sodium myo-inositol cotransporter gene and determine the effect on phenotype.

07/01/97-06/30/00 PI, American Diabetes Association, $150,000
American Diabetes Association, Osmoregulatory Na+/myo-inositol cotransporter (SLC5A3) gene and diabetic nephropathy
The purpose of this proposal was to link single nucleotide polymorphisms in the SLC5A3 gene with enhanced propensity for diabetic nephropathy.

06/01/00-05/31/04 PI, March of Dimes, $90,000
March of Dimes, Murine homozygous Na+/myo-inositol cotransporter (SLC5A3) deletion

model
The purpose of this proposal was to establish the abnormalities in myo-inositol transport, phosphatidylinositol metabolism and electrophysiological disturbances in murine knockout SLC5A3 models.

06/12/09-06/11/10 PI, Parents of Galactosemic Children (PGC) Organization, $30,000
Parents of Galactosemic Children (PGC) Organization Research Award, Health and Psychosocial Outcome in Adults with Classic Galactosemia"
The goal of the award is to perform the first prospective, cross-sectional study of adults with galactosemia with an emphasis on multiorgan complications including cognitive impairment, speech defects, neurologic abnormalities, psychiatric disease, gonadal dysfunction, fertility, bone mineral density, and social well-being.

10/01/08-09/30/13 PI, The Manton Foundation Senior Scientist Award, $140,000
The Manton Foundation
The goal of the award is to better understand the pathophysiology of hereditary galactosemia and develop new treatment strategies using multiple approaches including whole body galactose metabolism studies, employment of model systems such as the knockout SLC5A3 animal model and study of new treatment modalities.

07/01/13-06/30/2015 PI, The Galactosemia Foundation Award, $48,000
The Galactosemia Foundation, Inc. Research Award, Modifier Genes and Epigenetics Effects in Classic Galactosemia
The goal of the award is to prepare induced pluripotent stem cells from skin and blood cells derived from patients with classic galactosemia, allow them to differentiate into neural progenitor cells and neurons, and study the consequences of galactose stress on the transcriptome, methylome, and metabolome in neurons with a Q188R/Q188R genotype with and without CRISPR/Cas9 gene editing. The data that emerges from this work will be used to help inform our analysis of whole genome sequencing that is to be performed on each individual subject whose cells were used to create the experimental neurons.

01/15/2015-12/31/2016 Co-Investigator, NIH
NIH/NIAID R21AI113459-01A1 (PI:Notarangelo), Characterization of a novel combined immunodeficiency with skeletal dysplasia
The purpose of this project is to demonstrate that the synthesis of heparan sulfate is perturbed in cells derived from the patients with a EXTL3 gene defect.

03/15/2012-03/31/2019 Site Lead/Co-Investigator, NIH $51,803
NIH 2 R01HD058567 (PI:Mendel Tuchman), N-carbamylglutamate in the Treatment of Hyperammonemia: Developmental Outcome and Safety in Propionic Acidemia and Methylmalonic Acidemia
The goal of this study has 3 aims: 1. To determine whether N-carbamylglutamate (NCG) treatment of acute hyperammonemia in severe, neonatal onset propionic acidemia (PA) and methylmalonic acidemia (MMA) improves neurodevelopmental outcome, and whether it is safe. 2. To determine whether NCG treatment of acute hyperammonemia accelerates the resolution of hyperammonemia and clinical recovery in patients with severe PA and MMA and in those with partial CPS I deficiency (CPSD) and ornithine transcarbamylase deficiency (OTCD), and

whether it is safe. 3. To determine whether the effect of a 3-day NCG treatment on ureagenesis in metabolically stable patients is predictive of the outcomes observed in Aims 1 and 2.

07/01/2018 – 06/30/2020 Co-Investigator, NIH, $150,000
NIH 1 R21HD096355-01 (PI: Richard Goldstein), Genetics of Sudden Unexpected Death in Pediatrics
The major goal of this project is to discover the causes of sudden death in infants and children using whole exome sequencing and metabolomics analyses.

07/01/19-06/30/2020 PI, The Galactosemia Foundation Award, $35,000
The Galactosemia Foundation, Inc. Research Award, Establishing a brain organoid system to study neurological complications of classic galactosemia
The major goal of this proposal is to optimize generation of brain organoids for galactosemic iPSCs, and investigate their morphological, molecular and genetic differences compared to control organoids.

05/01/2021 04/30/2022, PI, The Galactosemia Foundation Award
The Galactosemia Foundation, Inc. Research Award
"Prevalence and Progression of Cognitive, Motor and Socioemotional Challenges Experienced by Adults with Classic Galactosemia"
The goal of this project is to better delineate the long-term outcome with patients with galactosemia who are over 30 years of age.

04/01/2014-6/30/2022 Co-Investigator, NIH, $140,066
NIH/NHGRI U01HG007690-04 (PI: Joseph Loscalzo), Center for Integrated Approaches to Undiagnosed Diseases
The objective of this proposal is to evaluate patients with undiagnosed diseases in the UDN as part of a joint clinical site with the Brigham and Women's Hospital and the Massachusetts General Hospital. Together with Devin Oglesbee and Ian Lanza, I am serving as the Co-Chair for the UDN Metabolomics Working Group.

07/01/2017 – 06/30/2020 Site Lead/Co-Investigator, NIH, $71,820
NIH/NICHHD 1R01NR016991 (PI: Jerry Vockley), Use of Home Phenylalanine Meter to Help Manage PKU
The major goal of this project is the testing of a new device that may be able to measure phenylalanine levels in the home.

08/25/2014-07/31/2022 Site Lead, NIH, $56,497
NIH U54HD061221 (PI: Gropman) Rare Diseases Clinical Research Consortia (RDCRC)
Project 1 – Rare Diseases Clinical Research Consortia (RDCRC) for the RDCR Network
The goals of this project are to delineate the natural history of the urea cycle disorders and study the nature of brain complications using MRI/MRS.

Current Funded Projects:

07/01/2019 – 06/30/2024 Site Lead/Co-Investigator, NIH, $115,049
NIH U54 (PI: Cary Harding), HyperPhe Consortium
The goals of the project are to determine the natural history of patients with hyperphenylalaninemia and perform clinical trials. I am serving as the Co-director of Career Development Core.

07/01/2019-06/30/2024 Site Lead/Co-Investigator, NIH, $51,806
NIH U54 (PI: Eva Morava), Frontiers in Congenital Disorders of Glycosylation
Natural History Project-The goals of the project are to determine the natural history of patients with CDGs and perform clinical trials. I am serving as the Co-director of Administrative Core.

Current Unfunded Projects:

PI, **Endogenous galactose production in patients with galactosemia**
The goal is to develop a new method to accurately measure whole body rates of endogenous galactose production. The new approach involves the use of stable isotopically-labeled glucose to assess the rate of conversion of glucose to galactose in the patient following steady-state labeling of plasma glucose.

PI, **Osteoporosis and galactosemia**
The goal is to determine whether decreased bone mineral content, an apparent diet-independent complication in girls and boys with galactosemia, is related to the use of a lactose-free soy formula or is a very early complication in galactosemia, i.e. is an integral feature of this inherited disorder and is independent of calcium and vitamin D intake.

PI, **Inositol metabolism in brain and during development**
The goal of this work is to determine the mechanism(s) whereby a reduction in neuronal myo-inositol concentrations disturbs homeostasis in the nervous system.

<u>Report of Local Teaching and Training</u>

1. Local contributions

a. Courses for medical/dental/Ph.D. students

1981 – 1995	Medicine 100, University of Pennsylvania			
			contact time	*prep time*
	Preceptor	4 medical students	16 hours/year for 14 years	2 hours/year for 14 years

2005 – 2006	Clinical Medicine (ICM1) course, Thomas Jefferson University

			contact time	*prep time*
	Preceptor	10 medical students	3 hours/month for 1 year	5 hours/year for 1 year

2007 – 2014	HST 160: Molecular Biology and Genetics in Medicine, Harvard Medical School			
			contact time	*prep time*
	Clinical mentor	1-2 students: 1-2 medical students & 1-2 students from MIT	2-4 hours/month for 3 months	1 hour/year for 1 year

2008 – 2009	HMS: Human Genetics Course			
			contact time	*prep time*
	Preceptor for Journal Club	10 medical students	2 hours/day for 1 day	2.5 hours/year for 1 year

2011 –	HMS: Human Genetics Course (IN755.0)			
			contact time	*prep time*
	Lecturer	1st year medical students	1.5 hours/day for 1 day	2.5 hours/year for 1 year

2011 –	HST 146: Human Biochemistry and Metabolic Diseases Course			
			contact time	*prep time*
	Lecturer ("frontiers" lecture, with an illustrative patient)	30 1st year medical students	2.5 hours/day for 1 day	2.5 hours/year for 1 year

2014 –	HST 146: Human Biochemistry and Metabolic Diseases Course			
			contact time	*prep time*
	Block leader for amino acids in the course	30 1st year medical students	1 hour/day for 1 day	2.5 hours/year for 1 year

Major Teaching and Clinical Responsibilities at the University of Pennsylvania:

1. Attending for Division of Genetics & Metabolism, The Children's Hospital of Philadelphia- 2 months/year
2. Metabolic Diseases Clinic, The Children's Hospital of Philadelphia - 1 afternoon/week
3. Attending for Division of Endocrinology and Diabetes, The Children's Hospital of Philadelphia - 1 month/year
4. Endocrinology/Diabetes Clinic, The Children's Hospital of Philadelphia-1 morning/week
5. Clinical Genetics Clinic, Hospital of the University of Pennsylvania - 1 morning/week

6. Senior Rounds, The Children's Hospital of Philadelphia - 1 morning/week
8. Member of the Nutrition Center of The Children's Hospital of Philadelphia
9. Supervisor of post-doctoral students
10. Preceptor for Medicine 303 course, Human Gene Therapy

Major Teaching and Clinical Responsibilities at the Jefferson Medical School:
Preceptor for the Introduction to Clinical Medicine (ICM1) course (first year medical students)

Lecturer, Neuroscience (first year Neuroscience graduate students)

Major Teaching and Clinical Responsibilities at the George Washington University School of Medicine:
1. Attending for Division of Genetics & Metabolism, Children's National Medical Center
2. Metabolic Diseases Clinic, Children's National Medical Center

b. Graduate Medical Courses

1981 – 2001	Genetics Fellows, University of Pennsylvania			
			contact time	*prep time*
	Conference Leader	6 Fellows	1 hour/month for 20 years	10 yours/year for 20 years

1985 – 1995	Pediatric Residents, University of Pennsylvania			
			contact time	*prep time*
	Faculty Facilitator	25 Residents	1 hour/month for 10 years	4 hours/year for 10 years

1998 – 2001	Endocrinology/Diabetes Fellows, University of Pennsylvania			
			contact time	*prep time*
	Conference Leader	4 Fellows	4 hours/year for 3 years	2 hours/year for 3 years

1999 – 2000	Cell and Molecular Biology Graduate Group, University of Pennsylvania			
			contact time	*prep time*
	Lecturer	20 Graduate Students	1 hour/year for 2 years	1.5 hours/year for 2 years

2005 – 2006	Neuroscience Graduate Students, Thomas Jefferson University			
			contact time	*prep time*
	Lecturer	15 Graduate Students	1 hour/year for 1 year(s)	2 hours/year for 1 year(s)

2011 – 2014	Harvard University Chemical Biology Ph.D. Program		
		contact time	*prep time*
	Member, Laura Ann Strittmatter thesis and defense committee	4 hours	2 hours

04/22/2013	Harvard University Chemical Biology Ph.D. Program		
		contact time	*prep time*
	Member, Steve Hershman defense committee	2 hours	1 hour

Local Invited Teaching Presentations

05/26/88 "Use of an endothelial cell culture system to study the problem of inositol insufficiency in diabetes", The Lankenau Medical Research Center Guest Lecture Series, Lankenau Medical Center, Philadelphia, PA

12/20/88 "Metabolic Diseases: Recognition and Guide to Use of Metabolic Screening Tests", Guest Lecture Series Thomas Jefferson University/Jefferson Medical College, Philadelphia, PA

04/17/91 "Congenital Chloride Diarrhea", Grand Rounds, Thomas Jefferson University/Jefferson Medical College, Philadelphia, PA

10/03/91 "Nutritional Therapy of Maple Syrup Urine Disease", Grand Rounds, Thomas Jefferson University/Jefferson Medical College, Philadelphia, PA

03/31/92 "Inborn Errors of Metabolism in the Neonate", Grand Rounds, Albert Einstein Medical Center, Philadelphia, PA

04/26/94 "Up regulation of endothelial inositol 1,4,5-trisphosphate signaling in experimental diabetes", Diabetes Center Seminar, Department of Medicine, Temple University, Philadelphia, PA

02/20/96 "Genetic Disorders in Medicine", Grand Rounds, Thomas Jefferson University/Jefferson Medical College, Philadelphia, PA

02/03/99 "Galactosemia", Grand Rounds, The Children's Hospital of Philadelphia, Philadelphia, PA

03/01/01 "An Approach to the Inherited Metabolic Diseases", Grand Rounds, Department of Pediatrics, Jefferson Medical College, Philadelphia, PA

03/29/05 "Hereditary Galactosemia", Grand Rounds, Department of Pediatric and Adolescent Medicine, Albert Einstein Medical Center, Philadelphia, PA

05/23/06 "Inositol and the Developing Mammalian Brain", Farber Institute for the Neuroscience Seminar Series, Thomas Jefferson University, Philadelphia, PA

06/04/06 "Investigating Metabolic Diseases," Brain Teasers: Delaware Valley Child Neuroscience Update, Alfred I. DuPont Hospital for Children, Nemours Children's Clinic, Philadelphia, PA

03/14/07 "Hereditary Galactosemia", Grand Rounds, Boston Children's Hospital, Boston, MA

07/23/07 "Galactosemia Update", Tufts-NEMC Genetics Rounds, Division of Genetics, New England Medical Center, Boston, MA

10/02/07 "Metabolic Diseases," Pediatric Grand Rounds, St. Joseph Hospital, Nashua, NH

11/07/07 "Metabolic Disorders: A New Age of Management," Boston Children's Hospital Fall Seminar, Advances in Pediatric Health Care, Norwood, MA

11/03/09 "Carnitine Transporter Deficiency Cardiomyopathy", Cardiology Grand Rounds, Department of Medicine, Boston Medical Center, Boston, MA

04/21/10 "When to Suspect a Metabolic Disorder", Pediatric Grand Rounds, Holy Family Hospital, Methuen, MA

12/01/10 "Galactosemia", Shire visiting lecture program, Lexington, MA

05/14/12 "Galactosemia: have we made a mistake in treatment?" Genetics Rounds, Division of Genetics, Tufts Medical Center, Boston, MA

10/22/13 "GI and Liver Manifestations of Metabolic Disease", GI Fellows Lecture Series sponsored by the Education Committee of the HMS Fellowship in Pediatric Gastroenterology and Nutrition, Boston Children's Hospital, Boston, MA

11/07/14 "How biochemical genetic diseases inform the science of human metabolism," Endocrine Grand Rounds, Beth Israel Deaconess Medical Center, Boston, MA

03/13/15 "Overview of the metabolic/biochemical genetic diseases", Pediatric Neurology Grand Rounds, Boston Medical Center, Boston, MA

04/30/15 Harvard University Genetic Disorder Project Symposium, Panelist, Cambridge, MA

02/24/16 "The metabolic infant", NICU Core Curriculum Lectures, Division of Neonatology, Boston Children's Hospital, Boston, MA

05/18/17 "Two siblings with identical genotypes but divergent mitochondrial phenotypes" MitoCase Conference, Massachusetts General Hospital, Boston, MA

04/27/18 "21-year-old female with chronic encephalopathy, bilateral optic neuropathy, basal ganglia lesions, dystonia and spasticity: Secondary mitochondropathies" MitoCase Conference, Massachusetts General Hospital, Boston, MA

03/08/19 "A lactic acidosis disorder" MitoCase Conference, Massachusetts General Hospital, Boston, MA

03/03/20 "Urea Cycle Disorders", Physician Advocacy Program Lecture, Boston University School of Medicine, Boston, MA

03/02/22 " Metabolic Disease" Boston Children's Hospital, Division of Gastroenterology, Boston, MA

01/05/23 "Galactosemia and Fetal Brain MRI/MRS", Neonatology Conference BIDMC, Boston, MA

d. Continuing Medical Education Courses	
The Children's Hospital of Philadelphia	
2000, 2001	Pediatric Update Lecturer: 200 participants, 1 hour contact time per year, 2 hours prep time per year
Boston Children's Hospital	
2007	Pediatric Update Group Leader: 130 participants, 1.5 hours contact time per year, 3 hours prep time per year

e. Advisory and Supervisory Responsibilities in Clinical or Laboratory Setting	
1981-2001	2 Fellows for 25 hrs/year, Biochemical Genetics Laboratory, The Children's Hospital of Philadelphia

	2001-2003	2 Fellows for 25 hrs/year, Biochemical Genetics Laboratory, Children's National Medical Center, Washington, D.C.
	2016-	1 Fellow for 4 supervisory hours/year, Leder Human Biology Clinical Course (Nelson Lamarchi) Boston Children's Hospital

f. Teaching Leadership Roles

	2006-2023	Director of Biochemical Genetics Training Program for residents and post-doctoral fellows, Boston Children's Hospital

g. Names of advisees or trainees

Training Duration	*Name*	*Current Position*
1981-1984	Dennis Cryer	Bristol-Myers Squibb Co., Princeton, NJ
1981-1984	Jerome Gorski	Chief, Division of Medical Genetics, Professor of Child Health, University of Missouri, Columbia, MO
1982-1985	David A.H. Whiteman	Medical Director, Shire Human Genetic Therapies Inc., Cambridge, MA and Zurich, Switzerland
1984-1987	Marvin Natowicz	Professor, Case Western Reserve University, Cleveland, OH
1984-1987	Rhonda Schnur	Professor, University of Medicine and Dentistry of New Jersey, Chief, Division of Genetics, Cooper Medical Center, Camden, NJ
1984-1987	Randall Heidenreich	Professor, University of New Mexico, Chief, Division of Genetics, Albuquerque, NM
1985-1987	Natalie Blagowidow	Medical Director, Genetics Prenatal Diagnostic Center, Harvey Institute for Human Genetics, Baltimore, MD
1985-1987	Alan Donnenfeld	Neonatologist, Main Line Perinatology, Wynnewood, PA
1985-1988	D. Holmes Morton	MacArthur award recipient, Clinic for Special Children, Strasburg, PA
1985-1988	Samuel Harold Sigal	Assistant Professor, Department of Medicine, New York-Presbyterian Hospital/ Weill Cornell Medical Center
1985-1989	Julie Neidich	Director, Biochemical Geneticist, Nichols Institute-Quest Diagnostics, San Juan Capistrano, CA
1985-1997	John C. Baker	Professor, Kaiser Permanente, Oakland, CA
1986-1989	Max Muenke	Chief, Medical Genetics Branch, National Human Genome Research Institute, NIH, Bethesda, MD
1987-1989	Deborah Driscoll	Chair, Department of Obstetrics/Gynecology, University of Pennsylvania

1988-1989	Fred Levine	Associate Professor, Center for Molecular Genetics, University of California San Diego
1988-1990	Samuel M. Rosenberg	Pulmonologist, Rockville, MD
1988-1989; 1991-1992	Carolyn A. Bay	Associate Professor, Chief, Division of Genetics, University of Kentucky
1989-1991	Nancy Rose	Obstetrician, Intermountain Health Care, LDS Hospital, Salt Lake City, UT
1990-1993	James B. Gibson	Associate Professor, Division of Genetics, University of Texas at San Antonio, Children's Hospital of Dallas
1990-1993	JoAnn Bergoffen	Chief, Dept. of Genetics, Kaiser Permanente Medical Group, San Jose, CA
1991-1994	Roy A. Johanson	Senior Research Associate, Department of Neurology, Thomas Jefferson University
1991-1992	Kara S. Ornstein	Undergraduate, University of Pennsylvania
1991-1993	Ousina Adewale	Geneticist, Chattanooga, TN
1991-1993	Ronald E. Barabas	Neurologist/ Geneticist, Child Neurology Associates, West Long Branch, NJ
1992-1995	Nathaniel H. Robin	Professor, University of Alabama at Birmingham, Director, Genetics Residency Programs, Birmingham, AL
1993-1996	Wadia Mulla	Clinical Geneticist, Department of Obstetrics and Gynecology, Christiana Care Health System, Newark, DE
1993-1996	Joan Pellegrino	Geneticist, SUNY Upstate Medical University, Syracuse, NY
1994-1996	Teresa Parrella	Metabolic Specialist, Children's Hospital, Turino, Italy
1994-1997	Anthony D. Lucente	Undergraduate, University of Pennsylvania
1994-1997	Ian Krantz	Associate Professor, University of Pennsylvania, Division of Genetics, The Children's Hospital of Philadelphia
1995-1997	Eric Roessler	Biochemical Geneticist, NHGRI, Bethesda, MD
1995-1998	Jeffrey Ming	Attending Physician, Division of Genetics, The Children's Hospital of Philadelphia
1995-1999	Karen Gripp	Associate Professor, Thomas Jefferson University, Chief, Division of Genetics, A.I. DuPont Hospital for Children
1996-1999	Karen McVeigh	Assistant Professor, University of Southern California School of Medicine, Children's Hospital Los Angeles
1996-1999	Katherine L. Nathanson	Associate Professor, University of Pennsylvania
1996-1999	Rosemarie Smith	Chief, Clinical Genetics, Maine Medical Center, Portland, ME
1996-2000	George Anadiotis	Biochemical Geneticist, Legacy Emanuel Hospital, Portland, OR
1997-2000	Shuang Wu	Bristol-Myers Squibb Co., Princeton, NJ

1997-1999	Cong Ning	Pediatric Endocrinologist, Shady Grove Adventist Hospital, Rockville, Maryland
1997-1999	Steffi F. Dreha - Kulaczewski	Faculty of Medicine, Departments of Pediatrics and Pediatric Neurology, Georg August University, Göttingen, Germany
1997-2000	David G. Brooks	Associate Director, Clinical Molecular Profiling, Merck Research Labs, West Point, PA
1997-2000	Sulagna Saitta	Associate Professor, University of Pennsylvania, Division of Genetics, The Children's Hospital of Philadelphia
1998-2001	Lynne Ierardi-Curto	Medical Geneticist, Laboratory Corporation of America (LabCorp), Northeast Division, Genetics Services, Raritan, NJ
1998-2001	Ayala Laufer-Cahana	Medical Geneticist, Wynnewood, PA
1999-2002	Charles P. Venditti	Director of the Organic Acid Research Unit, National Institutes of Health, Bethesda, MD
2000-2002	Andrea Kelly	Assistant Professor, University of Pennsylvania, Division of Endocrinology/ Diabetes, The Children's Hospital of Philadelphia, PA
2000-2002	Dina J. Zand	Assistant Professor, George Washington University School of Medicine, Children's National Medical Center, Washington DC
2000-2002	Ralph J. DeBerardinis	Assistant Professor of Pediatrics and Genetics, University of Texas Southwestern Medical Center, Dallas, TX (Co-recipient of the HMS 2008 William K. Bowes, Jr. Award in Medical Genetics)
2001-2002	Jaya Ganesh	Assistant Professor AC, University of Pennsylvania, Division of Rehabilitation and Biochemical Genetics-Metabolic Disease, The Children's Hospital of Philadelphia
2001-2002	Lynette Gillis	Assistant Professor, Vanderbilt University, Nashville, TN
2002-2003	Susan Sparks	Biochemical Geneticist, Children's National Medical Center, Washington, DC.
2002-2003	B.P. Brooks	Director, Ophthalmic Genetics Clinic, Children's National Medical Center & National Eye Institute, Bethesda, MD
2005-2006	Andrea Hunt	Medical Student, Jefferson Medical College; Resident in Pediatrics, University of Michigan, Mott's Children's Hospital
2006-2008	Philip James	Instructor, Harvard Medical School, Division of Genetics, Boston Children's Hospital, MA
2006-2007	Fowzan Alkuraya	2010 Bowes Awardee, Assistant Professor of Human Genetics, Alfaisal University College of Medicine, King Faisal Specialist Hospital and Research Center; Instructor in Pediatrics, Harvard Medical School

2006-2007	Carrie Schmid	Clinical Geneticist, San Antonio Military Medical Center, San Antonio, TX
2006-2009	Abidemi Adegbola	Instructor, Harvard Medical School, Division of Genetics, Massachusetts General Hospital, Boston, MA
2006-2009	Elliot Stolerman	Clinical Instructor in Pediatrics, University of Alabama, Burmingham, AL
2006-2009	Roman Yusupov	Attending Physician, Memorial Regional Hospital, Hollywood, FL
2006-2009	Vijay Hedge	Post Doctoral Fellow, Brigham and Women's Hospital, Mel B. Feany Laboratory, Boston, MA
2006-2008	Joseph Thakuria	Instructor, Harvard Medical School, Division of Genetics, Massachusetts General Hospital
2006-2008	Phillip James	Assistant Professor, University of Arizona, Phoenix Children's Hospital, Phoenix, AZ
2006-2010	Roberto Buccafusca	Graduate Student, Drexel University, Philadelphia, PA, Ph.D. degree awarded December 2010 (served as thesis advisor)
2008-2011	Yijun Li	Staff Scientist, New England Newborn Screening Public Health Department, Jamaica Plain, MA
2011-2013	Yuval Landau	Staff Neurologist, Tel Aviv, Israel
2013-2014	Hong Li	Emory University, Atlanta, GA
2014-2015	Ahmed Alfares	King Faisal Specialist Hospital and Research Centre, Saudi Arabia
2013-2015	Roy Peake	Instructor HMS, Department of Medicine, BCH
2014-2015	Lance Rodan	Instructor HMS, Department of Medicine, BCH
2015-2016	Amy Kritzer	Instructor HMS, Department of Medicine, BCH
2015-2016	Anne O'Donnell Luria	Instructor HMS, Department of Medicine, BCH, Broad Institute
2016-2017	Farrah Rajabi	Instructor HMS, Department of Medicine, BCH
2015-2017	Didem Demirbas Cakici	Postdoctoral Fellow, Berry Lab, BCH
2018-2019	Clara Hildebrant	Attending Physician, UNC Health
2018-2019	Joshua Baker	Attending, Physician, Lurie Children's Hospital
2018-2019	William Brucker	Attending Physician, Hasbro Children's Hospital
2019-2020	Chen-Han Wilfred Wu	Assistant Professor, Case Western Reserve University, Department of Genetics and Genome
2019-2020	Jasmine L. Knoll	Attending Physician, Phoenix Children's Hospital
2021-2022	Melinda Palma	Attending Physician, Boston Children's Hospital
2021-2022	Daniel Pomerantz	Postdoctoral Fellow, Beth Israel Deaconess Medical Center, Cummings Lab/Boston Children's Hospital lab

Regional, National, or International Contributions

a. Invited Presentations

01/15/84	"Relationship of Inositol, Phosphatidylinositol and Phosphatidic acid in CNS nerve-endings", International Chilton Conference on Inositol and Phospholipids, Dallas, TX
06/12/86	Maple Syrup Urine Disease, Guest Lecture Series, Wilmington Medical Center, Wilmington, DE
10/10/86	Hereditary Galactosemia, Guest Lecture Series, Wilmington Medical Center, Wilmington, DE
01/07/87	"An approach to the Hyperammonemic Newborn", Guest Lecture Series, Wilmington Medical Center, Wilmington, DE
06/12/87	"Hereditary Tyrosinemia", Guest Lecture Series, Wilmington Medical Center, Wilmington, DE
07/12/87	"Hyperammonemia", Grand Rounds, Cooper Medical Center, Camden, NJ
10/09/87	"Introduction to Screening for Metabolic Diseases", Guest Lecture Series, Wilmington Medical Center, Wilmington, DE
02/23/88	"Glucose, Myo-Inositol and Phosphoinositide Metabolism in Vascular Endothelial Cells" - Diabetic Complications Workshop on "Phosphoinositides and Na/K-ATPase in the Pathogenesis of Diabetic Complications", San Diego, CA
06/17/88	"Nutritional Therapy of MSUD", 1988 National Maple Syrup Urine Disease Symposium, Lancaster, PA
03/21/89	"Hereditary Galactosemia", Grand Rounds, Alfred I. du Pont Institute of the Nemours Foundation, Wilmington, DE
04/13/89	"The Role of Polyol and Inositol in Galactosemia" - NICHD and Children's Hospital of Los Angeles-sponsored conference, Galactosemia: New Frontiers in Research", Universal City, CA
06/18/92	"Inborn Errors of Metabolism in the Neonate", Neurology Lecture Series, A. I. du Pont Institute of the Nemours Foundation, Wilmington, DE
11/19/93	"The role of polyols in the pathophysiology of galactosemia" - International

Symposium on "Galactosemia - Facts and Unresolved Issues" Fulda, Germany

03/27/94 "Planning a prospective multicenter study of hereditary galactosemia", SERGG, Atlanta, GA.

07/13/94 "Hereditary Galactosemia", Grand Rounds, Doernbecher Children's Hospital, Oregon Health Sciences University, Portland, OR

07/27/94 "In vivo oxidation of ^{13}C-Galactose in patients with Galactosemia", Division of Genetics, Department of Pediatrics, Emory University School of Medicine, Atlanta, GA

04/18/97 "When and How to Test for a Metabolic Disease" Grand Rounds, Department of Pediatrics, Manmouth Medical Center, Long Branch, NJ

08/08/97 "Galactosemic complications and epigenetic phenomena", "Galactosemia – The 21st Century, Looking Forward", Annual Symposium of the Parents of Galactosemic Children Inc., Austin, TX

03/98 "An Overview of the Inborn Errors", Grand Rounds, Lehigh Valley Hospital, Wilkes Barre, PA

04/98 "Pediatric Presentation of Metabolic Diseases", Grand Rounds, York Hospital, York, PA

07/09/98 "Galactosemia," 20th Annual Scientific Meeting of the Human Genetic Society of Australasia, University of Melbourne, Melbourne, Australia

07/11/98 "Galactosemia", Lecture for Victoria Parents of galactosemic children, Children's Hospital, Melbourne, Australia

07/13/98 "Galactosemia", Lecture for New South Wales Parents of galactosemic children, New Children's Hospital, Sydney, Australia

07/13/98 "Secondary hyperammonemia and glutamine metabolism", Research Seminar, Division of Clinical and Biochemical Genetics, Department of Pediatrics, University of Sydney, New Children's Hospital, Sydney, Australia

12/15/98 "Epigenetic factors in the pathogenesis of galactosemia", Seminar Program, Department of Genetics, Yale University School of Medicine, New Haven, Connecticut

04/20/99 "Inborn Errors of Metabolism", Grand Rounds, Department of Pediatrics, South Jersey Hospital, Bridgeton, NJ

10/04/99 "An Overview of Biochemical Genetic Diseases", Visiting lecture, Department

of Pediatrics, The Children's Hospital, Beijing, The People's Republic of China

10/07/99 "Hereditary Galactosemia", Visiting lecture, Department of Pediatrics, The Children's Hospital, Shanghai, The People's Republic of China

06/16/00 "Growth Disturbances in Barth Syndrome", Barth Syndrome Symposium, Johns Hopkins University and the Kennedy-Krieger Institute, Baltimore, MD

03/30/01 "Galactosemia", Ross Metabolic Conference, Advances in Management of Inherited Metabolic Disorders, Costa Mesa, CA

04/09/01 "The Murine Sodium Myo-Inositol Cotransporter Gene Deletion Model", Research Institute Seminar Series, The McGill University-Montreal Children's Hospital, Montreal, Canada

06/22/01 "The Central Issues in Galactosemia" Annual Meeting of the Parents of Galactosemic Children, Inc., Atlanta, GA

06/30/01 "Nutritional Issues in Patients with UCDs" Annual Meeting of the National Urea Cycle Disorders Foundation, Houston, TX

06/30/01 "Pancreatitis", Annual Meeting of the Organic Acid Disorders Society, Houston, TX

07/16/01 "The importance of the SLC5A3 gene in the newborn mammal", Research Seminar Series, Children's National Research Institute, Children's National Medical Center, Washington, DC

07/30/01 "In vivo ^{13}C-galactose turnover and oxidative studies in Galactosemia", Guest Lecture, Department of Pediatrics, University of Kentucky, Lexington, KY

08/15/01 "SLC5A3 Gene", Research Seminar, Department of Biochemistry, University of Kentucky, Lexington, KY.

09/10/01 "Tandem Mass Spectrometry Beyond the Screening Laboratory", Enhancing the Implementation of Tandem Mass Spectrometry for Newborn Screening Laboratories, Wisconsin State Laboratory of Hygiene/CDC Symposium, University of Wisconsin, Center for Health Sciences, Madison WI.

02/19/02 "The Biochemical Genetics Perspective," Mitochondrial Disease Epidemiology Planning Workshop, United Mitochondrial Disease Foundation, Pittsburgh, PA

03/07/02 "Galactosemia", Pennsylvania Department of Health/March of Dimes Conference, "Integrating Genetics into Your Healthcare Practice", Harrisburg, PA

03/09/02 "Myo-inositol and The Control of Breathing," Featured Talk, Neonatology, Annual Meeting of the Eastern Society for Pediatric Research, Greenwich, CT

01/14/03 "Diagnosis of Amino Acid Disorders by MS/MS", 3rd MS/MS Program Implementation Meeting: Improving the Efficacy and Effectiveness of Tandem Mass Spectrometry Screening, The Genetic Diseases Branch, California Department of Health Services, Berkeley, CA

03/15/03 "Transplantation as a Cure for the Inborn Errors of Metabolism (IEMs)",Plenary Session, Annual Meeting of the American College of Medical Genetics, San Diego, CA

04/05/03 "Ataxia Telangiectasia", 13th Annual Pediatric Neurology Symposium, Children's National Medical Center, Washington, DC

05/20/03 "Loss of Murine Sodium/Myo-Inositol Cotransporter Leads to Brain Myo-Inositol Depletion and Central Apnea", "L. Ruth Guy Lectureship", Pathology Seminar Series, The Department of Pathology, The University of Texas Southwestern Medical Center, Dallas, TX

07/11/03 "Effect of Genotype on Acute and Chronic Complication in Galactosemia", Annual Meeting of Parents of Galactosemic Children, Inc., Reno, NV

10/03/03 "A New Treatment for Metabolic Disorders: Liver Transplantation", Annual Meeting of the Middle Atlantic Regional Human Genetics Network: (MARHGN) Issues in Newborn Screening 2003, Division of Genetics, Alfred I. duPont Hospital for Children, Wilmington, DE

10/10/03 "Acute Illness Management of Inherited Metabolic Disorders", SHS North America Metabolic Conference 2003, Boston, MA

08/31/04 "Endogenous galactose release in galactosaemia: latest research," Dieticians Meeting at the Society for the Study of Inborn Errors of Metabolism Annual Symposium 2004, Amsterdam, Netherlands

04/07/05 "Liver Transplantation in Metabolic Disorders: Biochemical and Clinical Outcome", Ross Metabolic Conference 2005, Savannah, GA

04/08/05 "Quantitative Assessment of Endogenous Galactose Production in Patients with GALT Deficiency", Ross Metabolic Conference 2005, Savannah, GA

07/25/05 "Hereditary Galactosemia", Guest Lecture, Children's Hospital of Pittsburgh, Pittsburgh, PA

11/17/05 "Quantitative Aspects of Galactose Metabolism, Endogenous Galactose Production and Disposal Pathways", International Symposium, Galactosaemia –

Facts and Unresolved Issues - 2005, Fulda, Germany

12/08/05 "Hereditary Galactosemia", Nemours Children's Clinic Education Conference, Jacksonville, FL

03/11/06 "Anti-Oxidant Treatment of Patients with Ataxia Telangiectasia", National Institutes of Health (NIH) Ataxia-Telangiectasia Clinical Research Workshop, March 10-11, 2006, North Bethesda Marriott, Bethesda, MD

04/28/06 "Acute Management of Metabolic Patients at Diagnosis and During Illness", 1st Annual International Metabolic Nutrition Conference, April 27-29, 2006, Emory Conference Center Hotel, Atlanta, GA

07/12/06 "Inborn Errors", Pennsylvania Department of Health Newborn Screening Advisory Committee Meeting, Harrisburg, PA

07/28/06 "Galactosemia Update", Biannual Conference of the Parents of Galactosemic Children, Inc., Philadelphia, PA

08/31/06 "Biochemical Genetics: An Overview" Grand Rounds, Alfred I. duPont Hospital for Children, Wilmington, DE

08/31/06 "The Spectrum of Mitochondrial Diseases", Neurology Conference, Alfred I duPont Hospital for Children, Wilmington, DE

11/06/06 "Galactosemia Revisited", Keynote Address, Annual Meeting of the New England Consortium of Metabolic Programs, Tower Hill Botanic Garden, Boylston, MA

04/23/07 "Hereditary Galactosemia: A Problem in Metabolomics", Annual Sigma Xi Distinguished Lecturer Presentation, UMDNJ- New Jersey Medical School, Newark, NJ

04/23/07 "Newborn Screening for Genetic Diseases and its Impact on Medicine in the U.S.", AOA Visiting Professor Program, UMDNJ- New Jersey Medical School, Newark, NJ

06/18/07 "Nutritional Strategies to Manage Methylmalonic Acidemia: A Critical Overview", NIH Conference, Methylmalonic Acidemia: Clinical and Scientific Advances, Bethesda, MD

09/03/07 "Metabolic Profiling", 62nd Nestle Nutrition Workshop: Personalized Nutrition for the Diverse Needs of Infants and Children, Helsinki, Finland

10/19/07 "Galactosemia and Amenorrhea in the Adolescent," NIH Symposium: the Menstrual Cycle and Adolescent Health Conference, Bolger Conference Center,

Rockville, MD

03/04/08 "Ask the Experts!" Panel session, Annual Meeting of the Society for Inherited Metabolic Diseases, Asilomar, CA

05/17/08 "Personalized Electronic Health Records," (in conjunction with Debra L. Weiner, M.D., Ph.D., Division of Emergency Medicine), Biannual Conference of the New England Connection for PKU and Allied Disorders Organization, Natick, MA

06/27/08 "Medical Management of MSUD," MSUD Symposium 2008, Columbus, OH

07/18/08 "Galactosemia: Neurologic Complications," Biannual Conference of the Parents of Galactosemic Children, Inc., Chicago, IL

10/03/08 "Translational Research: Galactosemia as a Model for Genetic Mechanisms of Primary Ovarian Insufficiency" (in conjunction with Catherine Gordon, M.D.), NIH Orphan Mechanisms of Primary Ovarian Insufficiency Symposium, Bolger Conference Center, Potomac, MD

10/11/08 "Genetic and Metabolic Unknowns," (in conjunction with Mira Irons, M.D.), American Academy of Pediatrics, 2008 National Conference and Exhibition, Boston, MA

05/04/09 "Myo-inositol Deficiency in Brain: A Role in the Pathophysiology of Galactosemia?", Maastrich University Workshop on Galactosemia, Maastrich, Netherlands

11/20/09 "Galactosemia Revisited", Lecture, Annual Meeting of the New England Consortium of Metabolic Programs, Tower Hill Botanic Garden, Boylston, MA

03/29/10 "Ask the Experts!" Panel session, Annual Meeting of the Society for Inherited Metabolic Disorders, Albuquerque, NM

04/07/10 "Neurometabolic Disorders", Philadelphia Neurology Society, Philadelphia, PA

04/16/10 "Galactosemia – and the Survey Says…", (In conjunction with Laurie Bernstein, MS RD, FADA), Genetic Metabolic Dieticians International Conference, Baltimore, MD

07/23/10 "Adults with Galactosemia Study", Keynote Address, Biannual Conference of the Parents of Galactosemic Children, Inc., Bloomington, MN

07/23/10 "Galactosemia: Neurologic Complications and Tremors", Biannual Conference of the Parents of Galactosemic Children, Inc., Bloomington, MN

01/14/11 "Galactosemia", Pediatric Grand Rounds, Rhode Island Hospital/Hasbro Children's Hospital, Providence, RI

03/17/11 "Galactosemia: When is it a newborn screening emergency?", Plenary Session, Annual Meeting of the American College of Medical Genetics, Vancouver, BC, Canada

04/02/12 "What's new in Galactosemia", Annual Meeting of the Society for Inherited Metabolic Disorders, Charlotte, NC

11/21/12 "Galactosemia", Guest Lecture, Center for Human Genetics, University Hospitals Case Medical Center, Cleveland, OH

01/29/14 "USA RedCap Registry," Galactosemias Network, Reference network for the Galactosemias group of rare disorders, Maastricht Meeting January 29 and 30, 2014, Maastricht University Medical Centre, The Netherlands

01/30/14 "Induced pluripotent stem cell model for GALT deficiency," Galactosemias Network, Reference network for the Galactosemias group of rare disorders, Maastricht Meeting January 29 and 30, 2014, Maastricht University Medical Centre, The Netherlands

07/18/14 "Modifier Genes and Epigenetic Effects in Galactosemia," General Session, Galactosemia Foundation 2014 Conference, Orlando, FL

07/18/14 "The new infant with galactosemia – What comes next?" Breakout Session, Galactosemia Foundation 2014 Conference, Orlando, FL

07/19/14 "Research Registry Database," Breakout Session, Galactosemia Foundation 2014 Conference, Orlando, FL

09/30/15 "Galactosemia and iPS Cells," Research Seminar, Oregon Health Sciences University (OHSU), Portland, OR

10/01/15 "Galactosemia," Genetics Grand Rounds, Oregon Health Sciences University (OHSU), Portland, OR

12/15/15 "Galactosemia," The Palmieri Lecture, Pediatric Grand Rounds, The Children's Hospital of Philadelphia, Philadelphia, PA

03/31/16 "Inborn Errors of Metabolism: Metabolomics", Succinic Semialdehyde Dehydrogenase Deficiency (SSADH) Symposium, Cambridge, MA

07/16/16 "How does galactosemia affect the brain?" General Session, Galactosemia Foundation 2016 Conference, Atlanta, GA

07/16/16 "GALT activity and phenotype", Breakout Session, Galactosemia Foundation 2016 Conference, Atlanta, GA

09/05/16 'Spasticity and functional assessment of patients with arginase 1 deficiency", Aeglea Biotherapeutics Inc. Symposium, Rome, Italy

09/23/16 "Galactosemia", Clinical Translational Seminar Series, Boler-Parseghian Center for Rare and Neglected Diseases, Department of Biological Sciences, University of Notre Dame, Notre Dame, IN

05/31/17 "Galactosemia", Pediatric Grand Rounds Westchester Medical Center, New York Medical College, Valhalla, NY

06/10/17 "Liver transplantation in Propionic Acidemia", The Propionic Acidemia Foundation, 2017 Warrior Wisdom Conference, Deerfield, IL

07/08/17 Panelist at NUCDF Family Conference, Washington DC

09/03/17 "Barriers to generating an international arginase deficiency database", IASMB Meeting, Rio de Janeiro, Brazil (with George Diaz, MD, PhD)

09/12/17 "Overview of Galactosemia", Garraham Hospital Conference, Buenos Aires, Argentina

09/12/17 "Duarte Galactosemia", Garraham Hospital Conference, Buenos Aires, Argentina

09/12/17 "Living with Galactosemia", Galactosemia Family Conference, Leloir Institute, Buenos Aires, Argentina

09/13/17 "Galactosemia: Luis Leloir led the way", 70th Anniversary Celebration, Leloir Institute, Buenos Aires, Argentina

09/19/17 "Inborn Errors of Metabolism in the Context of Precision Medicine", Precision Medicine Summit, New Research Building (NRB), Harvard Medical School, Boston, MA

05/19/18 "An Overview of Galactosemia", 2018 Summit Forum of Pediatric Endocrine and Inborn Metabolic Disease, Shanghai, China

07/13/18 "Galactosemia: What to expect in the 1st 9 years?" Galactosemia Conference, Denver, Colorado

07/14/18 "The Use of Stem Cells to Better Understand Disease Mechanisms in Galactosemia", Galactosemia Conference, Denver, Colorado

07/14/18 "Galactose-1-phosphate: Is it necessary?" Galactosemia Conference, Denver, Colorado

09/14/18 "Inborn Errors of Metabolism in the Context of Precision Medicine", Precision Medicine Summit, New Research Building (NRB), Harvard Medical School, Boston, MA

09/02/19 "Aldose reductase" GalNet Symposium, Rotterdam, The Netherlands

10/12/19 "Aldose reductase inhibition", Joint European Galactosemia Society (EGS)/The Galactosemia Network (GalNet) Annual Meeting, Amsterdam, The Netherlands

10/17/19 "The Biology of Galactosemia: A Molecular and Genetic Perspective", The American Society of Human Genetics Annual Meeting Educational Event, Houston, TX

01/15/20 "Galactosemia", UT Southwestern Medical Center Pediatric Grand Rounds, Dallas, TX

01/15/20 "An Overview of the Metabolic or Biochemical Genetics Diseases" UT Southwestern Medical Center Pediatric Noon Lecture, Dallas, TX

07/18/20 "Establishing a Brain Organoid System to Study Neurological Complications of Classic Galactosemia", Galactosemia Foundation Conference 2020, Virtual

07/19/20 "Living with Galactosemia as an adult", Galactosemia Foundation Conference 2020, Virtual

04/28/21 "Galactosemia: newborn screening, treatment and long term follow up", Latin American Society of Inborn Errors of Metabolism and Neonatal Research (SLEIMPN) Web Seminars, Organizer: Genetics and Metabolic diseases laboratory, INTA, Universidad de Chile, Santiago, Chile Virtual

06/11/21 "Metabolic Disorders Impacting the Pregrancy", The 13th Annual Prenatal Diagnosis and Maternal-Fetal/Obstetrics Confrerence, Philadelphia, Virtual

06/24/21 "Liver Transplant and Cardiomyopathy in Propionic Acidemia", Scientific Updates and Organic Acidemias and Homocystinurias- Webinar, June 24-25, 2021, NIH, Virtual event

12/22/21 "Galactosemia", FDA Lecture, Virtual event

01/17/22 "A brief overview of galactosemia", Galactosemia Foundation Webinar, Virtual event (with Judith Fridovich-Keil)

04/11/22 "An overview of Galactosemia", SIMD 43rd Annual Meeting, Presidential address, Rosen Shingle Creek Resort, Orlando, Florida

04/15/22 "Aminoacidopathies and transport disorders", International Center for Genetic Disease Symposium, Virtual event

05/05/22 "Role of vitamin B12 in inborn errors of metabolism", XII Congress of the Latin American Society for Inborn Errors of Metabolism and Newborn Screening, International Convention Center, Punta Cana, Dominican Republic

05/06/22 "mRNA Treatment of Methylmalonic Acidemia", XII Congress of the Latin American Society for Inborn Errors of Metabolism and Newborn Screening, International Convention Center, Punta Cana, Dominican Republic

05/29/22 "An overview of Galactosemia", XVI. International Metabolic Disorders and Nutrition Congress, Hatay, Turkey, virtual

06/25/22 "Organic Acid Diseases A Journey: 1978-2022", 2022 OAA/HCUA/PAF Conference Bethesda MD

06/28/22 "Recent Advancements in Developing New Treatments and Therapies for Metabolic Diseases", Robert Guthrie Symposium 2022, University at Buffalo, Oishei Children's Hospital

07/11/22 "Breath testing studies in subjects with PKU", PHEFREE Meeting, Vancouver, Washington

08/30/22 "Nucleic acid therapies, benefits and dilemmas", ESN/GalNet Meeting at SSIEM, Freiburg, Germany

09/26/22 "Myo-inositol therapy for GPI-anchor diseases", FCDGC Meeting, Mayo Clinic, Rochester, Minnesota

11/12/22 "GALT Enzyme assays:Where's the problem?", GalNet Annual Meeting, Amsterdam, Netherlands

11/17/23 "The Effect of Liver Transplant Upon Metabolic Control, Heart Failure, Outcomes in and Propionic Acidemia", The Central Pennsylvania Clinic 10-Year Anniversary Meeting, Belleville, PA

b. Professional and Educational Leadership Roles Related to Teaching

1990 Chairman, Diabetes and Metabolism Platform, Society for Pediatric Research (SPR)
2002 Chairman, Neonatology Platform, Eastern Society for Pediatric Research
2003 Chairman, Session on Carbohydrate Disorders, Ninth International Congress on

Inborn Errors of Metabolism (ICIEM)

2006 Chair, Committee to Evaluate "Genetics in Medicine" Education in the Jefferson Medical School curriculum

2006 Member, 2007 LCME Self Study, Educational Program Committee, Jefferson Medical College

2010 Chair, ACMG/SIMD Joint Plenary Session: Metabolic Causes of Autism and Neurodevelopmental Disabilities, Annual Meeting of the American College of Medical Genetics, Albuquerque, NM

2010 Co-Chair (with Susan Perlman, M.D., UCLA), Promising Therapeutic Approaches for A-T Session, ATW 2010 International Workshop on Ataxia-Telangiectasia, Redondo Beach, CA, April 11-14, 2010.

2010 Co-Chair (with Rene Santer, M.D., Ph.D., Hamburg), Disorders of Carbohydrate Metabolism, Annual meeting of the Society for the Study of Inborn Errors of Metabolism, Istanbul, Turkey, September 3, 2010.

2012 Co-Chair (with Jeff Milunsky, M.D., Boston University), Genetics/Inborn Errors of Metabolism, Annual Meeting of the Pediatric Academic Societies, Boston, MA, April 30, 2012.

2014 Moderator, Presentations of potential work packages from the different centres, Galactosemias Network, Reference network for the Galactosemias group of rare disorders, Maastricht Meeting January 30, 2014, Maastricht University Medical Centre, The Netherlands.

2017 Faculty, ACMG Genetics and Genomics Review Course, May 4-7, 2017, Tampa, FL

2018 Chair, Concurrent Scientific Session: It's All in the Brain: Neurometabolic Disorders Not To Miss, ACMG Annual Clinical Genetics Meeting, April 14, 2018, Charlotte, NC

2019 Co-Chair, ACMG-SIMD Joint Session: Could this really be metabolic? Dysmorphic features of IEMs, ACMG Annual Clinical Genetics Meeting, Society for Inherited Metabolic Disorders Meeting, April 6, 2019, Seattle, Washington

2019 Faculty, ACMG Genetics and Genomics Review Course, June 27-30, 2019, Tampa, FL

2021 Co-Chair, Gene Therapy in Metabolic Disorders, ACMG Annual Clinical Genetics Meeting, April 13-16, 2021, Virtual

2021 Faculty, ACMG Genetics and Genomics Review Course, May 17 – June 25, 2021, Virtual

2023 Faculty, Clinical Genomic Medicine and Genetic Counseling Training Program, International Center for Genetic Disease, March 8, 2024, Virtual

Report of Clinical Activities and Innovations:

Licensure and Certification:

1975 Pennsylvania

1982 New Jersey

2002 District of Columbia

2003 Maryland

2006	Delaware
2006	Massachusetts
1980	American Board of Pediatrics
1983	American Board of Pediatrics Subspecialty, Pediatric Endocrinology
1984	American Board of Medical Genetics, Clinical Biochemical Genetics

Practice Activities:

	1981-2001	Clinical Biochemical Genetics and/or Endocrinology/Diabetes, The Children's Hospital of Philadelphia Clinical Activity Description: Attending Physician for in-patient service, 1-5 months annually; out-patient metabolism and/or endocrinology clinic, 1-2 half-day sessions weekly; consultant in metabolic diseases for the Delaware Valley region which includes the Philadelphia metropolitan area. Patient Load: 4-10 patients/week plus inpatient service 1-5 months; significant fraction of total time spent in the Intensive Care Units (NICU, PICU, CICU) and Emergency Department as the primary physician caring for the metabolic patients with acute life-threatening emergenices (e.g. hyperammonemic coma secondary to urea cycle enzyme defects, hypoglycemia secondary to fatty acid oxidation defects and acute metabolic decomposition with brain edema secondary to maple syrup urine disease). Clinical Contributions: Developed a new, safer biochemical therapy that obviates the need for hemodialysis, i.e. modified branched-chain amino acid-free total parenteral nutrition, in acutely ill children with maple syrup urine disease (with or without brain edema). This was first described in the NEJM in 1991 (Berry et al, 324: 175) and is now an accepted treatment around the world. Other Relevant Information: One of the top five prime users of intravenous sodium benzoate, sodium phenylacetate and/or arginine hydrochloride to treat patients in the US (Enns et al, NEJM 356: 2282, 2007). One of the first clinical investigators to characterize the effect of liver transplantation on inborn errors of metabolism that involve mitochondrial enzyme defects (Maple Syrup Urine Disease, Methylmalonic Acidemia, Propionic Acidemia and Trifunctional Protein Deficiency) in the U.S. (e.g. Kaplan et al, Mol Genet Metab, 88: 322, 2006).

	2001-2003	Clinical Biochemical Genetics, Children's National Medical Center Clinical Activity Description: Chief of the Division of Metabolism (Clinical Biochemical Genetics), Director of the Metabolism clinical service (staff included four other attending physicians); Attending Physician for in-patient service, 2 months annually; out-patient metabolism clinic, 1 half-day session weekly; consultant in metabolic diseases for the Maryland-D.C.-Virginia region. Patient Load: 4-5 patients/ week plus in-patient service 2 months.

	2003-2006	Clinical Biochemical Genetics , Thomas Jefferson University Hospital Clinical Activity Description: Consultant for in-patient service, 12 months annually; out-patient metabolism clinic, one half-day session weekly. Patient Load: 1-3 patients/week plus inpatient service 12 months.

	2006-	Clinical Biochemical Genetics, Boston Children's Hospital Clinical Activity Description: Director of the Metabolism clinical service (staff includes three other attending physicians); Attending Physician for in-patient service, 2-3 months annually; out-patient metabolism 4 half-day sessions weekly; consultant in metabolic diseases for New England. Patient Load: 20 patients/ week plus in-patient service 2-3 months.

	2009-	New Neurometabolic Clinic in the Department of Neurology, Boston Children's Hospital. Clinical Activity Description: Patients with neurogenetic diseases are seen in conjunction with pediatric neurologist Patient Load: 2-4 patients on Thursday afternoon, twice per month.

Report of Scholarship:

Peer Reviewed Publication in print or other media

1. Foreman JW, Yudkoff M, Berry GT and Segal S. Acidosis associated with dietotherapy of Maple Syrup Urine Disease. J. Pediatr. 96: 62-64, 1980.
2. Berry GT, Yandrasitz JR and Segal S. Experimental galactose toxicity: Effects on synaptosomal phosphatidylinositol metabolism. J. Neurochem. 37: 888-891, 1981.
3. Yandrasitz JR, Berry GT and Segal S. HPLC of phospholipids with UV detection: Optimization of separation on silica. J. Chromatog. 225: 319-328, 1981.
4. Berry GT, Yandrasitz JR and Segal S. The effect of phentolamine stimulation on synaptosomal phosphatidylinositol in experimental galactose toxicity. Neurochem. Res. 7: 49-54, 1982.
5. Berry GT, Yandrasitz JR and Segal S. CMP-dependent phosphatidylinositol: myoinositol exchange activity in isolated nerve-endings. Biochem. Biophys. Res. Commun. 112: 817-821, 1983.
6. Yandrasitz JR, Berry GT and Segal S. High performance liquid chromatography of phospholipids: Quantitation by phosphate analysis. Anal. Biochem. 135: 239-243, 1983.
7. Bennett RH, Ludvigson P, DeLeon G and Berry GT. Large fiber sensory neuronopathy in autosomal dominant spinocerebellar degeneration. Arch. Neurol. 41: 175-178, 1984.
8. Yandrasitz JR, Berry GT and Segal S. Relationship of Inositol, Phosphatidylinositol and Phosphatidic acid in CNS Nerve Endings in Inositol and Phosphoinositides: Metabolism and Regulation (Beasdale, J.E. Eichberg. J. and Hauser, G., eds.) Humana Press, pp 601-619, 1985.

9. Berry GT, Yandrasitz JR, Cipriano VM, Hwang SM and Segal S. Phosphatidylinositol: myo-inositol exchange activity in intact nerve endings: substrate and cofactor dependence, nucleotide specificity, and effect on synaptosomal handling of myo-inositol. J. Neurochem. 46: 1073-1080, 1986.
10. Yandrasitz JR, Berry GT, Cipriano VM, Hwang SM and Segal S. Effect of Elevated Potassium on Phospholipid and Inositol Metabolism of Isolated Nerve Endings. Neurochem. Intl. 9: 295-304, 1986.
11. Berry GT, Yudkoff M, and Segal S. Isovaleric Acidemia: Medical and neurodevelopment effects of long-term therapy. J. Pediatr. 113: 58-64, 1988.
12. Heidenriech R, Natowicz M, Hanline B, Berman P, Kelley RI, Hillman RE and Berry GT. Acute extrapyramidal syndrome in methylmalonic acidemia: "metabolic stroke" involving the globus pallidus. J. Pediatr. 113: 1022-1027,1988.
13. Maddalena A, Sosnoski DM, Berry GT and Nussbaum RL. Mosaicism for an intragenic X chromosome deletion in a male with mild ornithine transcarbamylase deficiency. New Eng. J. Med. 319: 999-1003, 1988.
14. Michalski AJ, Berry GT and Segal S. Holocarboxylase synthetase deficiency: Nine-year follow-up of a case and a review of the literature. J. Inher. Metab. Dis. 12: 312-316, 1989.
15. Berlow S, Bachman RP, Berry GT, Donnell GN, Grix A, Levitsky LL, Hoganson G and Levy HL. Betaine Therapy in Homocystinemia. Brain Dysfunction 2: 10-24, 1989.
16. Berry GT, Heidenreich R, Kaplan P, Levine F, Mazur A, Palmieri M, Yudkoff M and Segal S. Branched-chain amino acid-free parenteral nutrition in the treatment of acute metabolic decompensation in patients with maple syrup urine disease. New Eng. J. Med. 324: 175-179, 1991.
17. Palmieri MJ, Berry GT, Player DA, Rogers S and Segal S. The concentration of red blood cell UDPglucose and UDPgalactose determined by HPLC. Anal. Biochem. 194: 388-393, 1991.
18. Batshaw ML and Berry GT. The Use of Citrulline as a Diagnostic Marker in the Prospective Treatment of Urea Cycle Disorders. J. Pediatr. 118: 914-917, 1991.
19. Kaplan P, Mazur A, Field M, Berlin JA, Berry GT, Heidenreich R, Yudkoff M and Segal S. Intellectual Outcome in Children with Maple Syrup Urine Disease. J. Pediatr. 119: 46-50, 1991.
20. Van Coster R, Lombes A, DeVivo DC, Chi TL, Dodson WE, Rothman S, Orrechio EJ, Grover W, Berry GT, Schwartz JF, Habib A and DiMauro S. Cytochrome-c Oxidase-Associated Leigh syndrome-Phenotypic Features and Pathogenetic speculations. J. Neuro. Sci. 104: 97-111, 1991.
21. Rosenberg SM, Berry GT, Yandrasitz JR and Grunstein MM. Maturational regulation of inositol 1,4,5-trisphosphate metabolism in rabbit airway smooth muscle. J. Clin. Invest. 88: 2032-2038, 1991.
22. Peipert JM, Stallings VA, Berry GT and Henstenburg JA. Infant obesity: Weight reduction with normal increase in linear growth and fat-free body mass. Pediatr. 89: 143-145, 1992.
23. Wehrli S, Palmieri MJ, Berry GT, Kirkman HN and Segal S. 31P-NMR Analysis of Red Blood Cell UDPGlucose and UDPGalactose: Comparison with HPLC and Enzymatic Methods. Anal. Biochem. 201: 105-110, 1992.
24. Berry GT, Palmieri MJ, Heales S, Leonard JV and Segal S. Red Blood Cell Uridine Sugar Nucleotide Levels in Patients with Classic Galactosemia and Other Metabolic Disorders. Metab. 41: 783-87, 1992.

25. Ornstein KS, McGuire EJ, Berry GT, Roth S and Segal S. Abnormal Galactosylation of Complex Carbohydrates in Cultured Fibroblasts from Patients with Galactose-1-Phosphate Uridyltransferase Deficiency. Pediatr. Res. 31: 508-511, 1992.
26. Stanley CA, Hale DE, Berry GT, Deleeuw S, Boxer J and Bonnefont J-P. A deficiency of carnitine-acylcarnitine translocase in the inner mitochondrial membrane. New Eng. J. Med. 327: 19-23, 1992.
27. Sigal SH, Yandrasitz JR and Berry GT. Kinetic evidence for compartmentalization of myo-inositol in hepatocytes. Metab. 2: 395-401, 1993.
28. Berry GT, Palmieri M, Gross KC, Acosta PB, Henstenburg JA, Mazur A, Reynolds R and Segal S. The effect of dietary fruits and vegetables on urinary galactitol excretion in galactose-1-phosphate uridyltransferase deficiency. J. Inher. Metab. Dis.16: 91-100, 1993.
29. Stanley CA, Berry GT, Bennett MJ, Willi SM, Treem WR and Hale DE. Renal Handling of Carnitine in Secondary Carnitine Deficiency Disorders. Pediatr. Res. 34: 89-97, 1993.
30. Giacoia GP, Berry GT. Acrodermatitis enteropathica-like syndrome secondary to isoleucine deficiency during treatment of Maple Syrup Urine Disease. Am. J. Dis. Child. 147: 954-956, 1993.
31. Berry GT. The role of polyol and myo-inositol in Hereditary Galactosemia. In: Galactosemia: New Frontiers in Research. Donnell G, De la Cruz F, Koch R, Levy HL, eds., NIH Publication 93-3438, 1993.
32. Berry GT, Johanson RA, Prantner JE, States B and Yandrasitz JR. Myo-inositol transport and metabolism in fetal-bovine aortic endothelial cells. Biochem. J. 295: 863-869, 1993.
33. Bergoffen J, Kaplan P, Hale DE, Bennett MJ and Berry GT. Marked elevation of urinary 3-hydroxydecanedioic acid in a malnourished infant with glycogen storage disease, mimicking long chain L-3-hydroxyacyl-CoA dehydrogenase deficiency. J. Inher. Metab. Dis. 16: 851-856 1993.
34. Palmieri MJ, Reynolds RA, Gibson JB, Berry GT and Segal S. The concentration of white blood cell UDPgalactose and UDPglucose determined by high performance liquid chromatography. Enz. & Prot. 47: 105-115, 1993.
35. Berry GT, Prantner JE, States B and Yandrasitz JR. The effect of glucose and galactose toxicity on myo-inositol transport and metabolism in human skin fibroblasts in culture. Pediatr. Res. 35: 141-147, 1994.
36. Gibson JB, Reynolds RA, Palmieri MJ, States B, Berry GT and Segal S. UDPHexoses in Leukocytes and Fibroblasts of Classic Galactosemics and Patients with other Metabolic Diseases. Pediatr. Res. 36: 613-618, 1994.
37. Berry GT, Mallee JJ, Kwon HM, Rim JS, Mulla WR, Muenke M, Spinner NB. The human osmoregulatory Na+/myo-inositol cotransporter gene: molecular cloning and localization to chromosome 21. Genomics 25: 507-513, 1995.
38. Gibson JB, Reynolds RA, Palmieri MJ, Berry GT, Elsas, LJ, Levy HL and Segal S. Comparison of Erythrocyte Uridine Sugar Nucleotide levels in Normals, Classic Galactosemics and Patients with other Metabolic Disorders. Metabolism 44: 597-604, 1995.
39. Berry GT, Baker L, Kaplan FS and Witzleben CL. Diabetes-like renal glomerular disease in Fanconi-Bickel syndrome. Pediatr. Nephrol. 9: 287-291, 1995.
40. Berry GT. The role of polyols in the pathophysiology of galactosemia. Eur. J. Pediatr. 154: S53-S64, 1995.

41. Rutledge SL, Berry GT, Stanley CA, VanHove JLK and Millington DS. Glycine and L-carnitine therapy in 3-methylcrotonyl-CoA carboxylase deficiency. J. Inher. Metab. Dis. 18: 299-305, 1995.
42. Gibson JB, Berry GT, Mazur AT, Palmieri MJ, Reynolds RA, and Segal S. Effect of Glucose and Galactose Loading in Normal Subjects on Red and White Blood Cell Uridine Diphosphate Sugars. Biochem. and Molec. Med. 55: 8-14, 1995.
43. Shih V, Fringer J, Mandell R, Kraus J, Berry GT, Heidenreich R, Korson M, and Levy H. A Missense Mutation (I278T) in the Cystathionine (-Synthase Gene Prevalent in Pyridoxine Responsive Homocystinuria and Associated with Mild Clinical Phenotype. Am. J. Hum. Genet. 57: 34-39, 1995.
44. Manfredi G, Schon EA, Moraes CT, Bonilla E, Berry GT, Sladky JT and DiMauro S. A new mutation associated with MELAS is located in a mitochondrial DNA polypeptide-coding gene. Neuromusc. Disord. 5: 391-398, 1995.
45. Bay CA, Berry GT, Glauser TA, Hayward JC, Wolf B, Sladky JT, Kaplan P. Reversible metabolic myopathy in biotinidase deficiency: Its possible role in causing hypotonia. J. Inher. Metab. Dis. 18: 701-704, 1995.
46. Berry GT, Nissim I, Lin Z, Mazur AT, Gibson JB and Segal S. Endogenous synthesis of galactose in normal man and patients with hereditary galactosemia. Lancet 346: 1073-1074, 1995.
47. Berry GT, Nissim I, Mazur AT, Singh R, Elsas LJ, Klein PD, Gibson JB, Segal S. In vivo oxidation of [13C]galactose in patients with galactose-1-phosphate uridyltransferase deficiency. Biochem. and Molec. Med. 56: 158-164, 1995.
48. Sumner AE, Chin MM, Abrahm JL, Berry GT, Gracely EJ, Allen RH and Stabler SP. Elevated levels of methylmalonic acid and total homocysteine reveal high prevalence of B12 deficiency after gastric surgery. Ann. Intern Med. 124: 469-476, 1996.
49. Gibson JB, Berry GT, Palmieri MJ, Reynolds RA, Mazur AT and Segal S. Sugar nucleotide concentrations in red blood cells of patients on protein- and lactose limited diets: Effect of galactose supplementation. Am. J. Clin. Nutr. 63: 704-708, 1996.
50. Mallee JJ, Parrella T, Kwon HM and Berry GT. Multiple comparison of primary structure of the osmoregulatory Na+/myo-inositol cotransporter from bovine, human and canine species. Mam. Gen. 7: 252, 1996.
51. Berry GT, Mallee JJ, Blouin JL and Antonarakis SE. The 21q22.1 STS marker, VNO2 (EST00541 cDNA), is part of the 3′ sequence of the human Na+/myo-inositol cotransporter (SLC5A3) gene. Cytogenet. and Cell Genet. 73: 77-78,1996.
52. Lubetkin EI, Lipson DA, Palevsky HI, Kotloff R, Morris J, Berry GT, Tino G, Rosato E.F., Berlin JA, Wurster AB, Kaiser LR, Lichtenstein GR. Gastrointestinal complications after orthotopic lung transplantation. Am. J. Gastro. 91: 2382-2389, 1996.
53. Zhou X-Y, van der Spoel A, Rottier R, Hale G, Willemsen R, Berry GT, Strisciuglio P, Andria G and d'Azzo A. Molecular and biochemical analysis of protective protein/cathepsin a mutations: correlation with clinical severity in galactosialidosis. Hum. Mol. Genet. 12: 1977-1987, 1996.
54. Lichtenstein GR, Kaiser LR, Tuchman M, Palevsky HI, Kotloff RM, O'Brien CB, Furth EE, Raps EC and Berry GT. Fatal hyperammonemia following orthotopic lung transplantation. Gastroent. 112: 236-240, 1997.

55. Weinzimer SA, Stanley CA, Berry GT, Yudkoff M, Tuchman M and Thornton PS. A syndrome of congenital hyperinsulinism and hyperammonemia. J. Pediatr. 130: 661-664, 1997.
56. Berry GT, Nissim I, Gibson JB, Mazur AT, Lin Z, Elsas LJ, Singh RH, Klein PD and Segal S. Quantitative assessment of whole body galactose metabolism in galactosemic patients. Eur. J. Pediatr. 156: S43-S49, 1997.
57. Tuchman M, Lichtenstein GR, Rajagopal BS, McCann MT, Furth EE, Bavaria J, Kaplan PB, Gibson JB and Berry GT. Hepatic glutamine synthetase deficiency in fatal hyperammonemia following lung transplantation. Ann. Intern. Med. 127: 446-449, 1997.
58. Verma A, Piccoli DA, Bonilla E, Berry GT, Di Mauro S and Moraes CT. A novel mitochondrial G8313A mutation associated with prominent initial gastrointestinal symptoms and progressive encephaloneuropathy. Pediatr. Res. 42: 448-454, 1997.
59. Wehrli S, Berry GT, Palmieri MJ, Mazur A, Elsas LJ, and Segal S. Urinary galactonate in patients with Galactosemia: Quantitation by NRM Spectroscopy. Pediatr. Res. 42: 855-861, 1997.
60. Mallee JJ, Atta MG, Lorica V, Rim JS, Kwon HM, Lucente AD, Wang Y and Berry GT. The structural organization of the human Na+/Myo-inositol cotransporter (SLC5A3) gene and characterization of the promoter. Genomics 46: 459-465, 1997.
61. Lam WWM, Wang ZJ, Zhao H, Berry GT, Kaplan P, Gibson JB, Kaplan BS, Bilaniuk LT, Hunter JV, Haselgrove JC, Zimmerman RA. 1H-MR Spectroscopy of the basal ganglia in childhood: a semi-quantitative analysis. Neuroradiol. 40: 315-323, 1998.
62. Rim JS, Atta MG, Dahl SC, Berry GT, Handler JS and Kwon HM. Transcription of the sodium/myo-inositol cotransporter gene is regulated by multiple tonicity-responsive enhancers spread over 50 kb in the 5′ flanking region. J. Biol. Chem. 273: 20615-20621, 1998.
63. Berry GT, Wehrli S, Reynolds R, Palmieri M, Frangos M, Williamson JR and Segal S. Elevation of Erythrocyte Redox Potential Linked to galactonate biosynthesis eliminated by Tolrestat. Metab. 47: 1423-1428, 1998.
64. Berry, GT and Bennett MJ. The detection of inborn errors of metabolism. Contemp. Pediatr. 15: 79-102,1998.
65. Berry GT, Bridges ND, Nathanson KL, Kaplan P, Clancy RR, Lichtenstein GR and Spray TL. Successful use of alternate waste nitrogen agents and hemodialysis in a patient with hyperammonemic coma following heart-lung transplantation. Arch. Neurol. 56: 481-484, 1999.
66. Yorek MA, Dunlap JA, Manzo-Fontes A, Bianchi R, Berry GT and Eichberg J. Abnormal myo-inositol and phospholipid metabolism in cultured fibroblasts from patients with ataxia-telangiectasia. Biochimica. Et. Biophysica. Acta. 1437: 287-300, 1999.
67. Palmieri M, Mazur A, Berry GT, Ning C, Wehrli S, Yager C, Reynolds R, Singh R, Muralidharan K, Langley S, Elsas L and Segal S. Urine and plasma galactitol in patients with galactose-1-phosphate uridyltransferase deficiency galactosemia. Metab. 48: 1294-1302, 1999.
68. Hadjigeorgiou GM, Kim SH, Fischbeck KH, Andreu AL, Berry GT, Bingham P, Shanske S Bonilla E and Di Mauro S. A new mitochondrial DNA mutation (A3288G) in the tRNAleu(UUR) gene associated with familial myopathy. J Neurol. Sci. 164: 153-157, 1999.
69. Berry GT, Wang Z, Dreha S, Finucane BM and Zimmerman RA. In Vivo Brain Myo-Inositol Levels in Children with Down syndrome. J. Pediatr. 135: 94-97, 1999.

70. Lichtenstein GR, Yang Y-X, Nunes FA, Lewis JD, Tuchman M, Tino G, Kaiser LR, Palecsky HI, Kotloff RM, Furth EF, Bavaria JE, Stecjer MM, Kaplan P, Berry GT. Fatal hyperammonemia after orthotopic lung transplantation. Ann. Intern. Med. 132: 283-287, 2000.
71. Ning C, Reynolds R, Chen J, Yager C, Berry GT, McNamara PD, Leslie N, and Segal S. Galactose metabolism by the mouse with galactose-1-phosphate uridyltransferase deficiency. Pediatr. Res. 48: 211-217, 2000.
72. Ierardi-Curto L, Kaplan P, Saitta S, Mazur A, Berry GT. The glutamine paradox in a neonate with propionic acidemia and severe hyperammonemia. J. Inherit. Metab. Dis. 23: 85-86, 2000.
73. McVeigh K, Mallee JJ, Lucente A, Barnoski BL, Wu S, Berry GT. Murine chromosome 16 telomeric region, syntenic with human chromosome 21q22, contains the osmoregulatory Na+/myo-inositol cotransporter (SLC5A3) gene. Cytogenet. & Cell Genet. 88:153-158, 2000.
74. Fukao T, Mitchell GA, Song X-Q, Nakamura H, Kassovska-Brainiva S, Orii KE, Wraith JE, Besley G, Wanders FJA, Niezen-Koning KE, Berry GT, Palmieri M, Kondo N. Succinyl-CoA:3-ketoacid CoA transferase (SCOT): Cloning of the human SCOT gene, tertiary structural modeling of the human SCOT monomer, and characterization of three pathogenic mutations. Gen. 68: 144-151, 2000.
75. Guerrero NV, Singh RH, Manatunga A, Berry GT, Steiner RD, Elsas L.J.: Risk factors for premature ovarian failure in females with galactosemia. J. Pediatr. 137: 833-841, 2000.
76. Ning C, Fenn PT, Blair IA, Berry GT, Segal S. Apparent galactose appearance in human galactosemia based on plasma [13C] galactose isotopic enrichment. Mol. Genet. and Metab. 70: 261-271, 2000.
77. Berry GT, Singh RH, Mazur AT, Guerrero N, Kennedy MJ, Chen J, Reynolds R, Palmieri MJ, Klein PB, Segal S, Elsas LJ. Galactose breath testing distinguishes variant and severe galactose-1-phosphate uridyltransferase genotypes. Pediatr. Res. 48: 23-328, 2000.
78. Anadiotis G, Ierardi-Curto L, Kaplan PB, Berry GT. Ornithine transcarbamylase deficiency and pancreatitis. J. Pediatr.138: 123-124, 2001.
79. Berry GT, Hunter JV, Wang Z, Dreha S, Mazur A, Brooks DG, Ning C, Zimmerman RA and Segal S. In vivo evidence of brain galactitol accumulation in an infant with galactosemia and encephalopathy. J. Pediatr. 138: 260-262, 2001.
80. Berry GT, Steiner RD. Long-term management of patients with urea cycle disorders. J. Pediatr. 138: S56-S60, 2001.
81. Ning C, Reynolds R, Chen J, Yager CT, Berry GT, Leslie N, Segal S. Galactose metabolism in mice with galactose-1-phosphate uridyltransferase deficiency: sucklings and 7-week old animals fed a high-galactose diet. Mol. Genet. and Metab. 72: 306-315, 2001.
82. Berry GT, Leslie N, Reynolds R, Yager CT, Segal S. Evidence for alternate galactose oxidation in a patient with deletion of the galactose-1-phosphate uridyltransferase gene. Mol. Genet. and Metab. 72: 316-321, 2001.
83. Licht DJ, Berry GT, Younkin D. Reversible subacute combined degeneration of the spinal cord in a 14-year old due to a strict vegan diet. Clin. Pediatr. 40: 413-415, 2001.
84. Berry GT, Fukao T, Mitchell GA, Mazur A, Ciafre M, Gibson J, Kondo N, Palmieri MJ: Neonatal hypoglycemia in severe succinyl-CoA: 3-oxoacid CoA transferase (SCOT) deficiency. J. Inherit. Metab. Dis. 24: 587-95, 2001.
85. Wang ZJ, Berry GT, Dreha SF, Zhao H, Segal S and Zimmerman RA. Proton

Magnetic Resonance Spectroscopy of Brain Metabolites in Galactosemia. Ann. Neurol. 2001.
86. Santer R, Groth S, Kinner M, Dombrowski A, Berry GT, Brodehl J, Leonard JV, Moses S, Norgren S, Skovby F, Schneppenheim R, Steinmann B, Schaub J. The mutation spectrum of the facilitative glucose transporter gene SLC2A2 (GLUT2) in patients with Fanconi-Bickel syndrome. Hum. Genet. 110: 21-29, 2002.
87. Matthews RP, Russo P, Berry GT, Piccoli DA, and Rand EB. Biliary atresia associated with a fatty acid oxidation defect. J. Pediatr. Gastroenterol. Nutr. 35: 624-8, 2002.
88. Tavani F, Zimmerman RA, Berry GT, Sullivan K, Gatti R, and Bingham P. Ataxia-Telangiectasia: The Pattern of Cerebellar Atrophy on MRI. Neuroradiol. 45: 315-319, 2003.
89. Pearl PL, Gibson KM, Acosta MT, Vezina LG, Theodore WH, Rogawski MA, Novotny EJ, Gropman A, Conry JA, Berry GT and Tuchman M. Clinical Spectrum of Succinic Semialdehyde Dehydrogenase Deficiency. Neurol. 60: 1413-1417, 2003.
90. Berry GT, Wu S, Buccafusca R, Ren J, Gonzales LW, Ballard P, Golden J, Stevens MJ and Greer JJ. Loss of murine Na+/myo-inositol cotransporter leads to brain myo-inositol depletion and central apnea. J. Biol. Chem. 92: 278, 18297-18302, 2003.
91. Jan W, Zimmerman RA, Wang ZJ, Berry GT, Kaplan PB and Kaye EM. MR diffusion imaging and MR spectroscopy of maple syrup urine disease during acute metabolic decompensation. Neuroradiol. 24:1471-1474, 2003.
92. Zand DJ, Simon EM, Pulitzer SB, Wang DJ, Wang ZJ, Rorke LB, Palmieri M and Berry GT. In Vivo Pyruvate Detected by MR Spectroscopy in Neonatal Pyruvate Dehydrogenase Deficiency. Am. J. Neuroradiol. 24: 1471-1474, 2003.
93. Venditti LN, Venditti CP, Berry GT, Kaplan PB, Kaye EM, Glick H, Stanley CA. Newborn screening by Tandem Mass Spectrometry for Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD): A Cost-Effective Analysis. Pediatr. 112:1005-1015, 2003.
94. Berry GT, Moate PJ, Reynolds RA, Yager CT, Ning C, Boston RC and Segal S. The Rate of *De Novo* Galactose Synthesis in Patients with Galactose -1-Phosphate Uridyltransferase (GALT) Deficiency. Mol. Genet. Metab. 81: 22-30, 2004.
95. Berry GT, Buccafusca R, Greer JJ, Eccleston E. Phosphoinositide Deficiency Due to Inositol Depletion is not a Mechanism of Lithium Action in Brain. Mol. Genet. Metab. 82: 87-92, 2004.
96. Berry GT, Reynolds RA, Yager CT, Segal S. Extended [^{13}C] Galactose Oxidation Studies in Patients with Galactosemia. Mol. Genet. Metab. 82: 130-136, 2004.
97. Venditti CP, Harris MC, Huff D, Peterside I, Munson D, Weber HS, Rome J, Kaye EM, Shanske S, Sacconi S, Tay S, DiMauro S, Berry GT. Congenital Cardiomyopathy and Pulmonary Hypertension: Another Fatal Variant of Cytochrome c Oxidase Deficiency. J.Inherit. Metab. Dis. 27: 735-739, 2004.
98. Berry GT, Baynes JW, Wells-Knecht KJ, Szwergold BS, Santer R. Elements of diabetic nephropathy in a patient with GLUT2 deficiency. Mol. Genet. Metab. 86: 473-477, 2005.
99. Openo KK, Schulz JM, Vargas CA, Orton CS, Epstein MP, Schnur RE, Scaglia F, Berry GT, Gottesman G, Ficicioglu C, Slonim AE, Shroer RJ, Chunli Y, Rangel VE, Kenan J, Lamance K, and Fridowich-Keil JL. Epimerase-deficiency galactosemia is not a binary condition. Amer.J.Hum.Genet. 78:89-102, 2006.

100. Kaplan P, Ficicoglu C, Mazur AT, Palmieri MJ, Berry GT. Liver transplantation is not curative for methylmalonic acidopathy caused by methylmalonyl-CoA mutase deficiency. Mol. Genet. Metab. 88: 322-326, 2006.
101. Shaldubina A, Johanson RA, O'Brien WT, Buccafusca R, Agam G, Belmaker RH, Klein PS, Bersudsky Y, Berry GT. SMIT haploinsufficiency causes brain inositol deficiency without affecting lithium-sensitive behavior. Mol. Genet. Metab. 88: 384-388, 2006.
102. Shaldubina A, Buccafusca R, Johanson R, Agam G, Belmaker RH, Berry GT, Bersudsky Y. Behavioral phenotyping of sodium-myo-inositol cotransporter heterozygous knockout mice with reduced brain inositol. Genes Brain Behav., 6:253-259, 2007.
103. Johanson RA, Buccafusca R, Shaw AM, Berry G T. Phosphatidylcholine removal from brain lipid extracts expands lipid detection and enhances phosphoinositide quantification by matrix-assisted laser desorption/ionization time-of-flight mass spectrometry (MALDI-TOF MS). Anal. Biochem. 362:155-67, 2007.
104. Enns GM, Berry SA, Berry GT, Rhead WJ, Hamosh A. Survival after treatment with phenylacetate and benzoate for urea-cycle disorders. New Eng. J. Med. 356: 2282-2292, 2007.
105. Tein I, Elpeleg O, Ben-Zeev B, Korman SH, Lossos A, Lev D, Lerman-Sagie T, Leshinsky-Silver E, Vockley G, Berry GT, Lamhownah AM, Matern D, Roe CR, Gregersen N. Short-chain acyl-CoA dehydrogenase gene mutation (c.319C>T) presents with clinical heterogeneity and is candidate founder mutation in individuals of Ashkenazi Jewish origin. Mol. Genet. Metab. 93: 179-189, 2008.
106. Bersudsky Y, Shaldubina A, Agam G, Berry GT, Belmaker RH. Homozygote inositol transporter knockout mice show a lithium-like phenotype. Bipolar Disord. 10: 453-459, 2008.
107. Buccafusca R, Venditti CP, Kenyon LC, Johanson RA, VanBockstaele E, Ren J, Pagliardini S, Minarcik J, Golden JA, Coady MJ, Greer JJ, Berry GT. Characterization of the null murine sodium/myo-inositol cotransporter 1 (Smit1 or Slc5a3) phenotype: myo-inositol rescue is independent of expression of its cognate mitochondrial ribosomal protein subunit 6 (Mrps6) gene and of phosphatidylinositol levels in neonatal brain. Mol. Genet. Metab. 95: 81-95, 2008.
108. Agam G, Bersudsky Y, Berry GT, Moechars D, Lavi-Avnon Y, Belmaker RH. Knockout mice in understanding the mechanism of action of lithium. Biochem. Soc. Trans. 37: 1121-1125, 2009.
109. Johanson RA, Berry GT. Brain phosphoinositide extraction, fractionation, and analysis by MALDI-TOF MS. Armstrong D ed., The Human Press Inc., Totowa, NJ, 579:189-200, 2009.
110. Ching MS, Shen Y, Tan WH, Jeste SS, Morrow EM, Chen X, Mukaddes NM, Yoo SY, Hanson E, Hundley R, Austin C, Becker RE, Berry GT, Driscoll K, Engle EC, Friedman S, Gusella JF, Hisama FM, Irons MB, Lafiosca T, LeClair E, Miller DT, Neessen M, Picker JD, Rappaport L, Rooney CM, Sarco DP, Stoler JM, Walsh CA, Wolff RR, Zhang T, Nasir RH, Wu BL. Boston Children's Hospital Genotype Phenotype Study Group. Deletions of NRXN1 (neurexin-1) predispose to a wide spectrum of developmental disorders. Am. J. Med. Genet. B Neuropsychiatr. Genet. 153B(4):937-947, 2010.
111. Shen Y, Dies KA, Holm IA, Bridgemohan C, Sobeih MM, Caronna EB, Miller KJ, Frazier JA, Silverstein I, Picker J, Weissman L, Raffalli P, Jeste S, Demmer LA, Peters HK, Brewster SJ, Kowalczyk SJ, Rosen-Sheidley B, McGowan C, Duda AW 3rd, Lincoln SA,

Lowe KR, Schonwald A, Robbins M, Hisama F, Wolff R, Becker R, Nasir R, Urion DK, Milunsky JM, Rappaport L, Gusella JF, Walsh CA, Wu BL, Miller DT; Autism Consortium Clinical Genetics/DNA Diagnostics Collaboration. Clinical genetic testing for patients with autism spectrum disorders. Pediatr. 125: 727-735, 2010.

112. Li Y, Ptolemy A, Harmonay L, Kellogg M, Berry GT. Quantification of Galactose-1-Phosphate Uridyltransferase Enzyme Activity by Liquid Chromatography-Tandem Mass Spectrometry. Clin. Chem., 56: 772-80, 2010.
113. Li Y, Ptolemy A, Harmonay L, Kellogg M, Berry GT. Ultra fast and sensitive liquid chromatography tandem mass spectrometry based assay for galactose-1-phosphate uridylyltransferase and galactokinase deficiencies. Mol. Genet. Metab.,102: 33-40, 2011.
114. Ptolemy AS, Li Y, Sanderson, T, Khwaja, O, Berry, GT, Kellogg M. A 9-month-old boy with seizures and discrepant urine tryptophan concentrations**.** Clin. Chem., 57: 545-8, 2011.
115. Berry, GT and Elsas, LJ. Introduction to the Maastricht workshop on galactosemia: lessons from the past and new directions in galactosemia. J Inherit Metab Dis., 34: 249-55, 2011.
116. Berry, GT. Is prenatal myo-inositol deficiency a mechanism of CNS injury in galactosemia? J Inherit Metab Dis., 34: 345-55, 2011.
117. Gubbels CS, Maurice-Stam H, Berry GT, Bosch AM, Waisbren S, Rubio-Gozalbo ME, Grootenhuis MA. Psychosocial developmental milestones in men with classic galactosemia. J Inherit Metab Dis., 34: 415-9, 2011.
118. Li Y, Cleary R, Kellogg M, Soul JS, Berry GT*, Jensen FE*. Sensitive isotope dilution liquid chromatography/tandem mass spectrometry method for quantitative analysis of bumetanide in serum and brain tissue. J Chromatogr B, 879(13-14):998-1002, 2011.
119. O'Brien WT, Huang J, Buccafusca R, Garskof J, Valvezan AJ, Berry GT, Klein PS. Essential role of glycogen synthase kinase-3 in β-arrestin-2 complex formation and lithium-sensitive behaviors in mice. J Clin Invest., 121:3756-62, 2011.
120. Jones MA, Ng BG, Bhide S, Chin E, Rhodenizer D, He P, Losfeld ME, He M, Raymond K, Berry G, Freeze HH, Hegde MR. DDOST Mutations Identified by Whole-Exome Sequencing Are Implicated in Congenital Disorders of Glycosylation. Am J Hum Genet., 90: 363-8, 2012.
121. Waisbren SE, Potter NL, Gordon CM, Green RC, Greenstein P, Gubbels CS, Rubio-Gozalbo E, Schomer D, Welt C, Anastasoaie V, D'Anna K, Gentile J, Guo C-Y, Hecht L, Jackson R, Jansma BM, Li Y, Lip V, Miller DT, Murray M, Power L, Quinn N, Rohr F, Shen Y, Skinder-Meredith A, Timmers I, Tunick R, Wessel A, Wu B-L, Levy H, Elsas L, Berry GT. The adult galactosemic phenotype. J Inherit Metab Dis., 35: 279-86, 2012. (2013 SSIEM Kowrower Award)
122. Green RC, Berg JS, Berry GT, Biesecker LG, Dimmock DP, Evans JP, Grody WW, Hegde MR, Kalia S, Korf BR, Krantz I, McGuire AL, Miller DT, Murray MF, Nussbaum RL, Plon SE, Rehm HL, Jacob HJ. Exploring Concordance and Discordance for Return of Incidental Findings from Clinical Sequencing. Genetics in Medicine, Genet Med. 14:405-10, 2012.
123. Jumbo-Lucioni PP, Garber K, Kiel J, Baric I, Berry GT, Bosch A, Burlina A, Chiesa A, Pico ML, Estrada SC, Henderson H, Leslie N, Longo N, Morris AA, Ramirez-Farias C, Scheweitzer-Krantz S, Silao CL, Vela-Amieva M, Waisbren S, Fridovich-Keil JL. Diversity of approaches to classic galactosemia around the world: a comparison of diagnosis, intervention, and outcomes. J Inherit Metab Dis., 35:1037-49, 2012.

124. Berry GT. Galactosemia: When is it a newborn screening emergency? Mol Genet Metab., 106:7-11, 2012.
125. Thakuria JV, Zaranek AW, Church GM, Berry GT. Back to the future: From genome to metabolome. Hum Mutat., 33:809-12, 2012.
126. Poduri A, Chopra SS, Neilan EG, Christina Elhosary P, Kurian MA, Meyer E, Barry BJ, Khwaja OS, Salih MA, Stödberg T, Scheffer IE, Maher ER, Sahin M, Wu BL, Berry GT, Walsh CA, Picker J, Kothare SV. Homozygous PLCB1 deletion associated with malignant migrating partial seizures in infancy. Epilepsia., 53:e146-50, 2012.
127. Liu Y, Xia B, Gleason TJ, Castaneda U, He M, Berry GT, Fridovich-Keil JL. N- and O-linked glycosylation of total plasma glycoproteins in galactosemia. Mol Genet Metab., 106:442-454, 2012.
128. Jamuar SS, Newton SA, Prabhu SP, Hecht L, Costas KC, Wessel AE, Harris DJ, Anselm I, Berry GT. Rhabdomyolysis, Acute Renal Failure, and Cardiac Arrest Secondary to Status Dystonicus in a Child with Glutaric Aciduria Type I. Mol Genet Metab., 106:488-490, 2012.
129. Zhang Y, Landau YE, Miller DT, Marsden D, Berry GT, Kellogg MD. Recurrent unexplained hyperammonemia in an adolescent with arginase deficiency. Clin Biochem., 45:1583-6, 2012.
130. Batey LA, Welt CK, Rohr F, Wessel A, Anastasoaie V, Feldman HA, Guo CY, Rubio-Gozalbo E, Berry GT, Gordon CM. Skeletal health in adult patients with classic galactosemia. Osteoporos Int., 24:501-9, 2013
131. Gubbels CS, Welt CK, Dumoulin J, Robben SGF, Gordon CM, Dunselman G, Rubio-Gozalbo ME, Berry GT. The male reproductive system in classic galactosemia: cryptorchidism and low semen volume. J Inherit Metab Dis., 36:779-86, 2013.
132. Cleary RT, Sun H, Huynh T, Manning SM, Li Y, Rotenberg A, Talos DM, Kahle KT, Jackson M, Rakhade SN, Berry GT, Jensen FE. Bumetanide enhances phenobarbital efficacy in a rat model of hypoxic neonatal seizures. PLoS One, 8(3): e57148, 2013.
133. Lieber DS, Calvo SE, Shanahan K, Slate NG, Liu S, Hershman SG, Gold NB, Chapman BA, Thorburn DR, Berry GT, Schmahmann JD, Borowsky ML, Mueller DM, Sims KB, and Mootha VK. Targeted exome sequencing of suspected mitochondrial disorders. Neurology, 80:1762-70, 2013.
134. Manoli I, Sysol JR, Lingli Li, Houillier P, Garone C, Wang C, Zerfas PM, Cusmano-Ozog K, Young S, Trivedi NS, Cheng J, Sloan JL, Chandler RJ, Abu-Asab M, Tsokos M, Elkahloun AG, Rosen S, Enns GM, Berry GT, Hoffmann V, DiMauro S, Schnermann J, and Venditti CP. Targeting proximal tubule mitochondrial dysfunction attenuates the renal disease of methylmalonic acidemia. Proc Natl Acad Sci. 110:13552-7, 2013.
135. van Erven B, Gubbels CS, van Golde RJ, Dunselman GA, Derhaag JG, de Wert G, Geraedts JP, Bosch AM, Treacy EP, Welt CK, Berry GT, and Estela Rubio-Gozalbo M. Fertility preservation in female classic galactosemia patients. Orphanet J Rare Dis. 8:107, 2013.
136. Kinney HC, McDonald AG, Minter ME, Berry GT, Poduri A, Goldstein RD. Witnessed sleep-related seizure and sudden unexpected death in infancy: a case report. Forensic Sci Med Pathol. 9:418-21, 2013.
137. Sankaran V, Joshi M, Agrawal A, Schmitz K, Towne M, Marinakis N, Markianos K, Berry GT, Agrawal P. Rare complete loss-of-function provides insight into a pleiotropic genome-wide association study locus. Blood. 2013 Nov 28;122(23):3845-7.

138. Toker L, Bersudsky Y, Plaschkes I, Chalifa-Caspi V, Berry GT, Buccafusca R, Moechars D, Belmaker RH, Agam G. Inositol-Related Gene Knockouts Mimic Lithium's Effect on Mitochondrial Function. Neuropsychopharmacology. 2014 Jan;39(2):319-28.
139. Niewczas MA, Sirich TL, Mathew AV, Skupien J, Mohney RP, Warram JH, Smiles A, Huang XP, Walker W, Byun J, Karoly ED, Kensicki EM, Berry GT, Bonventre JV, Pennathur S, Meyer TW, Krolewski AS. Uremic solutes and risk of end-stage renal disease in type 2 diabetes: metabolomic study. Kidney Int. 2014 May;85(5):1214-24.
140. Wessel AE, Mogensen KM, Rohr F, Erick M, Neilan E, Chopra S, Levy HL, Gray KJ, Wilkins-Haug L, Berry GT. Management of a woman with Maple Syrup Urine Disease during pregnancy, delivery, and lactation. JPEN J Parenter Enteral Nutr. 2015 Sep;39(7):875-9. doi: 10.1177/0148607114526451. Epub 2014 Mar 11. PMID: 24618664
141. Van Calcar SC, Bernstein LE, Rohr FJ, Yannicelli S, Berry GT, Scaman CH. Galactose content of legumes, caseinates and some hard cheeses: Implications for diet treatment for classic galactosemia. J Agric Food Chem. 2014 Feb 12;62(6):1397-402.
142. Li Y, Huang X, Harmonay L, Liu Y, Kellogg M, Fridovich-Keil JL, Berry GT. Liquid Chromatography Tandem Mass Spectrometry Enzyme Assay for UDP-Galactose 4'-Epimerase: Use of Fragment Intensity Ratio in Differentiation of Structural Isomers. Clin. Chem. 2014 May;60(5):783-90.
143. Abbott GW, Tai K, Neverisky D, Hansler A, Hu Z, Roepke TK, Lerner DJ, Chen Q, Liu L, Zupan B, Toth M, Haynes R, Huang X, Demirbas D, Buccafusca R, Gross SS, Kanda VA*, Berry GT*. KCNQ1, KCNE1, and NA^{+}-Coupled Solute Transporters Form Reciprocally Regulating Complexes that Affect Neuronal Excitability. Science Signaling, 2014 Mar 4;7(315):ra22.
144. Joshi M, Eagan J, Desai NK, Newton SA, Towne MC, Marinakis NS, Esteves KM, De Ferranti S, Bennett MJ, McIntyre A, Beggs AH, Berry GT*, Agrawal PB*. A compound heterozygous mutation in GPD1 causes hepatomegaly, steatohepatitis, and hypertriglyceridemia. European Journal of Human Genetics, advance online publication 2014 Feb 19; doi: 10.1038/ejhg.2014.8.
145. Camp KM, Parisi MA, Acosta PB, Berry GT, Bilder DA, Blau N, Bodamer OA, Brosco JP, Brown CS, Burlina AB, Burton BK, Chang CS, Coates PM, Cunningham AC, Dobrowolski SF, Ferguson JH, Franklin TD, Frazier DM, Grange DK, Greene CL, Groft SC, Harding CO, Howell RR, Huntington KL, Hyatt-Knorr HD, Jevaji IP, Levy HL, Lichter-Konecki U, Lindegren ML, Lloyd-Puryear MA, Matalon K, Macdonald A, McPheeters ML, Mitchell JJ, Mofidi S, Moseley KD, Mueller CM, Mulberg AE, Nerurkar LS, Ogata BN, Pariser AR, Prasad S, Pridjian G, Rasmussen SA, Reddy UM, Rohr FJ, Singh RH, Sirrs SM, Stremer SE, Tagle DA, Thompson SM, Urv TK, Utz JR, van Spronsen F, Vockley J, Waisbren SE, Weglicki LS, White DA, Whitley CB, Wilfond BS, Yannicelli S, Young JM. Phenylketonuria Scientific Review Conference: State of the Science and future research needs. Mol Genet Metab. 2014 Mar 6. pii: S1096-7192(14)00085-7. doi: 10.1016/j.ymgme.2014.02.013. Epub 2014 Mar 6. PMID: 24667081
146. Olson H, Shen Y, Avallone J, Sheidley BR, Pinsky R, Bergin AM, Berry GT, Duffy FH, Eksioglu Y, Harris DJ, Hisama FM, Ho E, Irons M, Jacobsen CM, James P, Kothare S, Khwaja O, Lipton J, Loddenkemper T, Markowitz J, Maski K, Megerian JT, Neilan E, Raffalli PC, Robbins M, Roberts A, Roe E, Rollins C, Sahin M, Sarco D, Schonwald A, Smith SE, Soul J, Stoler JM, Takeoka M, Tan WH, Torres AR, Tsai P, Urion DK, Weissman L, Wolff R, Wu BL, Miller DT, Poduri A. Copy number variation plays an

important role in clinical epilepsy. Ann Neurol. 2014 May 9; doi: 10.1002/ana.24178. Epub 2014 Jun 13. PMID: 24811917
147. Zsengellér ZK, Aljinovic N, Teot LA, Korson M, Rodig N, Sloan JL, Venditti CP, Berry GT, Rosen S. Methylmalonic acidemia: A megamitochondrial disorder affecting the kidney. Pediatr Nephrol. 2014 Nov;29(11):2139-46. doi: 10.1007/s00467-014-2847-y. Epub 2014 May 28. PMID: 24865477
148. Hecht LE, Wessel AE, Levy HL, Berry GT. The complexity of newborn screening follow-up in Phenylketonuria. JIMD Rep. 2014;17:37-9. doi: 10.1007/8904_2014_329. Epub 2014 Aug 26. PMID: 25155776
149. Tran TT, Liu Y, Zwick ME, Ramachandran D, Cutler DJ, Huang X, Berry GT, Fridovich-Keil JL. A de novo variant in galactose-1-P uridylytransferase (GALT) leading to classic galactosemia. JIMD Rep. 2015;19:1-6. doi: 10.1007/8904_2014_349. Epub 2015 Feb 15. PMID: 25681079
150. Smpokou P, Samanta M, Berry GT, Hecht L, Engle E, Lichter-Konecki U. Menkes Disease in Affected Females: The Clinical Disease Spectrum. Am J Med Genet A. 2015 Feb;167A(2):417-20. doi: 10.1002/ajmg.a.36853. Epub 2014 Nov 26. Review. PMID: 25428120
151. Van Calcar SC, Bernstein LE, Rohr FJ, Scaman CH, Yannicelli S, Berry GT. A re-evaluation of life-long severe galactose restriction for the nutrition management of classic galactosemia. Mol Genet Metab. 112:191-197. 2014
152. Coelho Al, Berry GT, Rubio-Gozalbo ME. Galactose metabolism and health. Curr Opin Clin Nutr Metab Care. 18:422-427, 2015.
153. Torres A, Newton SA, Crompton B, Borzutzky A, Neufeld EJ, Notarangelo L, Berry GT. CSF 5-Methyltetrahydrofolate Serial Monitoring to Guide Treatment of Congenital Folate Malabsorption Due to Proton-Coupled Folate Transporter (PCFT) Deficiency. JIMD Rep. 2015;24:91-6. Doi: 10.1007/8904_2015_445. Epub 2015 May 26. PMID: 26006721.
154. Balakrishnan B, Chen W, Tang M, Huang X, Cakici DD, Siddiqi A, Berry G, Lai K. Galactose-1 phosphate uridylyltransferase (GalT) gene: A novel positive regulator of the PI3K/Akt signaling pathway in mouse fibroblasts. Biochem Biophys Res Commun. 2016 Jan 29;470(1):205-12. doi: 10.1016/j.bbrc.2016.01.036. Epub 2016 Jan 8.PMID: 26773505
155. Ng BG, Shiryaev SA, Rymen D, Eklund EA, Raymond K, Kircher M, Abdenur JE, Alehan F, Midro AT, Bamshad MJ, Barone R, Berry GT, Brumbaugh JE, Buckingham KJ, Clarkson K, Cole FS, O'Connor S, Cooper GM, Van Coster R, Demmer LA, Diogo L, Fay AJ, Ficicioglu C, Fiumara A, Gahl WA, Ganetzky R, Goel H, Harshman LA, He M, Jaeken J, James PM, Katz D, Keldermans L, Kibaek M, Kornberg AJ, Lachlan K, Lam C, Yaplito-Lee J, Nickerson DA, Peters HL, Race V, Régal L, Rush JS, Rutledge SL, Shendure J, Souche E, Sparks SE, Trapane P, Sanchez-Valle A, Vilain E, Vøllo A, Waechter CJ, Wang RY, Wolfe LA, Wong DA, Wood T, Yang AC; University of Washington Center for Mendelian Genomics, Matthijs G, Freeze HH. ALG1-CDG: Clinical and molecular characterization of 39 unreported patients. Hum Mutat. 2016 Jul;37(7):653-60. doi: 10.1002/humu.22983. Epub 2016 Mar 21.
156. Anselm I, MacCuaig M, Prabhu SB, Berry GT. Disease Heterogeneity in Na+/Citrate Cotransporter Deficiency. JIMD Rep. 2016 Mar 10.
157. Rodan LH, Berry GT. N-Acetylcysteine Therapy in an Infant with Transaldolase Is Well Tolerated and Associated with Normalization of Alpha Fetoprotein Levels. JIMD Rep. 2016 Apr 30. [Epub ahead of print] PMID: 27130472

158. Beinvogl BC, Rosman NP, Baumer FM, Rodan LH, Forster CS, Kwon AH, Berry GT.A 10-month-Old with Intermittent Hypotonia and Paralysis. Pediatrics. 2016 Jul;138(1). pii: e20151896. doi: 10.1542/peds.2015-1896. Epub 2016 Jun 1.
159. Richardson A, Berry GT, Garganta C, Abbott M-A. Hydroxysteroid 17-beta dehydrogenase type 10 disease in siblings, JIMD Rep. 2017;32:25-32.
160. Smedemark-Margulies N, Brownstein CA, Vargas S, Tembulkar SK, Towne MC, Shi J, Gonzalez-Cuevas E, Liu KX, Bilguvar K, Kleiman RJ, Han MJ, Torres A, Berry GT, Yu TW, Beggs AH, Agrawal PB, Gonzalez-Heydrich J. A novel de novo mutation in ATP1A3 and childhood-onset schizophrenia. Cold Spring Harb Mol Case Stud. 2016 Sep;2(5):a001008.
161. Kinney HC, Poduri AH, Cryan JB, Haynes RL, Teot L, Sleeper LA, Holm IA, Berry GT, Prabhu SP, Warfield SK, Brownstein C, Abram HS, Kruer M, Kemp WL, Hargitai B, Gastrang J, Mena OJ, Haas EA, Dastjerdi R, Armstrong DD, Goldstein RD. Hippocampal Formation Maldevelopment and Sudden Unexpected Death across the Pediatric Age Spectrum. J Neuropathol Exp Neurol. 2016 Oct;75(10):981-997. PMID: 27612489
162. Rodan LH, El Achkar CM, Berry GT, Poduri A, Prabhu SP, Yang E, Anselm I. De Novo TUBB2A Variant Presenting With Anterior Temporal Pachygyria. J Child Neurol. 2017 Jan;32(1):127-131.
163. Rubio-Gozalbo ME, Bosch AM, Burlina A, Berry GT, Treacy EP; Steering Committee on behalf of all Galactosemia Network representatives. The galactosemia network (GalNet). J Inherit Metab Dis. 2017 Mar;40(2):169-170. PMID: 27837294
164. Welling L, Bernstein LE, Berry GT, Burlina AB, Eyskens F, Gautschi M, Grünewald S, Gubbels CS, Knerr I, Labrune P, van der Lee JH, MacDonald A, Murphy E, Portnoi PA, Õunap K, Potter NL, Rubio-Gozalbo ME, Spencer JB, Timmers I, Treacy EP, Van Calcar SC, Waisbren SE, Bosch AM; Galactosemia Network (GalNet). International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up. J Inherit Metab Dis. 2017 Mar;40(2):171-176. PMID: 27858262
165. Huang X, Bedoyan JK, Demirbas D, Harris DJ, Miron A, Edelheit S, Grahame G, DeBrosse SD, Wong LJ, Hoppel CL, Kerr DS, Anselm I, Berry GT. Succinyl-CoA synthetase (SUCLA2) deficiency in two siblings with impaired activity of other mitochondrial oxidative enzymes in skeletal muscle without mitochondrial DNA depletion. Mol Genet Metab. 2017 Mar;120(3):213-222. PMID: 27913098
166. Sade Y, Toker L, Kara NZ, Einat H, Rapoport S, Moechars D, Berry GT, Bersudsky Y, Agam G. IP_3-Accumulation and/or Inositol-Depletion - Two Downstream Lithium's Effects that May Mediate its Behavioral and Cellular Changes. Translational Psychiatry, 2016 Dec 6;6(12):e968.
167. Van Steenbergen A, Balteau M, Ginion A, Ferté L, Battault S, Ravenstein CM, Balligand JL, Daskalopoulos EP, Gilon P, Despa F, Despa S, Vanoverschelde JL, Horman S, Koepsell H, Berry GT, Hue L, Bertrand L, Beauloye C. Sodium-myoinositol cotransporter-1, SMIT1, mediates the production of reactive oxygen species induced by hyperglycemia in the heart. Sci Rep. 2017 Jan 27;7:41166.
168. Aldubayan S, Rodan L, Berry GT, Levy H. Acute Management Protocol for Organic Acidemias: Methylmalonic Acidemia (MMA) and Propionic Acidemia (PA). Pediatr Emerg Care. 2017 Feb;33(2):142-146.
169. Vockley J, Burton B, Berry GT, Longo N, Phillips J, Sanchez-Valle A, Tanpaiboon P, Grunewald S, Murphy E, Humphrey R, Mayhew J, Bowden A, Zhang L, Cataldo J, Marsden

DL, Kakkis E. UX007 for the Treatment of Long Chain-Fatty Acid Oxidation Disorders: Safety and Efficacy in Children and Adults following 24 Weeks of Treatment. Mol Genet Metab. 2017 Apr;120(4):370-377.
170. Aldubayan S, Rodan L, Berry GT, Levy H. Acute Illness Protocol for Fatty Acid Oxidation and Carnitine Disorders. Pediatr Emerg Care. 2017 Apr;33(4):296-301.
171. O'Donnell-Luria AH, Lin AP, Merugumala SK, Rohr F, Waisbren SE, Lynch R, Tchekmedyian V, Goldberg AD, Bellinger A, McFaline-Figueroa JR, Simonc T, Gershanik EF, Levy BD, Cohen DE, Samuels MA, Berry GT, Frank NY. Brain MRS glutamine as a biomarker to guide therapy of hyperammonemic coma. Mol Genet Metab. *2017 May;121(1):9-15.*
172. van Erven B, Berry GT, Cassiman D, Connolly G, Forga M, Gautschi M, Gubbels CS, Hollak CEM, Janssen MC, Knerr I, Labrune P, Langendonk JG, Õunap K, Thijs A, Vos R, Wortmann SB, Rubio-Gozalbo ME. Fertility in adult women with classic galactosemia and primary ovarian insufficiency. Fertil Steril. 2017 Jul;108(1):168-174. PMID: 28579413
173. Haynes RL, Frelinger AL 3rd, Giles EK, Goldstein RD, Tran H, Kozakewich HP, Haas EA, Gerrits AJ, Mena OJ, Trachtenberg FL, Paterson DS, Berry GT, Adeli K, Kinney HC, Michelson AD. High serum serotonin in sudden infant death syndrome. Proc Natl Acad Sci U S A. 2017 Jul 18;114(29):7695-7700. PMID: 28674018
174. Wojcik MH, Wierenga KJ, Rodan LH, Sahai I, Ferdinandusse S, Genetti CA, Towne MC, Peake RWA, James PM, Beggs AH, Brownstein CA, Berry GT, Agrawal PB. Beta-ketothiolase deficiency presenting with metabolic stroke after a normal newborn screening in two individuals. JIMD Rep. 2017 Jul 20. doi: 10.1007/8904_2017_45. PMID: 28726122
175. Rajabi F, Rodan LH, Jonas MM, Soul JS, Ullrich NJ, Wessel A, Waisbren SE, Tan WH, Berry GT. Liver Failure as the Presentation of Ornithine Transcarbamylase Deficiency in a 13-Month-Old Female. JIMD Rep. 2017 Sep 9. doi: 10.1007/8904_2017_55. PMID: 28887792
176. Vanoevelen JM, van Erven B, Bierau J, Huang X, Berry GT, Vos R, Coelho AI, Rubio-Gozalbo ME. Impaired fertility and motor function in a zebrafish model for classic galactosemia. J Inherit Metab Dis. 2017 Sep 14. doi: 10.1007/s10545-017-0071-1PMID: 28913702
177. Aldubayan S, Rodan L, Berry GT, Levy H. Acute Illness Protocol for Maple Syrup Urine Disease (MSUD). Pediatr Emerg Care. 2018 Jan;34(1):64-67. doi: 10.1097/PEC.0000000000001299.
178. Aldubayan S, Rodan L, Berry GT, Levy H. Acute Illness Protocol for Urea Cycle Disorders. Pediatric Emergency Care, 2017 Nov 14. doi: 10.1097/PEC.0000000000001298.
179. Demirbas D, Coelho AI, Rubio-Gozalbo ME, Berry GT. Hereditary Galactosemia. Metabolism: Clinical and Experimental, Review, 2018 Jan 30. pii: S0026-0495(18)30033-7. doi: 10.1016/j.metabol.2018.01.025.
180. Hall PL, Lam C, Alexander JJ, Asif G, Berry GT, Ferreira C, Freeze HH, Gahl WA, Nickander KK, Sharer JD, Watson CM, Wolfe L, Raymond KM. "Urine oligosaccharide screening by MALDI-TOF for the identification of NGLY1 deficiency". Mol Genet Metab. 2018 Mar 10. pii: S1096-7192(18)30071-4. doi: 10.1016/j.ymgme.2018.03.002.
181. Rodan LH, Hauptman M, D'Gama AM, Qualls AE, Cao S, Tuschl K, Al-Jasmi F, Hertecant J, Hayflick SJ, Wessling-Resnick M, Yang ET, Berry GT, Gropman A, Woolf AD, Agrawal PB. "Novel founder intronic variant in SLC39A14 in two families causing

Manganism and potential treatment strategies." Mol Genet Metab. 2018 Apr 6. pii: S1096-7192(18)30049-0. doi: 10.1016/j.ymgme.2018.04.002.
182. Torres A, Brownstein CA, Tembulkar SK, Graber K, Genetti C, Kleiman RJ, Sweadner KJ, Mavros C, Liu KX, Smedemark-Margulies N, Maski K, Yang E, Agrawal PB, Shi J, Beggs AH, D'Angelo E, Lincoln SH, Carroll D, Dedeoglu F, Gahl WA, Biggs CM, Swoboda KJ, Berry GT, Gonzalez-Heydrich J. "*De novo ATP1A3* and compound heterozygous *NLRP3* mutations in a child with autism spectrum disorder, episodic fatigue and somnolence, and muckle-wells syndrome." Mol Genet Metab Rep. 2018 Jun 15;16:23-29. doi: 10.1016/j.ymgmr.2018.06.001. eCollection 2018 Sep.
183. Vockley J, Burton B, Berry GT, Longo N, Phillips J, Sanchez-Valle A, Tanpaiboon P, Grunewald S, Murphy E, Bowden A, Chen W, Chen CY, Cataldo J, Marsden D, Kakkis E. Results from a 78-week, single-arm, open-label Phase 2 study to evaluate UX007 in pediatric and adult patients with severe long-chain fatty acid oxidation disorders (LC-FAOD). J Inherit Metab Dis. 2018 Jul 9. doi: 10.1007/s10545-018-0217-9.
184. Rodan LH, Qi W, Ducker GS, Demirbas D, Laine R, Yang E, Walker MA, Eichler F, Rabinowitz JD, Anselm I*, Berry GT*. "5,10-methenyltetrahydrofolate synthetase deficiency causes a neurometabolic disorder associated with microcephaly, epilepsy, and cerebral hypomyelination", Mol Genet Metab. 2018 Jun 15. pii: S1096-7192(18)30114-8. doi: 10.1016/j.ymgme.2018.06.006.
185. Almontashiri NAM, Demirbas D, Berry GT, Peake RWA. (2018) Hyperammonemia in a Child Presenting with Growth Delay, Short Stature, and Diarrhea. Clin Chem. 64(8):1260-1262.
186. Mullikin D, Pillai N, Sanchez R, O'Donnell-Luria AH, Kritzer A, Tal L, Almannai M, Berry GT, Gambello MJ, Li H, Graham B, Srivaths L, Sutton VR, Grimes A. Megaloblastic Anemia Progressing to Severe Thrombotic Microangiopathy in Patients with Disordered Vitamin B12 Metabolism: Case Reports and Literature Review. J Pediatr. 2018 Jul 26. pii: S0022-3476(18)30879-5. doi: 10.1016/j.jpeds.2018.06.054.
187. Metz KA, Teng X, Coppens I, Lamb HM, Wagner BE, Rosenfeld JA, Chen X, Zhang Y, Kim HJ, Meadow ME, Wang TS, Haberlandt ED, Anderson GW, Leshinsky-Silver E, Bi W, Markello TC, Pratt M, Makhseed N, Garnica A, Danylchuk NR, Burrow TA, Jayakar P, McKnight D, Agadi S, Gbedawo H, Stanley C, Alber M, Prehl I, Peariso K, Ong MT, Mordekar SR, Parker MJ, Crooks D, Agrawal PB, Berry GT, Loddenkemper T, Yang Y, Maegawa GHB, Aouacheria A, Markle JG, Wohlschlegel JA, Hartman AL, Hardwick JM. KCTD7 deficiency defines a distinct neurodegenerative disorder with a conserved autophagy-lysosome defect. Ann Neurol. 2018 Nov;84(5):766-780.
188. Rodan LH, Anyane-Yeboa K, Chong K, Wassink-Ruiter K, Wilson A, Smith L, Kothare SV, Rajabi F, Ni M, DeBerardinis RJ, Poduri A, Berry GT. Gain-of-function variants in the *ODC1* gene cause a syndromic neurodevelopmental disorder associated with macrocephaly, alopecia, dysmorphic features, and neuroimaging abnormalities. Am J Med Genet A. 2018 Nov 26.
189. Haskovic M, Derks B, van der Ploeg L, Trommelen J, Nyakayiru J, van Loon LJC, Mackinnon S, Yue WW, Peake RWA, Zha L, Demirbas D, Qi W, Huang X, Berry GT, Achten J, Bierau J, Rubio-Gozalbo ME, Coelho AI. Arginine does not rescue p.Q188R mutation deleterious effect in classic galactosemia. Orphanet J Rare Dis. 2018 Nov 26;13(1):212.

190. Almontashiri NAM, Berry GT, Majzoub J, Peake RWA. Abnormal Glycerol Metabolism in a Child with Global Developmental Delay, Adrenal Insufficiency, and Intellectual Disability. Clin Chem. 2018 Dec;64(12):1785-1787.
191. Gartner V, Markello TC, Macnamara E, De Biase A, Thurm A, Joseph L, Beggs A, Schmahmann JD, Berry GT, Anselm I, Boslet E, Tifft CJ, Gahl WA, Lee PR. Novel variants in SPTAN1 without epilepsy: An expansion of the phenotype. Am J Med Genet A. 2018 Dec;176(12):2768-2776.
192. Ah Mew N, Cnaan A, McCarter R, Choi H, Glass P, Rice K, Scavo L, Gillespie CW, Diaz GA, Berry GT, Wong D, Konczal L, McCandless SE, Coughlin Ii CR, Weisfeld-Adams JD, Ficicioglu C, Yudkoff M, Enns GM, Lichter-Konecki U, Gallagher R, Tuchman M. Conducting an investigator-initiated randomized double-blinded intervention trial in acute decompensation of inborn errors of metabolism: Lessons from the N-Carbamylglutamate Consortium. Transl Sci Rare Dis. 2018 Dec 20;3(3-4):157-170.
193. Demirbas D, Huang X, Daesety V, Feenstra S, Haskovic M, Qi W, Gubbels CS, Hecht L, Levy HL, Waisbren SE, Berry GT. The ability of an LC-MS/MS-based erythrocyte GALT enzyme assay to predict the phenotype in subjects with GALT deficiency. Mol Genet Metab. 2019 Apr;126(4):368-376.
194. Vockley J, Burton B, Berry GT, Longo N, Phillips J, Sanchez-Valle A, Tanpaiboon P, Grunewald S, Murphy E, Bowden A, Chen W, Chen CY, Cataldo J, Marsden D, Kakkis E. Results from a 78-week, single-arm, open-label phase 2 study to evaluate UX007 in pediatric and adult patients with severe long-chain fatty acid oxidation disorders (LC-FAOD). J Inherit Metab Dis. 2019 Jan;42(1):169-177.
195. Posset R, Gropman AL, Nagamani SCS, Burrage LC, Bedoyan JK, Wong D, Berry GT, Baumgartner MR, Yudkoff M, Zielonka M, Hoffmann GF, Burgard P, Schulze A, McCandless SE, Garcia-Cazorla A, Seminara J, Garbade SF, Kölker S; Urea Cycle Disorders Consortium and the European Registry and Network for Intoxication Type Metabolic Diseases Consortia Study Group. Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Ann Neurol. 2019 Jul;86(1):116-128.
196. Rubio-Gozalbo ME, Haskovic M, Bosch AM, Burnyte B, Coelho AI, Cassiman D, Couce ML, Dawson C, Demirbas D, Derks T, Eyskens F, Forga MT, Grunewald S, Häberle J, Hochuli M, Hubert A, Huidekoper HH, Janeiro P, Kotzka J, Knerr I, Labrune P, Landau YE, Langendonk JG, Möslinger D, Müller-Wieland D, Murphy E, Õunap K, Ramadza D, Rivera IA, Scholl-Buergi S, Stepien KM, Thijs A, Tran C, Vara R, Visser G, Vos R, de Vries M, Waisbren SE, Welsink-Karssies MM, Wortmann SB, Gautschi M, Treacy EP, Berry GT. The natural history of classic galactosemia: lessons from the GalNet registry. Orphanet J Rare Dis. 2019 Apr 27;14(1):86.
197. Demirbas D, Harris DJ, Arn PH, Huang X, Waisbren SE, Anselm I, Lerner-Ellis JP, Wong LJ, Levy HL, Berry GT. Phenotypic variability in deficiency of the α subunit of succinate-CoA ligase. JIMD Rep. 2019 Mar 14;46(1):63-69.
198. Bedoyan JK, Hecht L, Zhang S, Tarrant S, Bergin A, Demirbas D, Yang E, Shun HK, Grahame GJ, DeBrosse SD, Hoppel CL, Kerr DS, Berry GT. A novel null mutation in the pyruvate dehydrogenase phosphatase catalytic subunit gene (PDP1) causing pyruvate dehydrogenase complex deficiency. JIMD Rep. 2019 Jun 17;48(1):26-35. doi: 10.1002/jmd2.12054.
199. Verheijen J, Wong SY, Rowe JH, Raymond K, Stoddard J, Delmonte OM, Bosticardo M, Dobbs K, Niemela J, Calzoni E, Pai SY, Choi U, Yamazaki Y, Comeau AM, Janssen E,

Henderson L, Hazen M, Berry G, Rosenzweig SD, Aldhekri HH, He M, Notarangelo LD, Morava E. Defining a new immune deficiency syndrome: MAN2B2-CDG. J Allergy Clin Immunol. 2019 Nov 24. pii: S0091-6749(19)31598-2. doi: 10.1016/j.jaci.2019.11.016. PMID:31775018

200. Haskovic M, Coelho AI, Bierau J, Vanoevelen JM, Steinbusch LKM, Zimmermann LJI, Villamor-Martinez E, Berry GT, Rubio-Gozalbo ME. Pathophysiology and targets for treatment in hereditary galactosemia: A systematic review of animal and cellular models. J Inherit Metab Dis. 2019 Dec 6. doi: 10.1002/jimd.12202. [Epub ahead of print] Review. PMID:31808946
201. Brucker WJ, Croteau SE, Prensner JR, Cullion K, Heeney MM, Lo J, McAlvin JB, Peeler K, Shah N, Yee CSK, Berry GT, Bodamer O. An emerging role for endothelial barrier support therapy for congenital disorders of glycosylation. J Inherit Metab Dis. 2020 Feb 16. doi: 10.1002/jimd.12225. [Epub ahead of print] PMID:32064623
202. Rech ME, McCarthy JM, Chen CA, Edmond JC, Shah VS, Bosch DGM, Berry GT, Williams L, Madan-Khetarpal S, Niyazov D, Shaw-Smith C, Kovar EM, Lupo PJ, Schaaf CP. Phenotypic expansion of Bosch-Boonstra-Schaaf optic atrophy syndrome and further evidence for genotype-phenotype correlations. Am J Med Genet A. 2020 Apr 10. doi: 10.1002/ajmg.a.61580. PMID: 32275123
203. Berry GT, Blume ED, Wessel A, Singh T, Hecht L, Marsden D, Sahai I, Elisofon S, Ferguson M, Kim HB, Harris DJ, Demirbas D, Almuqbil MA, Nyhan WL. The re-occurrence of cardiomyopathy in propionic acidemia after liver transplantation. JIMD Rep. 2020 Apr 8;54(1):3-8. doi: 10.1002/jmd2.12119.
204. Stergachis AB, Mogensen KM, Khoury CC, Lin AP, Peake RW, Baker JJ, Barkoudah E, Sahai I, Sweetser DA, Berry GT, Krier JB. A retrospective study of adult patients with non-cirrhotic hyperammonemia J Inherit Metab Dis. 2020 Jul 26. doi: 10.1002/jimd.12292.
205. M. Estela Rubio-Gozalbo ME, Derks B, Das AM, Meyer U, Moslinger D, Couce L, Empain A, Ficicioglu C, Palacios NJ, De Pelegrin MD, Rivera IA, Scholl-Burgi S, Bosch AM, Cassiman D, Demirbas D, Gautschi M, Knerr I, Labrune P, Skouma A, Verloo P, Worthmann SB, Treacy EP, Timson DJ, Berry GT. Galactokinase deficiency: lessons from the GalNet registry. Genet Med. 2020 Aug 18. doi: 10.1038/s41436-020-00942-9.
206. Shayota BJ, Donti TR, Xiao J, Gijavanekar C, Kennedy AD, Hubert L, Rodan L, Vanderpluym C, Nowak C, Bjornsson HT, Ganetzky R, Berry GT, Pappan KL, Sutton VR, Sun Q, Elsea SH. Untargeted metabolomics as an unbiased approach to the diagnosis of inborn errors of metabolism of the non-oxidative branch of the pentose phosphate pathway. Mol Genet Metab. 2020 Aug 5:S1096-7192(20)30182-7.
206. Banu Ahtam B, Berry GT, Prabhu SP, Greenstein PE, Afacan O, Waisbren SE, Brown M, Schomer D, Anastasoaie V, Petrides S, Grant E. Identification of neuronal structures and pathways corresponding to clinical functioning in galactosemia. JIMD, 2020, *in press*.
207. Vockley J, Burton B, Berry G, Longo N, Philips J, Sanches-Valle A, Chapman K, Tanpaiboon P, Grunewald S, Murphy E, Lu X, Cataldo J. Effects of triheptanoin (UX007) in patients with long-chain fatty acid oxidation disorders: Results from an open-label, long-term extension study. JIMD, 2020, *in press*.
208. D'Gama A, England E, Madden JA, Shi J, Chao KR, Wojcik MH, Torres AR, Tan WH, Berry GT, Prabhu SP, Agrawal PB. Exome sequencing identifies novel missense and deletion variants in RTN4IP1 associated with Optic Atrophy, Global Developmental Delay,

Epilepsy, Ataxia, and Choreoathetosis. Am J Med Genet A. 2021 Jan;185(1):203-207. doi: 10.1002/ajmg.a.61910. Epub 2020 Oct 9. PMID: 33037779
209. Ficicioglu C, Demirbas D, Derks B, Shashidhar PG, Timson DJ, Rubio-Gozalbo ME, Berry G. [13C]-galactose breath test in a patient with galactokinase deficiency and spastic diparesis. JIMD Reports, Feb 2021; https://doi.org/10.1002/jmd2.12205.
210. McGraw CM, Mahida S, Jayakar P, Koh HY, Taylor A, Resnick T, Rodan L, Schwartz MA, Ejaz A, Sankaran VG, Berry G, Poduri A. Uridine-responsive epileptic encephalopathy due to inherited variants in CAD: A Tale of Two Siblings. Ann Clin Transl Neurol. 2021 Mar;8(3):716-722. doi: 10.1002/acn3.51272.
211. Stergachis AB, Krier JB, Merugumala SK, Berry GT, Lin AP. Clinical utility of brain MRS imaging of patients with adult-onset non-cirrhotic hyperammonemia. Mol Genet Metab Rep. 2021 Mar 13;27:100742. doi: 10.1016/j.ymgmr.2021.100742. eCollection 2021 Jun.
212. Godfrey D, Torres A, Heidary G, Zahoor H, Lee A, Berry G, Engle E. A 7-year old female with arthrogryposis multiplex congenita, Duane retraction syndrome, and Marcus Gunn phenomenon due to a ZC4H2 gene mutation: a clinical presentation of the Wieacker-Wolff syndrome. Ophthalmic Genet. 2021 May 5:1-3. doi: 10.1080/13816810.2021.1923040.
213. Waisbren SE, Tran C, Demirbas D, Gubbels CS, Hsiao M, Daesety V, Berry GT. Transient developmental delays in infants with Duarte-2 variant galactosemia. Mol Genet Metab. 2021 Jul 30:S1096-7192(21)00760-5. doi: 10.1016/j.ymgme.2021.07.009. PMID: 34391645.
214. Rana M, Cuttin K, Berry GT, Torres A. Paroxysmal hyperthermia, dysautonomia and rhabdomyolysis in a patient with Lesch–Nyhan syndrome. JIMD Reports, *JIMD Rep.* 2021 Sep 28;62(1):30-34.
215. Koh HY, Haghighi A, Keywan C, Alexandrescu S, Plews-Ogan E, Haas EA, Brownstein CA, Vargas SO, Haynes RL, Berry GT, Holm IA, Poduri AH, Goldstein RD. Genetic Determinants of Sudden Unexpected Death in Pediatrics. Genet Med. 2022 Apr;24(4):839-850. doi: 10.1016/j.gim.2021.12.004.
216. Tiwary H, Hecht LE, Brucker WJ, Berry GT, Rodig NM. The development of end stage renal disease in two patients with PMM2-CDG. JIMD Rep. 2022 Jan 10;63(2):131-136. doi: 10.1002/jmd2.12269. PMID: 35281664; PMCID: PMC8898725
217. Zhao B, Madden JA, Lin J, Berry GT, Wojcik MH, Zhao X, Brand H, Talkowski M, Lee EA, Agrawal PB. A neurodevelopmental disorder caused by a novel de novo SVA insertion in exon 13 of the SRCAP gene. Eur J Hum Genet. 2022 Jun 30. doi: 10.1038/s41431-022-01137-3. Epub ahead of print. PMID: 35768521
218. Fridovich-Keil JL, Berry GT. Pathophysiology of long-term complications in classic galactosemia: What we do and do not know. Mol Genet Metab. 2022 Jul 9;137(1-2):33-39. doi: 10.1016/j.ymgme.2022.07.005. Epub ahead of print. PMID: 35882174.
219. Derks B, Demirbas D, Arantes RR, Banford S, Burlina AB, Cabrera A, Chiesa A, Couce ML, Dionisi-Vici C, Gautschi M, Grünewald S, Morava E, Möslinger D, Scholl-Bürgi S, Skouma A, Stepien KM, Timson DJ, Berry GT, Rubio-Gozalbo ME. Galactose epimerase deficiency: lessons from the GalNet registry. Orphanet J Rare Dis. 2022 Sep 2;17(1):331. doi: 10.1186/s13023-022-02494-4. PMID: 36056436; PMCID: PMC9438182.
220. Katler QS, Stepien KM, Paull N, Patel S, Adams M, Balci MC, Berry GT, Bosch AM, DeLaO A, Demirbas D, Edman J, Ficicioglu C, Goff M, Hacker S, Knerr I, Lancaster K, Li H, Mendelsohn BA, Nichols B, de Rezende Pinto WBV, Rocha JC, Rubio-Gozalbo ME,

Saad-Naguib M, Scholl-Buergi S, Searcy S, de Souza PVS, Wittenauer A, Fridovich-Keil JL. A multinational study of acute and long-term outcomes of Type 1 galactosemia patients who carry the S135L (c.404C > T) variant of GALT. J Inherit Metab Dis. 2022 Sep 12. doi: 10.1002/jimd.12556. Epub ahead of print. PMID: 36093991.

221. Derks B, Rivera-Cruz G, Hagen-Lillevik S, Vos EN, Demirbas D, Lai K, Treacy EP, Levy HL, Wilkins-Haug LE, Rubio-Gozalbo ME, Berry GT. The hypergonadotropic hypogonadism conundrum of classic galactosemia, Human Reproduction Update, 2022; dmac041, https://doi.org/10.1093/humupd/dmac041
222. Vockley J, Burton BK, Berry G, Longo N, Phillips J, Sanchez-Valle A, Chapman KA, Tanpaiboon P, Grunewald S, Murphy E, Lu X, Rahman S, Ray K, Reineking B, Pisani L, Ramirez AN. Triheptanoin for the treatment of long-chain fatty acid oxidation disorders: Final results of an open-label, long-term extension study. J Inherit Metab Dis. 2023 Sep;46(5):943-955. doi: 10.1002/jimd.12640. Epub 2023 Jun 19. PMID: 37276053.
223. Vos EN, Demirbas D, Mangel M, Gozalbo MER, Levy HL, Berry GT. The treatment of biochemical genetic diseases: From substrate reduction to nucleic acid therapies. Mol Genet Metab. 2023 Nov;140(3):107693. doi: 10.1016/j.ymgme.2023.107693. Epub 2023 Aug 30. PMID: 37716025 Review.
224. Panis B, Vos EN, Barić I, Bosch AM, Brouwers MCGJ, Burlina A, Cassiman D, Coman DJ, Couce ML, Das AM, Demirbas D, Empain A, Gautschi M, Grafakou O, Grunewald S, Kingma SDK, Knerr I, Leão-Teles E, Möslinger D, Murphy E, Õunap K, Pané A, Paci S, Parini R, Rivera IA, Scholl-Bürgi S, Schwartz IVD, Sdogou T, Shakerdi LA, Skouma A, Stepien KM, Treacy EP, Waisbren S, Berry GT, Rubio-Gozalbo ME. Brain function in classic galactosemia, a galactosemia network (GalNet) members review. Front Genet. 2024 Feb 15;15:1355962. doi: 10.3389/fgene.2024.1355962. eCollection 2024. PMID: 38425716
225. Manoli I, Sysol JR, Head PE, Epping MW, Gavrilova O, Crocker MK, Sloan JL, Koutsoukos SA, Wang C, Ktena YP, Mendelson S, Pass AR, Zerfas PM, Hoffmann V, Vernon HJ, Fletcher LA, Reynolds JC, Tsokos MG, Stratakis CA, Voss SD, Chen KY, Brown RJ, Hamosh A, Berry GT, Chen XS, Yanovski JA, Venditti CP. Lipodystrophy in methylmalonic acidemia associated with elevated FGF21 and abnormal methylmalonylation. JCI Insight. 2024 Feb 22;9(4):e174097. doi: 10.1172/jci.insight.174097. PMID: 38271099

Peer-reviewed Consortium Publications

1. Burrage LC, Reynolds JJ, Baratang NV, Phillips JB, Wegner J, McFarquhar A, Higgs MR, Christiansen AE, Lanza DG, Seavitt JR, Jain M, Li X, Parry DA, Raman V, Chitayat D, Chinn IK, Bertuch AA, Karaviti L, Schlesinger AE, Earl D, Bamshad M, Savarirayan R, Doddapaneni H, Muzny D, Jhangiani SN, Eng CM, Gibbs RA, Bi W, Emrick L, Rosenfeld JA, Postlethwait J, Westerfield M, Dickinson ME, Beaudet AL, Ranza E, Huber C, Cormier-Daire V, Shen W, Mao R, Heaney JD, Orange JS; University of Washington Center for Mendelian Genomics; Undiagnosed Diseases Network, Bertola D, Yamamoto GL, Baratela WAR, Butler MG, Ali A, Adeli M, Cohn DH, Krakow D, Jackson AP, Lees M, Offiah AC, Carlston CM, Carey JC, Stewart GS, Bacino CA, Campeau PM, Lee B. Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. Am J Hum Genet. 2019 Mar 7;104(3):422-438.

2. Nicoli ER, Weston MR, Hackbarth M, Becerril A, Larson A, Zein WM, Baker PR 2nd, Burke JD, Dorward H, Davids M, Huang Y, Adams DR, Zerfas PM, Chen D, Markello TC, Toro C, Wood T, Elliott G, Vu M; Undiagnosed Diseases Network, Zheng W, Garrett LJ, Tifft CJ, Gahl WA, Day-Salvatore DL, Mindell JA, Malicdan MCV. Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. Am J Hum Genet. 2019 Jun 6;104(6):1127-1138.
3. Frésard L, Smail C, Ferraro NM, Teran NA, Li X, Smith KS, Bonner D, Kernohan KD, Marwaha S, Zappala Z, Balliu B, Davis JR, Liu B, Prybol CJ, Kohler JN, Zastrow DB, Reuter CM, Fisk DG, Grove ME, Davidson JM, Hartley T, Joshi R, Strober BJ, Utiramerur S; Undiagnosed Diseases Network; Care4Rare Canada Consortium, Lind L, Ingelsson E, Battle A, Bejerano G, Bernstein JA, Ashley EA, Boycott KM, Merker JD, Wheeler MT, Montgomery SB. Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. Nat Med. 2019 Jun;25(6):911-919.
4. Shashi V, Geist J, Lee Y, Yoo Y, Shin U, Schoch K, Sullivan J, Stong N, Smith E, Jasien J, Kranz P; Undiagnosed Diseases Network, Lee Y, Shin YB, Wright NT, Choi M, Kontrogianni-Konstantopoulos A. Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Hum Mutat. 2019 May 5. doi: 10.1002/humu.23760.
5. Bhatia A, Mobley BC, Cogan J, Koziura ME, Brokamp E, Phillips J, Newman J; Undiagnosed Diseases Network (UDN), Moore SA, Hamid R; Members of the Undiagnosed Diseases Network. Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clin Imaging. 2019 Jun 21;58:108-113. doi: 10.1016/j.clinimag.2019.06.010.
6. Kanca O, Andrews JC, Lee PT, Patel C, Braddock SR, Slavotinek AM, Cohen JS, Gubbels CS, Aldinger KA, Williams J, Indaram M, Fatemi A, Yu TW, Agrawal PB, Vezina G, Simons C, Crawford J, Lau CC; Undiagnosed Diseases Network, Chung WK, Markello TC, Dobyns WB, Adams DR, Gahl WA, Wangler MF, Yamamoto S, Bellen HJ, Malicdan MCV. De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. Am J Hum Genet. 2019 Aug 1;105(2):413-424. doi: 10.1016/j.ajhg.2019.06.014. Epub 2019 Jul 18.
7. Geng LN, Kohler JN, Levonian P; Members of the Undiagnosed Diseases Network, Bernstein JA, Ford JM, Ahuja N, Witteles R, Hom J, Wheeler M. Genomics in medicine: a novel elective rotation for internal medicine residents. Postgrad Med J. 2019 Oct;95(1128):569-572. doi: 10.1136/postgradmedj-2018-136355. Epub 2019 Aug 22. PMID:31439813
8. Johnson BV, Kumar R, Oishi S, Alexander S, Kasherman M, Vega MS, Ivancevic A, Gardner A, Domingo D, Corbett M, Parnell E, Yoon S, Oh T, Lines M, Lefroy H, Kini U, Van Allen M, Grønborg S, Mercier S, Küry S, Bézieau S, Pasquier L, Raynaud M, Afenjar A, Billette de Villemeur T, Keren B, Désir J, Van Maldergem L, Marangoni M, Dikow N, Koolen DA, VanHasselt PM, Weiss M, Zwijnenburg P, Sa J, Reis CF, López-Otín C, Santiago-Fernández O, Fernández-Jaén A, Rauch A, Steindl K, Joset P, Goldstein A, Madan-Khetarpal S, Infante E, Zackai E, Mcdougall C, Narayanan V, Ramsey K, Mercimek-Andrews S, Pena L, Shashi V; Undiagnosed Diseases Network, Schoch K, Sullivan JA, Pinto E Vairo F, Pichurin PN, Ewing SA, Barnett SS, Klee EW, Perry MS, Koenig MK, Keegan CE, Schuette JL, Asher S, Perilla-Young Y, Smith LD, Rosenfeld JA,

Bhoj E, Kaplan P, Li D, Oegema R, van Binsbergen E, van der Zwaag B, Smeland MF, Cutcutache I, Page M, Armstrong M, Lin AE, Steeves MA, Hollander ND, Hoffer MJV, Reijnders MRF, Demirdas S, Koboldt DC, Bartholomew D, Mosher TM, Hickey SE, Shieh C, Sanchez-Lara PA, Graham JM Jr, Tezcan K, Schaefer GB, Danylchuk NR, Asamoah A, Jackson KE, Yachelevich N, Au M, Pérez-Jurado LA, Kleefstra T, Penzes P, Wood SA, Burne T, Pierson TM, Piper M, Gécz J, Jolly LA. Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor β Signaling. Biol Psychiatry. 2019 Jun 29. pii: S0006-3223(19)31479-9. doi: 10.1016/j.biopsych.2019.05.028. PMID:31443933

9. Accogli A, Calabretta S, St-Onge J, Boudrahem-Addour N, Dionne-Laporte A, Joset P, Azzarello-Burri S, Rauch A, Krier J, Fieg E, Pallais JC; Undiagnosed Diseases Network, McConkie-Rosell A, McDonald M, Freedman SF, Rivière JB, Lafond-Lapalme J, Simpson BN, Hopkin RJ, Trimouille A, Van-Gils J, Begtrup A, McWalter K, Delphine H, Keren B, Genevieve D, Argilli E, Sherr EH, Severino M, Rouleau GA, Yam PT, Charron F, Srour M. De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Collosum, Axon, Cardiac, Ocular, and Genital Defects. Am J Hum Genet. 2019 Oct 3;105(4):854-868. doi: 10.1016/j.ajhg.2019.09.005. PMID:31585109
10. Holt JM, Wilk B, Birch CL, Brown DM, Gajapathy M, Moss AC, Sosonkina N, Wilk MA, Anderson JA, Harris JM, Kelly JM, Shaterferdosian F, Uno-Antonison AE, Weborg A; Undiagnosed Diseases Network, Worthey EA. VarSight: prioritizing clinically reported variants with binary classification algorithms. BMC Bioinformatics. 2019 Oct 15;20(1):496. doi: 10.1186/s12859-019-3026-8. PMID:31615419
11. Zielonka M, Kölker S, Gleich F, Stützenberger N, Nagamani SCS, Gropman AL, Hoffmann GF, Garbade SF, Posset R; Urea Cycle Disorders Consortium (UCDC) and the European Registry and Network for Intoxication type Metabolic Diseases (E-IMD) Consortia Study Group. Early prediction of phenotypic severity in Citrullinemia Type 1. Ann Clin Transl Neurol. 2019 Sep;6(9):1858-1871. doi: 10.1002/acn3.50886. Epub 2019 Aug 30. PMID: 31469252
12. Mao D, Reuter CM, Ruzhnikov MRZ, Beck AE, Farrow EG, Emrick LT, Rosenfeld JA, Mackenzie KM, Robak L, Wheeler MT, Burrage LC, Jain M, Liu P, Calame D, Küry S, Sillesen M, Schmitz-Abe K, Tonduti D, Spaccini L, Iascone M, Genetti CA, Koenig MK, Graf M, Tran A, Alejandro M. De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation.; Undiagnosed Diseases Network, Lee BH, Thiffault I, Agrawal PB, Bernstein JA, Bellen HJ, Chao HT. Am J Hum Genet. 2020 Apr 2;106(4):570-583. doi:10.1016/j.ajhg.2020.02.016. Epub 2020 Mar 19. PMID: 32197074
13. Posset R, Garbade SF, Gleich F, Gropman AL, de Lonlay P, Hoffmann GF, Garcia-Cazorla A, Nagamani SCS, Baumgartner MR, Schulze A, Dobbelaere D, Yudkoff M, Kölker S, Zielonka M; Urea Cycle Disorders Consortium (UCDC); European registry and network for Intoxication type Metabolic Diseases (E-IMD). Long-term effects of medical management on growth and weight in individuals with urea cycle disorders. Sci Rep. 2020 Jul 20;10(1):11948.
14. Schneeberger PE, Kortüm F, Korenke GC, Alawi M, Santer R, Woidy M, Buhas D, Fox S, Juusola J, Alfadhel M, Webb BD, Coci EG, Abou Jamra R, Siekmeyer M, Biskup S, Heller C, Maier EM, Javaher-Haghighi P, Bedeschi MF, Ajmone PF, Iascone M, Peeters H, Ballon K, Jaeken J, Rodríguez Alonso A, Palomares-Bralo M, Santos-Simarro F,

Meuwissen MEC, Beysen D, Kooy RF, Houlden H, Murphy D, Doosti M, Karimiani EG, Mojarrad M, Maroofian R, Noskova L, Kmoch S, Honzik T, Cope H, Sanchez-Valle A; Undiagnosed Diseases Network, Gelb BD, Kurth I, Hempel M, Kutsche K. Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder.Brain. 2020 Aug 6:awaa204. doi: 10.1093/brain/awaa204.

15. Schoch K, Esteves C, Bican A, Spillmann R, Cope H, McConkie-Rosell A, Walley N, Fernandez L, Kohler JN, Bonner D, Reuter C, Stong N, Mulvihill JJ, Novacic D, Wolfe L, Abdelbaki A, Toro C, Tifft C, Malicdan M, Gahl W, Liu P, Newman J, Goldstein DB, Hom J, Sampson J, Wheeler MT; Undiagnosed Diseases Network, Cogan J, Bernstein JA, Adams DR, McCray AT, Shashi V. Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genet Med. 2021 Feb;23(2):259-271. doi: 10.1038/s41436-020-00984-z. Epub 2020 Oct 23. PMID: 33093671
16. Studwell CM, Kelley EG; Undiagnosed Diseases Network, Sinsheimer JS, Palmer CGS, LeBlanc K. Family genetic result communication in rare and undiagnosed disease communities: Understanding the practice. J Genet Couns. 2020 Oct 27. doi: 10.1002/jgc4.1329. Online ahead of print. PMID: 33108040
17. Meissner LE, Macnamara EF, D'Souza P, Yang J, Vezina G; Undiagnosed Diseases Network, Ferreira CR, Zein WM, Tifft CJ, Adams DR. DYRK1A pathogenic variants in two patients with syndromic intellectual disability and a review of the literature. Mol Genet Genomic Med. 2020 Dec;8(12):e1544. doi: 10.1002/mgg3.1544. Epub 2020 Nov 7. PMID: 33159716
18. Ferdinandusse S, McWalter K, Te Brinke H, IJlst L, Mooijer PM, Ruiter JPN, van Lint AEM, Pras-Raves M, Wever E, Millan F, Guillen Sacoto MJ, Begtrup A, Tarnopolsky M, Brady L, Ladda RL, Sell SL, Nowak CB, Douglas J, Tian C, Ulm E, Perlman S, Drack AV, Chong K, Martin N, Brault J, Brokamp E, Toro C, Gahl WA, Macnamara EF, Wolfe L; Undiagnosed Diseases Network, Waisfisz Q, Zwijnenburg PJG, Ziegler A, Barth M, Smith R, Ellingwood S, Gaebler-Spira D, Bakhtiari S, Kruer MC, van Kampen AHC, Wanders RJA, Waterham HR, Cassiman D, Vaz FM. An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids. Genet Med. 2020 Nov 26. doi: 10.1038/s41436-020-01027-3. Online ahead of print. PMID: 33239752
19. Kobren SN, Baldridge D, Velinder M, Krier JB, LeBlanc K, Esteves C, Pusey BN, Züchner S, Blue E, Lee H, Huang A, Bastarache L, Bican A, Cogan J, Marwaha S, Alkelai A, Murdock DR, Liu P, Wegner DJ, Paul AJ; Undiagnosed Diseases Network, Sunyaev SR, Kohane IS. Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. Genet Med. 2021 Feb 12. doi: 10.1038/s41436-020-01084-8. Online ahead of print. PMID: 33580225
20. Marbach F, Stoyanov G, Erger F, Stratakis CA, Settas N, London E, Rosenfeld JA, Torti E, Haldeman-Englert C, Sklirou E, Kessler E, Ceulemans S, Nelson SF, Martinez-Agosto JA, Palmer CGS, Signer RH; Undiagnosed Diseases Network, Andrews MV, Grange DK, Willaert R, Person R, Telegrafi A, Sievers A, Laugsch M, Theiß S, Cheng Y, Lichtarge O, Katsonis P, Stocco A, Schaaf CP. Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. Med. 2021 Apr 8. doi: 10.1038/s41436-021-01152-7.
21. Kyle JE, Stratton KG, Zink EM, Kim YM, Bloodsworth KJ, Monroe ME; Undiagnosed Diseases Network, Waters KM, Webb-Robertson BM, Koeller DM, Metz TO. A resource

of lipidomics and metabolomics data from individuals with undiagnosed diseases. Sci Data. 2021 Apr 21;8(1):114. doi: 10.1038/s41597-021-00894-y.
22. Cope H, Barseghyan H, Bhattacharya S, Fu Y, Hoppman N, Marcou C, Walley N, Rehder C, Deak K, Alkelai A; Undiagnosed Diseases Network, Vilain E, Shashi V. Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Mol Genet Genomic Med. 2021 May 6:e1665. doi: 10.1002/mgg3.1665.
23. Sturrock BRH, Macnamara EF, McGuire P, Kruk S, Yang I, Murphy J; Undiagnosed Diseases Network, Tifft CJ, Gordon-Lipkin E. Progressive cerebellar atrophy in a patient with complex II and III deficiency and a novel deleterious variant in SDHA: A Counseling Conundrum. Mol Genet Genomic Med. 2021 May 7:e1692. doi: 10.1002/mgg3.1692.
24. Baldridge D, Wangler MF, Bowman AN, Yamamoto S; Undiagnosed Diseases Network, Schedl T, Pak SC, Postlethwait JH, Shin J, Solnica-Krezel L, Bellen HJ, Westerfield M. Model organisms contribute to diagnosis and discovery in the undiagnosed diseases network: current state and a future vision. Orphanet J Rare Dis. 2021 May 7;16(1):206. doi: 10.1186/s13023-021-01839-9. PMID: 33962631
25. Panneerselvam S, Wang J, Zhu W, Dai H, Pappas JG, Rabin R, Low KJ, Rosenfeld JA, Emrick L, Xiao R, Xia F, Yang Y, Eng CM, Anderson A, Chau V, Soler-Alfonso C, Streff H, Lalani SR, Mercimek-Andrews S; Undiagnosed Diseases Network; DDD Study, Bi W. PPP3CA truncating variants clustered in the regulatory domain cause early-onset refractory epilepsy. Clin Genet. 2021 May 8. doi: 10.1111/cge.13979.
26. Brokamp E, Koziura ME, Phillips JA 3rd, Tang LA, Cogan JD, Rives LC, Robertson AK, Duncan L, Bican A, Peterson JF, Newman JH, Hamid R, Bastarache L; Undiagnosed Diseases Network. One is the loneliest number: genotypic matchmaking using the electronic health record. Genet Med. 2021 Jul 6. doi: 10.1038/s41436-021-01179-w.
27. Rodan LH, Spillmann RC, Kurata HT, Lamothe SM, Maghera J, Jamra RA, Alkelai A, Antonarakis SE, Atallah I, Bar-Yosef O, Bilan F, Bjorgo K, Blanc X, Van Bogaert P, Bolkier Y, Burrage LC, Christ BU, Granadillo JL, Dickson P, Donald KA, Dubourg C, Eliyahu A, Emrick L, Engleman K, Gonfiantini MV, Good JM, Kalser J, Kloeckner C, Lachmeijer G, Macchiaiolo M, Nicita F, Odent S, O'Heir E, Ortiz-Gonzalez X, Pacio-Miguez M, Palomares-Bralo M, Pena L, Platzer K, Quinodoz M, Ranza E, Rosenfeld JA, Roulet-Perez E, Santani A, Santos-Simarro F, Pode-Shakked B, Skraban C, Slaugh R, Superti-Furga A, Thiffault I, van Jaabrsveld RH, Vincent M, Wang HG, Zacher P; Undiagnosed Diseases Network, Rush E, Pitt G, Au PYB, Shashi V. Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genet Med. 2021 Jun 23. doi: 10.1038/s41436-021-01232-8. Online ahead of print. PMID: 34163037
28. McConkie-Rosell A, Schoch K, Sullivan J, Spillmann RC, Cope H, Tan QK, Palmer CGS; Undiagnosed Disease Network, Hooper SR, Shashi V. Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples.J Genet Couns. 2021 Jun 11. doi: 10.1002/jgc4.1451. Online ahead of print. PMID: 34115423
29. Ravenscroft TA, Phillips JB, Fieg E, Bajikar SS, Peirce J, Wegner J, Luna AA, Fox EJ, Yan YL, Rosenfeld JA, Zirin J, Kanca O; Undiagnosed Diseases Network, Benke PJ, Cameron ES, Strehlow V, Platzer K, Jamra RA, Klöckner C, Osmond M, Licata T, Rojas S, Dyment D, Chong JSC, Lincoln S, Stoler JM, Postlethwait JH, Wangler MF, Yamamoto

S, Krier J, Westerfield M, Bellen HJ. Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. Genet Med. 2021 Jun 10. doi: 10.1038/s41436-021-01216-8. Online ahead of print. PMID: 34113007

30. Kohler JN, Kelley EG, Boyd BM, Sillari CH, Marwaha S; Undiagnosed Diseases Network, Wheeler MT. Genetic counselor roles in the undiagnosed diseases network research study: Clinical care, collaboration, and curation. J Genet Couns. 2021 Aug 10. doi: 10.1002/jgc4.1493. PMID: 34374469
31. Lerner S, Eilam R, Adler L, Baruteau J, Kreiser T, Tsoory M, Brandis A, Mehlman T, Ryten M, Botia JA, Ruiz SG, Garcia AC, Dionisi-Vici C, Ranucci G, Spada M, Mazkereth R, McCarter R, Izem R, Balmat TJ, Richesson R; Members of the UCDC, Gazit E, Nagamani SCS, Erez A. ASL expression in ALDH1A1+ neurons in the substantia nigra metabolically contributes to neurodegenerative phenotype. Hum Genet. 2021 Aug 21. doi: 10.1007/s00439-021-02345-5. PMID: 34417872
32. Borja N, Bivona S, Peart LS, Johnson B, Gonzalez J, Barbouth D, Moore H, Guo S; Undiagnosed Disease Network, Bademci G, Tekin M. Genome sequencing reveals novel noncoding variants in PLA2G6 and LMNB1 causing progressive neurologic disease. Mol Genet Genomic Med. 2022 Apr;10(4):e1892. doi: 10.1002/mgg3.1892. Epub 2022 Mar 5. PMID: 35247231; PMCID: PMC9000935
33. Shankar SP, Grimsrud K, Lanoue L, Egense A, Willis B, Hörberg J, AlAbdi L, Mayer K, Ütkür K, Monaghan KG, Krier J, Stoler J, Alnemer M, Shankar PR, Schaffrath R, Alkuraya FS, Brinkmann U, Eriksson LA, Lloyd K, Rauen KA; Undiagnosed Diseases Network. A novel DPH5-related diphthamide-deficiency syndrome causing embryonic lethality or profound neurodevelopmental disorder. Genet Med. 2022 Jul;24(7):1567-1582. doi: 10.1016/j.gim.2022.03.014. Epub 2022 Apr 28. PMID: 35482014
34. Barish S, Senturk M, Schoch K, Minogue AL, Lopergolo D, Fallerini C, Harland J, Seemann JH, Stong N, Kranz PG, Kansagra S, Mikati MA, Jasien J, El-Dairi M, Galluzzi P; Undiagnosed Diseases Network, Ariani F, Renieri A, Mari F, Wangler MF, Arur S, Jiang YH, Yamamoto S, Shashi V, Bellen HJ. The microRNA processor DROSHA is a candidate gene for a severe progressive neurological disorder. Hum Mol Genet. 2022 Aug 25;31(17):2934-2950. doi: 10.1093/hmg/ddac085. PMID: 35405010; PMCID: PMC9433733.
35. Kozycki CT, Kodati S, Huryn L, Wang H, Warner BM, Jani P, Hammoud D, Abu-Asab MS, Jittayasothorn Y, Mattapallil MJ, Tsai WL, Ullah E, Zhou P, Tian X, Soldatos A, Moutsopoulos N, Kao-Hsieh M, Heller T, Cowen EW, Lee CR, Toro C, Kalsi S, Khavandgar Z, Baer A, Beach M, Long Priel D, Nehrebecky M, Rosenzweig S, Romeo T, Deuitch N, Brenchley L, Pelayo E, Zein W, Sen N, Yang AH, Farley G, Sweetser DA, Briere L, Yang J, de Oliveira Poswar F, Schwartz IVD, Silva Alves T, Dusser P, Koné-Paut I, Touitou I, Titah SM, van Hagen PM, van Wijck RTA, van der Spek PJ, Yano H, Benneche A, Apalset EM, Jansson RW, Caspi RR, Kuhns DB, Gadina M, Takada H, Ida H, Nishikomori R, Verrecchia E, Sangiorgi E, Manna R, Brooks BP, Sobrin L, Hufnagel RB, Beck D, Shao F, Ombrello AK, Aksentijevich I, Kastner DL; Undiagnosed Diseases Network. Gain-of-function mutations in ALPK1 cause an NF-κB-mediated autoinflammatory disease: functional assessment, clinical phenotyping and disease course of patients with ROSAH syndrome. Ann Rheum Dis. 2022 Oct;81(10):1453-1464. doi: 10.1136/annrheumdis-2022-222629. Epub 2022 Jul 22. PMID: 35868845; PMCID: PMC9484401.

36. Bainbridge MN, Mazumder A, Ogasawara D, Abou Jamra R, Bernard G, Bertini E, Burglen L, Cope H, Crawford A, Derksen A, Dure L, Gantz E, Koch-Hogrebe M, Hurst ACE, Mahida S, Marshall P, Micalizzi A, Novelli A, Peng H; Rady Children's Institute for Genomic Medicine, Rodriguez D, Robbins SL, Rutledge SL, Scalise R, Schließke S, Shashi V, Srivastava S, Thiffault I, Topol S; Undiagnosed Disease Network, Qebibo L, Wieczorek D, Cravatt B, Haricharan S, Torkamani A, Friedman J. Endocannabinoid dysfunction in neurological disease: neuro-ocular DAGLA-related syndrome. Brain. 2022 Oct 21;145(10):3383-3390. doi: 10.1093/brain/awac223. PMID: 35737950; PMCID: PMC9586540.
37. Scharre S, Posset R, Garbade SF, Gleich F, Seidl MJ, Druck AC, Okun JG, Gropman AL, Nagamani SCS, Hoffmann GF, Kölker S, Zielonka M; Urea Cycle Disorders Consortium (UCDC) and the European registry and network for Intoxication type Metabolic Diseases (E-IMD) Consortia Study Group. Predicting the disease severity in male individuals with ornithine transcarbamylase deficiency. Ann Clin Transl Neurol. 2022 Nov;9(11):1715-1726. doi: 10.1002/acn3.51668. Epub 2022 Oct 10. PMID: 36217298; PMCID: PMC9639638.
38. Miller IM, Yashar BM; Undiagnosed Disease Network; Macnamara EF. Continuing a search for a diagnosis: the impact of adolescence and family dynamics. Orphanet J Rare Dis. 2023 Jan 9;18(1):6. doi: 10.1186/s13023-022-02598-x. PMID: 36624503; PMCID: PMC9830697.
39. Morimoto M, Bhambhani V, Gazzaz N, Davids M, Sathiyaseelan P, Macnamara EF, Lange J, Lehman A, Zerfas PM, Murphy JL, Acosta MT, Wang C, Alderman E; Undiagnosed Diseases Network; Reichert S, Thurm A, Adams DR, Introne WJ, Gorski SM, Boerkoel CF, Gahl WA, Tifft CJ, Malicdan MCV. Bi-allelic ATG4D variants are associated with a neurodevelopmental disorder characterized by speech and motor impairment. NPJ Genom Med. 2023 Feb 10;8(1):4. doi: 10.1038/s41525-022-00343-8. PMID: 36765070; PMCID: PMC9918471.
40. Spillmann RC, Tan QK, Reuter C, Schoch K; Undiagnosed Diseases Network; Kohler J, Bonner D, Zastrow D, Alkelai A, Baugh E, Cope H, Marwaha S, Wheeler MT, Bernstein JA, Shashi V; Undiagnosed Diseases Network. A concurrent dual analysis of genomic data augments diagnoses: Experiences of 2 clinical sites in the Undiagnosed Diseases Network. Genet Med. 2023 Apr;25(4):100353. doi: 10.1016/j.gim.2022.12.001. Epub 2022 Dec 5. PMID: 36481303.
41. McConkie-Rosell A, Spillmann RC, Schoch K, Sullivan JA, Walley N, McDonald M; Undiagnosed Diseases Network; Hooper SR, Shashi V. Unraveling non-participation in genomic research: A complex interplay of barriers, facilitators, and sociocultural factors. J Genet Couns. 2023 Apr 2. doi: 10.1002/jgc4.1707. Epub ahead of print. PMID: 37005744.
42. Rosenfeld LE, LeBlanc K, Nagy A, Ego BK; Undiagnosed Diseases Network; McCray AT. Participation in a national diagnostic research study: assessing the patient experience. Orphanet J Rare Dis. 2023 Apr 10;18(1):73. doi: 10.1186/s13023-023-02695-5. PMID: 37032333; PMCID: PMC10084693.
43. Srivastava S, Shaked HM, Gable K, Gupta SD, Pan X, Somashekarappa N, Han G, Mohassel P, Gotkine M, Doney E, Goldenberg P, Tan QKG, Gong Y, Kleinstiver B, Wishart B, Cope H, Pires CB, Stutzman H, Spillmann RC; Undiagnosed Disease Network; Sadjadi R, Elpeleg O, Lee CH, Bellen HJ, Edvardson S, Eichler F, Dunn TM. SPTSSA

variants alter sphingolipid synthesis and cause a complex hereditary spastic paraplegia. Brain. 2023 Apr 19;146(4):1420-1435. doi: 10.1093/brain/awac460. PMID: 36718090.
44. Tepe B, Macke EL, Niceta M, Weisz Hubshman M, Kanca O, Schultz-Rogers L, Zarate YA, Schaefer GB, Granadillo De Luque JL, Wegner DJ, Cogne B, Gilbert-Dussardier B, Le Guillou X, Wagner EJ, Pais LS, Neil JE, Mochida GH, Walsh CA, Magal N, Drasinover V, Shohat M, Schwab T, Schmitz C, Clark K, Fine A, Lanpher B, Gavrilova R, Blanc P, Burglen L, Afenjar A, Steel D, Kurian MA, Prabhakar P, Gößwein S, Di Donato N, Bertini ES; Undiagnosed Diseases Network; Wangler MF, Yamamoto S, Tartaglia M, Klee EW, Bellen HJ. Bi-allelic variants in INTS11 are associated with a complex neurological disorder. Am J Hum Genet. 2023 May 4;110(5):774-789. doi: 10.1016/j.ajhg.2023.03.012. Epub 2023 Apr 12. PMID: 37054711; PMCID: PMC10183469.
45. Andrews JC, Mok JW, Kanca O, Jangam S, Tifft C, Macnamara EF, Russell BE, Wang LK; Undiagnosed Diseases Network; Nelson SF, Bellen HJ, Yamamoto S, Malicdan MCV, Wangler MF. De novo variants in MRTFB have gain-of-function activity in Drosophila and are associated with a novel neurodevelopmental phenotype with dysmorphic features. Genet Med. 2023 Jun;25(6):100833. doi: 10.1016/j.gim.2023.100833. Epub 2023 Mar 31. PMID: 37013900.
46. Pujol-Giménez J, Mirzaa G, Blue EE, Albano G, Miller DE, Allworth A, Bennett JT, Byers PH, Chanprasert S, Chen J, Doherty D, Folta AB, Gillentine MA, Glass I, Hing A, Horike-Pyne M, Leppig KA, Parhin A, Ranchalis J, Raskind WH, Rosenthal EA, Schwarze U, Sheppeard S, Strohbehn S, Sybert VP, Timms A, Wener M; University of Washington Center for Mendelian Genomics (UW-CMG)a, Undiagnosed Diseases Network (UDN); Bamshad MJ, Hisama FM, Jarvik GP, Dipple KM, Hediger MA, Stergachis AB. Dominant-negative variant in SLC1A4 causes an autosomal dominant epilepsy syndrome. Ann Clin Transl Neurol. 2023 Jun;10(6):1046-1053. doi: 10.1002/acn3.51786. Epub 2023 May 16. PMID: 37194416; PMCID: PMC10270265.
47. Halley MC, Young JL, Tang C, Mintz KT, Lucas-Griffin S, Maghiro AS, Ashley EA, Tabor HK; Undiagnosed Diseases Network. Genomics Research with Undiagnosed Children: Ethical Challenges at the Boundaries of Research and Clinical Care. J Pediatr. 2023 Jun 2:113537. doi: 10.1016/j.jpeds.2023.113537. Epub ahead of print. PMID: 37271495.
48. Mohajeri A, Vaseghi-Shanjani M, Rosenfeld JA, Yang GX, Lu H, Sharma M, Lin S, Salman A, Waqas M, Sababi Azamian M, Worley KC, Del Bel KL, Kozak FK, Rahmanian R, Biggs CM, Hildebrand KJ, Lalani SR, Nicholas SK, Scott DA, Mostafavi S, van Karnebeek C, Henkelman E, Halparin J, Yang CL, Armstrong L; Undiagnosed Diseases Network; Care4Rare Canada Consortium; Turvey SE, Lehman A. Dominant negative variants in IKZF2 cause ICHAD syndrome, a new disorder characterised by immunodysregulation, craniofacial anomalies, hearing impairment, athelia and developmental delay. J Med Genet. 2023 Jun 14:jmg-2022-109127. doi: 10.1136/jmg-2022-109127. Epub ahead of print. PMID: 37316189.
49. Borja N, Borjas-Mendoza P, Bivona S, Peart L, Gonzalez J, Johnson BK, Guo S, Yusupov R; Undiagnosed Diseases Network; Bademci G, Tekin M. H4C5 missense variant leads to a neurodevelopmental phenotype overlapping with Angelman syndrome. Am J Med Genet A. 2023 Jul;191(7):1911-1916. doi: 10.1002/ajmg.a.63193. Epub 2023 Mar 29. PMID: 36987712; PMCID: PMC10286100.

50. Morleo M, Venditti R, Theodorou E, Briere LC, Rosello M, Tirozzi A, Tammaro R, Al-Badri N, High FA, Shi J; Undiagnosed Diseases Network; Telethon Undiagnosed Diseases Program; Putti E, Ferrante L, Cetrangolo V, Torella A, Walker MA, Tenconi R, Iascone M, Mei D, Guerrini R, van der Smagt J, Kroes HY, van Gassen KLI, Bilal M, Umair M, Pingault V, Attie-Bitach T, Amiel J, Ejaz R, Rodan L, Zollino M, Agrawal PB, Del Bene F, Nigro V, Sweetser DA, Franco B. De novo missense variants in phosphatidylinositol kinase PIP5KIγ underlie a neurodevelopmental syndrome associated with altered phosphoinositide signaling. Am J Hum Genet. 2023 Aug 3;110(8):1377-1393. doi: 10.1016/j.ajhg.2023.06.012. Epub 2023 Jul 13. PMID: 37451268
51. Niggl E, Bouman A, Briere LC, Hoogenboezem RM, Wallaard I, Park J, Admard J, Wilke M, Harris-Mostert EDRO, Elgersma M, Bain J, Balasubramanian M, Banka S, Benke PJ, Bertrand M, Blesson AE, Clayton-Smith J, Ellingford JM, Gillentine MA, Goodloe DH, Haack TB, Jain M, Krantz I, Luu SM, McPheron M, Muss CL, Raible SE, Robin NH, Spiller M, Starling S, Sweetser DA, Thiffault I, Vetrini F, Witt D, Woods E, Zhou D; Genomics England Research Consortium; Undiagnosed Diseases Network; Elgersma Y, van Esbroeck ACM. HNRNPC haploinsufficiency affects alternative splicing of intellectual disability-associated genes and causes a neurodevelopmental disorder. Am J Hum Genet. 2023 Aug 3;110(8):1414-1435. doi: 10.1016/j.ajhg.2023.07.005. PMID: 37541189
52. Shashi V, Schoch K, Ganetzky R, Kranz PG, Sondheimer N, Markert ML, Cope H, Sadeghpour A, Roehrs P, Arbogast T, Muraresku C; Undiagnosed Diseases Network; Tyndall AV, Esser MJ, Woodward KE, Ping-Yee Au B, Parboosingh JS, Lamont RE, Bernier FP, Wright NAM, Benseler SM, Parsons SJ, El-Dairi M, Smith EC, Valdez P, Tennison M, Innes AM, Davis EE. Biallelic variants in ribonuclease inhibitor (RNH1), an inflammasome modulator, are associated with a distinctive subtype of acute, necrotizing encephalopathy. Genet Med. 2023 Sep;25(9):100897. doi: 10.1016/j.gim.2023.100897. Epub 2023 May 13. PMID: 37191094
53. Koop K, Yuan W, Tessadori F, Rodriguez-Polanco WR, Grubbs J, Zhang B, Osmond M, Graham G, Sawyer S, Conboy E, Vetrini F, Treat K, Płoski R, Pienkowski VM, Kłosowska A, Fieg E, Krier J, Mallebranche C, Alban Z, Aldinger KA, Ritter D, Macnamara E, Sullivan B, Herriges J, Alaimo JT, Helbig C, Ellis CA, van Eyk C, Gecz J, Farrugia D, Osei-Owusu I, Adès L, van den Boogaard MJ, Fuchs S, Bakker J, Duran K, Dawson ZD, Lindsey A, Huang H, Baldridge D, Silverman GA, Grant BD, Raizen D; Undiagnosed Diseases Network; van Haaften G, Pak SC, Rehmann H, Schedl T, van Hasselt P. Macrocephaly and developmental delay caused by missense variants in RAB5C. Hum Mol Genet. 2023 Oct 17;32(21):3063-3077. doi: 10.1093/hmg/ddad130. PMID: 37552066
54. Posset R, Garbade SF, Gleich F, Scharre S, Okun JG, Gropman AL, Nagamani SCS, Druck AC, Epp F, Hoffmann GF, Kölker S, Zielonka M; Urea Cycle Disorders Consortium (UCDC); European registry and network for Intoxication type Metabolic Diseases (E-IMD) Consortia Study Group. Severity-adjusted evaluation of liver transplantation on health outcomes in urea cycle disorders. Genet Med. 2023 Dec 3;26(4):101039. doi: 10.1016/j.gim.2023.101039. Online ahead of print. PMID: 38054409
55. Ward SK, Wadley A, Tsai CA, Benke PJ, Emrick L, Fisher K, Houck KM, Dai H; Undiagnosed Diseases Network; Guillen Sacoto MJ, Craigen W, Glaser K, Murdock DR, Rohena L, Diderich KEM, Bruggenwirth HT, Lee B, Bacino C, Burrage LC, Rosenfeld JA. De novo missense variants in ZBTB47 are associated with developmental delays,

hypotonia, seizures, gait abnormalities, and variable movement abnormalities. Am J Med Genet A. 2024 Jan;194(1):17-30. doi: 10.1002/ajmg.a.63399. Epub 2023 Sep 25. PMID: 37743782

56. Paul MS, Michener SL, Pan H, Chan H, Pfliger JM, Rosenfeld JA, Lerma VC, Tran A, Longley MA, Lewis RA, Weisz-Hubshman M, Bekheirnia MR, Bekheirnia N, Massingham L, Zech M, Wagner M, Engels H, Cremer K, Mangold E, Peters S, Trautmann J, Mester JL, Guillen Sacoto MJ, Person R, McDonnell PP, Cohen SR, Lusk L, Cohen ASA, Le Pichon JB, Pastinen T, Zhou D, Engleman K, Racine C, Faivre L, Moutton S, Denommé-Pichon AS, Koh HY, Poduri A, Bolton J, Knopp C, Julia Suh DS, Maier A, Toosi MB, Karimiani EG, Maroofian R, Schaefer GB, Ramakumaran V, Vasudevan P, Prasad C, Osmond M, Schuhmann S, Vasileiou G, Russ-Hall S, Scheffer IE, Carvill GL, Mefford H; Undiagnosed Diseases Network; Bacino CA, Lee BH, Chao HT. A syndromic neurodevelopmental disorder caused by rare variants in PPFIA3. Am J Hum Genet. 2024 Jan 4;111(1):96-118. doi: 10.1016/j.ajhg.2023.12.004. PMID: 38181735
57. Lu J, Toro C, Adams DR; Undiagnosed Diseases Network; Moreno CAM, Lee WP, Leung YY, Harms MB, Vardarajan B, Heinzen EL. LUSTR: a new customizable tool for calling genome-wide germline and somatic short tandem repeat variants. BMC Genomics. 2024 Jan 26;25(1):115. doi: 10.1186/s12864-023-09935-9. PMID: 38279154
58. Jeffries L, Mis EK, McWalter K, Donkervoort S, Brodsky NN, Carpier JM, Ji W, Ionita C, Roy B, Morrow JS, Darbinyan A, Iyer K, Aul RB, Banka S, Chao KR, Cobbold L, Cohen S, Custodio HM, Drummond-Borg M, Elmslie F, Finanger E, Hainline BE, Helbig I, Hewson S, Hu Y, Jackson A, Josifova D, Konstantino M, Leach ME, Mak B, McCormick D, McGee E, Nelson S, Nguyen J, Nugent K, Ortega L, Goodkin HP, Roeder E, Roy S, Sapp K, Saade D, Sisodiya SM, Stals K, Towner S, Wilson W; Deciphering Developmental Disorders; Genomics England Research Consortium; Undiagnosed Disease Network; Khokha MK, Bönnemann CG, Lucas CL, Lakhani SA. Biallelic CRELD1 variants cause a multisystem syndrome, including neurodevelopmental phenotypes, cardiac dysrhythmias, and frequent infections. Genet Med. 2024 Feb;26(2):101023. doi: 10.1016/j.gim.2023.101023. Epub 2023 Nov 7. PMID: 37947183
59. Forghani I, Lang SH, Rodier MJ, Bivona SA; Undiagnosed Diseases Network; Morales AA, Zuchner S, Bademci G, Tekin M. EFEMP1 haploinsufficiency causes a Marfan-like hereditary connective tissue disorder. Am J Med Genet A. 2024 Feb 13:e63556. doi: 10.1002/ajmg.a.63556. Online ahead of print. PMID: 38348595
60. Donkervoort S, Mohassel P, O'Leary M, Bonner DE, Hartley T, Acquaye N, Brull A, Mozaffar T, Saporta MA, Dyment DA, Sampson JB, Pajusalu S, Austin-Tse C, Hurth K, Cohen JS, McWalter K, Warman-Chardon J, Crunk A, Foley AR; Undiagnosed Diseases Network; Mammen AL, Wheeler MT, O'Donnell-Luria A, Bönnemann CG. Recurring homozygous ACTN2 variant (p.Arg506Gly) causes a recessive myopathy. Ann Clin Transl Neurol. 2024 Mar;11(3):629-640. doi: 10.1002/acn3.51983. Epub 2024 Feb 4. PMID: 38311799
61. Scala M, Tomati V, Ferla M, Lena M, Cohen JS, Fatemi A, Brokamp E, Bican A, Phillips JA 3rd, Koziura ME, Nicouleau M, Rio M, Siquier K, Boddaert N, Musante I, Tamburro S, Baldassari S, Iacomino M, Scudieri P; Undiagnosed Diseases Network; Rosenfeld JA, Bellus G, Reed S, Al Saif H, Russo RS, Walsh MB, Cantagrel V, Crunk A, Gustincich S, Ruggiero SM, Fitzgerald MP, Helbig I, Striano P, Severino M, Salpietro V, Pedemonte N, Zara F. De novo variants in DENND5B cause a neurodevelopmental disorder. Am J Hum

Genet. 2024 Mar 7;111(3):529-543. doi: 10.1016/j.ajhg.2024.02.001. Epub 2024 Feb 21. PMID: 38387458

62. Dohrn MF, Bademci G, Rebelo AP, Jeanne M, Borja NA, Beijer D, Danzi MC, Bivona SA, Gueguen P, Zafeer MF; Undiagnosed Diseases Network; Tekin M, Züchner S. Recurrent ATP1A1 variant Gly903Arg causes developmental delay, intellectual disability, and autism. Ann Clin Transl Neurol. 2024 Mar 19. doi: 10.1002/acn3.51963. Online ahead of print. PMID: 38504481
63. Ezell KM, Tinker RJ, Furuta Y, Gulsevin A, Bastarache L, Hamid R, Cogan JD, Rives L, Neumann S, Corner B, Kozuria M, Phillips JA 3rd; Undiagnosed Diseases Network. Undiagnosed Disease Network collaborative approach in diagnosing rare disease in a patient with a mosaic CACNA1D variant. Am J Med Genet A. 2024 Mar 21:e63597. doi: 10.1002/ajmg.a.63597. Online ahead of print.PMID: 38511854
64. Pucel J, Briere LC, Reuter C, Gochyyev P; Undiagnosed Diseases Network; LeBlanc K. Exome and genome sequencing in a heterogeneous population of patients with rare disease: Identifying predictors of a diagnosis. Genet Med. 2024 Mar 1;26(6):101115. doi: 10.1016/j.gim.2024.101115. Epub ahead of print. PMID: 38436216.
65. Sen K, Izem R, Long Y, Jiang J, Konczal LL, McCarter RJ; Members of the Urea Cycle Disorders Consortium (UCDC); Gropman AL, Bedoyan JK. Are asymptomatic carriers of OTC deficiency always asymptomatic? A multicentric retrospective study of risk using the UCDC longitudinal study database. Mol Genet Genomic Med. 2024 Apr;12(4):e2443. doi: 10.1002/mgg3.2443. PMID: 38634223; PMCID: PMC11024633.
66. Kohler JN, Legro NR, Baldridge D, Shin J, Bowman A, Ugur B, Jackstadt MM, Shriver LP, Patti GJ, Zhang B, Feng W, McAdow AR, Goddard P, Ungar RA, Jensen T, Smith KS, Fresard L, Alvarez R, Bonner D, Reuter CM, McCormack C, Kravets E, Marwaha S, Holt JM; Undiagnosed Diseases Network; Worthey E, Ashley EA, Montgomery SB, Fisher P, Postlethwait J, De Camilli P, Solnica-Krezel L, Bernstein JA, Wheeler MT. Loss of function of FAM177A1, a Golgi complex localized protein, causes a novel neurodevelopmental disorder. Genet Med. 2024 May 16:101166. doi: 10.1016/j.gim.2024.101166. Epub ahead of print. PMID: 38767059.
67. Rael VE, Yano JA, Huizar JP, Slayden LC, Weiss MA, Turcotte EA, Terry JM, Zuo W, Thiffault I, Pastinen T, Farrow EG, Jenkins JL, Becker ML, Wong SC, Stevens AM, Otten C, Allenspach EJ, Bonner DE, Bernstein JA, Wheeler MT, Saxton RA; Undiagnosed Diseases Network; Liu B, Majer O, Barton GM. Large-scale mutational analysis identifies UNC93B1 variants that drive TLR-mediated autoimmunity in mice and humans. J Exp Med. 2024 Aug 5;221(8):e20232005. doi: 10.1084/jem.20232005. Epub 2024 May 23. PMID: 38780621; PMCID: PMC11116816.

Letters

1. Berry GT, Freeze HH, Morava E. Is X-linked, infantile onset ALG13-related developmental and epileptic encephalopathy a congenital disorder of glycosylation? Epilepsia. 2021 Feb;62(2):335-336. doi: 10.1111/epi.16817. Epub 2021 Feb 11. PMID: 33576051

Non-peer reviewed scientific or medical publications/materials in print or other media

1. Berry GT. Disorders of Amino Acid Metabolism. In: Pediatric Gastrointestinal Disease. Walker WA, Durie PR, Hamilton JR, Walker-Smith JA and Walkins JB, eds., B.C. Decker, Inc. Philadelphia, PA, pp. 943-957, 1991.

2. Batshaw M and Berry GT. Inborn errors of amino acid and organic acid metabolism. In: Principles of perinatal-neonatal metabolism. Cowett RM, ed., Springer-Verlag, Inc., New York, pp. 426-444, 1991.
3. Thornton P, Berry GT and Stanley CA. Disorders of Intermediary Metabolism: Organ systems failure. In: Pediatric Critical Care. Holbrook P, ed., W.B. Saunders, Orlando, FL, pp. 725-740, 1993.
4. Segal S and Berry GT. Disorders of Galactose Metabolism. In: The Metabolic and Molecular Bases of Inherited Disease (Seventh Edition). Scriver CR, Beaudet AL, Sly WS and Valle D, eds., Vol. I, McGraw-Hill, New York, pp. 967-1000, 1995.
5. Bennett MJ and Berry GT. Use of the Clinical Laboratory for the Evaluation of Metabolic Disease. In: The Pathology of the Developing Human Nervous System. Duckett S, ed., Williams & Wilkins, Baltimore, MD, pp. 823-829, 1995.
6. Berry GT. Methylmalonic acidemia. In: Neurobase (Second Edition). Gilman S, Goldstein GW and Waxman SG, eds., Arbor Publishing, La Jolla, CA, 1995.
7. Berry GT. Disorders of Carbohydrate Metabolism. In: Principles of Child Neurology. Berg BO, ed., McGraw Hill, Inc., New York, pp 969-978, 1996.
8. Berry GT and Segal S. Disorders of Carbohydrate Metabolism. In: Textbook of Internal Medicine (Third Edition). Kelley WN, ed., Lippincott, Philadelphia, PA, pp 2268-2270, 1996.
9. Berry GT. Metabolism. In: Pediatric Secrets (Second Edition). Polin RA, Ditmer MF, eds., Hanley & Belfus Inc., Philadelphia, PA, Mosby, St. Louis, MO, pp 309-326, 1996.
10. Berry GT. Disorders of Amino Acid Metabolism. In: Pediatric Gastrointestinal Disease (Second Edition). Walker WA, Durie PR, Hamilton JR, Walker-Smith JA and Watkins JB, eds., B.C. Decker Inc., Hamilton, Ontario, Canada L8N 3K7 pp1137-1154, 1996.
11. Gibson JB, Piccoli D and Berry GT. Pathophysiology of Metabolic Diseases of the Liver. In: Fetal and Neonatal Physiology (Second Edition). Polin R and Fox W, eds., W.B. Saunders Company, Philadelphia, PA.
12. Berry GT. Inborn errors of amino acid and organic acid metabolism. In: Principles of perinatal-neonatal metabolism (Second Edition). Cowett RM, ed., Springer-Verlag, Inc., New York.
13. Bennett MJ and Berry GT. Use of the Clinical Laboratory for the Evaluation of Metabolic Disease. In: Pediatric Neurologic Pathology (Second Edition). Duckett, S ed., Williams & Wilkins, Baltimore, MD.
14. Berry GT. Methylmalonic acidemia. In: Neurobase (Third Edition). Gilman S., Goldstein GW and Waxman SG, eds., Arbor Publishing, La Jolla, CA, 1999.
15. Berry GT and Cohn RM. Disorders of porphyrin, purine and pyrimidine metabolism. In: Current Pediatric Therapy 16th edition. Burg FD, Ingelfinger JR, Polin RA, Wald ER, eds., W. B. Saunders Company, Philadelphia, PA, pp 797-803, 1998.
16. Berry GT. Introduction to the Metabolic and Biochemical Genetic Diseases, Chapter 24, pp 239-244 and Inborn Errors of Carbohydrate, Ammonia, Amino Acid, and Organic Acid Metabolism, Chapter 25, pp 245-274, Part IV, Genetics and Metabolism. In: Schaeffer and Avery's Diseases of the Newborn (Seventh Edition). Taeusch HW, Ballard RA eds., W.B. Saunders Company, Philadelphia, PA, 1998.
17. Berry GT and Fortina P. Genetic Polymorphisms in the Mitochondrial Genome. In: Genetic Polymorphisms and Susceptibility to Disease. Miller MS, Cronin MT, eds., Taylor & Francis, London, England, pp 207-229, 2000.

18. Berry GT and Yudkoff M. Metabolism. In: Pediatric Secrets (Third Edition). Polin RA, Ditmer MF, eds., Hanley & Belfus Inc., Philadelphia, PA, Mosby, St. Louis, MO, pp 389-407, 2001.
19. Berry GT. Endocrinology and Metabolism. In: Fetal and Neonatal Secrets. Polin RA, Spitzer AR, eds., Hanley & Belfus, Inc., Philadelphia, PA, pp 97-98, 2001.
20. Berry GT. Disorders of Amino Acid Metabolism. In: Pediatric Gastrointestinal Disease (Third Edition). Walker WA, Durie PR, Hamilton JR, Walker-Smith JA and Watkins JB, eds., B.C. Decker Inc., Hamilton, Ontario, Canada L8N 3K7, pp1072 - 1087, 2000.
21. Berry GT. Inborn Errors of Metabolism, "Galactosemia". In: The NORD Guide to Rare Disorders. Lippincott, Williams & Wilkins, Philadelphia, PA, pp 446, 2003.
22. Anadiotis GA and Berry GT. Galactose-1-Phosphate Uridyltransferase Deficiency (Galactosemia). In: eMedicine. Steiner R.D, Konop R, Youssoufian H, Petry PD and Buehlers B, eds., Omaha, Ne 2003.
23. Berry GT. Metabolism. In: Pediatric Secrets (Fourth Edition). Polin R., Ditmer MF, eds., Hanley & Belfus Inc., Philadelphia, PA, Mosby, St. Louis, MO, pp 309-326, 2004.
24. Gibson JB and Berry GT. Pathophysiology of Metabolic Diseases of the Liver. In: Fetal and Neonatal Physiology (Third Edition). Polin R, Fox W, and Abman, eds., W.B. Saunders Company, Philadelphia, PA.
25. Venditti CP and Berry GT. Methylmalonic acidemia. In: Neurobase (Fourth Edition). Gilman S, Goldstein GW and Waxman SG, eds., Arbor Publishing, La Jolla, CA, 2004.
26. Berry GT, Segal S and Gitzelman R. Disorders of Galactose Metabolism. In: Inborn Metabolic Diseases – Diagnosis and Treatment (Fourth Edition). Fernandes J, Saudubray M, van den Berghe G, Walter JH, eds., Springer-Verlag, Inc., New York, NY. 2006.
27. Venditti CP and Berry GT. Inborn Errors of Metabolism for the Gastroenterologist, Chapter 24. In: Clinical Nutrition: A Guide for Gastroenterologists. Buchman A, ed., Slack, Inc. Thorofare, NJ, 2005.
28. Berry GT. Introduction to the Metabolic and Biochemical Genetic Diseases, Chapter 21 and Inborn Errors of Carbohydrate, Ammonia, Amino Acid, and Organic Acid Metabolism, Chapter 22, Part VI, Congenital Metabolic Problems, Ballard RA and Berry GT, section eds. In: Schaeffer and Avery's Diseases of the Newborn (Eighth Edition). Taeusch HW, Ballard RA, Gleason CA, eds., W.B. Saunders Company, Philadelphia, PA, 2005.
29. Venditti CP and Berry GT. Treatment of Acute Metabolic Emergencies in the Newborn Period. In: Current Pediatric Therapy 18th edition. Burg FD, Ingelfinger JR, Polin RA, Gershon AA, eds., W.B. Saunders Company, Philadelphia, PA, 2006.
30. Anadiotis GA and Berry GT. Galactose-1-Phosphate Uridyltransferase Deficiency (Galactosemia). In: eMedicine. Steiner RD, Konop R, Youssoufian H, Petry PD and Buehlers B, eds., Omaha, NE 2006.
31. Venditti CP and Berry GT. Inborn Errors of Metabolism and the Liver. In: Nutrition in Pediatrics, Part 2, Chapter 46, 4th edition. Walker, WA, ed., B.C. Decker, Inc., Hamilton, Ontario, Canada 2007
32. Berry GT. Metabolic Profiling. In: Personalized Nutrition for the Diverse Needs of Infants and Children, Nestlé Nutrition Workshop Series, Pediatric Program, Vol. 62. Bier DM, German JB, Lönnerdal B, eds. Nestec Ltd., S. Karger AG, Basel, Switzerland, 2008.
33. Berry GT. Galactosemia and Amenorrhea in the Adolescent. NYAS, 1135: 112-117, 2008.

34. Berry GT. Galactosemia. In: Neurobase (Fourth Edition). Gilman S, Goldstein GW and Waxman SG, eds., Arbor Publishing, La Jolla, CA, 2009.
35. Berry GT and Walter JH. Disorders of Galactose Metabolism. In: Inborn Metabolic Diseases – Diagnosis and Treatment (Fifth Edition). Saudubray JM, van der Berghe G, Walter JH, eds., Springer-Verlag, Inc., New York, NY, 2016.
36. Cederbaum S and Berry GT. Inborn Errors of Carbohydrate, Ammonia, Amino Acid, and Organic Acid Metabolism, Chapter 22, Part VI, Congenital Metabolic Problems. In: Schaeffer and Avery's Diseases of the Newborn (Ninth Edition). Gleason CA and Devaskar SU, eds., W.B. Saunders Company, Philadelphia, PA, 2012.
37. Thomas JA, Greene CL and Berry GT. Lysosomal Storage, Peroxisomal, and Glycosylation Disorders and Smith-Lemli-Opitz Syndrome Presenting in the Neonate, Chapter 23, Part VI, Congenital Metabolic Problems. In: Schaeffer and Avery's Diseases of the Newborn (Ninth Edition). Gleason CA and Devaskar SU, eds., W.B. Saunders Company, Philadelphia, PA, 2012.
38. Rubio-Gozalbo ME, Panis B and Berry GT. Growth in classical galactosemia. In: The Handbook of Growth and Growth Monitoring in Health and Disease. Preedy VR, ed., Springer-Verlag, Inc., New York, NY, 2012.
39. Anadiotis GA and Berry GT. Galactose-1-Phosphate Uridyltransferase Deficiency (Galactosemia). In: eMedicine. Steiner R.D, Konop R, Youssoufian H, Petry PD and Buehlers B, eds., Omaha, NE 2015.
40. Venditti CP and Berry GT. Methylmalonic acidemia. In: Neurobase (Fourth Edition). Gilman S, Goldstein GW and Waxman SG, eds. Arbor Publishing, La Jolla, CA, 2015.
41. Berry GT. Galactosemia. In: Neurobase (Fourth Edition). Gilman S, Goldstein GW and Waxman SG, eds., Arbor Publishing, La Jolla, CA, 2015.
42. Berry GT. Disorders of Galactose Metabolism. In: Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease (Fifth Edition). Rosenberg RN and Pascual JM, eds., Academic Press, Elsevier, Amsterdam, The Netherlands, 2015.
43. Berry GT, Walter JH and Friedovich-Keil J. Disorders of Galactose Metabolism. In: Inborn Metabolic Diseases – Diagnosis and Treatment (Sixth Edition). Saudubray JM, van der Berghe G, Walter JH, eds., Springer-Verlag, Inc., New York, NY, 2016.
44. Berry GT. Classic Galactosemia and Clinical Variant Galactosemia. 2000 Feb 4 [Updated 2017 March 9]. In: Pagon RA, Adam MP, Bird TD, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1518/
45. Thomas JA, Lam C, Berry GT. Lysosomal Storage, Peroxisomal, and Glycosylation Disorders and Smith-Lemli-Opitz Syndrome Presenting in the Neonate, Chapter 23, Part VI, Congenital Metabolic Problems. In: Schaeffer and Avery's Diseases of the Newborn (Tenth Edition). Gleason CA and Devaskar SU, eds., W.B. Saunders Company, Philadelphia, PA, 2017.
46. Venditti CP and Berry GT. Methylmalonic acidemia. In: Neurobase (Fifth Edition). Gilman S, Goldstein GW and Waxman SG, eds. Arbor Publishing, La Jolla, CA, 2015.
47. Berry GT. Galactosemia. In: Neurobase (Fifth Edition). Gilman S, Goldstein GW and Waxman SG, eds., Arbor Publishing, La Jolla, CA, 2015.
48. Demirbas D, Brucker W, Berry GT. Inborn errors of metabolism with hepatopathy: metabolism defects of galactose, fructose, and tyrosine". In: Pediatric Clinics of North

America: Inborn Errors of Metabolism. El-Hattab A and Sutton VR, eds. 2018 Apr;65(2):337-352. doi: 10.1016/j.pcl.2017.11.008. Review.

49. Demirbas D, Berry GT. Disorders of Galactose Metabolism. In: Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease (6th Edition). Rosenberg RN and Pascual JM, eds., Academic Press, Elsevier, Amsterdam, The Netherlands, 2019, in press.
50. Berry GT. Classic Galactosemia and Clinical Variant Galactosemia. 2000 Feb 4 [Updated 2021 March 9]. In: Pagon RA, Adam MP, Bird TD, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1518/
51. Berry GT, Walter JH and Friedovich-Keil J. Disorders of Galactose Metabolism. In: Inborn Metabolic Diseases – Diagnosis and Treatment (Seventh Edition). Saudubray JM, van der Berghe G, Walter JH, eds., Springer-Verlag, Inc., New York, NY, 2021.
52. Berry GT, Demirbas D, Volpe J. Amino acids. In: Volpe's Neurology of the Newborn (7th edition). Joseph J. Volpe & Terrie E. Inder, eds. Elsevier, 2025.
53. Berry GT, Demirbas D, Volpe J. Organic acids. In: Volpe's Neurology of the Newborn (7th edition). Joseph J. Volpe & Terrie E. Inder, eds. Elsevier, 2025.

Abstracts (those not followed by full publication and only since 2013):

1. Demirbas D, Huang X, Cianci A, Fitzgerald K, DeVine A, Schlaeger T, Sahin M, Daley GQ, Berry GT. An induced pluripotent stem cell model of GALT deficiency recapitulates the galactosemia biochemical phenotype. 12th International Congress of Inborn Errors of Metabolism (ICIEM), Barcelona, Spain. September 3-6, 2013.
2. Berry GT, Newton SA, Bennett MJ. SCHAD deficiency with lethal hepatic phenotype may only be diagnosed through enzyme analysis. 12th International Congress of Inborn Errors of Metabolism (ICIEM), Barcelona, Spain. September 3-6, 2013.
3. Demirbas D, Huang X, Li X, Cianci A, Fitzgerald K, DeVine A, Niederst E, Sahin M, Schlaeger T, He M, Daley GQ, Berry GT. iPS Cell-Based Modeling of Classic Galactosemia, Society for Inherited Metabolic Disorders Annual Meeting, Ponte Vedra, FL, April 4, 2016.
4. Demirbas D, Huang X, Li X, Cianci A, Fitzgerald F, Devine A, Niederst E, Sahin M, Schlaeger T, He M, Daley GQ, Berry GT. "iPSC-based modeling of GALT deficiency to uncover pathophysiology of neurological complications in classic galactosemia" Keystone Symposia: Rare and Undiagnosed Diseases: Discovery and Models of Precision Therapy, Boston, MA, March 5—8, 2017.
5. Demirbas D, Huang X, Feenstra S, Berry GT. "A Mass Spectrometry Based Measurement of GALT Activity May Distinguish Between Classic and Clinical Variant Galactosemia" ACMG Annual Clinical Genetics Meting, Phoenix, AZ, March 2017
6. Demirbas D, Brucker WJ, Levy HL, Berry GT. "Adult galactosemia revisited: A case of a sexagenarian with early autism and late Parkinsonism", Society for Inherited Metabolic Disorders Annual Meeting, San Diego, CA, March 11-14, 2018.
7. Brucker W, Shen L, Almbuquil M, Sacharow S, Berry GT, O'Donnell A. Dysmorphology as a Guide to Uncover Acute Thrombosis Risk: A Case Report of Facial Purpura Fulminans in a patient with PMM2 deficiency (CDG-Ia) associated with

Streptococcus Pneumoniae Sepsis, 40th Annual David W. Smith Workshop on Malformations and Morphogenesis, Snowbird, Utah

Narrative Report:

Research Activities:

The focus of research activities has always been on the mechanism of disease in the biochemical genetic disorder, hereditary galactosemia. Research has been divided between 1.) clinical research involving infants, children and adults with galactosemia and 2.) basic science research on polyol (e.g. galactitol and myo-inositol) metabolism, especially in the brain and during fetal development.

a. Clinical Research:
As an unexplained problem in the management of patients with galactosemia is the chronic elevation of galactose metabolites in blood and urine, despite employment of appropriate diet therapy, I performed studies on hidden sources of galactose in the diet (Berry G.T., et al. The effect of dietary fruits and vegetables on urinary galactitol excretion in galactose-1-phosphate uridyltransferase deficiency. J. Inher. Metab. Dis.16: 91-100, 1993) and documented that there is de novo synthesis of galactose in man (Berry GT, Nissim I, Lin Z, Mazur AT, Gibson JB and Segal S. Endogenous synthesis of galactose in normal man and patients with hereditary galactosemia. Lancet 346: 1073-1074, 1995.). Using stable isotopically-labeled galactose in whole body metabolism studies that were focused on the conversion of [1-^{13}C] galactose to $^{13}CO_2$ and H_2O in vivo, I published genotype- phenotype correlations with prognostic implications. In other in vivo studies, I demonstrated that a non-GALT pathway probably exists in patients, allowing for alternate galactose oxidation. These studies were performed in an infant who has no GALT mRNA and protein, due to a 5.2 kb deletion of almost all of the exons in the GALT gene. My biochemical genetics fellow, Yijun Li, created the first state-of-the-art GALT enzyme assay using stable isotopically –labeled galactose-1-phosphate and LC-MS/MS (Li Y, et al., Quantification of Galactose-1-Phosphate Uridylytransferase Enzyme Activity by Liquid Chromatography-Tandem Mass Spectrometry. Clin. Chem., 56: 772-80, 2010). With Dr. Susan Waisbren, I performed the first prospective comprehensive adult galactosemic CTSU study in Boston in August, 2009 involving 34 subjects. The Boston Globe covered this event. The unprecedented event resulted in several publications including one that received the 2013 SSIEM Garrod Award (Waisbren S., et al. The adult galactosemic phenotype. J Inherit Metab Dis. 35: 279-86, 2012.

b. Basic science research:
I have been studying mammalian myo-inositol and galactitol metabolism since 1979. Both are polyol metabolites whose metabolism may be perturbed in the galactosemic condition. I have studied the kinetics of sodium dependent myo-inositol transport in fetal bovine aortic endothelial cells and human fibroblasts (

Berry GT, et al., Myo-inositol transport and metabolism in fetal-bovine aortic endothelial cells. Biochem. J. 295: 863-869, 1993; Berry GT, et al., The effect of glucose and galactose toxicity on myo-inositol transport and metabolism in human skin fibroblasts in culture. Pediatr. Res. 35:141-147, 1994). The latter was performed under conditions of galactose stress and galactitol accumulation. My laboratory also cloned the human, bovine and murine Na^+/ myo-inositol cotransporter genes. The first murine SLC5A3 or SMIT1 knockout model was published in 2003 (Berry GT, et al., Loss of murine Na+/myo-inositol cotransporter leads to brain myo-inositol depletion and central apnea. J. Biol. Chem. 92:278, 2003). The delineation of this unique model that simulates non-syndromic central congenital hypoventilation is the subject of current studies in my laboratory. The lethal phenotype may be rescued by treating the pregnant carrier female with myo-inositol (Buccafusca, et al., Characterization of the null murine sodium/myo-inositol cotransporter 1 (Smit1 or Slc5a3) phenotype: myo-inositol rescue is independent of expression of its cognate mitochondrial ribosomal protein subunit 6 (Mrps6) gene and of phosphatidylinositol levels in neonatal brain. Mol. Genet. Metab. 95:81-95, 2008). My laboratory assisted in the discovery that the SMIT1 protein may couple with other proteins in the nervous system that play a role in the regulation of membrane potential and myo-inositol flux (Abbott GW, et al., KCNQ1, KCNE1, and NA+-Coupled Solute Transporters Form Reciprocally Regulating Complexes that Affect Neuronal Excitability. Science Signaling. 7, ra22, 2014).

Teaching Activities:

1. Between 1981 and 2001, I taught the genetics fellows, residents and medical students at the Children's Hospital of Philadelphia and the University of Pennsylvania School of Medicine. Between 1996 and 2001, I lectured to the graduate students in the Cell and Molecular Biology graduate courses at the University of Pennsylvania. Between 1998 and 2001, I served as the Preceptor for Medicine 303 course, Human Gene Therapy, at the University of Pennsylvania. Between 2001 and 2003, I trained the NIH genetics fellows, the Children's National Medical Center residents, and the medical students of George Washington University School of Medicine. Between 2003 and 2006, I trained the residents and medical students at the Thomas Jefferson University Hospital and the Jefferson Medical College. From 2005 to 2006, I served as a preceptor for the Introduction to Clinical Medicine (ICM1) course for first year Jefferson medical students. Between 2006 and 2016, I taught the genetic fellows, residents, and medical students at the Boston Children's Hospital. I am the Director of the Biochemical Genetics Training Program at the Harvard Medical School. As such, I lecture many times a year to the residents and fellows in the Harvard Medical School Genetics Training Program (HMSGTP) year-long course and assist in the preparation of the final examination. I, along with several patients and/or their families, provide " clinic session" lectures during the Genetics course for the first year HMS medical students. I provide lectures and, along with several patients and/or their families, provide " clinic session" lectures in the HST 146 Human Biochemistry and Metabolic Diseases course,

and assist with the final examination questions. As of 2014, I am the Block Leader for amino acids in HST 146: Human Biochemistry and Metabolism course.

2. In 2006, I served as the Chair of the Committee to evaluate "Genetics in Medicine" education in the Jefferson Medical School curriculum. As a consequence, a "white paper" was submitted to the Dean of the Medical School. In 2006, I served as a member of the LCME Self Study, Educational Program Committee at the Jefferson Medical College.

Clinical activities:

I am the Director of the Metabolism Program at the Boston Children's Hospital. Clinical responsibilities include the more than 500 different biochemical genetic diseases as well as the newborn screening program for metabolic diseases in New England. Our Program follows a cohort of 400 patients with phenylketonuria (PKU), and in 2006-2018 served as the consultative service for the diagnosis and/or treatment of approximately 1,400 non-PKU patients during the course of each year.

Goals for current academic year:

My objectives include:

1.Continue to improve the infrastructure of the Metabolism Program at Boston Children's Hospital.

2.Expand the biochemical genetics diagnostic laboratory testing program to improve diagnostic capabilities, and patient care and research and development. This will also enhance the educational experience for the fellows enrolled in the HMSGTP Genetics training program.

3. Continue to develop new galactosemia enzyme, molecular and analyte testing at Boston Children's Hospital.
4. Initiate studies at Boston Children's Hospital on *in vivo* galactose metabolism in patients in the CTSU setting.
5. Initiate the first international study on the role of modifier genes and epigenetic effects on outcome of patients with galactosemia: delineation of the phenotype in and collection of genomic DNA from 2000 patients recruited in North America and Europe including detailed characterization of 150 adults with galactosemia at the Boston Children's Hospital over the next ten years; and, creation of a Biorepository at the Manton Center for Orphan Disease Research at Boston Children's Hospital.
6. I am now serving as the Co-Chair for the Metabolomics Working Group of Undiagnosed Diseases Network (UDN) (NIH/NHGRI U01HG007690).