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Position: Medical Director, Parexel International

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EDUCATION

1979 - 1981	Bryn Mawr College, Bryn Mawr, PA		
1982 - 1984	Yale College, New Haven, CT	B.S.	Cum Laude, Molecular Biophysics and Biochemistry
1985 - 1997	University of California, San Francisco	M.D.	
1989 - 1996	University of California, San Francisco	Ph.D.	Biochemistry
1998 - 1999	University of Washington	Intern	Pediatrics
1999 - 2001	University of Washington	Resident	Pediatrics
2001 - 2002	Stanford University	Postdoctoral Fellow	Genetics
2002 - 2004	Stanford University	Fellow	Medical Genetics
2004 - 2005	Stanford University	Fellow	Clinical Biochemical Genetics
2005 - 2006	University of California, San Francisco	Postdoctoral Fellow	Biopharmaceutical Sciences

LICENSES, CERTIFICATION

1998 Medical Licensure, Washington

2001 Medical Licensure, California

2001 Board Certification, Pediatrics (re-certified in 2009 and 2015)

2005 Board Certification, Clinical Genetics and Genomics (re-certified in 2015)

2005 Board Certification, Clinical Biochemical Genetics (re-certified in 2015)

2005 Medical Licensure, Colorado

2014 Medical Licensure, California

PRINCIPAL POSITIONS HELD

2006 - 2013	University of Colorado School of Medicine	Assistant Professor	Pediatrics
2013 - 2014	University of Colorado School of Medicine	Associate Professor	Pediatrics
2015 - 2016	University of California, San Francisco	Associate Professor of Clinical Pediatrics	Pediatrics
2016 - 2023	University of California, San Francisco	Professor of Clinical Pediatrics	Pediatrics
2023 - present	University of California, San Francisco	Professor Emeritus	Pediatrics
2023 - present	Parexel International	Medical Director	

OTHER POSITIONS HELD CONCURRENTLY

2008 - 2014	University of Colorado School of Medicine	Faculty Member, Graduate School
2010 - 2014	University of Colorado School of Medicine	Medical Director, Graduate Program in Genetic Counseling
2011 - 2014	University of Colorado School of Medicine	Member, Human Medical Genetics Program
2015 - present	University of California, San Francisco	Member, Institute of Human Genetics
2015 - 2023	University of California, San Francisco	Medical Director, Metabolism Clinic
2016 - 2023	University of California, San Francisco	Director, Section of Biochemical Genetic Medicine

HONORS AND AWARDS

1981	Truman Scholar from New York State	Harry S. Truman Scholarship Foundation. This is a living memorial to President Harry Truman and is awarded for demonstrated interest in Public Service.
1984	Distinction in the Major, Molecular Biophysics and Biochemistry	Yale College

1984	Henry Fellow	The Charles and Julia Henry Fund and Yale College. This is for one year of post-graduate study at Oxford or Cambridge for one Yale College Senior. This was awarded for study at Cambridge University in History and Philosophy of Science in 1984 to 1985.
2009	Best Doctor	Best Doctors
2009	Junior Faculty Trainee	Urea Cycle Disorders Consortium, Rare Disease Clinical Research Network, funded by the National Institutes of Health. This was awarded to provide support for coursework in Clinical Sciences and Clinical Research.
2010	Visiting Professor	University of New Mexico, Department of Pediatrics
2018	Chan Zuckerberg Biohub Intercampus Research Award Team Investigator	Chan Zuckerberg Biohub. This is a nonprofit medical research organization which has awarded three-year grants to support cutting-edge biomedical research from seven teams of scientists, physicians, and engineers, with each team including faculty members from the University of California, San Francisco; the University of California, Berkeley; and Stanford University. Our project is Machine learning for interpreting rare variation in comprehensive newborn screening and pharmacogenetics.
2020	Paper describing NBSeq research results named one of the Top Ten Papers in Genomic Medicine in 2020	National Human Genetics Research Institute of the NIH

KEYWORDS/AREAS OF INTEREST

Biochemical genetics, small RNAs, urea cycle defects, primary and secondary carnitine deficiency, carnitine transport defect, genotype-phenotype correlations, organic acidemias, pyridoxine-dependent epilepsy, infantile epileptic encephalopathy, liver disease, newborn screening, DNA testing in newborn screening, lysosomal storage disorders, in utero therapy, gene therapy

MEMBERSHIPS

2004 - present Society for Inherited Metabolic Disorders

2008 - present Society for the Study of Inborn Errors of Metabolism

2017 - present American College of Medical Genetics and Genomics

2017 - present American Society of Human Genetics

SERVICE TO PROFESSIONAL ORGANIZATIONS

2008 - 2008	New York and Mid-Atlantic Consortium for Genetic and Newborn Screening Services	Invited participant, Newborn Screening Standardization and Diagnostics Work Group
2011 - 2011	National Urea Cycle Disorders Foundation	Local physician host and organizer of the 2011 Family Foundation Conference
2011 - 2011	Pediatric Academic Societies	Moderator, session on Genetic Basis of Disease, joint meeting with The Asian Society for Pediatric Research
2011 - 2011	American Board of Medical Genetics	Served on Standards Committee for 2011 Medical and Clinical Biochemical Genetics Board Examinations
2011 - 2011	American Board of Medical Genetics	Item writer for 2012 Medical and Clinical Biochemical Genetics Board Examinations
2012 - 2012	American College of Medical Genetics	Co-moderator, session on Signs and Symptoms of Metabolic Disease, joint session with the Society for Inherited Metabolic Disorders
2015 - 2015	American Board of Medical Genetics	Item writer for 2015 Medical and Clinical Biochemical Genetics Board Examinations
2016 - 2016	American Board of Medical Genetics	Item writer for 2016 Medical and Clinical Biochemical Genetics Board Examinations
2016 - 2016	American College of Medical Genetics and Genomics	Member, committee to update and revise ACMGG standards and guidelines document for organic acid analysis

2016 - 2016	Western Society for Pediatric Research	Co-moderator, Genetics Session
2017 - 2017	American Board of Medical Genetics and Genomics	Item writer for 2018 Medical and Clinical Biochemical Genetics Board Examinations
2020 - 2023	Society for Inherited Metabolic Disorders, Wellness Committee	Member
2022 - present	American College of Medical Genetics and Genomics, Workforce Development and Optimization Committee	Member
2022 - 2023	National Organization for Rare Diseases, Rare Disease Center of Excellence	Local Site Director

SERVICE TO PROFESSIONAL PUBLICATIONS

2003 - present Ad hoc referee for American Journal of Human Genetics

2003 - present Ad hoc referee for American Journal of Medical Genetics

2004 - present Ad hoc referee for Molecular Genetics and Metabolism

2005 - present Ad hoc referee for European Journal of Pediatrics

2008 - present Ad hoc referee for Genetics in Medicine

2009 - present Ad hoc referee for Epilepsia

2009 - present Ad hoc referee for Journal of Inherited Metabolic Disease

2015 - present Ad hoc referee for The New England Journal of Medicine

2015 - present Ad hoc referee for Pediatrics

2015 - present Ad hoc referee for Canadian Journal of Neurologic Science

2015 - present Ad hoc referee for Journal of Pediatrics

2015 - present Ad hoc referee for Orphan Drugs: Research and Reviews

2015 - present Ad hoc referee for Pediatric Research

2015 - present Ad hoc referee for Journal of Pediatric Gastroenterology and Nutrition

2015 - present Ad hoc referee for Neurogenetics

2015 - present Ad hoc referee for Neurology

2016 - present Ad hoc referee for Human Mutation

2016 - present Ad hoc referee for Pediatric Transplantation

2016 - present Ad hoc referee for Expert Opinion on Therapeutic Targets

2016 - present Ad hoc referee for Pediatric Neurology

2017 - present Ad hoc referee for JAMA Neurology

2018 - present Ad hoc referee for GeneReviews

2020 - present Ad hoc referee for International Journal of Molecular Sciences

2021 - present Ad hoc referee for Orphanet Journal of Rare Diseases

INTERNATIONAL INVITED PRESENTATIONS

2003	25th Annual David W. Smith Workshop on Malformations and Morphogenesis, Vancouver, British Columbia	Platform
2013	Urea Cycle Disorders Satellite Symposium to the 12th International Congress of Inborn Errors of Metabolism, Barcelona, Spain	Invited Talk

NATIONAL INVITED PRESENTATIONS

1993	Gordon Conference on the Molecular Biology of Ciliated Protozoa, Henniker, NH	Platform
1995	FASEB Meeting on the Molecular Biology of Ciliated Protozoa, Copper Mountain, CO	Platform
2002	American Society of Human Genetics, Baltimore, MD	Platform
2009	Children's Health Improvement through Laboratory Diagnostics, Salt Lake City, UT	Invited Talk
2010	Society for Inherited Metabolic Disorders, Albuquerque, NM	Invited Talk
2011	Urea Cycle Disorders Symposium, Society for Inherited Metabolic Disorders, Asilomar, CA	Invited Talk
2012	American College of Medical Genetics, Charlotte, North Carolina	Invited Talk
2015	Satellite Symposium on Late-onset urea cycle defects, American College of Medical Genetics, Salt Lake City, UT	Speaker and Chair
2018	Satellite Symposium on Non-coding Mutations in Late-onset Metabolic Disease, American College of Medical Genetics, Charlotte, NC	Speaker
2018	Rady Children's Institute, Frontiers in Pediatric Genomic Medicine Conference, Newborn Screening in the NICU and the Nursery: The Critical Importance of DNA Sequence, San Diego	Invited Talk
2019	Society for Inherited Metabolic Disorders, Seattle, WA, Late-breaking abstract session, DNA sequencing in newborn screening	Platform

REGIONAL AND OTHER INVITED PRESENTATIONS

2002	Northern California Clinical Genetics Exchange, Stanford, CA
2003	Western Society for Pediatric Research, Carmel, CA
2004	Western Society for Pediatric Research, Carmel, CA

2004	Conference Regarding Treatment of Late-Treated PKU, San Jose, CA
2007	Grand Rounds, Department of Pediatrics, University of Colorado School of Medicine, Aurora, CO
2010	Grand Rounds, Department of Pediatrics, University of New Mexico, Albuquerque, New Mexico
2010	Visiting Professor, University of New Mexico, Department of Pediatrics, Division of Neonatology, Albuquerque, New Mexico
2011	Grand Rounds, Department of Pediatrics, Division of Medical Genetics, Stanford University, Stanford, CA
2011	National Urea Cycle Disorders Foundation Family Conference, Denver, CO
2012	National Urea Cycle Disorders Foundation Family Conference, Washington, DC
2012	Propionic Acidemia Foundation Annual Meeting, Denver, CO
2014	Grand Rounds, Department of Pediatrics, Section of Clinical Genetics and Metabolism, University of Colorado School of Medicine, Aurora, CO
2015	Grand Rounds, UCSF Benioff Children’s Hospital Oakland, Urea Cycle Defects
2018	Advances in Pediatrics, Newborn Screening for X-linked Adrenoleukodystrophy, Hurler and Pompe, San Francisco
2018	Grand Rounds, UCSF Benioff Children’s Hospital Oakland, Inborn Errors of Metabolism: What to do and When to do it
2019	Northern California Genetics Exchange, UC Davis, Evaluating the Potential of Screening Exome Sequencing for Newborn Screening
2022	Human Genomics Seminar Series, UC Davis, Acid Sphingomyelinase Deficiency: Clinical Characteristics and Treatment

CONTINUING EDUCATION AND PROFESSIONAL DEVELOPMENT ACTIVITIES

2012 Liver Transplantation for Metabolic Disease, Children's Hospital of Pittsburgh, Pittsburgh, PA

RESEARCH AWARDS - PAST

1. U54 HD061221 Junior faculty trainee
   NIH/NICHD 8/1/2009 7/31/2011
   Urea Cycle Disorders Rare Disease Clinical Research Consortium

2. U54 HD061221-11 Local Site PI Batshaw (PI)
   NIH/NCATS Our site in Colorado was funded through philanthropy 09/01/2008 08/31/2019
   Urea Cycle Disorders NIH Rare Disease Clinical Research Consortium

3. RO1HD058567 Local Site PI Tuchman (PI)
   NIH/NICHD 03/15/2012 02/28/2017
   N-Carbamylglutamate in the treatment of hyperammonemia

4. U54 HD061221-11 UCSF Local Site PI
   Philanthropy, National Urea Cycle Disorder Consortium 1/1/2018 12/31/2018
   Urea Cycle Disorders NIH Rare Disease Clinical Research Consortium
   Natural History Study of Urea Cycle Disorders
   Local site PI

5. U19HD077627-04 Consultant Puck (PI)
   NIH 09/01/2017 08/31/2019
   Sequencing of Newborn Blood Spot DNA to Improve and Expand Newborn Screening - Supplement
   Assessing role of DNA sequencing in newborn screening for inborn errors
   Co-Investigator

6. 1U10HG009599-01 Consultant Koenig, Slavotinek, Norton (PI)
   NIH
   P3EGS
   Sequencing in under-represented groups
   Content expert in inborn errors

7. 16-10041 PI Gallagher (PI)
CALIF H&W Dept of Public Health 0701/2017 06/30/2018
Evaluation and follow-up of patient with abnormal newborn screens for inborn errors of metabolism.
See title
PI

8. 301OTC01 PI Gallagher (PI)
Dimension Therapeutics, Inc 06/14/2017 09/30/2021
A Phase 1/2, Open-Label Safety and Dose-Finding Study of Adeno-Associated Virus (AAV) Serotype 8 (AAV8)-Mediated Gene Transfer of Human Ornithine Transcarbamylase (OTC) in Adults with Late-Onset OTC Deficiency
Assess the safety and efficacy and effect of ureagenesis of an AAV8 mediated OTC gene delivered to patient liver cells.
Local Site Principal Investigator

9. UX007-CL202 PI Gallagher (PI)
Ultragenyx Pharmaceutical Inc. 0601/2016 09/30/2021
UX007-CL202: An Open-label Long-Term Safety and Efficacy Extension Study in Subjects with Long-Chain Fatty Acid Oxidation Disorders (LC-FAOD) Previously Enrolled in UX007 or Triheptanoin Studies
This is an extension study of an alternative oil in the dietary treatment of long-chain fat defects to assess effect on heart and muscle
Local site Principal Investigator

10. VTS301 PI Gallagher (PI)
Vtesse, Inc 0525/2016 09/30/2021
VTS301: A Phase 2b/3 Prospective, Randomized, Double-Blind, Sham-Controlled Trial of 2-hydroxypropyl-β-cyclodextrin
This is a clinical trial study of this medication for Neimann-Pick C to assess safety and efficacy in affected patients.
Local site Principal Investigator

11. NA Co-Investigator Brenner (PI)
Chan Zuckerberg Biohub 10/31/2018 10/30/2021
Machine learning for interpreting rare variation in comprehensive newborn screening and pharmacogenetics
Team of experts in computational biology, pharmacogenomics, computer science and ethics with goal to develop algorithms for interpreting variants relevant to newborn screening and pharmacogenomics
Metabolic disease content expert

12. 1 R21 HG011805-01 PI Gallagher (PI)
NIH-NHGRI 07/01/2021 06/30/2023
Toward DNA Sequencing as a Primary Newborn Screen for Treatable Disorders not Amenable to Current Screening
We propose to improve DNA sequencing as a newborn screening test by increasing the sensitivity and specificity to an acceptable level for those early-onset treatable disorders for which there is no alternative screening test. Given the large number of early-onset treatable conditions for which there is no test, or for which current testing continues to miss cases, DNA sequencing has the potential to have a large public health impact. Current newborn screening for multiple inborn errors of metabolism by tandem mass spectrometry is able to detect 40-50 disorders and is considered to be among the ten greatest public health successes of the first decade of the 21st century by the Centers for Disease Control; DNA based newborn screening could greatly increase the number of disorders identified and lead to timely and disease-specific interventions, thereby further decreasing death and disability due to treatable, early-onset recessive disorders.
Metabolic disease content expert, liaison with newborn screening program, team leader, contribute to design of study and analysis and interpretation of data.

13. 21-10157 PI Gallagher (PI)
California H and W Dept of Public Health Genetic Disease Screening Program 07/01/2021 06/30/2024
Evaluation and follow-up of patient with abnormal newborn screens for inborn errors of metabolism
Evaluation and follow-up of patient with abnormal newborn screens for inborn errors of metabolism
I direct the Metabolic Special Care Center and supervise the evaluation and management of patients referred for abnormal newborn screens

PEER REVIEWED PUBLICATIONS

1. Dror Y, **Gallagher R**, Wara D, Colombe BW, Merino A, Benkerrou M, Cowan MJ. Immune reconstitution in severe combined immunodeficiency disease after lectin-treated, T-cell-depleted haplocompatible bone marrow transplantation. Blood. 1993 Apr 15, 81(8):2021-30
2. Lee MS, **Gallagher RC**, Bradley J, Blackburn EH. In vivo and in vitro studies of telomeres and telomerase. Cold Spring Harbor Symposium on Quantitative Biology. 1993;58:707-18

3. Pan WJ, **Gallagher RC**, Blackburn EH. Replication of an rRNA gene origin plasmid in the Tetrahymena thermophila macronucleus is prevented by transcription through the origin from an RNA polymerase I promoter. Molecular and Cellular Biology. 1995 Jun;15(6):3372-81
4. **Gallagher RC**, Blackburn EH. A promoter region mutation affecting replication of the Tetrahymena ribosomal DNA minichromosome. Molecular and Cellular Biology. 1998 May;18(5):3021-33
5. **Gallagher RC**, Pils B, Albalwi M, Francke U. Evidence for the role of PWCR1/HBII-85 C/D box small nucleolar RNAs in Prader-Willi syndrome. American Journal of Human Genetics. 2002 Sep;71(3):669-78. Epub 2002 Jul 31
6. Toriello HV, Carey JC, Addor MC, Allen W, Burke L, Chun N, Dobyns W, Elias E, **Gallagher R**, Hordijk R, Hoyme G, Irons M, Jewett T, LeMerrer M, Lubinsky M, Martin R, McDonald-McGinn D, Neumann L, Newman W, Pauli R, Seaver L, Tsai A, Wargowsky D, Williams M, Zackai E. Toriello-Carey syndrome: delineation and review. American Journal of Medical Genetics. 2003 November 15; 123A(1):84-90
7. Zweier C, Thiel CT, Dufke A, Crow YJ, Meinecke P, Suri M, Ala-Mello S, Beemer F, Bernasconi S, Bianchi P, Bier A, Devriendt K, Dimitrov B, Firth H, **Gallagher RC**, Garavelli L, Gillessen-Kaesbach G, Hudgins L, Kääriäinen H, Karstens S, Krantz I, Mannhardt A, Medne L, Mücke J, Kibaek M, Krogh LN, Peippo M, Rittinger O, Schulz S, Schelley SL, Temple IK, Dennis NR, Van der Knaap MS, Wheeler P, Yerushalmi B, Zenker M, Seidel H, Lachmeijer A, Prescott T, Kraus C, Lowry RB, Rauch A. Clinical and mutational spectrum of Mowat-Wilson syndrome. European Journal of Medical Genetics. 2005 Apr-Jun;48(2):97-111. Epub 2005 Feb 25
8. **Gallagher RC**, Cowan TM, Goodman SI, Enns GM. Glutaryl-CoA dehydrogenase deficiency and newborn screening: retrospective analysis of a low excretor provides further evidence that some cases may be missed. Molecular Genetics and Metabolism. 2005 Nov;86(3):417-20. Epub 2005 Sep 23
9. Urban TJ, **Gallagher RC**, Brown C, Castro RA, Lagpacan LL, Brett CM, Taylor TR, Carlson EJ, Ferrin TE, Burchard EG, Packman S, Giacomini KM. Functional genetic diversity in the high-affinity carnitine transporter OCTN2 (SLC22A5). Molecular Pharmacology. 2006 Nov;70(5):1602-11. Epub 2006 Aug 24
10. Adam MP, Schelley S, **Gallagher R**, Brady AN, Barr K, Blumberg B, Shieh JT, Graham J, Slavotinek A, Martin M, Keppler-Noreuil K, Storm AL, Hudgins L. Clinical features and management issues in Mowat-Wilson syndrome. American Journal of Medical Genetics. 2006 December 15; 140(24):2730-2741
11. Longo N, Schrijver I, Vogel H, Pique LM, Cowan TM, Pasquali M, Steinberg GK, Hedlund GL, Ernst SL, **Gallagher RC**, Enns GM. Progressive cerebral vascular degeneration with mitochondrial encephalopathy. American Journal of Medical Genetics. 2008 Feb 1;146(3):361-7
12. Shchelochkov OA, Li FY, Geraghty MT, **Gallagher RC**, Van Hove JL, Lichter-Konecki U, Fernhoff PM, Copeland S, Reimschisel T, Cederbaum S, Lee B, Chinault AC, Wong LJ. High-frequency detection of deletions and variable rearrangements at the ornithine transcarbamylase (OTC) locus by oligonucleotide array CGH. Molecular Genetics and Metabolism. 2009 Mar;96(3):97-105. Epub 2009 Jan 12
13. **Gallagher RC**, Van Hove JL, Scharer G, Hyland K, Plecko B, Waters PJ, Mercimek-Mahmutoglu S, Stockler-Ipsiroglu S, Salomons GS, Rosenberg EH, Struys EA, Jakobs C.

Folinic acid-responsive seizures are identical to pyridoxine-dependent epilepsy. Annals of Neurology. 2009 May;65(5):550-6. 2009 Jan 13 (E-pub)

14. Rosenberg EH, Struys EA, Hyland K, Plecko B, Waters PJ, Mercimek-Mahmutoglu S, Stockler-Ipsiroglu S, **Gallagher RC**, Scharer G, Van Hove JL, Jakobs C, Salomons GS. Mutation detection in DNA isolated from cerebrospinal fluid and urine: clinical utility and pitfalls of multiple displacement amplification Molecular Genetics and Metabolism. 2009 Aug;97(4):312-4. Epub 2009 May 13
15. Seminara J, Tuchman M, Krivitzky L, Krischer J, Lee HS, LeMons C, Baumgartner M, Cederbaum S, Diaz GA, Feigenbaum A, **Gallagher RC**, Harding CO, Kerr DS, Lanpher B, Lee B, Lichter-Konecki U, McCandless SE, Merritt JL, Oster-Granite ML, Seashore MR, Stricker T, Summar M, Waisbren S, Yudkoff M, Batshaw ML. Establishing a consortium for the study of rare diseases: The Urea Cycle Disorders Consortium. Molecular Genetics and Metabolism. 2010;100 Suppl 1:S97-105. Epub 2010 Feb 10
16. Van Hove JL, Saenz MS, Thomas JA, **Gallagher RC**, Lovell MA, Fenton LZ, Shanske S, Myers SM, Wanders RJ, Ruiter J, Turkenburg M, Waterham HR. Succinyl-CoA ligase deficiency: a mitochondrial hepatoencephalomyopathy. Pediatric Research. 2010 Aug;68(2):159-64
17. Quadros EV, Lai SC, Nakayama Y, Sequeira JM, Hannibal L, Wang S, Jacobsen DW, Fedosov S, Wright E, **Gallagher RC**, Anastasio N, Watkins D, Rosenblatt DS. Positive newborn screen for methylmalonic aciduria identifies the first mutation in TCblR/CD320, the gene for cellular uptake of transcobalamin-bound vitamin B(12). Human Mutation. 2010 Aug;31(8):924-9
18. Scharer G, Brocker C, Vasiliou VK, Creadon-Swindell G, **Gallagher RC**, Spector E, Van Hove JLK. The genotypic and phenotypic spectrum of pyridoxine-dependent epilepsy due to mutations in ALDH7A1. Journal of Inherited Metabolic Disease. 2010 Oct;33(5):571-81
19. Kronn D, Mofidi S, Braverman N, Harris K; Diagnostics Guidelines Work Group. Diagnostic guidelines for newborns who screen positive in newborn screening. Genetics in Medicine. 2010 Dec;12(12 Suppl):S251-5. PubMed PMID: 21150371. (I am one of 33 listed collaborators.)
20. Wright EL, Van Hove JL, Thomas J; Mountain States Metabolic Consortium. Mountain states genetics regional collaborative center's metabolic newborn screening long-term follow-up study: a collaborative multi-site approach to newborn screening outcomes research. Genetics in Medicine. 2010 Dec;12(12 Suppl):S228-41. PubMed PMID: 21150369. (I am one of 16 listed collaborators.)
21. Bireley WR, Van Hove JL, **Gallagher RC**, Batshaw ML; Hepatocellular carcinoma in a gene therapy research subject with ornithine transcarbamylase deficiency. Molecular Genetics and Metabolism. 2012 Feb;105(2):263-5
22. Wilson JM, Shchelochkov OA, **Gallagher RC**, Batshaw ML; Hepatocellular carcinoma in a gene therapy research subject with ornithine transcarbamylase deficiency. Molecular Genetics and Metabolism. 2012 Feb;105(2):263-5
23. Mokhtarani M, Diaz GA, Rhead W, Lichter-Konecki U, Bartley J, Feigenbaum A, Longo N, Berquist W, Berry SA, **Gallagher R**, Bartholomew D, Harding CO, Korson MS, McCandless SE, Smith W, Vockley J, Bart S, Kronn D, Zori R, Cederbaum S, Dorrani N, Merritt JL 2nd, Sreenath-Nagamani S, Summar M, Lemons C, Dickinson K, Coakley DF, Moors TL, Lee B, Scharschmidt BF.Urinary Phenylacetylglutamine as Dosing Biomarker

for Patients with Urea Cycle Disorders. Molecular Genetics and Metabolism. 2012 Nov;107(3):308-14

24. Nagamani SC, Shchelochkov OA, Mullins MA, Carter S, Lanpher BC, Sun Q, Kleppe S, Erez A, O'Brian Smith E, Marini JC; Members of the Urea Cycle Disorders Consortium, Lee B. A randomized controlled trial to evaluate the effects of high-dose versus low-dose of arginine therapy on hepatic function tests in argininosuccinic aciduria. Mol Genet Metab. 2012 Nov;107(3):315-21 (I am one of 20 listed collaborators.)
25. Ah Mew N, Krivitzky L, McCarter R, Batshaw M, Tuchman M; Urea Cycle Disorders Consortium of the Rare Diseases Clinical Research Network. Clinical outcomes of neonatal onset proximal versus distal urea cycle disorders do not differ. J Pediatr. 2013 Feb;162(2):324-9 (I am one of 22 listed collaborators.)
26. Diaz GA, Krivitzky LS, Mokhtarani M, Rhead W, Bartley J, Feigenbaum A, Longo N, Berquist W, Berry SA, **Gallagher R**, Lichter-Konecki U, Bartholomew D, Harding CO, Cederbaum S, McCandless SE, Smith W, Vockley G, Bart SA, Korson MS, Kronn D, Zori R, Merritt JL 2nd, C S Nagamani S, Mauney J, Lemons C, Dickinson K, Moors TL, Coakley DF, Scharschmidt BF, Lee B. Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. Hepatology, 2013 Jun;57(6):2171-9
27. Mokhtarani M, Diaz GA, Rhead W, Berry SA, Lichter-Konecki U, Feigenbaum A, Schulze A, Longo N, Bartley J, Berquist W, **Gallagher R**, Smith W, McCandless SE, Harding C, Rockey DC, Vierling JM, Mantry P, Ghabril M, Brown RS Jr, Dickinson K, Moors T, Norris C, Coakley D, Milikien DA, Nagamani SC, Lemons C, Lee B, Scharschmidt BF. Elevated phenylacetic acid levels do not correlate with adverse events in patients with urea cycle disorders or hepatic encephalopathy and can bepredicted based onthe plasma PAA to PAGN ratio. Molecular Genetics and Metabolism. 2013 Dec;110(4):446-53
28. **Gallagher RC**, Lam C, Wong D, Cederbaum S, Sokol RJ. Significant Hepatic Involvement in Ornithine Transcarbamylase Deficiency. Journal of Pediatrics, 2014 Apr;164(4):720-725
29. van Karnebeek CD, Stockler-Ipsiroglu S, Jaggumantri S, Assmann B, Baxter P, Buhas D, Bok LA, Cheng B, Coughlin CR 2nd, Das AM, Giezen A, Al-Hertani W, Ho G, Meyer U, Mills P, Plecko B, Struys E, Ueda K, Albersen M, Verhoeven N, Gospe SM Jr, **Gallagher RC**, Van Hove JK, Hartmann H. Lysine-restricted diet as adjunct therapy for pyridoxine-dependent epilepsy: The PDE consortium consensus recommendations. Journal of Inherited Metabolic Disease Reports, Apr 2014 [Epub ahead of print].
30. Lee B, Diaz GA, Rhead W, Lichter-Konecki U, Feigenbaum A, Berry SA, Le Mons C, Bartley J, Longo N, Nagamani SC, Berquist W, **Gallagher RC**, Bartholomew D, Harding CO, Korson MS, McCandless SE, Smith W, Cederbaum S, Wong D, Merritt JL II, Schulze A, Vockley J, Kronn D, Zori R, Summar M, Milikien DA, Marino M, Coakley DF, Mokhtarani M, Members Of The UCDC, Scharschmidt BF. Blood ammonia and glutamine as predictors of hyperammonemic crises in urea cycle disorder patients. Genetics in Medicine, Accepted, August 2014
31. Burrage LC, Jain M, Gandolfo L, Lee BH; Members of the Urea Cycle Disorders Consortium, Nagamani SC Sodium phenylbutyrate decreases plasma branched-chain amino acids in patients with urea cycle disorders. Mol Genet Metab. 2014 Sep-Oct;113(1-2):131-5. doi: 10.1016/j.ymgme.2014.06.005. Epub 2014 Jul 3

32. Batshaw ML, Tuchman M, Summar M, Seminara J; Members of the Urea Cycle Disorders Consortium. A longitudinal study of urea cycle disorders. Mol Genet Metab. 2014 Sep-Oct;113(1-2):127-30. doi: 10.1016/j.ymgme.2014.08.001. Epub 2014 Aug 10.

33. Chatfield KC, Coughlin CR, Friederich MW, **Gallagher RC**, Hesselberth JR, Lovell MA, Ofman R, Swanson MA, Thomas JA, Wanders RJ, Wartchow EP, Van Hove JL. Mitochondrial energy failure in HSD10 disease is due to defective mtDNA transcript processing. Mitochondrion. 2015 Mar; 21:1-10. PMID: 25575635.

34. Coughlin CR, van Karnebeek CD, Al-Hertani W, Shuen AY, Jaggumantri S, Jack RM, Gaughan S, Burns C, Mirsky DM, **Gallagher RC**, Van Hove JL. Triple therapy with pyridoxine, arginine supplementation and dietary lysine restriction in pyridoxine-dependent epilepsy: Neurodevelopmental outcome. Mol Genet Metab. 2015 Sep-Oct; 116(1-2):35-43. PMID: 26026794

35. Ferdinandusse S, Friederich MW, Burlina A, Ruiter JP, Coughlin CR, Dishop MK, **Gallagher RC**, Bedoyan JK, Vaz FM, Waterham HR, Gowan K, Chatfield K, Bloom K, Bennett MJ, Elpeleg O, Van Hove JL, Wanders RJ. Clinical and biochemical characterization of four patients with mutations in ECHS1. Orphanet J Rare Dis. 2015; 10(1):79. PMID: 26081110.

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ADDITIONAL RELEVANT INFORMATION

2006-2014 I was a local site principal investigator at the University of Colorado for industry sponsored studies that lead to the FDA approval of a novel therapy for urea cycle disorders, glycerol phenylbutyrate. These trials were: HPN-006 A Phase 3, Randomized, Double-Blind, Cross-Over, Active-Controlled Study of the Efficacy and Safety of HPN-100, Glyceryl tri-(4-phenylbutyrate), for the Treatment of Adults with Urea Cycle Disorders; HPN-007 A Phase 2, Open-Label Study of the Safety of HPN-100 for the Long-Term Treatment of Urea Cycle Disorders; and HPN-011 Long Term Use of HPN-100 in Urea Cycle Disorders, Sponsor, Hyperion Therapeutics Inc. I was a co-investigator at the University of Colorado for multiple industry trials including: A Phase 2, Open-label Dose-Finding Study to Evaluate the Safety, Efficacy and Tolerability of Multiple Subcutaneous Doses of rAvPAL-PEG In subjects with PKU (PAL-002); Long-term Extension of a Phase 2, Open-label Dose-Finding Study to Evaluate the Safety, Efficacy and Tolerability of Multiple Subcutaneous Doses of rAvPAL-PEG in subjects with PKU (PAL-003); A Phase 2, Open-Label Study to Evaluate the Safety, Tolerability, and Efficacy of Subcutaneous Dose Levels of rAvPAL-PEG Administered Daily in Subjects with Phenylketonuria Protocol PAL-004; A Phase 2, multi-center, Open-Label, Dose-Finding Study to Evaluate Safety, Efficacy, and Tolerability of Subcutaneously administered rAvPAL-PEG in Patients with PKU for 24 Weeks Protocol 165-205; A Three-Part, Phase 3, Randomized, Double-Blind, Placebo-Controlled, Four-Arm, Discontinuation Study to Evaluate the Efficacy and Safety of Subcutaneous Injections of BMN 165 Self Administered by Adults With Phenylketonuria; PKU-DOS, PKU Demographics, Outcomes and Safety Registry; A double-blind, placebo-controlled, randomized study to evaluate the safety and therapeutic effects of sapropterin dihydrochloride on neuropsychiatric symptoms in subjects with phenylketonuria, PKU-016; A Phase 3, Open-Label, Randomized, Multi-Center Study to Assess the Safety and Tolerability of an Induction, Titration, and Maintenance Dose Regimen of BMN 165 Self Administered by Adults with Phenylketonuria Not Previously Treated with BMN 165; A Multicenter, Open-label BMN 110 US Expanded Access Program to Provide BMN 110 to Patients Diagnosed with MPS IVA (110-503), Sponsor BioMarin Pharmaceutical Inc. A Phase 4, Open-Label, Prospective Study in Patients with Pompe Disease to Evaluate the Efficacy and Safety of Alglucosidase Alfa at the 4000L Scale, Sponsor, Genzyme