Urea Cycle Disorders Consortium (UCDC) Publications

*Indicates that the publication results from a trainee project

Publications Resulting from 5101:

Ah Mew N, Krivitzky L, McCarter R, Batshaw M, Tuchman M. Clinical outcomes of neonatal onset proximal versus distal urea cycle disorders do not differ. J Pediatr. 2013 Feb;162(2):324-329.e321. PMID: 22901741, PMCID: PMC4440324

Batshaw ML, Tuchman M, Summar ML, Seminara J. A longitudinal study of urea cycle disorders. Mol Genet Metab. 2014 Sep-Oct;113(1-2):127-130. PMID: 25135652, PMCID: PMC4178008

Burrage LC, Jain M, Gandolfo L, Lee BH, Nagamani SCS. Sodium phenylbutyrate decreases plasma branched-chain amino acids in patients with urea cycle disorders. Mol Genet Metab. 2014 Sep-Oct;113(0):131–135. PMID: 25042691, PMCID: PMC4177960

Burrage LC, Sun Q, Elsea SH, Jiang M, Nagamani SCS, Frankel AE, Stone E, Alters SE, Johnson DE, Rowlinson SW, Georgiou G, Members of the Urea Cycle Disorders Consortium, Lee BH. Human recombinant arginase enzyme reduces plasma arginine in mouse models of arginase deficiency. Hum Mol Genet. 2015;24(22):6417-6427. PMID: 26358771, PMCID: PMC5007608

Gallagher RC, Lam C, Wong D, Cederbaum S, Sokol RJ. Significant hepatic involvement in patients with ornithine transcarbamylase deficiency. J Pediatr. 2014 Apr;164(4):720-725.e726. PMID: 24485820, PMCID: PMC4070427

Jain-Ghai S, Nagamani SCS, Blaser S, Siriwardena K, Feigenbaum A. Arginase I Deficiency: Severe infantile presentation with hyperammonemia: more common than reported? Molecular Genetics and Metabolism. 2011;104:107–111. PMID: 21802329. PMCID: PMC3171515

Kolker S, Dobbelaere D, Haberle J, Burgard P, Gleich F, Summar ML, Hannigan S, Parker S, Chakrapani A, Baumgartner MR, and on behalf of the E-IMD Consortium. Networking across borders for individuals with organic acidurias and urea cycle disorders: The E-IMD Consortium. JIMD Reports. 2015;22:29-38. PMID: 25701269, PMCID: PMC4486274

Krivitzky LS, Babikian T, Lee HS, Thomas NH, Burke-Paull KL, Batshaw ML. Intellectual, adaptive, and behavioral functioning in children with urea cycle disorders. Pediatric Research. 2009 Jul;66(1):96-101. PMID: 19287347, PMCID: PMC2746951

Krivitzky LS, Walsh KS, Fisher EL, Berl MM. Executive functioning profiles from the BRIEF across pediatric medical disorders: Age and diagnosis factors. Child Neuropsychol. 2016;22(7):870–888. PMID: 26143938, PMCID: PMC4703575

Mc Guire PJ, Lee HS, Members of the UCDC, Summar ML. Infectious precipitants of acute hyperammonemia are associated with indicators of increased morbidity in patients with Urea Cycle Disorders. J Pediatr. 2013 Dec;163(6):1705–1710.e1. PMID: 24084106, PMCID: PMC3958925

Morgan TM, Schlegel C, Edwards KM, Welch-Burke T, Zhu Y, Sparks R, Summar ML, the Urea Cycle Disorders Consortium. Vaccines are not associated with metabolic events in children with urea cycle disorders. Pediatrics. 2011 May;127(5): e1147–e1153; PIMD: 21482610, PMCID: PMC3387867

Patrick TB, Richesson R, Andrews JE, Folk LC. SNOMED CT coding variation and grouping for “other findings” in a longitudinal study on urea cycle disorders. AMIA Annu Symp Proc. 2008 Nov;6:11-5. PIMD: 18998949, PMCID: PMC2656069

Posset R, Garbade SF, Boy N, Burlina AB, Dionisi-Vici C, Dobbelaere D, Garcia-Cazorla A, de Lonlay P, Teles EL, Vara R, Ah Mew N, Batshaw ML, Baumgartner MR, McCandless SE, Seminara J, Summar ML, Hoffmann

GF, Kölker S, Burgard P; on behalf of the UCDC and the E-IMD consortium. Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disorders – a successful strategy for clinical research of rare diseases. J Inherit Metab Dis. 2018 Jul 4. PMID: 29974348

Richesson RL, Lee HS, Cuthbertson D, Lloyd J, Young K, Krischer JP. An automated communication system in a contact registry for persons with rare diseases: Scalable tools for identifying and recruiting clinical research participants. Contemp Clin Trials. 2009 Jan;30(1):55-62. PIMD: 18804556, PMCID: PMC2640948

Seminara J, Tuchman M, Krivitzky L, Krischer J, Lee HS, Le Mons 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. Mol Genet Metab. 2010;100 Suppl 1:S97-S105. PMID: 20188616, PMCID: PMC2858794

Shapiro E, Bernstein J, Adams HR, Barbier AJ, Buracchio T, Como P, Delaney KA, Eichler F, Goldsmith JC, Hogan M, Kovacs S, Mink JW, Odenkirchen J, Parisi MA, Skrinar A, Waisbren SE, Mulberg AE. Neurocognitive clinical outcome assessments for inborn errors of metabolism and other rare conditions. Mol Genet Metab. 2016 Jun;118(2):65-69. PMID: 27132782, PMCID: PMC4895194

Summar ML, Koelker S, Freedenberg D, Le Mons C, Haberle J, Lee HS, Kirmse B, the European Registry and Network for Intoxication Type Metabolic Diseases (E-IMD), Members of the Urea Cycle Disorders Consortium (UCDC). The incidence of urea cycle disorders. Mol Genet Metab. 2013 Sep-Oct;110(1-2):179-180. PMID: 23972786, PMCID: PMC4364413

Tuchman M, Lee B, Lichter-Konecki U, Summar ML, Yudkoff M, Cederbaum SD, Kerr DS, Diaz GA, Seashore MR, Lee HS, Krischer JP, Batshaw ML, the Urea Cycle Disorders Consortium of the Rare Diseases Clinical Research Network. Cross-sectional multi-center study of patients with urea cycle disorders in the United States. Mol Genet Metab. 2008;94:397-402. PMID:18562231, PMCID: PMC2640937

Waisbren SE, Cuthbertson D, Burgard P, Holbert A, McCarter R, Cederbaum S, Members of the Urea Cycle Disorders Consortium. Biochemical markers and neuropsychological functioning in distal urea cycle disorders. J Inherit Metab Dis. 2018 Jul;41(4):657-667. PMID: 29423830, PMCID: PMC6041144

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Waisbren SE, He J, McCarter R. Assessing psychological functioning in metabolic disorders:Validation of the adaptive behavior assessment system, second edition (ABAS-ii), and the behavior rating inventory of executive function (BRIEF) for identification of individuals at risk. JIMD Reports. 2015;21:35–43. PMID: 25712381, PMCID: PMC4470946

Wilson JM, Shchelochkov OA, Gallagher RC, Batshaw ML. Hepatocellular carcinoma in a research subject with ornithine transcarbamylase deficiency. Mol Genet Metab. 2012 Feb;105(2):263-265. PMID: 22129577, PMCID: PMC3273986

Publications Resulting from 5102:

Nagamani SCS, 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 Liver Function Tests in Argininosuccinic Aciduria**.** Mol Genet Metab. 2012 Nov;107(3):315-21. Epub 2012 Sep 17. PMID: 23040521. PMCID: PMC3483446

Publications Resulting from 5104 and 5107:

*Gropman AL. Brain imaging in urea cycle disorders. Mol Genet Metab. 2010;100 Suppl 1:S20-30. PMID: 20207564, PMCID: PMC3258295

Gropman AL. Expanding the diagnostic and research toolbox for inborn errors of metabolism: the role of magnetic resonance spectroscopy. Mol Genet Metab. 2005 Sep-Oct;86(1-2):2-9. PMID: 16276565

Gropman AL. Patterns of brain injury in inborn errors of metabolism. Semin Pediatr Neurol. 2012 Dec;19(4):203-210. PMID: 23245553, PMCID: PMC3758694

Gropman AL, Batshaw ML. Cognitive outcome in urea cycle disorders. Mol Genet Metab. 2004 Apr;81 Suppl 1:S58-62. PMID: 15050975, Full Text (with PubMed access)

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Gropman AL, Gertz B, Shattuck K, Kahn IL, Seltzer R, Krivitsky L, VanMeter J. Diffusion tensor imaging detects areas of abnormal white matter microstructure in patients with partial ornithine transcarbamylase deficiency. AJNR Am J Neuroradiol. 2010 Oct;31(9):1719-1723. PMID: 20488904, PMCID: PMC3758695

Gropman AL, Prust M, Breeden A, Fricke S, VanMeter J. Urea cycle defects and hyperammonemia: effects on functional imaging. Metab Brain Dis. 2013 Jun;28(2):269-275. PMID: 23149878, PMCID: PMC3594356

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Gropman AL, Shattuck K, Prust MJ, Seltzer RR, Breeden AL, Hailu A, Rigas A, Hussain R,VanMeter J. Altered neural activation in ornithine transcarbamylase deficiency during executive cognition: an fMRI study. Hum Brain Mapp. 2013 Apr;34(4):753-761. PMID: 22110002, PMCID: PMC3338900

*Gropman AL, Summar ML, Leonard JV. Neurological implications of urea cycle disorders. J Inherit Metab Dis. 2007 Nov;30(6):865-879. PMID: 18038189, PMCID: PMC3758693

Oldham MS, VanMeter JW, Shattuck KF, Cederbaum SD, Gropman AL. Diffusion tensor imaging in arginase deficiency reveals damage to corticospinal tracts. Pediatr Neurol. 2010 Jan;42(1):49-52. PMID: 20004862, PMCID: PMC3758690

Pacheco-Colón Ileana, Fricke Stanley and Gropman Andrea. “Role of brain imaging for demonstrating ammonia-induced changes”. In Current Approach to Hyperammonemia. Johannes Häberle (ed). eBook, Future Medicine. 2014.

Pacheco-Colón I, Washington SD, Sprouse C, Helman G, Gropman AL, VanMeter JW. Reduced functional connectivity of default mode and set-maintenance networks in ornithine transcarbamylase deficiency. PLoS ONE. 2015;10(6):e0129595. PMID: 26067829, PMCID: PMC4466251

Prust MJ, Gropman AL, Hauser N. New frontiers in neuroimaging applications to inborn errors of metabolism. Mol Genet Metab. 2011 Nov;104(3):195-205. PMID: 21778100, PMCID: PMC3758691

Sprouse C, King J, Helman G, Pacheco-Colón I, Shattuck K, Breeden A, Seltzer R, VanMeter JW, Gropman AL. Investigating neurological deficits in carriers and affected patients with ornithine transcarbamylase deficiency. Mol Genet Metab. 2014 Sep-Oct;113(1-2):136-141. PMID: 24881970, PMCID: PMC4458385

Publications Resulting from 5105:

Ah Mew N, McCarter R, Daikhin Y, Lichter-Konecki U, Nissim I, Yudkoff M, Tuchman M. Augmenting ureagenesis in patients with partial carbamyl phosphate synthetase 1 deficiency with N-carbamyl-L-glutamate. J Pediatr. 2014 Aug;165(2):401-403.e3. PMID: 24880889, PMCID: PMC4111993.

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Ah Mew N, Yudkoff M, Tuchman M. Stable isotopes in the diagnosis and treatment of inherited hyperammonemia. J Pediatr Biochem. 2014 Jan 1;4(1):57-63. PMID: 24634704, PMCID: PMC3952002

Caldovic L, Morizono H, Daikhin Y, Nissim I, McCarter RJ, Yudkoff M, Tuchman M. Restoration of ureagenesis in N-acetylglutamate synthase deficiency by N-carbamylglutamate. J Pediatr. 2004 Oct;145(4):552-554. PMID: 15480384, Full Text (with PubMed access)

Heibel SK, Ah Mew N, Caldovic L, Daikhin Y, Yudkoff M, Tuchman M. N-carbamylglutamate enhancement of ureagenesis leads to discovery of a novel deleterious mutation in a newly defined enhancer of the NAGS gene and to effective therapy. Hum Mutat. 2011 Oct;32(10):1153-1160. PMID: 21681857, PMCID: PMC3976964

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Tuchman M, Caldovic L, Daikhin Y, Horyn O, Nissim I, Korson M, Burton B, Yudkoff M. N-carbamylglutamate markedly enhances ureagenesis in N-acetylglutamate deficiency and propionic acidemia as measured by isotopic incorporation and blood biomarkers. Pediatr Res. 2008 Aug;64(2): 213–217. PMID: 18414145, PMCID: PMC2640836

*Yudkoff M, Ah Mew N, Daikhin Y, Horyn O, Nissim I, Nissim I, Payan I, Tuchman M. Measuring in vivo ureagenesis with stable isotopes. Mol Genet Metab. 2010;100 Suppl 1:S37-41. PMID: 20338795, PMCID: PMC2858793

Publications Resulting from Trainee Projects:

*Atwal PS, Medina CR, Burrage LC, Sutton VR. Nineteen-year follow-up of a patient with severe glutathione synthetase deficiency. J Hum Genet. 2016 Jul;61(7):669-672. PMID: 26984560, PMCID: PMC4961564

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*Burrage LC, Charng WL, Eldomery MK, Willer JR, Davis EE, Lugtenberg D, Zhu W, Leduc MS, Akdemir ZC, Azamian M, Zapata G, Hernandez PP, Schoots J, de Munnik SA, Roepman R, Pearring JN, Jhangiani S, Katsanis N, Vissers LE, Brunner HG, Beaudet AL, Rosenfeld JA, Muzny DM, Gibbs RA, Eng CM, Xia F, Lalani

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*Eeds AM, Mortlock D, Wade-Martins R, Summar ML. Assessing the functional characteristics of synonymous and nonsynonymous mutation candidates by use of large DNA constructs. Am J Hum Genet. 2007 Apr;80(4):740-750. PMID: 17357079, PMCID: PMC1852709

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*Jain-Ghai S, Nagamani SCS, Blaser S, Siriwardena K, Feigenbaum A. Arginase I deficiency: severe infantile presentation with hyperammonemia: more common than reported? Mol Genet Metab. 2011 Sep-Oct;104(1-2):107-111. PMID: 21802329, PMCID: PMC3171515

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*Nagamani SCS, Campeau PM, Shchelochkov OA, Premkumar MH, Guse K, Brunetti-Pierri N, Chen Y, Sun Q, Tang Y, Palmer D, Reddy AK, Li L, Slesnick TC, Feig DI, Caudle S, Harrison D, Salviati L, Marini JC, Bryan NS, Erez A, Lee B. Nitric-oxide supplementation for treatment of long-term complications in argininosuccinic aciduria. Am J Hum Genet. 2012 May 4;90(5):836-846.PMID: 22541557, PMCID: PMC3376491

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Chapters Resulting from Trainee Projects:

*EA Crombez, SD Cederbaum. Urea cycle disorders. In: Schapira AHV (ed) Neurology and Clinical Neuroscience. Mosby 2007, chapter 110, pp1469-1476.

*Lindsay C. Burrage, Brendan Lee, and Sandesh C.S. Nagamani; Urea Cycle Disorders, Rudolph Pediatrics, Chapter 141, p 23rd Edition, McGraw Hill, 2018

UCDC Consortium Publications:

Batshaw ML, Groft SC, Krischer JP. Research into rare diseases of childhood. JAMA. 2014 May 7;311(17):1729-1730. PMID: 24794360, Full Text (with PubMed access)

Nagamani SCS, Agarwal U, Tam A, Azamian M, McMeans A, Didelija IC, Mohammad MA, Marini JC. A randomized trial to study the comparative efficacy of phenylbutyrate and benzoate on nitrogen excretion and ureagenesis in healthy volunteers. Genet Med. 2018 Jul; 20(7): 708–716. PMID: 29693650, PMCID: PMC5924481

Publications Directly Resulting from UCDC-Related Activities

Ah Mew N, Caldovic L. N-acetylglutamate synthase deficiency: an insight into the genetics, epidemiology, pathophysiology, and treatment. Appl Clin Genet. 2011 Aug 24;4:127-35. PMID: 23776373, PMCID: PMC3681184

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