Urea Cycle Disorders Consortium

Abstracts Presented at Conferences

1. Lichter-Konecki U, Cabrera-Luque J, Moses L, Gallo V. Identifying astrocyte functions altered during hyperammonemic encephalopathy. Paper presented at: Society for Neuroscience Annual Meeting; November 12-16, 2005; Washington, DC.
2. Tuchman M. Collaborative Investigations of Urea Cycle Disorders: The Importance of Research Networks in the Study of Rare Diseases. Paper presented at: American Society of Human Genetics Annual Meeting; October 23-27, 2007; San Diego, CA.
3. Gropman A. 13C MRS study of ornithine transcarbamylase deficiency (OTCD). Paper presented at: Society for Inherited Metabolic Disorders Meeting; March, 2008; Asilomar, CA.
4. Kahn I, Seltzer R, Van Meter J, Gropman A. Diffusion tensor imaging detects areas of abnormal white matter microstructure in patients with partial ornithine transcarbamylase deficiency (OTCD). Paper presented at: Society for Inherited Metabolic Disorders Meeting; March, 2008; Asilomar, CA.
5. Tuchman M. Setting up Multi-Institutional Network Research in Rare Diseases: The Urea Cycle Consortium. Paper presented at: Society for Inherited Metabolic Disorders Meeting; March, 2008; Asilomar, CA
6. Lee B. Urea cycle disorders best practices and new developments: clinical presentation, laboratory diagnosis, and chronic management. Paper presented at: American College of Medical Genetics Annual Clinical Genetics Meeting; March 12-16, 2008; Phoenix, AZ.
7. Lichter-Konecki U. A Rare Disease Clinical Research Consortium for the collaborative investigation of UCDs Paper presented at: Pediatric Academic Societies Annual Meeting; May, 2009; Baltimore, MD.
8. Batshaw M. The Urea Cycle Disorders Consortium. Paper presented at: National Urea Cycle Disorders Foundation; July, 2009; Pasadena, California.
9. Batshaw M. The Urea Cycle Disorders Consortium. Paper presented at: Satellite Symposium to the 11th International Congress on Inborn Errors of Metabolism; August, 2009; La Jolla, California.
10. Batshaw M. Setting up multi-institutional research network in rare disease: The Urea Cycle Disorders Consortium. Paper presented at: Institute of Medicine Committee on Accelerating Rare Diseases Research and Orphan Product Development; November 23, 2009.
11. Gropman A, Shattuck K, Prust M, et al. Increased dorsolateral prefrontal cortex activation in OTCD during working memory. Paper presented at: National Urea Cycle Disorders Foundation Annual Conference; July 9-11, 2010; Cambridge, MA.
12. Krivitzky L, Waisbren S. Neuropsychological Functioning in Rare Diseases; Research Challenges and Potential Solutions. Paper presented at: 39th Annual Meeting of the International Neuropsychological Society; February 2-5, 2011; Boston, MA.

13. Breedan A, Prust M, Krivitzky L, Gropman A. Cognitive Tests Sensitive to the Neurological Impairment in Ornithine Transcarbamylase Deficiency (OTCD). Paper presented at: 34th Annual Meeting of the Society for Inherited Metabolic Disorders; February 27 - March 2, 2011; Asilomar, CA.

14. Lichter U. Update on the urea cycle disorders registry. Paper presented at: 34th Annual Meeting of the Society for Inherited Metabolic Disorders; February 27 - March 2, 2011; Asilomar, CA.

15. Shattuck K, Prust M, Seltzer R, et al. Increased Dorsolateral Prefrontal Cortex Activation in Ornithine Transcarbamylase Deficiency (OTCD) During Working memory: An fMRI study. Paper presented at: Society for Inherited Metabolic Disorders Annual Conference; February 27 - March 2, 2011; Asilomar, CA.

16. Shattuck K, Prust M, Seltzer R, Hailu A, vanMeter J, Gropman A. Altered Neural Activation in ornithine Transcarbamylase Deficiency during working memory. Paper presented at: Society for Inherited Metabolic Disorders Annual Conference; February 27 - March 2, 2011; Asilomar, CA.

17. Batshaw M. The Urea Cycle Disorders Consortium (UCDC). Paper presented at: 4th Annual Rare Disease Day February 28, 2011; Bethesda, MD.

18. Gropman A, Shattuck K, Prust M, et al. Increased dorsolateral prefrontal cortex activation in OTCD during working memory. Paper presented at: 11th International Congress of the European Society for Magnetic Resonance in Neuropediatrics; March 24-26, 2011; Amsterdam, The Netherlands.

19. Batshaw M. Data Quality Challenges of Multisite Clinical Trials Panel. Paper presented at: Quality Data from Pediatric Clinical Trials Meeting; October 21, 2011; Bethesda, MD.

20. Diaz G, Krivitzky L, Mokhtarani M, et al. Ammonia (NH3) Control and Improved Neurocognitive Outcome Among Urea Cycle Disorder (UCD) Patients Treated with Glycerol Phenylbutyrate (GPB). Paper presented at: American College of Medical Genetics Conference; March 27-31, 2012; Charlotte, NC.

21. Diaz G, Krivitzky L, Mokhtarani M, et al. Ammonia (NH3) Control and Improved Neurocognitive Outcome Among Urea Cycle Disorder (UCD) Patients Treated with Glycerol Phenylbutyrate (GPB). Paper presented at: Society for Inherited Metabolic Disorders Annual Conference March 31 - April 2, 2012; Charlotte, NC.

22. Ludwig W, Ellenbogan A, Gropman A. MDFiber Tracts in the Corpus Callosum Correlate with Scores on Behavioral Tasks and Glutamine Levels in Patients with OTCD, initial study (selected for travel award). Paper presented at: Society for Inherited Metabolic Disorders Annual Conference; March 31 - April 2, 2012; Charlotte, NC.

23. Mokhtarani M, Diaz G, Rhead W, et al. Elevated Phenylacetic Acid (PAA) Levels Appear Linked to Neurological Adverse Events in Healthy Adults But Not in Urea Cycle Disorder (UCD) Patients.

Paper presented at: Society for Inherited Metabolic Disorders Annual Conference; March 31 - April 2, 2012; Charlotte, NC.

24. Monteleone J, Mokhtarani M, Diaz G, et al. Population PK Analysis of Glycerol Phenylbutyrate (GPB) and Sodium Phenylbutyrate (NaPBA) in Adult and Pediatric Patients with Urea Cycle Disorders (UCD). Paper presented at: Society for Inherited Metabolic Disorders Annual Conference; March 31 - April 2, 2012; Charlotte, NC.

25. Mokhtarani M, Diaz G, Rhead W, et al. Urinary Phenlyacetateylglutamine Appears to Be a More Useful Marker than Metabolite Blood Levels for Therapeutic Monitoring of Phenylacetic Acid (PAA) Prodrugs. Paper presented at: Society for Inherited Metabolic Disorders Annual Conference; March 31 - April 2, 2012; Charlotte, NC.

26. Diaz G, Mokhtarani M, Rhead W, et al. Ammonia (NH3) amino acid and hyperammonemic crises (HACS) in pediatric and adult patients with urea cycle disorders (UCDS) during dosing with sodium phenylbutyrate (NaPBA) vs. Glycerol phenylbutyrate (GPB). Paper presented at: Garrod Association Symposium; May 31 - June 1, 2013; Sherbrooke, Canada.

27. Lee B, Mokhtarani M, Diaz G, et al. Optimizing ammonia (NH3) control in urea cycle disorder (UCD) patients: short and long-term implications. Paper presented at: Garrod Association Symposium; May 31 - June 1, 2013; Sherbrooke, Canada.

28. Diaz G, Mokhtarani M, Rhead W, et al. Ammonia (NH3) amino acid and hyperammonemic crises (HACS) in pediatric and adult patients with urea cycle disorders (UCDS) during dosing with sodium phenylbutyrate (NaPBA) vs. Glycerol phenylbutyrate (GPB. Paper presented at: 31st Annual Meeting of the Southeastern Regional Genetics Group™, Inc July 18-20, 2013; Asheville, NC.

29. Lee B, Mokhtarani M, Diaz G, et al. Optimizing ammonia (NH3) control in urea cycle disorder (UCD) patients: short and long-term implications. Paper presented at: 12th International Congress of Inborn Errors of Metabolism; September 3-6, 2013; Barcelona, Spain.

Conference Proceedings

1. 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.* 2008:11-15.

2. Bhavsar S, Khalidi N, Carette S, et al. Venothromboembolism in Large Vessel Vasculitis. Paper presented at: American College of Rheumatology (ACR/ARHP) Annual Meeting; October 2014, 2014; Boston, MA.

3. Carmona F, Mackie S, Martin J, et al. An Immunochip Study Confirms a Strong Contribution of HLA Class I and II Genes in the Susceptibility to Giant Cell Arteritis. Paper presented at: American College of Rheumatology (ACR/ARHP) Annual Meeting2014; Boston, MA.

4. Mecoli C, Wang F, Pappas C, et al. The Relationship of ARMS2 Genotype with Idiopathic Inflammatory Vasculitis. Paper presented at: American College of Rheumatology (ACR/ARHP) Annual Meeting2014; Boston, MA.

5. A R-P, Warner R, Cuthbertson D, et al. Biomarkers of Disease Activity in Vasculitis. Paper presented at: American College of Rheumatology (ACR/ARHP) Annual Meeting2014; Boston, MA.
6. Sreih A, Ezzeddine R, Fan J, et al. The Role of Macrophage Migration Inhibitory Factor (MIF) and MIF Gene Polymorphisms in the Pathogenesis of Granulomatosis with Polyangiitis. Paper presented at: American College of Rheumatology (ACR/ARHP) Annual Meeting)2014; Boston, MA.
7. Sy A, Dehghan N, Khalidi N, et al. Vasculitis and Inflammatory Bowel Diseases: A Study of 32 Patients with Both Conditions and Systematic Review of the Literature. Paper presented at: American College of Rheumatology (ACR/ARHP) Annual Meeting2014; Boston, MA.

Journal Articles

1. Kleppe S, Mian A, Lee B. Urea Cycle Disorders. *Curr. Treat. Options Neurol.* Jul 2003;5(4):309-319. PMID: 12791198
2. Gropman AL, Batshaw ML. Cognitive outcome in urea cycle disorders. *Mol. Genet. Metab.* Apr 2004;81 Suppl 1:S58-62. PMID: 15050975
3. Mian A, McCormack WM, Jr., Mane V, Kleppe S, Ng P, Finegold M, O'Brien WE, Rodgers JR, Beaudet AL, Lee B. Long-term correction of ornithine transcarbamylase deficiency by WPRE-mediated overexpression using a helper-dependent adenovirus. *Mol. Ther.* Sep 2004;10(3):492-499. PMID: 15336649
4. Caldovic L, Morizono H, Daikhin Y, et al. Restoration of ureagenesis in N-acetylglutamate synthase deficiency by N-carbamylglutamate. *J. Pediatr.* Oct 2004;145(4):552-554. PMID: 15480384
5. Scaglia F, Brunetti-Pierri N, Kleppe S, Marini J, Carter S, Garlick P, Jahoor F, O'Brien W, Lee B. Clinical consequences of urea cycle enzyme deficiencies and potential links to arginine and nitric oxide metabolism. *J. Nutr.* Oct 2004;134(10 Suppl):2775S-2782S; discussion 2796S-2797S. PMID: 15465784
6. Crombez EA, Cederbaum SD. Hyperargininemia due to liver arginase deficiency. *Mol. Genet. Metab.* Mar 2005;84(3):243-251. PMID: 15694174
7. Gropman AL. Expanding the diagnostic and research toolbox for inborn errors of metabolism: the role of magnetic resonance spectroscopy. *Mol. Genet. Metab.* Sep-Oct 2005;86(1-2):2-9.
8. Summar ML, Barr F, Dawling S, et al. Unmasked adult-onset urea cycle disorders in the critical care setting. *Crit. Care Clin.* Oct 2005;21(4 Suppl):S1-8. PMID: 16227111
9. Lanpher B, Brunetti-Pierri N, Lee B. Inborn errors of metabolism: the flux from Mendelian to complex diseases. *Nat. Rev. Genet.* Jun 2006;7(6):449-460. PMID: 16708072
10. 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.* Apr 2007;80(4):740-750. PMID: 17357079, PMCID: PMC1852709
11. Gropman AL, Summar M, Leonard JV. Neurological implications of urea cycle disorders. *J. Inherit. Metab. Dis.* Nov 2007;30(6):865-879. PMID: 18038189, PMCID: PMC3758693
12. Gropman AL, Seltzer RR, Yudkoff M, Sawyer A, VanMeter J, Fricke ST. 1H MRS allows brain phenotype differentiation in sisters with late onset ornithine transcarbamylase deficiency

(OTCD) and discordant clinical presentations. *Mol. Genet. Metab.* May 2008;94(1):52-60. PMID: 18262815, PMCID: PMC2486377

13. Tuchman M, Caldovic L, Daikhin Y, et al. N-carbamylglutamate markedly enhances ureagenesis in N-acetylglutamate deficiency and propionic acidemia as measured by isotopic incorporation and blood biomarkers. *Pediatr. Res.* Aug 2008;64(2):213-217. PMID: 18414145, PMCID: PMC2640836

14. Tuchman M, Lee B, Lichter-Konecki U, Summar ML, Yudkoff M, Cederbaum SD, Kerr DS, Diaz GA, Seashore MR, Lee HS, McCarter RJ, Krischer JP, Batshaw ML. Cross-sectional multicenter study of patients with urea cycle disorders in the United States. *Mol. Genet. Metab.* Aug 2008;94(4):397-402. PMID: 18562231, PMCID: PMC2640937

15. Brunetti-Pierri N, Clarke C, Mane V, Palmer DJ, Lanpher B, Sun Q, O'Brien W, Lee B. Phenotypic correction of ornithine transcarbamylase deficiency using low dose helper-dependent adenoviral vectors. *J. Gene Med.* Aug 2008;10(8):890-896. PMID: 18563850, PMCID: PMC2766563

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17. Gropman AL, Fricke ST, Seltzer RR, Hailu A, Adeyemo A, Sawyer A, van Meter J, Gaillard WD, McCarter R, Tuchman M, Batshaw M. 1H MRS identifies symptomatic and asymptomatic subjects with partial ornithine transcarbamylase deficiency. *Mol. Genet. Metab.* Sep-Oct 2008;95(1-2):21-30. PMID: 18662894, PMCID: PMC3724938

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transcarbamylase deficiency. *AJNR Am. J. Neuroradiol.* Oct 2010;31(9):1719-1723. PMID: 19287347, PMCID: PMC2746951

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