Urea Cycle Disorders Consortium (UCDC) Publications

*Indicates that the publication results from a trainee project

Publications Resulting from 5101:


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


Publications Resulting from 5102:
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, 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. 2008; 94(1):52-60. PMID: 18262815, PMCID: PMC2486377


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


**Publications Resulting from 5105:**


**Publications Resulting from Trainee Projects:**


*Pferdehirt R, Jain M, Blazo MA, Lee B, Burrage LC. Catel-manzke syndrome: further delineation of the phenotype associated with pathogenic variants in TGDS. Molecular Genetics And Metabolism Reports. 2015 Sep 1;4:89-91. PMID: 26366375, PMCID: PMC4563870


**Chapters Resulting from Trainee Projects:**


**UCDC Consortium Publications:**


**Publications Directly Resulting from UCDC-Related Activities**


Dobrowolski SF, Ellingson C, Caldovic L, Tuchman M. Streamlined assessment of gene variants by high resolution melt profiling utilizing the ornithine transcarbamylase gene as a model system. Hum Mutat. 2007;28:1133-1140. PMID: 17565723


MacArthur RB, Altim catal A, Tuchman M. Pharmacokinetics of sodium phenylacetate and sodium benzoate following intravenous administration as both a bolus and continuous infusion to healthy adult volunteers. Mol Genet Metab. 2004;Suppl:67-73. PMID: 15050977, Full Text (with PubMed access)


Nettesheim S, Kölker S, Karall D, Häberle J, Posset R, Hoffmann GF, Heinrich B, Gleich F, Garbade SF; Arbeitsgemeinschaft für Pädiatrische Stoffwechselstörungen (APS); European registry and network for Intoxication type Metabolic Diseases (E-IMD); Erhebungseinheit für Seltene Pädiatrische Erkrankungen in Deutschland (ESPED); Austrian Metabolic Group; Swiss Paediatric Surveillance Unit (SPSU). Incidence, disease onset and short-term outcome in urea cycle disorders - cross-border surveillance in Germany, Austria and Switzerland. Orphanet J Rare Dis. 2017;12:111. PMID: 28619060, PMCID: PMC5472961


Shi D, Allewell NM, Tuchman M. From genome to structure and back again: A family portrait of the transcarbamylases. Int J Mol Sci. 2015;16(8):18836-18864. PMID: 26274952, PMCID: PMC4581275


Summar ML, Endo F, Kölker S. On the creation, utility and sustaining of rare diseases research networks: Lessons learned from the Urea Cycle Disorders Consortium, the Japanese Urea Cycle Disorders Consortium, the European Registry and Network for Intoxication Type Metabolic Diseases. Mol Genet Metab. 2014 Sep-Oct;113(1-2):105-8. PMID: 25261246, PMCID: PMC4868037


Tuchman M. Hyperammonemia: are the burdens too grave? Case study. Ethics Intellect Disabil. 2004;8:1,3. PMID: 15835081


