Pediatrics 2017 Annual Report – Division Highlights Template

Genetics & Metabolism

DIVISION DESCRIPTION

The Division of Genetics and Metabolism provides comprehensive services to help patients and families cope medically and emotionally with a wide range of genetic disorders.

We have maintained a commitment to making these services available statewide through over 30 years of partnership with the Waisman Center on the UW campus and the State Division of Public Health, and through leadership of the Wisconsin State Genetics System.

Research interests include the genetic bases of inherited disorders and congenital anomalies, molecular techniques for newborn screening, diagnosis of nonsyndromic intellectual disability, fetal alcohol spectrum disorders, and the genetic causes of vertebral anomalies and bone dysplasias.

2017 HIGHLIGHTS

- **Jessica Scott Schwoerer, MD**; **Gregory Rice, MD**; **Mei Baker, MD**; **Jennifer Laffin, PhD, FACMG**; and **Ashley Kuhl, MS, CGC**, were part of a group who received a 2017 UW-Madison Community-University Partnership Award (link to YIR article on faculty awards) from Chancellor Rebecca Blank for their work on the Wisconsin Plain Community Project.

RECENT PUBLICATIONS

Adamsheck HC, **Petty EM**, Hong J, **Baker MW**, Brilliant MH, Mailick MR. Is Low FMR1 CGG repeat length in males correlated with family history of BRCA-associated cancers? An exploratory analysis of medical records. J Genet Couns. 2017 Dec;26(6):1401-1410. doi: 10.1007/s10897-017-0116-5. Epub 2017 Jun 30. PubMed PMID: 28667565.

Agather A, Rietzler J, **Reiser CA**, **Petty EM**. Working with the Hmong population in a genetics setting: Genetic counselor perspectives. J Genet Couns. 2017 Dec;26(6):1388-1400. doi: 10.1007/s10897-017-0117-4. Epub 2017 Jun 28. PubMed PMID: 28660354.

Calkins KL, DeBarber A, **Steiner RD**, Flores MJ, Grogan TR, Henning SM, Reyen L, Venick RS. Intravenous fish oil and pediatric intestinal failure-associated liver disease: changes in plasma phytosterols, cytokines, and bile acids and erythrocyte fatty acids. JPEN J Parenter Enteral Nutr. 2017 May 1:148607117709196. doi: 10.1177/0148607117709196. [Epub ahead of print] PubMed PMID: 28521607. *

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Hendee K, Wang LW, Reis LM, **Rice GM**, Apte SS, Semina EV. Identification and functional analysis of an ADAMTSL1 variant associated with a complex phenotype including congenital glaucoma, craniofacial, and other systemic features in a three-generation human pedigree. Hum Mutat. 2017 Nov;38(11):1485-1490. doi:10.1002/humu.23299. Epub 2017 Aug 1. PubMed PMID: 28722276; PubMed Central PMCID: PMC5638704.

Krieger M, Agather A, Douglass K, **Reiser CA**, **Petty EM**. Working with the Hmong population in a genetics setting: An interpreter perspective. J Genet Couns. 2017 Sep 24. doi: 10.1007/s10897-017-0153-0. [Epub ahead of print] PubMed PMID: 28942494.

Kuhl A, van Calcar S, **Baker M**, Seroogy CM, **Rice G**, **Scott Schwoerer J**. Development of carrier testing for common inborn errors of metabolism in the Wisconsin Plain population. Genet Med. 2017 Mar;19(3):352-356. doi: 10.1038/gim.2016.104. Epub 2016 Aug 11. PubMed PMID: 27513192. **

Lee CS, Fu H, Baratang N, Rousseau J, Kumra H, Sutton VR, Niceta M, Ciolfi A, Yamamoto G, Bertola D, Marcelis CL, Lugtenberg D, Bartuli A, Kim C, Hoover-Fong J, Sobreira N, **Pauli R**, Bacino C, Krakow D, Parboosingh J, Yap P, Kariminejad A, McDonald MT, Aracena MI, Lausch E, Unger S, Superti-Furga A, Lu JT; Baylor-Hopkins Center for Mendelian Genomics, Cohn DH, Tartaglia M, Lee BH, Reinhardt DP, Campeau PM. Mutations in fibronectin cause a subtype of spondylometaphyseal dysplasia with "corner fractures". Am J Hum Genet. 2017 Nov 2;101(5):815-823. doi: 10.1016/j.ajhg.2017.09.019. PubMed PMID: 29100092; PubMed Central PMCID: PMC5673654.

Legare JM, Seaborg K, **Laffin J**, Giampietro PF. Diaphanospondylodysostosis and ischiospinal dysostosis, evidence for one disorder with variable expression in a patient who has survived to age 9 years. Am J Med Genet A. 2017 Oct;173(10):2808-2813. doi: 10.1002/ajmg.a.38395. Epub 2017 Aug 17. PubMed PMID: 28815954.

Mailick M, Hong J, Greenberg J, Dawalt LS, **Baker MW**, Rathouz PJ. FMR1 genotype interacts with parenting stress to shape health and functional abilities in older age. Am J Med Genet B Neuropsychiatr Genet. 2017 Jun;174(4):399-412. doi: 10.1002/ajmg.b.32529. Epub 2017 Apr 13. PubMed PMID: 28407408; PubMed Central PMCID: PMC5435525.

McCrory NM, Edick MJ, Ahmad A, Lipinski S, **Scott Schwoerer JA**, Zhai S, Justice K, Cameron CA, Berry SA, Pena LD; Inborn Errors of Metabolism Collaborative. Comparison of methods of initial ascertainment in 58 cases of propionic acidemia enrolled in the Inborn Errors of Metabolism information system reveals significant differences in time to evaluation and symptoms at presentation. J Pediatr. 2017 Jan;180:200 205.e8. doi: 10.1016/j.jpeds.2016.09.050. Epub 2016 Oct 21. PubMed PMID: 27776753; PubMed Central PMCID: PMC5183466.

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Petty E, Golden RN. Embracing innovation in medical education. WMJ. 2017 Aug;116(3):179-180. PubMed PMID: 29323838.

Reynolds KK, Juusola J, **Rice GM**, Giampietro PF. Prenatal presentation of Mabry syndrome with congenital diaphragmatic hernia and phenotypic overlap with Fryns syndrome. Am J Med Genet A. 2017 Oct;173(10):2776-2781. doi: 10.1002/ajmg.a.38379. Epub 2017 Aug 17. PubMed PMID: 28817240.

Salen G, **Steiner RD**. Epidemiology, diagnosis, and treatment of cerebrotendinous xanthomatosis (CTX). J Inherit Metab Dis. 2017 Nov;40(6):771-781. doi: 10.1007/s10545-017-0093-8. Epub 2017 Oct 4. Review. PubMed PMID: 28980151.

Starr SR, Reed DA, Essary A, Hueston W, Johnson CD, Landman N, Meurer J, Miller B, Ogrinc G, **Petty EM**, Raymond J, Riley W, Gabriel S, Maurana C. Science of health care delivery as a first step to advance undergraduate medical education: A multi-institutional collaboration. Healthc (Amst). 2017 Sep;5(3):98-104. doi: 10.1016/j.hjdsi.2017.01.003. Epub 2017 Mar 23. PubMed PMID: 28342917.

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**ePub only; no print citation available when report was compiled*
*** Publication had previously appeared in 2016 report as an ePub*

GRANT SUPPORT

Faculty/Role	Funding Agency	Title
Baker, Mei Wang (PI)	DHHS, PHS, Centers for Disease Control & Prevention	Establishing a newborn screening process for early identification of infants with Pompe disease
Baker, Mei Wang (co-PI) with Farrell, Philip (co-PI)	Legacy of Angels	A prospective study of newborn screening for cystic fibrosis using a novel IRT/next generation sequencing method
Baker, Mei Wang (PI)	UWF - University of Wisconsin Foundation	NBS SMA biogen
Baker, Mei Wang (co-PI) with Farrell, Philip (co-PI) and Lai, Huichuan J (co-I)	Legacy of Angels	Assessing the added value of whole genome sequencing in cystic fibrosis newborn screening
Baker, Mei Wang; Laffin, Jennifer; Rice, Gregory; Scott Schwoerer, Jessica (collaborators) with Seroogy, Christine (PI) and Wald, Ellen (co-I)	UWF - Wisconsin Partnership - MERC	Improved Health Care Delivery to Wisconsin Amish Infants
Pauli, Richard M (co-PI) with Legare, Janet (co-PI)	Johns Hopkins University and and Biomarin Pharmaceutical, Inc.	Multi-center achondroplasia patient registry
Petty, Elizabeth (PI)	Medical College Of Wisconsin	The Robert D. and Patricia E. Kern

		Institute for the Transformation of Medical Education: National transformation network
Petty, Elizabeth (PI) with Carrel, Aaron (co-I)	UWF - Wisconsin Partnership - MERC	Engaging clinicians in online social learning to close knowledge gaps in community health: pilot focus on obesity and mental health care
Petty, Elizabeth (co-I) with Remington, Patrick (PI) and Navsaria, Dipesh (Tier 3 Director)	HRSA	Training in primary care medicine-interdisciplinary and interprofessional graduate joint degree program
Rice, Gregory M (PI)	Biomarin Pharmaceutical Inc	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 ASCEND)
Scott Schwoerer, Jessica A (PI)	Wisconsin Dept of Health Services	Congenital disorders formual
Scott Schwoerer, Jessica A (PI)	Wisconsin Dept of Health Services	Congenital disorders program
Steiner, Robert D (PI)	Circumvent Pharmaceuticals Inc.	Development of N-tert-(Butyl)hydroxylamine (NtBuHA) as a therapeutic agent for treating infantile neuronal ceroid lipofuscinosis
Wargowski, David S (PI)	DHHS, PHS, Centers for Disease Control & Prevention	FASD practice and implementation center for pediatrics
Wargowski, David S (PI)	Wisconsin Dept of Health Services	Clinical genetics unit, CY17
Wargowski, David S (PI)	Wisconsin Dept of Health Services	Congenital disorders, FY17