Department of Pediatrics
UNIVERSITY OF WISCONSIN
SCHOOL OF MEDICINE AND PUBLIC HEALTH

2018 Annual Report
Division of Genetics and 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 bone dysplasias.

2018 HIGHLIGHTS

- **Mei Baker, MD, FACMG**, received a one-year, $211,247 grant from Cure SMA (Spinal Muscular Atrophy) for her project, "A State-wide Spinal Muscular Atrophy (SMA) Newborn Screening Pilot in Wisconsin." Dr. Baker and collaborators will pilot the implementation of a statewide newborn screening for SMA using a multiplexing real-time PCR technology to incorporate identification of the disease into an ongoing screening process for severe combined immunodeficiency.

  Dr. Baker and Christine Seroogy, MD, also received a three-year, $120,000 Baldwin Wisconsin Idea Endowment Grant for their project, “Development and Implementation of Rapid Genetic Test to Improve Health Outcomes in Wisconsin Plain Newborns.” They aim to develop and implement state-of-the-art genetic testing for Wisconsin Amish and Mennonite newborns to improve early diagnosis and treatment for disorders frequently found in those communities.

- The Master of Genetic Counselor Studies program, led by **Catherine A. Reiser, MS, CGC,** has increased its class size from six to eight students and added two leadership positions: a clinical education coordinator and a research coordinator. Student clinical opportunities also now include Aurora for telehealth experiences and Marshfield Clinic as an option for the second year of training.

  In addition, the program’s Adam Rennebohm Perinatal Bereavement Conference, which was made possible by a generous endowment from the Rennebohm family in honor of their son, Adam, who had a diagnosis of Trisomy 13, marked its 10th year in 2018. Through outreach to other accredited genetic counseling programs, this conference has trained over 300 genetic counselors about families’ needs following the death of an infant.

- **Jessica Scott Schwoerer, MD,** and Christine Seroogy, MD, hosted the 6th Annual Translational Medicine in Plain Populations Conference, held at the Waisman Center on July 30-31, 2018.

- **Robert D. Steiner, MD, FAAP, FACMG,** was selected as the new editor-in-chief of The American College of Medical Genetics' (ACMG) peer-reviewed journal, *Genetics in Medicine*, after serving as the journal's deputy editor since 2011 and a section editor since 2006.

RECENT PUBLICATIONS

Abdel-Khalik J, Crick PJ, Yutuc E, DeBarber AE, Duell PB, **Steiner RD**, Laina I, Wang Y, Griffiths WJ. Identification of 7α,24-dihydroxy-3-oxocholest-4-en-26-oic and 7α,25-dihydroxy-3-oxocholest-4-en-26-oic acids in human cerebrospinal fluid and plasma. *Biochimie*. 2018 Oct;153:86-98. doi: 10.1016/j.biochi.2018.06.020. Epub 2018 Jun 28. PubMed PMID: 29960034; PubMed Central PMCID: PMC6171785.

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GRANT SUPPORT

Principal Investigator	Sponsor	Title	Co-Investigators
Baker, Mei W	Cure Spinal Muscular Atrophy	A State-wide spinal muscular atrophy newborn screening pilot in Wisconsin	

Baker, Mei W	Health Resources & Services Administration (HRSA)	Establishing an electronic communication system for newborn screening testing results and confirmatory testing information	
Farrell, Philip M	Legacy of Angels	Assessing the added value of whole genome sequencing in cystic fibrosis newborn screening	Genetics: Mei Baker
Mailick, Marsha R (Waisman Center)	National Institutes of Health (NIH)	FMR1 premutation phenotypes in population-based & clinically-ascertained samples	Genetics: Mei Baker
Legare, Janet M	Biomarin Pharmaceutical, Inc. & Johns Hopkins University	Multi-center achondroplasia patient registry	Genetics: Rich Pauli
Petty, Elizabeth M	Kern Family Foundation & Medical College of Wisconsin	The Robert D. and Patricia E. Kern Institute for the Transformation of Medical Education: National transformation network	GPAM: Gwen McIntosh
Petty, Elizabeth M	Wisconsin Partnership Program	Engaging clinicians in online social learning to close knowledge gaps in community health: Pilot focus on obesity and mental health care	
Scott Schwoerer, Jessica A	Wisconsin Department of Health Services	Congenital disorders	Genetics: Gregory Rice
Steiner, Robert D	National Institutes of Health (NIH) & Circumvent Pharmaceuticals, Inc.	Development of N-tert-(Butyl)hydroxylamine as a therapeutic agent for treating infantile neuronal ceroid lipofuscinosis	
Wargowski, David	Centers for Disease Control & Prevention (CDC)	FASD practice and implementation center for pediatrics	