



Genome Sciences Seminar

Wednesday, 1.17.18 | 3:30 | Foege Auditorium



William A. Gahl, MD, PhD

Clinical Director, NHGRI

Director, NIH Undiagnosed Diseases Program

<https://www.genome.gov/10005723/gahl--group/>

“The NIH Undiagnosed Diseases Program and Network”

Gahl Lab:

Since 2010, the section has been a training ground for world experts in the clinical aspects of cystinosis, alkaptonuria, Chediak-Higashi disease, Hermansky-Pudlak syndrome (HPS), gray platelet syndrome, Hutchinson-Gilford Progeria syndrome, GNE myopathy, albinism, autosomal recessive polycystic kidney disease, Joubert disease and other ciliopathies, and Erdheim-Chester disease. In aggregate, more than 500 patients with these very rare disorders have been evaluated in just the past five years to accrue cross-sectional and longitudinal data on the natural histories of these diseases for use in future interventional studies. Section investigators and collaborators have already obtained Food and Drug Administration approval for oral and topical cysteamine for nephropathic cystinosis. They have completed clinical trials of the TGF-beta inhibitor; pirfenidone, for the pulmonary fibrosis of HPS; and nitisinone, for the ochronosis of alkaptonuria. Nitisinone blocks the production of homogentisic acid, which accumulates in alkaptonuria and forms polymers that bind to and destroy connective tissue. Ongoing therapeutic trials address mitochondrial and oxidation-reduction disorders using an investigational drug (EPI-743), the histiocytosis of Erdheim-Chester disease using dabrafenib and trametinib, the myopathy of cystinosis using recombinant human growth hormone, and GNE myopathy using N-acetylmannosamine. This sugar is an intermediate in the sialic acid synthetic pathway distal to the block in sialic acid synthesis that constitutes the basic defect in GNE myopathy; sialic acid is necessary for muscle function because it interacts with the critical muscle protein, alpha-dystroglycan. All of the patients involved in these investigations are enrolled in one of 10 active institutional review board-approved clinical protocols managed by the section.

Refreshments served outside the Auditorium at 3:20pm

Questions? Contact Brian Giebel at bgiebel@uw.edu or visit the Seminar website at <http://www.gs.washington.edu/news/seminars.htm>

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