

GOLD Annual General Meeting 2007.

GOLD's Annual General Meeting will be held at 11.30am -1pm on Thursday 25th October, during the American Society of Human Genetics conference in San Diego.

We are pleased and honoured to have as our Guest Lecturer Dr William Gahl, who will speak on the topic of "Advocacy for Rare Diseases".

William A. Gahl, M.D., Ph.D.

Dr Gahl is Clinical Director, National Human Genome Research Institute. He has a distinguished career as a medical and biochemical geneticist, focused especially on bringing new therapeutic approaches to the treatment of rare diseases. His group investigates a number of rare metabolic disorders, including lysosomal storage diseases, specifically sialic acid disorders and cystinosis, for which he runs a reference laboratory. Dr. Gahl's laboratory elucidated the basic defect of cystinosis and demonstrated the safety and efficacy of cysteamine (β -mercaptoethylamine) therapy, a treatment that depletes cells of cystine. This therapy, along with kidney transplantation, has changed the life course of many cystinosis patients from one filled with debilitating complications to one marked by chronic, yet manageable symptoms. His group is following approximately 125 pre- and post-transplant cystinosis patients to track their clinical course, identify additional mutations, and document any complications of their cysteamine therapy.

Dr. Gahl emphasizes that the challenge of developing new and improved therapies for rare disorders is a partnership between researchers, patients and society as a whole. He has commented: "The test of a society is how it treats its most helpless and disadvantaged members. People with rare genetic diseases give humanity so much, scientifically and spiritually, that we owe them a huge debt of gratitude. In fact, they make us more human."