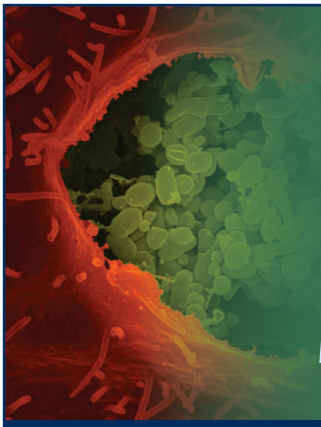

Rare and Undiagnosed Diseases: Discovery and Models of Precision Therapy

March 5–8, 2017 | Fairmont Copley Plaza | Boston, Massachusetts | USA

Scientific Organizers:
William A. Gahl, National Human Genome Research Institute, National Institutes of Health, USA
Christoph Klein, Ludwig-Maximilians-University Munich, Germany

Of the 7000 known rare diseases, only a fraction have their molecular and mechanistic bases delineated, and many other diseases have yet to be discovered. Characterization of these disorders will reveal new biochemical pathways and cell biological processes, point to potential drug targets, and provide hope for millions of affected individuals. This meeting will address approaches and best practices in pursuing rare and undiagnosed disorders, limitations in data sharing as a barrier to new disease discovery, and examples of successful descriptions of new diseases, mechanisms, and treatments. Specific aims include fostering the development of expertise in rare diseases and the definition of their natural histories; an international network of scientists and physicians collaborating on new disease discovery by sharing phenotypic and sequence data; and new therapeutic approaches based upon rare and new disease mechanisms. Through this meeting, participants should become familiar with rare and undiagnosed disease programs, acquire insights into new disease mechanisms, learn about potential therapeutic targets, and establish collaborations that enhance rare disorder expertise and new disease discovery. The meeting will bring together physicians who are expert in rare disorders with scientists who know metabolic pathways and mechanisms, advancing understanding and therapy. A desired outcome of the meeting includes developing lists of clinical and basic research experts for synergistic collaborations, and creating an international organization to perpetuate the joint investigation of rare and new diseases.

Session Topics:
- CRISPR/cas9; iPS; Embryonic Stem Cells
- Mechanisms of Rare Disease
- New Disease Discovery
- The Needs of Rare and Undiagnosed Disease Patients
- Therapeutics for Rare Diseases
- Late-Breaking Discoveries and Technological Underpinnings of Rare and Undiagnosed Diseases Research

plus a workshop

***Scholarship Application & Discounted Abstract Deadline:* November 2, 2016**
***Abstract Deadline:* December 6, 2016**
***Discounted Registration Deadline:* January 10, 2017**

Note: Scholarships and Underrepresented Trainee Scholarships are available for graduate students and postdoctoral fellows and are awarded based on the abstract submitted.
Upper image of Q fever bacteria courtesy of NIAID

Meeting Hashtag: #KSraredis
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