Abstract Title: Pompe disease - An exemplar for diagnosis, treatment and prevention

Author Name: Dr. Priya Kishnani, Professor of Pediatrics, Division Chief of Medical Genetics

Author Institute: Duke University Medical Center, Dept. of Pediatrics, Division of Medical Genetics, Durham, US

Abstract: Since the advent of enzyme replacement therapy for Pompe disease in 2006 the natural history and understanding of this once lethal disease has changed significantly. Long term outcomes of patients with Pompe disease including central nervous system aspects of this disease will be discussed. It is now recognized that early initiation of enzyme therapy improves clinical outcomes. Another barrier that has been recognized is the development of antidrug antibodies (ADA). Whilst babies with infantile onset Pompe disease (IOPD) who are cross reactive immunological material (CRIM) negative are most at risk for development of ADA's, there is a recognition that a subset of CRIM positive babies and also adults with LOPD who develop ADA's. There are also situations when patients can have a break in tolerance. The application of newborn screening, an increased dose of ERT and immune modulation has changed the landscape and outcome of children with Pompe disease. The role of intrauterine replacement therapy is also emerging. All these topics will be discussed.

Area of expertise: Glycogen Storage Disease, Lysosomal Storage Disease, Inborn Errors of Metabolism