



Friday, May 31, 2024 | The New York Academy of Medicine

1216 5th Ave, New York, NY 10029

All sessions located on the 3rd floor Library Reading Room unless otherwise noted. Agenda topics, speakers and timing is subject to change.

7:45a - 8:45a	Registration Check-in and Breakfast
8:35a -8:45a	Welcome – Frederick Kaskel MD, PhD, FAAP, FASN
8:45a - 9:15a	Cystinosis: Basic Defect and Genetics – Bill Gahl, MD, PhD
9:15a -9:45a	Cell Biology of the mTOR Pathway in Cystinosis – Jess Thoene, MD
9:45a - 10:15a	Neuromarkers of Sensory and Cognitive Function in Cystinosis – Sophie Molholm, PhD
10:15a-10:45a	Gastrointestinal Issues in Cystinosis – Adam C. Stein, MD
10:45a - 11:00a	Break
11:00a - 12:00p	Panel: Patient and Caregivers, Challenges within Rare Disease Jonathan Dicks, CRN President and VP Development, cystinosis caregiver Shannon Henderson, RN, living with cystinosis Christina Morris, Adult Leadership Advisory Board Member, living with cystinosis Herberth Sigler, CRN Board Member, cystinosis caregiver Joshua Zaritsky, MD, PhD (moderator)
12:00p - 12:15p	Orthopedic Issues from Childhood to Adulthood – Melinda S. Sharkey, MD
12:15p - 12:30p	Clinical Trial Readiness for Myopathy and Dysphagia in Adults with Cystinosis – Reza Seyedsadjadi, MD
12:30p - 1:00p	Ophthalmic Concerns Related to Cystinosis – Rachel Bishop, MD, MPH
1:00p - 2:00p	Lunch – Pharmaceutical Considerations and Collaboration in Rare Disease Leadiant Biosciences Recordati Rare Diseases Amgen Rare Disease
2:00p - 3:00p	Panel: Transitioning from Pediatric to Adult Nephrology James Drakakis, DO Ladan Golestaneh, MD, MS Larry Greenbaum, MD, PhD, FAAP (moderator) Rupesh Raina, MD, FACP, FAAP Cybele Ghossein, MD Jack Greeley, living with cystinosis Laura Krummenacker, living with cystinosis
3:00p - 3:30p	Engagement and Support in Rare Disease: Cystinosis Research Network as a Model Jonathan Dicks, CRN President, VP Development and cystinosis caregiver Marybeth Krummenacker, CRN VP Education & Awareness, cystinosis caregiver Kristina Sevel, MSN, RN, CRN VP Research, cystinosis caregiver Frederick Kaskel MD, PhD, FAAP, FASN (moderator)
3:30p - 3:45p	Break
3:45p - 4:15p	Supporting Mental Health of People Living with Rare and Chronic Disease Maya Doyle, MSW, PhD, LCSW Chelsea Meschke, LMSW, cystinosis caregiver
4:15p - 4:45p	The Potential of Moderna mRNA Technology to Treat Cystinosis – Paul Goodyer, MD
4:45p - 5:15p	Novel Genetic Treatments of Rare Disease and Applications to Common Disease John M. Greally, MD, PhD Melissa Wasserstein, MD
5:15p - 5:45p	Stem Cell Gene Therapy Clinical Trial Results in Cystinosis – Stephanie Cherqui, PhD
5:45p - 7:15p	Wrap-up Reception – Hors d'oeuvres and cocktails/beverages served *Located in the Periodicals Room, 3rd floor*