



CYSTINOSIS
RESEARCH NETWORK

CYSTINOSIS SYMPOSIUM

A Rare Disease Model for Comprehensive Care

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 - Lesli King, Eileen Wall Recordati Rare Diseases - France Lebel, MSc Amgen Rare Disease - Andrea Atherton, MS, CGC
2:00p - 3:00p	Panel: Transitioning from Pediatric to Adult Nephrology James Drakakis, DO Ladan Golestaneh, MD, MS Larry Greenbaum, MD, PhD, FAAP (moderator) 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	Mental Health Across the Lifespan with Rare and Chronic Disease: A Focus on Medical Trauma 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	Advanced Screening and Diagnostic Approaches for Rare Conditions at the New York Center for Rare Diseases 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*

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