

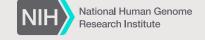
# Scientific Updates on Organic Acidemias and Homocystinurias

Webinar | June 24-25, 2021

#### Thursday, June 24

## **Scientific session 1: Methylmalonic and Propionic Acidemias**

10:00 - 10:05 a.m.	Introduction	
10:05 - 10:30 a.m.	Genomic Therapies for Methylmalonic and Propionic Acidemia	Charles P. Venditti, M.D., Ph.D.
10:30 - 11:00 a.m.	Liver Transplantation in Methylmalonic and Propionic Acidemia	Jerry Vockley, M.D., Ph.D.
11:00 - 11:30 a.m.	MMA and PA: First Guidelines Revision and Towards Novel Evidence	Patrick Forny, M.D., Ph.D.
11:30 - 12:00 p.m.	Role of Mitophagy in Methylmalonic Acidemia	Olivier Devuyst, M.D., Ph.D.
12:00 - 12:30 p.m.	Lunch Break	
12:30 - 12:50 p.m.	Acyl Modifications in MMA	Pamelasara Head, Ph.D.
12:50 - 1:20 p.m.	Mechanisms of Cardiomyopathy in Propionic Acidemia	Maria Fernandez-Velasco, Ph.D.
1:20 - 1:40 p.m.	Liver Transplant and Cardiomyopathy in Propionic Acidemia	Gerard Berry, M.D.
1:40 - 2:00 p.m.	Liver Neoplasms in Organic Acidemias: An Emerging Complication	Stephanie Grunewald, Ph.D.
2:00 - 2:30 p.m.	Break	
2:30 - 2:50 p.m.	Machine Learning for Biomarker Data Mining in Organic Acidemias	Oleg Shchelochkov, M.D.
2:50 - 3:10 p.m.	Hepatocyte-Based Models and Small Molecule Therapies for MMA/PA	Kimberly A. Chapman, M.D., Ph.D.
3:10 - 3:30 p.m.	Neonatal Gene Therapy in Animal Models of Organic Acidemias	Randy Chandler, Ph.D.
3:30 - 4:00 p.m.	Pave-GT Program for Organic Acidemias	PJ Brooks, Ph.D.



**Panel Discussion** 

4:00 - 4:30 p.m.



# **Scientific Updates on Organic Acidemias and Homocystinurias**

Webinar



### Friday, June 25

## Scientific Session 2: Combined Methylmalonic Acidemia and Homocystinuria

10:00 - 10:05 a.m.	Introduction	
10:05 - 10:30 a.m.	Remethylation Defects	David S. Rosenblatt, M.D.
10:30 - 10:45 a.m.	High-Dose B12 Therapy for cblC Deficiency	Irini Manoli, M.D., Ph.D.
10:45 - 11:10 a.m.	Pathophysiology of Manifestations of Methionine Synthesis Defects	ean Louis Gueant, M.D., D.Sc.
11:10 - 11:35 a.m.	Mouse Models of cbIC, cbIX	Ross A. Poché, Ph.D.
11:35 - 12:00 p.m.	The Transcobalamin Receptor: Insights from Patients, Populations	Lawrence Brody, Ph.D.
12:00 - 12:30 p.m.	Lunch Break	

## **Scientific Session 3: Homocystinuria**

12:30 - 1:00 p.m.	Updates on Classic HCU Diagnosis and Management	Harvey Levy, M.D.
1:00 - 1:30 p.m.	Update on the Activities of the EHOD Collaboration	Martina Huemer, M.D.
1:30 - 2:00 p.m.	Newborn Screening for HCU	Marzia Pasquali, Ph.D.
2:00 - 2:30 p.m.	Treatment of CBS Deficient Mice by AAV-Mediated Gene Therapy	Warren Kruger, Ph.D.
2:30 - 3:00 p.m.	Break	
3:00 - 3:20 p.m.	Enzyme Replacement for Homocystinuria	Tomas Majtan, Pharm.D., Ph.D.
3:20 - 3:40 p.m.	How Elucidating Regulatory Cross-Talk Between Folate and Thiol Metabolism Can Lead to Improved Treatment in Classical Homocystinuria: A Possible Route to the End of Methionine Restriction?	Ken Maclean, Ph.D.
3:40 - 4:10 p.m.	Testing All Possible Amino Acid Changes in the Homocystinuria Proteins CBS and MTHFR	Frederick (Fritz) Roth, Ph.D.
4:10 - 4:30 p.m.	Panel Discussion	



