

24-25 May | Maggio 2023

LOWE SYNDROME RESEARCH MEETING

The Telethon Institute of Genetics and Medicine, Pozzuoli, Italy &
Grand Hotel Serapide (Evening Event)



Speakers

Claudio Aguilar, USA

Antonija Jurak Begonja, HR

Antonella De Matteis, IT

Arnaud Echard, FR

Francesco Emma, IT

Kai Erdmann, UK

Jenny Gallop, UK

Paul James, UK

Jeri Kubicki, USA

Herb Lachman, USA

Martin Lowe, UK

Robert Nussbaum, USA

Raghu Padinjat, IN

Leopoldo Staiano, IT

Yang Sun, USA

Vidhu Thaker, USA

Andrew Thomas, UK

Meeting Sponsors



Organizers

Leopoldo Staiano, TIGEM, Italia

Jenny Gallop, University of Cambridge, UK

Jeri Kubicki, Lowe Syndrome Association, USA

The meeting is invitation only. A hybrid option is available by Zoom. Please email zimbardi@tigem.it for more information.

Day 1: Wednesday 24th May | Maggio 2023

AGENDA: LOWE SYNDROME RESEARCH MEETING 2023

Time:	Topic:	Detail:
12:30 - 14:00	Registration	TIGEM auditorium
12:30 - 13:50	<i>Buffet light lunch</i>	
13:55	Dial in to Zoom for 14:00 start	
14:00 - 14:10	Welcome & brief introduction	Annamaria Dinelli, AISLO, Italy Paul James, LST, UK
Session 1	Moving towards a therapy: From translational studies to clinical management of patients with Lowe Syndrome	Chair: Leopoldo Staiano
14:10 - 14:40	A pipeline for drug repositioning in Lowe Syndrome	Antonella De Matteis, TIGEM, Italy
14:40-15:10	Understanding the effects of phosphatidylinositol (4,5) bisphosphate lipid alterations on kidney and brain cells and how to ameliorate them	Jenny Gallop, Gurdon Institute, University of Cambridge, UK
15:10-15:40	Establishing the role of OCRL in the hypothalamus based on transcript specific expression	Vidhu Thaker, Columbia University, New York, USA
15:40-16:10	<i>Coffee break</i>	Outside auditorium
16:10-16:40	Ocular presentations of Lowe syndrome—challenges and advances	Yang Sun, Stanford University, California, USA
16:40-17:10	Renal Fanconi syndrome and its measurement in clinical trials	Francesco Emma, Bambino Gesù Hospital, Rome, Italy
17:10-17:30	<i>Refreshment Break</i>	Outside auditorium
Session II	Patient Advocacy and Engagement in understanding the unmet needs and progression to clinical trials	Chair: Robert Nussbaum
17.30-18.00	Strength in Unity: The LSA's Commitment to Community, Family Voices, and Research Advancement	Jeri Kubicki, Lowe Syndrome Association, USA
18:00-18:30	Showing there can be Positives	Paul James, Living with Lowes, UK
18.30-19.00	To be confirmed	Andrew Thomas, Lowe Syndrome Trust, UK
19.00	Shuttle to Grand Hotel Serapide	
	<i>Networking and Buffet dinner at the Grande Hotel Serapide</i>	

Day 2: Thursday 25th May | Maggio 2023

AGENDA: LOWE SYNDROME RESEARCH MEETING 2023

Time:	Topic:	Detail:
09:00	Shuttle to TIGEM	
Session III	From genetic mutation to phenotypic effect	Chair: Jenny Gallop
09:30-10:00	Using zebrafish to model Lowe syndrome	Martin Lowe, University of Manchester, UK
10:00-10:30	Modeling Lowe Syndrome using induced pluripotent stem cells (iPSC)	Herb Lachman, Albert Einstein, New York, USA
10:30-11:00	Rab35/OCRL control PI(4,5)P2 and F-actin both during endocytosis and cytokinesis	Arnaud Echard, Institut Pasteur, France
11:00-11:30	<i>Coffee break</i>	Outside auditorium
11:30-12:00	Bleeding tendency in Lowe syndrome: OCRL controls cytoskeletal rearrangements during platelet adhesion	Antonija Jurak Begonja, University of Rijeka, Croatia
12:00-12:30	Phenotypic and Biochemical Abnormalities Displayed by Conformationally Affected OCRL1 Patient's Variants	Claudio Aguilar, Purdue University, Indiana, USA (remote)
12:30-14:00	<i>Lunch break</i>	Campus canteen or garden
Session IV	Recent discoveries on the cell biology of Lowe Syndrome using novel in vitro and in vivo models	Chair: Arnaud Echard
14:00-14:30	Kidney organoids as a model to study novel roles of OCRL associated with the progressive decline of kidney function in Lowe Syndrome	Leopoldo Staiano, TIGEM, Italy
14:30-15:00	Cellular mechanisms underlying the neurodevelopmental phenotype of Lowe syndrome	Raghu Padinjat, National Centre for Biological Sciences, Bangalore, India
15:00-15:30	Organ on a chip technology to model Lowe syndrome	Kai Erdmann, University of Sheffield, UK
15:30-16:00	Perspectives, Key Next Steps and Research Priorities	Robert Nussbaum, Lowe Syndrome Association and Invitae, USA
16:00-16:15	Closing Remarks	Leopoldo Staiano / Jenny Gallop

Refreshments available outside the auditorium