Agenda Lowe Syndrome Research Meeting 2023

Day 1 **Wednesday 24th May**

Time	Topic	Detail
12:30-14:00	**Registration**	TIGEM auditorium
12:30-13:50	*Buffet light lunch*	
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 I	**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 discovery 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	*Coffee break*	*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 Gesu Hospital, Rome, Italy
17:10-17.40	To be confirmed	To be confirmed
17:40-18:00	*Refreshment Break*	*Outside auditorium*
Session II	**Patient Advocacy and Engagement in understanding the unmet needs and progression to clinical trials**	**Chair: Robert Nussbaum**
18.00-18.30	Strength in Unity: The LSA's Commitment to Community, Family Voices, and Research Advancement	Jeri Kubicki, Lowe Syndrome Association, USA
18:30-19:00	Showing there can be Positives	Paul James, Living with Lowes, UK
19.00-19.30	To be confirmed	Andrew Thomas, Lowe Syndrome Trust, UK
19.30	Shuttle to Grand Hotel Serapide	
20.30	*Networking and Buffet dinner at the Grande Hotel Serapide*	

Day 2 **Thursday 25th May**

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	Coffee break	*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	*Lunch break*	*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	