

The Third International Symposium of the [Lowe Syndrome Trust](#) 'Molecular and Clinical Advances in Lowe Syndrome [click here for invitation](#)

[Royal Society, December 3rd 2010](#)

09.00h Registration, tea and coffee

09.30h Welcome - **Lorraine Thomas**, Founder and Chair of the Lowe Syndrome Trust, London, UK

MORNING SESSION *Chair: Prof. Robert Unwin*

09.40h **Dr Detlef Bockenhauer** (Paediatric Nephrology and Institute of Child Health, UCL, UK):

'Clinical aspects of Lowe syndrome'

10.10h **Prof. Atul Mehta** (Lysosomal Storage Diseases Unit and Centre for Haematology, UCL, UK):

'Advances in lysosomal storage diseases'

10.40h **Prof. Albert Ong** (Academic unit of Nephrology, University of Sheffield, UK):

'How many ways to form a cyst?'

11.10h **Prof. Joel Lunardi** (Laboratoire de Biochimie de l'ADN, Grenoble, France):

'Correlations between mutations of the OCRL1 gene and the clinical and biochemical phenotypes'

11.40h Coffee break

11.50h **Dr Geneviève Baujat** (AP-HP Hôpital Necker, Génétique Pédiatrique, Paris, France):

'Retrospective evaluation of 49 Lowe patients: clinical variability and potential high risk of bleeding'

12.10h **Dr Dominique Lasne** (AP-HP Hôpital Necker, Laboratoire d'Hématologie, Faculté de Pharmacie, Paris, France):

'Bleeding disorders in Lowe syndrome due to primary haemostasis disorders'

12.30h **Prof. Robert Nussbaum** (University of California at San Francisco, USA):

'Mouse model for renal tubulopathy in Lowe syndrome and Dent disease type 2'

13.15h Lunch

AFTERNOON SESSION Chair Prof. Shamshad Cockcroft

14.15h **Prof. Pietro De Camilli** (Biological and Biomedical Sciences, Yale University, USA):

'Molecular interactions of OCRL in the endocytic pathway'

14.45h **Dr Antonella De Matteis** (Department of Cell Biology and Oncology, Consorzio Mario Negri Sud, Italy):

'Impaired endosomal trafficking induced by the loss of function of OCRL'

15.15h **Dr Claudio Aguillar** (Department of Biological Sciences, Perdue University, USA):

'Cellular phenotypes of Lowe syndrome patient's cells'

15.45h Tea and Coffee

Chair Prof. Philip Beales

16.00h **Dr Martin Lowe** (School of Biological Sciences, University of Manchester, UK):

'OCR L1 binding proteins and membrane traffic'

16.30h **Dr Tim Levine** (Department of Cell Biology, Institute of Ophthalmology, UCL, UK):

'Lowe Syndrome protein OCRL1 supports maturation of polarized epithelial cells'

17.00h **Dr Rudiger Woscholski** (Division of Cell and Molecular biology, Imperial College London, UK):

'PIP2 dependent pathways'

Chair Prof. Robert Unwin

17.30h Guest lecture: **Prof. Raj Thakker** (Nuffield Department of Medicine, University of Oxford, UK):

'Insights into mechanisms of proximal renal tubular reabsorption and transport - lessons from Dent's disease and Lowe's syndrome'

18.30h Close of meeting (followed by drinks/nibbles)