

The 5th International Symposium of the Lowe Syndrome Trust

'Molecular and Clinical Advances in Lowe Syndrome'

Royal Society, London, 12th December, 2014



The 5th International *Lowe Syndrome Trust* Symposium was held on the 12th December 2014 at the Royal Society, Pall Mall, London. The meeting drew together science and medical researchers into Lowe syndrome as well as world renowned experts in stem cells, and was followed by a drinks reception hosted by *Lowe Syndrome Trust* patron Penny Lancaster-Stewart.

The scientific programme brought together Lowe syndrome researchers from the US, UK and around Europe, many funded by the LST, to present latest research findings, discuss ideas and form collaborations. It also featured excellent presentations on exciting new stem cell technologies that could be used to develop novel treatments. The first talks described how mouse and zebrafish animal models have been developed to study Lowe syndrome. Such models will become invaluable in understanding how the disease works and for testing potential drugs. This was followed by exciting research into drugs to treat Lowe syndrome. The next step is to test these drugs in the animal models, with a view to eventually developing clinical trials in patients.

Eminent guest speakers Bruce Conklin (*UCSF*) and Juan Carlos Izpisua Belmonte (*Salk Institute*) described how cutting-edge stem cell technologies are being used to study and potentially treat rare genetic disorders, such as Lowe syndrome. This rapidly evolving field offers great potential and could revolutionize treatment of genetic disorders. There were further talks looking at the function of the enzyme that is faulty in Lowe syndrome (OCRL1) – what it does and why losing it causes such problems in cells, in particular in kidneys and the nervous system. In addition there was a session on the latest research into the behavioural and neurological problems seen in Lowe syndrome and how understanding these better could improve patient support.

The drinks reception was a great end to the day, where researchers were joined by many guests and supporters to discuss their work. Penny Lancaster-Stewart gave a moving speech about why she is a patron of the *Lowe Syndrome Trust*, and together with Dr Martin Lowe, received a cheque for £10,000 from the *Hospital Trust* to help fund future research. Immense progress continues to be made in understanding Lowe syndrome. This is now leading to potential drugs, with several candidates already undergoing testing. This rapid progress is largely due to the funding provided by the *Lowe Syndrome Trust* in the UK. They should be acknowledged not only for their outstanding efforts in supporting scientific research, but also for the support they provide for Lowe syndrome patients and their families.





Lowe Syndrome is a devastating genetic disorder that affects boys, causing multiple physical and mental handicaps, including cataracts at birth, muscle weakness, kidney problems, cysts, brittle bones, arthritis, poor growth, mental impairment with behavioural problems and epilepsy. Sadly this often leads to a short life expectancy.

" Fantastic science, some on Nobel prize level. It was a privilege to have been part of it "

Dr Detlef Bockenhauer



The Lowe Syndrome Trust

www.lowetrust.com

"Thank you so much for such a fantastic symposium... it allowed me to establish possible collaborations with other scientists and doctors trying to advance research"

Professor Juan Carlos Izpisua Belmonte

"Thank you for hosting such a terrific meeting. I was very impressed by the progress you have made in a short time"

Professor Bruce Conklin

"Thank you to the Lowe Syndrome Trust for organising another high-quality scientific meeting. I am particularly pleased about the advance in research fostered by the Trust, with most talks clearly pointing out the opportunities to screen for potential drugs to remedy the symptoms of Lowe Syndrome. The LST's impact goes well beyond the monetary value of the raised funds for research, with many scientific careers and outputs now being influenced by the Trust's funding and visibility. Since the LST came into being we have seen a doubling in publications that are addressing the target protein of Lowe syndrome, OCRL; this achievement is certainly due to your effort and determination in setting-up and establishing the top research-focussed charity on Lowe syndrome and Dent's disease. I think you can be proud of what you have achieved" **Dr Rudiger Woscholski**