LOWE SYNDROME TRUST PRESS RELEASE

April 2012

The Lowe Syndrome Trust awards a Lowe Syndrome research grant of £80,612.00 to Dr Martin Lowe at Manchester University.
The genetic basis for Lowe Syndrome is a defective gene OCRL1 that results in the deficiency of an enzyme Phosphatidylinositol 4,5-bisphosphate-5-phosphatase (OCRL1).Lowe's oculocerebrorenal syndrome is a disorder affecting the brain, eyes , kidneys and bones.

Dr Lowe said "I am delighted to receive this grant from the Lowe Syndrome Trust. The Trust is doing an amazing job in supporting research into Lowe syndrome, a severe disorder that affects the brain, eyes and kidneys of young boys. Impairment of kidney function ultimately leading to renal failure is a major cause of morbidity in Lowe syndrome patients. Unfortunately, we currently have a poor appreciation of the mechanisms involved. To better understand how the renal defects of Lowe syndrome are brought about, we have generated a zebrafish model that appears to faithfully recapitulate the human disease. We will use this model and exploit existing technologies to investigate the mechanisms responsible for the aforementioned kidney defects. Information gained from these studies will then be used to generate a reporter zebrafish strain for high-throughput screening for drugs to treat Lowe syndrome."

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The Lowe Syndrome Trust
Registered charity 1081241
0207 7948858
www.lowetrust.com