

# LOWE SYNDROME TRUST

December 2009



The Lowe Syndrome Trust was founded in March 2000 to raise funding to support vital research into Lowe Syndrome, an incurable children's disease. Lowe Syndrome is a devastating disease, affecting the brain, eyes, kidneys, bones and muscles. Children are born with cataracts leaving the child blind or partially sighted, other symptoms include weak muscles, mental and physical impairment, kidney wasting, scoliosis, arthritis, swelling of the joints and neurological aspects of the disease include seizures. Sadly, the life expectancy for Lowe children is short due to the severity of the disease. 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.



*Care today... Cure tomorrow*

The Lowe Syndrome Trust  
Registered charity 1081241  
0207 7948858  
[www.lowetrust.com](http://www.lowetrust.com)

## Research funded to date includes:

Great Ormond Street Children's Hospital  
Imperial College London  
Dundee University  
University College London  
Institute of Ophthalmology  
Addenbrooke's Cambridge  
Manchester University  
Purdue University (USA)  
University of California (USA)  
Howard Hughes, Yale University (USA)  
University of California (Berkeley) USA