

LOWE SYNDROME PRESS RELEASE

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The Lowe Syndrome Trust awards a Lowe Syndrome research grant of £79,000 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 second grant from the Lowe Syndrome Trust. Lowe syndrome (LS) is an inherited disorder that primarily affects the brain, kidneys and eyes. LS patients also have problems with blood clotting and tend to have a fair complexion, indicating reduced pigmentation. Lowe syndrome arises from mutation of a single gene, called OCRL1, but how this leads to the symptoms of the syndrome is currently unclear. To better understand the mechanisms involved, we have generated a transgenic zebrafish lacking OCRL1. Interestingly, these zebrafish have reduced pigmentation, as seen in humans. This application aims to investigate how this lack of pigmentation is brought about. We hypothesise that defects in the formation of a special type of compartment inside the pigment cells of the body are responsible. This will be investigated using both zebrafish embryos and human tissue culture cells. Our findings will have broad significance, since the pigment containing compartments are analogous to related compartments found in most cells of the body, including those of the immune system that participate in blood clotting. It is hoped in the future our studies will allow better design of therapeutics to treat LS. The zebrafish line we have generated is amenable to high throughput screening, and monitoring pigment offers a rapid and convenient screening method for identifying new drugs.



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