

LOWE SYNDROME PRESS RELEASE

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The Lowe Syndrome Trust awards a Lowe Syndrome research grant to R Claudio Aguilar of Purdue University USA and Professor Philip Beales Institute of Child Health UK

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.



R Claudio Aguilar said “Thanks to funds provided by the Lowe Syndrome Trust, the Aguilar (Purdue-US) and Beales (UCL Institute of Child Health-UK) labs discovered striking similarities between the defects displayed by cells from patients suffering of Lowe Syndrome and other developmental diseases such as Bardet-Biedl syndrome. These findings suggest that these patients have defects in common biochemical pathways.

The current project aims to test the existence of a common biochemical pathway to these illnesses that can be targeted by therapeutics. This innovative view can have a major impact on the patients' well-being. Specifically, breakthroughs in terms of discoveries or design of therapeutic approaches made in the field of other developmental diseases could be immediately capitalized by researchers working on Lowe syndrome.

Therefore, drugs capable of counteracting cellular defects in other developmental diseases could be beneficial for Lowe patients as well. Specifically, the effect of agents proven to suppress the cellular deficiencies of Bardet-Biedl syndrome will be tested on Lowe syndrome patient's cells.

In summary, this project will set the foundations for the synergistic action of investigators working on different Cerebro-Renal diseases. In fact, this proposal is an example of collaboration between one lab focused on Lowe Syndrome and another on Bardet-Biedl Syndrome.

The Lowe Syndrome Trust
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