Might fish provide Lowe-down on boyhood disease?

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Dr. Martin Lowe and Oscar, 14, who suffers from Lowe syndrome. Credit: Ed Swinden

Scientists have been awarded £72,000 to study zebrafish in a bid to understand the causes of an incurable genetic disorder in humans. The University of Manchester team will use the model organism to investigate Lowe syndrome, an inherited complaint affecting only boys.

“Lowe syndrome is a rare disorder that produces cataracts of the eyes, defects in brain development and kidney problems in young male sufferers,” said Dr Martin Lowe, who will head the research.

“Life expectancy is short due to complications associated with the disease, which can cause blindness, arthritis, rickets, mental impairment,

development delay, tooth and bone decay and kidney failure."

The research – funded by the Lowe Syndrome Trust – will focus on one particular gene, OCRL1, which scientists have identified as being a key factor in the cause of the condition.

"Lowe syndrome arises from a mutation in OCRL1, which is a gene found on the male X-chromosome involved in degrading fat-soluble molecules in the body called lipids," said Dr Lowe, who is based in the Faculty of Life Sciences.

"Although significant progress has been made to increase our understanding of OCRL1, we still do not know what processes it regulates. Furthermore, we have not been able to deduce how loss of OCRL1 brings about the physical changes associated with Lowe syndrome."

One of the difficulties earlier studies have faced is finding a suitable model system to explore the mechanisms underlying the disease. But in a pilot study, Dr Lowe and his team found that OCRL1 works in a similar manner in zebrafish as it does in humans.

He said: "Zebrafish offer a number of advantages over other model systems and we plan to extend our earlier analysis to further scrutinise the role of OCRL1 in development, focusing initially on the brain but also examining the other tissues affected in Lowe syndrome.

"In the long term it is hoped that zebrafish will serve as a model system for experimenting with chemicals that suppress the symptoms of Lowe syndrome in the hope of one day finding a cure."

The research is being funded by the Lowe Syndrome Trust, which was set up in June 2000 by Lorraine Thomas after her son, Oscar, now aged

14, was diagnosed with the condition in 1999.

No government support or UK research of the syndrome was available at that time and, for the last seven years, Lorraine has devoted her life to raising money for the charity.

Lorraine said: "The Lowe Syndrome Trust is delighted to award a grant to The University of Manchester to further research into this rare disease. Sadly, due to lack of awareness and funding, many children suffering from this disorder only live until their teenage years.

"The objective of the Trust is to fund medical research that will eventually lead to the development of drugs to better regulate the metabolic imbalance of the disease and eventually find a cure."

Since starting the charity Lorraine has persuaded many celebrities to back her cause, including television presenter Jonathan Ross.

Jonathan said: "As a trustee I am delighted that we are able to fund the Manchester project. We hope that this research will entice more interest into the disease from research scientists worldwide."

Source: University of Manchester