Scientists study the behaviour of cells from patients suffering the Lowe syndrome.

A team led by Dr. Claudio Aguilar at Purdue University (Indiana, USA) is set to study the cellular consequences of a genetic disease called Lowe syndrome. This illness is caused by a deficiency in an enzyme called OCRL1 that leads to developmental defects primarily affecting eye, brain and kidney function. The research, intended to provide clues about the still unknown mechanisms of the disease, will be funded by the UK-based Lowe Syndrome Trust.

"Our laboratory recently found that cells from Lowe syndrome patients are deficient for crawling and spreading on biological surfaces. Since these processes play a crucial role during embryo development, we believe that this faulty behaviour may contribute to the onset of the disease. Thanks to the support provided by the Lowe Syndrome Trust, we are going to be able to investigate the causes of these abnormalities".

"Biochemical experiments conducted by our team further indicate that OCRL1 interacts with the cellular machinery that dictates how cells relate to their environment. Thus, our research will also be directed towards gaining insight about how patient cells sense their surroundings and absorb nutrients. We will assess the functionality of *intake* routes in patient cells as these paths are key to the success of therapeutic countermeasures".

"This research, made possible by generous support provided by the Lowe Syndrome Trust, will help us to gather precious information about the cellular manifestations of this illness. Ultimately, we hope that a better understanding of the underlying mechanism will help to design new therapeutic approaches to fight this debilitating disease".


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