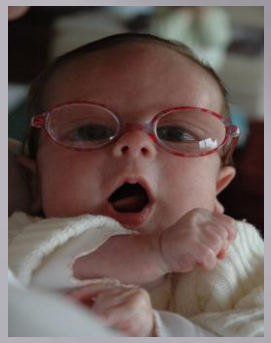

LOWE SYNDROME TRUST PRESS RELEASE MAY 2012

The Lowe Syndrome Trust awards a Lowe Syndrome research grant of £84,000 to Professor Robert Kleta , together with Professor Robert Unwin and Dr Detlef Bockenhauer at The Royal Free Hospital/UCL/Great Ormond Street Children's Hospital

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.

Professor Kleta said "**The study of rare diseases helps to understand how our body works and how we can help making it better for the affected individual. Even though significant progress in such understanding has been made, especially within the past 10 years (completion of the Human Genome Project), many important details are still far from being understood.**

Such an important medical problem is Lowe syndrome. Patients unfortunately develop a special form of kidney failure (renal Fanconi syndrome) which can make their bones weak and prone to fractures, unfortunately later the kidneys can fail all together. So far, many approaches have been made including genetically modified mice, which have been helpful, however yet not sufficiently enough to understand why the kidneys fail.

In order to understand this important and potentially life threatening problem related to Lowe syndrome, i.e., failing kidneys with need for dialysis or transplantation, we suggest to study also rare genetic forms of such kidney failure, familial renal Fanconi syndromes and their molecular relationship to Lowe Syndrome.

In our specialised clinics we see Lowe Syndrome patients, patients with similar kidney problems as in Lowe syndrome and other familial cases of Renal Fanconi syndromes. We have done extensive genetic studies in families with inherited Renal Fanconi syndromes to identify the genes at fault and were successful in doing so. However, just identifying the genetic defect is not really helpful for the patients. We now propose to, in collaboration with experts within London, the UK and the rest of the world, to work with a PhD student on understanding what exactly is going wrong with the body, i.e. the kidney in particular and how this relates to the development of kidney problems in patients with Lowe syndrome.

Understanding what goes wrong in the kidney in these patients will be instrumental to then understand why things go wrong in the kidneys of patients with Lowe syndrome, because it looks alike and is clearly somewhat related to each other. Potential and likely outcomes include more precise diagnostics, application of either known or new medicines to alleviate or cure their problem, potentially even to prevent developing it depending on the exact nature of the problem"

Professor Robert Kleta

Professor Robert Unwin

Dr Detlef Bockenhauer

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