

## Jonathan Ross, Trustee of the Lowe Syndrome Trust presents research grant to Addenbrooke's Hospital, Cambridge.

5th November 2005

Jonathan Ross, Trustee of the Lowe Syndrome Trust, a small voluntary charity, today met Dr Anthony Norden, from the Department of Clinical Biochemistry Addenbrooke's Hospital.

Jonathan was delighted to hand over a cheque to Dr Norden which will enable research to continue in the underfunded area of genetic disorders such as Lowe Syndrome.

Anthony has worked in the field for about 15 years and made steady progress towards understanding kidney disease in these patients. But for the help of charity LST, work would have almost ground to a halt and it would be impossible to develop the recent advances made. Hopefully understanding the basic process of kidney protein handling will help develop treatments to protect Lowe patients from worsening kidney disease and even from developing it in the first place'.



Lowe Syndrome is a rare incurable disorder that affects only boys and produces congenital cataracts in the lens of both eyes, muscle weakness, weak bones, kidney and brain development problems. Sadly life expectancy is short. The disease was first recognised in 1952 by Dr Charles Lowe, and is caused by a gene mutation which makes a defective version of an enzyme named OCRL1, which is needed for normal function of tissues like the lens, brain and kidney, although the reasons for this are still quite unclear.

The Lowe Syndrome Trust was set up by Lorraine Thomas as a voluntary charity in June 2000 when her son, Oscar (then aged 5), was diagnosed with the disease.



*Mrs Thomas, chair of the Trust says: "We were devastated when Oscar was diagnosed with the syndrome at the age of 5. All of the children are partially sighted or blind due to cataracts and some never ever walk. Even Lowe children (boys only) who are doing quite well with the disease, sadly deteriorate with the condition and most die in their teens. I am so grateful for the research being carried out by Dr Norden. The aim of the Trust is to raise awareness and funding for research projects that will one day lead to treatments and some increased hope for these boys".*

*Jonathan Ross said "I have been with the charity since its official launch back in 2000 at one of my favourite events – a Ladies Luncheon! I have seen the amazing results in raising awareness of the disease and funding for medical research.. This will be the sixth project we have funded in five years, which is a tremendous achievement. I hope, as do all the families affected, that it will lead to better lives for the children diagnosed."*