



Beefy's Charity Foundation & the BDFA give boost to Batten disease research

Batten disease is the most common inherited neurodegenerative disease of children and young adults in the UK. It develops in a healthy child, and causes progressive deterioration and loss of vision as well as epilepsy and a gradual decline in cognitive and physical abilities. There are currently no effective treatments available, and it is always fatal. UK researchers from the UCL Institute of Ophthalmology and the MRC Laboratory for Molecular Cell Biology are currently working to improve the sight of affected children. The BDFA already fund a graduate PhD student working with Dr. Sara Mole, who leads research on Batten disease at UCL, and Prof Robin Ali at the UCL Institute of Ophthalmology. This team is developing a treatment that involves inserting healthy copies of the missing Batten disease gene into the cells of the retina to help them to function normally.

Dr Sara Mole says *"This work is a culmination of twenty years research on Batten disease which began by identifying the first gene causing this disease in 1995, and thirteen more since then. We are still working to understand what these genes do but that does not stop us from developing new ways to bring therapies to help those families affected by this devastating and distressing disease"*.

The BDFA are committed to continuing to support this vital work to bring the goal of a treatment closer. The generous funding from Beefy's Charity Foundation has allowed us to continue to fund this work for a further year.

Andrea West (Chief Executive of the BDFA) comments:

This project is a high priority for us as a Charity and the generous funding from Beefy's Charity Foundation has enabled us to extend the vital work of Dr Mole, and Professor Ali's team as we endeavor to improve the lives of children and families living with this devastating disease.

Professor Robin Ali together with eye surgeon Mr. James Bainbridge have already successfully treated patients with a condition called Leber's congenital amaurosis (LCA), another rare inherited eye disease, in the first clinical trial of its kind in the world.. This treatment has been shown to be safe and can improve sight. The findings were a landmark for gene therapy technology and could have a significant impact on future treatments for many eye diseases, including Batten disease.

Commenting on the findings, Professor Ali said: *"Showing for the first time that gene therapy can work in patients with eye disease was a very significant*

milestone. The first trial established proof of principle of gene therapy for inherited retinal disease and paved the way for the development of gene therapy approaches for a broad range of eye disorders, including Batten disease for which there is no other treatment. Gene therapy is still an experimental treatment and not yet generally available to patients.”

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Notes for Editors

1. Members of the media wanting further information, or to arrange an interview with Professor Robin Ali or Dr Sara Mole, please contact ?? in the UCL Media Relations Office on tel: +44 (0)20 7679 9739, mobile: +44 (0)7990 675 947, out of hours: +44 (0)7917 271 364, e-mail: andreawest@bdfa-uk.org.uk 01252 416110

Please do not use these contact details if you are a member of the public. Information about Batten disease and the role of the BDFA in raising awareness, supporting families and funding research can be found on BDFA web site. www.bdfa-uk.org.uk

2. *The team is supported by funding from the Department of Health, The Wellcome Trust, The Medical Research Council, The Special Trustees of Moorfields Eye Hospital, Batten Disease Family Association (BDFA)*

3. Robin Ali is Professor of Human Molecular Genetics at UCL Institute of Ophthalmology and Head of Division of Molecular Therapy. James Bainbridge is a Wellcome Trust Advanced Fellow at UCL Institute of Ophthalmology and Consultant Ophthalmologist at Moorfields Eye Hospital. Dr Sara Mole is a Reader in Molecular Cell Biology at the UCL Institute of Child Health, with her laboratories in the MRC Laboratory for Molecular Cell Biology.

4. The Batten Disease Family Association (BDFA) is a national charity which aims to support families, raise awareness and fund research into the group of devastating neurodegenerative diseases commonly known as Batten Disease. The BDFA works with children and young adults, families and professionals across the UK. www.bdfa-uk.org.uk

5. The UCL Institute of Ophthalmology and Moorfields Eye Hospital NIHR Biomedical Research Centre was established in April 2007, funded by the Department of Health through the National Institute for Health Research (NIHR). It is one of 12 NHS-university partnerships that have been awarded Biomedical Research Centre status, following an international peer reviewed competition based on an outstanding international reputation for medical research and expertise, and experience of translating that research into the clinical setting.

It conducts translational research designed to take advances in basic medical research from the laboratory to the clinic, enabling patients to benefit more quickly from new scientific breakthroughs.

6. UCL Institute of Ophthalmology is one of a number of specialised research centres linked to UCL (University College London) and is, together with Moorfields Eye Hospital, one of the leading centres for eye research. The Institute scored 5*A (the highest possible rating) in the last Research

Assessment Exercise and is committed to a multi-disciplinary research portfolio that furthers an understanding of the eye and visual system, linked with clinical investigations targeted to specific problems in the prevention and treatment of eye disease. The combination of the Institute's research resource with the resources of Moorfields Eye Hospital, which has the largest ophthalmic patient population in the Western World, opens the way for advances at the forefront of vision research.