

Gene Therapy in Cystinosis: “Development of an in vivo gene repair kit: towards a cure for cystinosis”

In September 2008, we received substantial funding from Cystinosis Foundation Ireland and the HRB to determine if a novel gene repair strategy can be developed as a possible treatment for cystinosis. This gene repair technology was first described in June 2005, and our research laboratories at University College Cork have been working since early 2006 to develop this technology as a tool to correct the gene defect found in some cystic fibrosis patients.

Our initial aim in adapting this technology for cystinosis is to design and synthesise two proteins called ZFNs that can recognise a unique target sequence within the cystinosin gene, and measure the ability of these ZFN proteins to cleave this sequence accurately. Candidate sequences have been identified and ZFN production will commence shortly. We also have been optimising another novel assay to help identify the frequency of gene repair. This work is being conducted as part of a final year Genetics BSc project.

It is hoped to have as many as four undergraduate students working alongside the Cystinosis Ireland-funded PhD student in the laboratory in 2009. Indeed, 10% of UCC's Physiology and Genetics BSc 3rd year students are already undertaking library projects in the area of cystinosis; some may well go on to do some laboratory based research and make a practical contribution to this work.

Whilst too early for any of our research data to be published, Patrick has published details of some of the techniques to be used in the June 2008 issue of the journal *Infection Disorders - Drug Targets*. A book chapter outlining some of our plans is to be published in the 2009.