

JOINT RESEARCH FUNDING ANNOUNCEMENT

For the first time Cystinosis Ireland (CI) and Cystinosis Foundation UK (CFUK) have collaborated to jointly fund important research into cystinosis to a total value of €250,000.

The first research project is entitled **“Analysis and exploration of novel treatment approaches for muscular disease in cystinosis”**, which will be led by Principal Investigator **Professor Elena Levtchenko**. The research project comprises a combined total investment of **€100,000** from Cystinosis Ireland and CFUK (€50,000 each) over the next two years.

The second research project is entitled **“Treating ocular cystinosis by contact lenses”**, which will be led by Principal Investigator **Professor Anuj Chauhan**. The research project comprises a combined total investment of **€150,000** from Cystinosis Ireland and CFUK (€75,000 each) over the next two years.

Please find more details about the projects below:

JOINT RESEARCH FUNDING ANNOUNCEMENT

CYSTINOSIS IRELAND AND CYSTINOSIS FOUNDATION UK JOIN FORCES TO CO-FUND IMPORTANT RESEARCH TO ANALYSE AND EXPLORE NOVEL TREATMENT APPROACHES FOR MUSCULAR DISEASE IN CYSTINOSIS

Cystinosis Ireland and Cystinosis Foundation UK (CFUK) are delighted to announce they will jointly fund a research project entitled **“Analysis and exploration of novel treatment approaches for muscular disease in cystinosis”**, which will be led by Principal Investigator **Professor Elena Levtchenko**, Professor, Department of Pediatric Nephrology, University Hospitals Leuven and KU Leuven, Belgium and co-applicants **Dr Rik Gijssbers**, Laboratory for Viral Vector Technology & Gene Therapy, Department of Pharmaceutical and Pharmacological Sciences, KU Leuven, Belgium; **Professor Maurilio Sampaolesi**, Translational Cardiomyology Lab, Department of Development and Regeneration, KU Leuven, Belgium; **Professor Liesbeth De Waele**, Pediatric Neurology, Department of Development and Regeneration, KU Leuven, Belgium; **Dr Mirian Janssen**, Department of Internal Medicine and Metabolic Diseases, Radboud University Medical Centre, Nijmegen, NL; **Professor Bert van den Heuvel**, Department of Pediatric Nephrology, University Hospitals Leuven and KU Leuven, Belgium.

Cystinosis is a very rare inherited genetic disease that causes the build-up of cystine, an amino acid which is normally present in very small amounts in every single cell of a healthy person. The excess cystine forms sharp crystals that damage the body’s cells. Many of the body’s organs are affected by cystinosis including the kidneys and the eyes in particular. In more recent years, muscle weakness has become an increasingly important complication experienced by adult cystinosis patients who are now living into their 20s and beyond. The condition develops even when patients adhere well to the cysteamine drug regimen. The muscle weakness is progressive and has a significant impact on daily life, ranging from being unable to do

basic daily tasks (such as opening jars, dressing etc) to serious life threatening difficulties in swallowing and breathing.

The causes of muscle weakness in cystinosis patients are not well understood. This project aims to develop important research tools that will be used by researchers to study the cellular, molecular and physiological mechanisms that cause muscle weakness in cystinosis patients. These tools and the scientific knowledge that they will help to generate, will subsequently be used to look for new therapies that might address this specific aspect of the cystinosis disease.

The research project comprises a combined total investment of €100,000 from Cystinosis Ireland and CFUK (€50,000 each) over the next two years.

In selecting this project for co-funding, the Boards of Cystinosis Ireland and CFUK agreed that this is a scientifically significant research study, focused on an aspect of the cystinosis disease that is extremely relevant and important to the cystinosis community.

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CYSTINOSIS IRELAND AND CYSTINOSIS FOUNDATION UK JOIN FORCES

TO CO-FUND IMPORTANT RESEARCH INTO TREATING OCULAR CYSTINOSIS USING CONTACT LENSES

Cystinosis Ireland and Cystinosis Foundation UK (CFUK) are delighted to announce they will jointly fund a research project entitled “**Treating ocular cystinosis by contact lenses**”, which will be led by Principal Investigator **Professor Anuj Chauhan**, Professor, Department Head Chemical and Biological Engineering, Colorado School of Mines, USA, and co-applicant **Professor Uday Kompella**, Professor Pharmaceutical Sciences, University of Colorado, Denver, USA.

Cystinosis is a very rare inherited genetic disease that causes the build-up of cystine, an amino acid which is normally present in very small amounts in every single cell of a healthy person. The excess cystine forms sharp crystals that damage the body’s cells. Many of the body’s organs are affected by cystinosis including the kidneys and the eyes in particular. The treatment of ocular complications in patients with cystinosis is particularly challenging.

Oral cysteamine, the only current treatment available to cystinosis patients, does not cross into the cornea and cannot, therefore, prevent the accumulation of cystine crystals in the eyes. As a result, cystinosis patients are currently required to observe an onerous eye-drop dosing regimen (between 4-12 times a day depending on the type of eye-drops used) from a very young age and throughout their lives in order to reduce the levels of cystine in the eye. In addition, the preservatives used in the current formulations cause significant eye irritation which also make their use painful and difficult. Not surprisingly, compliance with this regimen is usually poor despite the potential for very serious consequences which include severe photophobia and even blindness. Added to these difficulties, current eyedrop/geldrop formulations must be stored in a fridge for a maximum of just 5 days. Thus, storage and cost are major hurdles.

As patient-focused charities, both Cystinosis Ireland and CFUK are acutely aware that many patients (particularly young children) and their families struggle to adhere to the current eyedrop therapy.

Professor Chauhan’s and Kompella’s project, which involves a combined total investment of €150,000 from Cystinosis Ireland and CFUK (€75,000 each) over the next two years, aims to develop a more effective system to deliver cysteamine treatment to the eyes using a patented contact lenses technology developed by Professor Chauhan. These contact lenses already show potential to be superior to the current eye drops/geldrops in delivering cysteamine to the eyes more effectively and with reduced side effects. In addition, the cysteamine-containing contact lenses are capable of being stored at room temperature for extended periods of time.

In selecting this project for co-funding, the Boards of Cystinosis Ireland and CFUK believe that Professors Chauhan’s and Kompella’s research will be very important step forward in addressing a key strategic objective for both organisations.

This proposal builds upon a number of previous research projects led by Professor Chauhan that have been supported by Cystinosis Ireland including a major co-funded research project (funded jointly by the

Irish national health research funding agency and Cystinosis Ireland) and more recently a 100% Cystinosis Ireland funded research project.

What is Cystinosis?

Cystine is a naturally occurring amino acid that is present in very small amounts in every single cell of a healthy person. When large amounts of cystine accumulate, they form crystals which can cause damage to the cells. Healthy people have special mechanisms in each cell that remove cystine when it becomes too much. However, people with the very rare disease cystinosis have a defect in their genes which means that these mechanisms do not work. This results in a lifelong illness for persons with cystinosis where, without treatment, cystine accumulates to dangerous levels resulting in crystals forming in the cells causing damage to many organs and tissues. The kidneys and eyes are especially affected by damage caused by cystine crystals but other organs and tissues such as muscles, thyroid, pancreas, testes and central nervous system are also affected.

About Cystinosis Ireland

Cystinosis Ireland was founded in 2003 and is an Irish registered charity, created by volunteers – family members and family friends of those living with cystinosis. Its purpose is dedicated to raising money to support research in cystinosis both in Ireland and all over the world. The charity also provides ongoing support and assistance to people living with cystinosis and in particular to the families of newly diagnosed children. In addition, Cystinosis Ireland also plays an important role in advocating for and providing support to those living with cystinosis on the island of Ireland.

Cystinosis Ireland is one of the main patient organisations in Europe driving research into this rare disease. Since its establishment, Cystinosis Ireland has supported research projects focused on all aspects of this disease to the value of over €2.6 million. These research projects focus on the advancement of treatments for cystinosis, an acceleration of our basic understanding of the cystinosis disease mechanisms at molecular, cellular, organ and whole body levels and developing new disease models.

Cystinosis Ireland is a founding member of Cystinosis Network Europe. In April 2020, on behalf of CNE, Cystinosis Ireland hosted the first ever Virtual International Cystinosis Conference for the global cystinosis community. The one day conference attracted an audience of more than 600 participants from 49 different countries in 20 time zones across the world and was simultaneously translated into 7 different languages. Cystinosis Ireland is also host of the Dublin Cystinosis Workshop which for its 6th and 7th editions in 2020 and 2021 were held virtually. This scientific workshop attracts scientific researchers, academics, clinicians and other professionals from countries from New Zealand to Europe and the Americas who shared their latest research and developments in the field of cystinosis.

Through its partnerships, workshops and international conferences, Cystinosis Ireland shares research findings, discusses drug access programmes, reviews challenges being faced by the greater community and works towards finding a cure for this challenging rare disease.

About Cystinosis Foundation UK (CFUK)

CFUK was founded in 1998 by Jonathan Terry. In concert with the Trustees that he assembled, Jonathan established the aims for the Foundation and wrote the Constitution. CFUK provides support to all diagnosed with cystinosis, their families and relatives; provides relevant, accessible, and up to date information about the disorder; and works with cystinosis related charities worldwide to co-ordinate research support, maintain awareness of new treatments and seek to understand more about metabolic disorders in general.

CFUK achieved charity status in March 1999 and a launch conference was held that April in Leeds.

Since that time significant funds have been raised by the Foundation to support research related to cystinosis, many patients and parents have been helped with advice and support, and awareness of cystinosis among the medical community has been raised.

Since 2004, CFUK has raised over £1 million towards the exciting pro-drug research started by the late Professor Roz Anderson at the University of Sunderland. This pro-drug could have a huge impact on the treatment of cystinosis and CFUK continues to work closely with Professor Herbie Newell who is now managing the project.

CFUK has low overheads and is staffed solely by volunteers who work around full-time jobs, families and other commitments to deliver the key aims of:

IMPROVING patients' outcomes, treatments and research

INFORMING patients, families and healthcare professionals

SHARING stories, experience and answers.