cystinosis ireland

30 Merrion Square North, Dublin 2, D02 VE40
01 678 5758
www.cystinosis.ie
facebook.com/cystinosisIrl
twitter.com/CystinosisIrl
Instagram.com/cystinosisireland
mail@cystinosis.ie

Research Summary:

To improve life for people with cystinosis, we need two things:

1. A better version of the current treatment, cysteamine, something easier to take and with fewer side effects.
2. Extra therapies that target other damage happening inside the cells that cysteamine alone cannot solve, but which still lead to kidney failure over time.

To address this, Professor Newell and the late Professor Roz Anderson (University of Sunderland, UK) developed **CF10**, an improved form of cysteamine. CF10 works just as well to remove cystine from cells but avoids many of cysteamine's biggest problems, including bad taste, smell, and stomach irritation. CF10 can potentially be given at much higher doses, increasing its potential benefit.

Earlier research from our group showed that cystinosis cells have a fault in a waste-clearing system called autophagy, which cysteamine does not fix. However, when we combined cysteamine with Everolimus (an existing FDA-approved drug that boosts autophagy), all these defects were corrected in the lab.

The goal of this study was to test whether combining CF10 with Everolimus would protect the kidneys better than CF10 alone in a rat model of cystinosis.

In our research, we first compared CF10 to cysteamine in cystinosis rats. We tested two ways of giving the drugs, either in jelly pills or directly into the stomach. Both methods worked equally well at lowering cystine levels in the kidney. At a lower dose, CF10 was just as effective as cysteamine at reducing cystine, improving kidney function, and reducing signs of Fanconi syndrome (a complication of cystinosis that causes the kidneys to lose nutrients and minerals). Importantly, CF10 achieved this with fewer side effects. These results played a key role in helping move CF10 into clinical trials in people with cystinosis.

We then tested a higher dose of CF10 given daily for six months, which continued to show good results. At the same time, we also studied Everolimus, a drug that can improve kidney health in different ways. At first, when given at a higher dose, Everolimus helped but also caused problems like weight loss and slow wound healing. By adjusting to a lower daily dose, we were able to keep the benefits while avoiding these side effects.

Finally, we tested the combination of high-dose CF10 with low-dose Everolimus. This combination worked better than CF10 alone. It lowered cystine more effectively, improved Fanconi syndrome symptoms, and offered stronger protection to kidney tissue.

In summary:

- CF10 is a highly promising new treatment for cystinosis.
- Combining CF10 with Everolimus could offer even stronger protection against kidney damage and improve long-term quality of life for patients.

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- A Phase I clinical trial of CF10 in patients is now approved and patient treatment is expected to start by 2028.

Scientific outputs:

Publications:

There are two publications in preparation based on this work.

Clinical trial:

The results from objectives 1 and 2 were used in the application to begin a CF10 clinical trial in the UK under the guidance of Prof Herbie Newell. Funding for this Phase I trial has now been approved, and patient treatment is expected to start by 2028.

Oral presentations:

This work was presented to the cystinosis community and scientists at a number of conferences including;

- Cystinosis Research Foundation, Day of Hope - invited speaker April 2024 -Newport Beach, California
- Dublin Cystinosis Ireland Workshop (DCW), invited recorded speaker April 2024 - Dublin
- Clinical Trial Steering committee meeting for CF10 - April 2024 - Sunderland
- Cystinosis Network Europe invited speaker, July 2024 - Manchester
- Stem Cell and Regenerative Medicine conference - Oral presentation - October 2024 – Berlin
- Invited to give a seminar at KU Leuven – September 2024 – Belgium.
- Invited to speak at Cork University Hospital Nephrology Journal Club - December 2024 - CUH, Cork

PhD awarded

Pang Yuk Cheung successfully defended her PhD thesis on September 10th, 2025. Some of her PhD work resulted in the work presented here.