

Cystinosis Research - heading in to a cure

The search for a cure for cystinosis is not unlike a game of golf. Full of great plans and aspirations, but often takes a lot longer than planned, and the result is never quite what one expects.

The first nine holes of cystinosis research have been tricky. We're over par, and it's taken a long time. That said, we've scored a couple of birdies, not least finding the gene which causes the disease, and Cystagon™, the drug which saves the lives of children with this disease.

However, as we head in on the back-nine, we face a long and complex path to the club house in our quest for a cure for cystinosis which would prevent renal failure, and the current need for medication every six hours, every day, for life.

In the last 18 months, the Cork-based Cystinosis Ireland funded research group headed by Dr Patrick Harrison have been working towards the development of a gene repair kit for cystinosis.

There are many challenges to repairing a gene, the first is be able to find it within a cell that also contains 25,000 other genes. The second is to target it without damaging those other 25,000 genes.

To date, Patrick's group have made good progress on these first two steps, and have set up collaborations with groups in Italy, Belgium and the Netherlands at the 2nd International Cystinosis Research Symposium in University of California, Irvine, in April this year to make further advances.

Also at this meeting, the Cystinosis Gene Therapy Consortium was formally established, to investigate ways to add a functional copy of the gene to a patient's bone marrow cells as a potential treatment. Patrick's research could potentially enhance the goal of the consortium by offering the possibility of permanently repairing these cells, rather than simply patching them up prior to transplantation.

With recent advances, the potential for a cure is no longer just a

dream, it will be a reality. The question is no longer "if", it is becoming "when"...

... but "when" requires more collaboration on research and funding.

Patrick has been invited to the speak at the forthcoming 6th International Cystinosis Conference in Italy, in September.