PROFILE

James Ennis

LIVING WITH CYSTINOSIS

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My name is James Ennis. I am 29 years old and I live with cystinosis. I have to take 30 tablets a day. The most important one is Cystagon which I must take every 6 hours to keep the cystine levels from building up. I also use Cysteamine eye-drops four times daily to prevent crystal build up in my eyes. I have had a kidney transplant.

I am a qualified joiner and worked in kitchen manufacturing. I had to give up work because of bad leg pain which severely affected my mobility. I have had surgery and physiotherapy but I am no better and now use a walking stick.

I try not to let it get me down. I became involved with Cystinosis Ireland around two years ago and became the Northern Ireland Representative. I am also a member of the Cystinosis Community Advisory Board, which is part of Cystinosis Network Europe. The thing that gets me up every day and keeps me positive is knowing that there is a lot of research being done into better treatments and cures for cystinosis worldwide. I can see a glimmer of hope for the cystinosis community.

RARE DISEASE

About Cystinosis

Cystinosis is caused by an inherited, recessive gene where the amino acid, cystine, accumulates in all organs, muscles and bones. Cystinosis is ultra-rare and has an incidence of 1:250,000. Children are usually diagnosed between 6 months and 3 years, but misdiagnosis is common. Patients present with excess thirst and urination, bone pain from rickets and loss of essential nutrients from the kidneys. The kidneys are the first organs affected, with kidney transplant inevitable, even with good treatment adherence. The sooner a child is diagnosed and the quicker they start treatment, the better the outcome.

The available current treatment slows deterioration but does not halt it. Two formulations of the treatment exists: Immediate-release, taken every six hours which means waking up every night and delayed-release taken every 12 hours, but is not yet widely available worldwide. Both formulations cause severe side effects and patients find it difficult to adhere to the regime. Gene therapy is at early stage clinical trial. Cystinosis Ireland (www.cystinosis.ie) supports families, raises awareness and raises money to fund research.