

Fifth International Cystinosis Conference 2008

June 27-28, Dublin, Ireland

This article focuses on the conference itself, taken from the perspective of an attendee. For a more medical orientated review please see the accompanying conference review by John Terry. The views stated in this article are the views of individuals and as such should not be taken as medical advice.

On June 27 & 28, 2008, over 200 delegates, ranging from medical practitioners, researchers, Cystinosis sufferers, their families and charity organisers, attended the Fifth International Cystinosis Conference at the Hilton Hotel, in Dublin, Ireland. Delegates originated from all corners of the globe – Britain, Australia, America, Egypt and Mexico to name but a few countries.

The key aim of the conference was to allow individuals and organisations to share their knowledge across the Cystinosis community to further our understanding and treatment of the condition. This year the theme of the conference was “from dreams to reality”. In the 1960s, Cystinosis was almost unheard of, with little, if any, effective treatment available and 10 years ago, at the first international conference, no abstracts for original research were submitted.

How much further on are we today?

This year there were 20 abstracts and to date over 1,000 papers have been written on Cystinosis. We now have an effective, if less than perfect, treatment in Cysteamine and, although no cure has yet been found, there is substantial research being undertaken to improve the treatments available and ultimately work towards a cure. Despite Cystinosis being so rare, the dreams are slowly becoming a reality.

Day One



Dr Jerry Schneider

Whilst there was pre-conference registration on the Thursday evening, the conference formally started on the Friday morning with an opening lecture by Dr Jerry Schneider, USA. Dr Schneider has been at the forefront of Cystinosis research since the 1960s and gave a brief history of his involvement in the field and available treatments.

His presentation opened with a stark image of a three-year-old Cystinotic taken in the 1960s showing the affects of Cystinosis when left untreated. It reminds us of how far we really have come, and how fortunate we are to have Cysteamine today. Dr Schneider stressed the importance of taking Cysteamine regularly and at the correct dosage for it to have a positive effect.¹ He also discussed his current research with Dr Dohill to

develop a “delayed release” form of Cysteamine to allow administration every 12 hours (as opposed to 6 hours), which is one of several projects world wide² seeking to improve the administration of Cysteamine. Dr Schneider is hoping for trials in the USA next year. Other projects were also discussed that are looking at curing Cystinosis via the introduction of bone marrow with “non-Cystinotic” genes, although these are in the very early stages.

Following Dr Schneider’s presentation, the conference split into two parallel sessions. “The Basis of Cystinosis” was a highly scientific session chaired by Dr William van’t Hoff³ designed to update professionals on current research. My wife and I attended the family session, “Living With Cystinosis”.

A panel, including nurses, an education psychologist, dietician, physiotherapist, Cystinotic and mother of a Cystinotic, took questions from families about the day-to-day realities of living with Cystinosis. Questions ranged from how to minimise bad breath caused by medicines, the impact of Cystinosis on education, to dealing with siblings of children with Cystinosis. It is impossible to comment on all the questions and answers given during this

¹ A figure of 1.3/g/m²/day every 6 hours was quoted, but please consult you GP for your own needs.

² The Cystinosis Foundation UK is funding research at Sunderland University into pro drugs. For more information see the Sunderland University presentation available at <http://www.cystinosis.org.uk/conference2008>.

³ Dr William van’t Hoff, who works at Great Ormond Street Hospital, London, is a member of the Cystinosis International Committee and a renowned expert in the field of Cystinosis.

lively session, but it was interesting to hear what parents had tried in order to deal with Cystinosis. Even trying simple ideas, such as taking milk, yogurt or pro-biotic drinks before medicines to help reduce vomiting, can make a big difference to daily life.

In the afternoon, the younger people with Cystinosis were invited on a bowling trip, with the strict condition that no parents were allowed to attend. For the rest, a plenary session allowed for 8 oral presentations on original research, with question and answer sessions after each one. Again, in such a short article it is impossible to review all these presentations. However it is worth noting, that whilst most of these presentations were very technical in nature, each was required to include at least one slide that would be more accessible to those not medically trained.

The first day of the conference ended with a poster session from researchers, support groups, industry and others, and a table of professionals were present to speak to families/patients on an individual and informal basis. The poster presentations included one by Roz Anderson's team at Sunderland University and one by Don Cairns' team at The Robert Gordon University in Aberdeen.

The evening was free for us to have time to relax, spend time with others and absorb the day's events.

Day Two

Day two began with "The Family Perspective" consisting of two talks given by Cystinosis sufferer Anne-Claire Panisset, of the Cystinosis Foundation France, and Sue Maguire of the Cystinosis Foundation Ireland who has two daughters with Cystinosis. The aim of the session was to provide an opportunity for professionals to learn from those who live with Cystinosis.

Anne-Claire gave a very honest and brave assessment of herself growing up with Cystinosis. At 18 Anne-Claire was still being seen by a paediatrician and asked to be transferred to the adult services. By her own admission she wasn't as ready as she thought for this responsibility and began skipping hospital appointments and medicines. Within 4 months she lost her transplanted kidney. Anne-Claire knew her meds were important, but at the time it was more important to her that she make her own decisions even if they were wrong decisions. Fortunately, Anne-Claire is doing much better these days, but her story shows the potential difficulties of transitioning between child and adult services.



Anne-Claire Panisset

Sue Maguire's talk focused on experiences of dealing with medical professionals and there were several themes that emerged during the conference that were highlighted in Sue's talk. Hospitals were often sticklers for routine and bureaucracy, with her family often having to go through full admission procedures every time they turned up at hospital. Sue talked of the need for fast track admissions for regular hospital attendees and the grouping of appointments to save disruption to family routine, time off school, etc. She also spoke of the need for professionals to be accurate, to "tell it like it is" – Cysteamine should be taken every 6 hours, not 4 times a day – but also respect the knowledge of patients and their families who live with Cystinosis 24/7, when many medical professionals are not even aware of the condition.

However, Sue's talk was also inspiring as she talked about her family's challenges and two phrases stick in the mind: "*we made Cystinosis fit around our lives, not the other way around*" and "*laughing beats crying*".

The morning continued with parallel sessions for professionals, young persons, and children and parents. The professional session consisted of several presentations on specific aspects of Cystinosis, from childhood medical issues, adult medical issues, academic performance of Cystinotics, specific kidney problems and transplantation. The young persons' session with Dr Gahl from the USA allowed those aged between 13 and early 20's to ask questions on any issue, without the presence of parents. Dr William van't Hoff also led a question and answer session with children and their families in the children's room.



Dr van't Hoff during the children's session.

The final afternoon was a busy one. A plenary session included presentations from professionals on the treatment of Cystinosis, including available treatments, monitoring cystine levels in blood, unusual features in Cystinosis and a review of on-going research around the world by Professor Don Cairns of the Robert Gordon University, Aberdeen.⁴ The conference was also fortunate enough to include a speech from Irish Minister for Health and Children, Mary Harney. This was followed by a keynote speech by Dr William van't Hoff, who also chaired a question & answer session with a panel of experts.



Jean Hotz presents Dr Schneider with a lifetime achievement award

However, one of the highlights of the conference came at the very end, when Jean Hotz of the Cystinosis Foundation USA presented Dr Jerry Schneider with a lifetime achievement award. Jean, whose grandchild has Cystinosis, started the very first Cystinosis Foundation in the 1960s when Dr Schneider first became involved in the field of Cystinosis. Through the hard work of individuals such as Jean and Dr Schneider, our dreams are slowly becoming reality.

That evening saw the conference close with a barbeque – a last opportunity to spend time with new friends before travelling home.

I attended the conference as part of a family holiday to Ireland, with my wife and one-year-old daughter. Childcare was provided by the conference organisers and buffet lunch was made available by the hard working staff at the Hilton Hotel. This meant my wife and I could concentrate on the conference itself with minimal distraction and our daughter enjoyed her time with the other children.

The Fifth International Conference was the first Cystinosis conference I have attended and it was a resounding success from all perspectives, with much of this due to the excellent organisation of the Cystinosis Foundation Ireland. It has been announced that the next conference will be held in Italy in 2010 and we are already considering working our family holiday around the conference again, if possible.

Matt Blackham

Website Manager, Cystinosis Foundation UK

Attendee of the International Conference, Dublin, 2008

⁴ Professor Cairns's project at Robert Gordon University is sponsored by the Cystinosis Foundation Ireland, but also receives financial assistance for equipment from the Cystinosis Foundation UK.