cystinosis ireland

Cystinosis is a rare disease where all organs and muscles deteriorate due to an abnormal accumulation of an amino acid. The kidneys are first affected, causing eventual kidney failure, dialysis and transplant in virtually all cases. Overtime, all the other organs and muscles deteriorate. There is a treatment to slow this deterioration process but it causes severe side effects with many patients finding it difficult and sometimes impossible to take. It must be taken every 6 hours which means waking up children every single night. Better access to treatments is key to finding a way to live with this disease.

What is a Rare Disease?

In Europe, a disease or disorder is defined as rare (or orphan) when it affects less than 1 in 2,000 people. There are more than 6,000 known rare diseases. 80% of these are of genetic origin, are present throughout a person's entire life, even if symptoms do not appear until adulthood, and are often chronic and life-threatening. One rare disease may affect only a handful of patients across Europe, while another may affect as many as 250,000 patients.

"Rare is not so rare – rare disease patients are many"

While an individual disease might be labelled as "rare", the total number of persons in Europe suffering from one of the over 6,000 different identified rare diseases is estimated at over 30 million (6% to 8 % of the European population), which equates to **more than 280,000 people in Ireland living with a rare disease**.

Rare diseases are characterised by a wide diversity of symptoms and signs that vary not only from disease to disease but also from patient to patient suffering from the same disease. Relatively common symptoms can often hide underlying rare diseases, leading to misdiagnosis. Rare diseases not only affect the person diagnosed, they also impact families, friends, carers and society as a whole.

Most rare diseases have no cure, so living with a rare disease is an ongoing learning experience for patients and their families.

What we are seeking:

Access to medicines

- More Government funding made available so the HSE can approve treatments/drugs already available to patients in other countries. These treatments are inaccessible to Irish patients simply because of money reasons.
- A fair and transparent process with appropriate assessment criteria for orphan drugs.
- An accountable person within the DoH/HSE to be responsible for a RD budget (this does not exist at present).
- "The 2013 Act requires the HSE to have regard to both the clinical benefits and cost effectiveness but it does not include provision for a different rule set when assessing orphan drugs." *(reference Parliamentary Questions 397 – 401 and 403 to Minister Simon Harris)*
- Collaboration with other European countries to have joint negotiations with pharmaceutical companies on common and fair pricing.

Soft Opt-Out Organ Donation

- We welcome the Minister Harris's commitment that the Human Tissue Bill will be brought to Committee in this term but are keen to ensure that adequate resources and staffing are put in place to allow "soft opt-out" organ donation and transplant services to be as effective as possible.

Access to genetic services

- Waiting lists are increasing year on year. The overall waiting list is currently in excess of 2800. This in an increase of 400 from 2017.
- The waiting list for people waiting more than 18 months is more than 500, an increase of 200 from 2017.
- The waiting list for 1 year has gone up by nearly 300.
- Records being complied by the Genetics Service indicate that 21 people, and more key relatives, have died while on the waiting list.

Families benefit from attendance at a genetics counselling service whatever the outcome of the consultation. The role of the geneticist is not to effect action but more to inform and allow the family to make their own decisions. More details can be found in the attached report by Dr Sally Ann Lynch.

Oireachtas Champions for Rare Diseases

- We are seeking commitment from Champions for rare diseases to promote the potential for novelty and innovation across all parties and government departments.
- Knowledge and awareness – eg tweeting on Rare Disease Day, 28 February 2019.
- All government departments need to understand the implications of rare diseases and how the innovation and research agenda essential to care for rare disease patients will have benefit across the whole system.

Additional resources

An Easy Guide to Rare Diseases in Ireland, Medical Research Charities Group
http://www.mrcg.ie/assets/68/8868A257-03ED-0FEE-1FF23C3DDFE10E46_document/Easy_Guide_ind.pdf

Wide disparity of clinical genetics services and EU rare disease research funding across Europe
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https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4796048/