Egyptian Group for Orphan Renal Diseases

EGORD

EGORD is a national health group that was established in April 2003. It is the only group of its kind in Egypt. It is a health group dedicated to the enhancement of the lives of all persons affected by rare "*orphan*" renal diseases.

☺ What did we achieve so far for cystinosis?

Cystinosis was the first disease to care for. It all started with a 6 years old girl referred to us diagnosed as Fanconi's syndrome. She was only receiving supportive therapy and no definitive diagnosis was offered to the family. As we have no WBC cystine assay, we had to resort to slit lamp examination for
corneal cystine crystals. Thanks to our pediatric ophthalmologists we managed to diagnose her as nephropathic cystinosis.
That was just the beginning; at that point we felt we need more awareness of the disease among both medical and public communities as well as getting the medications. With the endless support from Prof. Ramzi El-Baroudy, head of Center for Pediatric Nephrology & Transplantation (CPNT), enormous help of Mrs Jean Hotz, president of cystinosis foundation and the care of Mrs Samantha Parker and Dr Tony Zbeidy at Orphan Europe we managed to take our first steps. We had an Arabic brochure about cystinosis in collaboration with Cystinosis Foundation and Orphan Europe.
Later we managed to raise some funds to get the cystagon. As for cysteamine eye drops, many thanks to Dr Willam Gahl for helping us as to how to formulate it and to Dr Maged George for locally formulating the eye drops as a donation from Luna Medicals. Dr William Gahl also helped our ophthalmologist team with the corneal cystine crystal scoring (CCCS) which is currently used to score children while on treatment.

As the word was spreading we managed to diagnose six children living with cystinosis but unfortunately already had CRF or even on dialysis. Our main focus was trying to diagnose patients at an earlier age to prevent or at least retard renal failure. It worked well and we got our youngest kids, 9 and 17 months old with normal renal functions and normal slit lamp (diagnosed with WBC cystine assay by Isa Bernardini at Dr Gahl's laboratory). In addition to those on dialysis, we now have 13 more children with nephropathic cystinosis that started cystagon while still maintaining kidney functions.

EGORD is also encouraging and carrying out research work to study pattern of cystinosis in Egypt.

We keep detecting new patients through Cystinosis Awareness Progtam [CAP], health education and relentless efforts to try and diagnose children at an early age so we can start them on treatment early and save them the whole issue of renal failure and dialysis problems. In a country with over 75 million population and high rate of consanguineous marriages, we expect to find a lot more out there.

We hate to see them off the medication, after what we had achieved so far, just because there is no enough funding. That would be the worst scenario which we wish, hope and would fight not to live it.

Neveen A Soliman, MD

Professor of Pediatrics, Cairo University
Coordinator of the Egyptian Group
For Orphan Renal Diseases (EGORD)
Email: neveen.soliman@gmail.com
neveen.soliman@hotmail.com

EGORD Administration

egord2007@yahoo.com