Cystinosis Patient Journey

ERKNet

European Reference Network
for rare or low prevalence complex diseases

Note: Cystinosis is complex, no clinician can know all symptoms

Note: A well treated child with cystinosis can live a normal life

1 … first symptom...
2 … diagnosis...
3 … more symptom...
4 … surgery...
5 … adulthood...

1. First Symptoms
Glucosuria, but no diabetes. Not all symptoms start at the same time.
Renal Fanconi syndrome causing: Polyuria, vomiting, constipation or diarrhoe, general malaise and fatigue, excessive thirst, growth retardation, rickets.
Photophobia; nutritional problems; "weak" bones; some children stop walking, toilet trained children lose this ability again (because of the amount of fluid).

Needs: Listen to parents, believe what they tell about their children.
Do urine tests (glucose) and blood tests (loss of electrolytes; dehydration) even in very young children.

Ideally: Early diagnosis is crucial.
Treatment to slow down the progress of the disease can be given from day 1 is available and has to be prescribed and paid by insurances in all countries.

2. Diagnosis
Cystine measurement out of white blood cells to be done in a trained laboratory.
Genetic testing.
Newborn screening.

Needs: Give well informed the right diagnosis. Explain about the disease, not only give its name.
Give information about patient organisation and recommend strongly to get in contact.

Ideally: Nephrologic pediatrist cooperates with a specialized interdisciplinary center for cystinosis, but stays the main partner of the patient (family).
No long stay in the clinic, no examinations that are not necessary.

3. More symptoms
Learning disablilities caused by: chronic fatigue, visual-spatial problems, some fine motor skills limitations. BUT: Normal intelligence, other cognitive functions are usually preserved.
Delayed puberty, infertility in men. And many more …

Needs: Care coordination in a multidisciplinary team. This is a metabolic disease with many different health problems involved. Free access to medicines and further treatment.

Ideally:
Psychosocial as well as medical care.
Openness to new ideas is important, therefore accept patients' solutions in dealing with daily life problems

4. Treatment & Surgery
At the moment there is only one medication (Cysteamin) to slow down progression of cystinosis.
Dialysis and kidney transplantation can be avoided for a long time.
Patient can fall dehydrated and creatinine rises very fast again when fluids are restricted after transplantation.

Needs: A multidisciplinary approach is important, including nephrologist, ophthalmologist, internist in metabolic diseases and if required, an endocrinologist, a social worker and/or neurologist.

Ideally: Transition from paediatric to adult care is a precarious process, which is best done in a cystinosis center of expertise. Information on the use of cysteamine is crucial.
Surgery: Think of dehydration first and give fluids intravenous all the time while a patient is not allowed to drink.

5. Adulthood
Pregnancy: women can have children, they are not affected by cystinosis.
Cystinosis can show many severe symptoms in patients not well treated from the beginning.

Ideally:
A multi-disciplinary team of experienced specialists working hand in hand in one clinic seeing many patients is the best for all patients.