Cystinosis Patient Journey

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 paediatrist 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 disabilities 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 (Cysteamine) 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.

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

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

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