Rare Renal Diseases – Common Patient Journey

1. First Symptom
   - Sometimes disease is silent.

2. Diagnosis
   - A high proportion of kidney failure are reaching unnoticed till emergency stage due to lack of systematic survey.
   - Early diagnosis, early treatment and/or renal protection. Patients need to anticipate the progression of the disease.
   - Professionals should consider INFORMATION to PATIENTS and not restrict to information on the disease when diagnosis is announced. Promote clinical survey associate to biobanks to monitor the clinical history on a long-term basis. Promote Patients Engagement in surveys for prognosis development.

3. Monitoring
   - Patients should be informed and prepared as to what to do and how to seek support when crisis occurs.
   - As part of the INFORMATION to PATIENTS a special section on monitoring the disease a glossary of the main indicators with the explanation linked to the renal function with perhaps a differentiation by pathology.
   - Guidelines should encompass special sections for medical crisis and complications.

4. Treatment
   - Pediatric patients on dialysis are mostly rare/genetic diseases
   - Monitor the pediatric patients after transplant
   - Offer to all patients the largest choice possible options for dialysis.
   - Produce INFORMATION TO PATIENTS on Dialysis Pathways (booklet). Better information about structures for holidays.

5. Quality of Life
   - Generalise surveys on rare renal population for QoL:
     - Develop knowledge and adjust for policies that favour a better QoL.
     - Presymptomatic genetic screening
     - Share (and explain) information regarding the risk level of having a child with a severe renal disease before conception.
     - Offer to the parents the access to foetus genetic testing
     - Re-enforce (or create strong) links between nephrologist and obstetricians
     - Train obstetricians on kidney diseases

**Note:** Information to the general population for regular blood and urine sample

**Ideally:** Reduce the time lag before undiagnosed patients are directed towards a specialized/excellence centres.

Raise capacity of General Physicians (GP) to deal with rare diseases (initial training on RD)

Create local regional advisory cells on rare diseases “GPs for RD”, for guiding local physicians and patients in case of suspicion.