Rare Renal Diseases – Common Patient Journey

ERKNet

European Reference Network
for rare or low prevalence complex diseases

... diagnosis ... ... treatment ...

First symptom ... ... monitoring ... ...quality of life...

1. First Symptom

Sometimes disease is silent.

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

2. Diagnosis

A high proportion of kidney failure are reaching unnoticed till emergency stage due to lack of systematic survey

Note: Early diagnosis, early treatment and/or renal protection.
Patients need to anticipate the progression of the disease.

Ideally:
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

Note: Patients should be informed and prepared as to what to do and how to seek support when crisis occurs.

Ideally:
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

Dialysis:
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

Transplantation:
Living donor transplant to be considered as the first choice for ESRD.
Remove legal and practical barriers for cross match donation.
Promote organ donation.
Booklet on INFORMATION TO PATIENTS on Transplant Pathways

5. Quality of Life

.. 6. Pre-symptomatic genetic screening

Ideally:
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