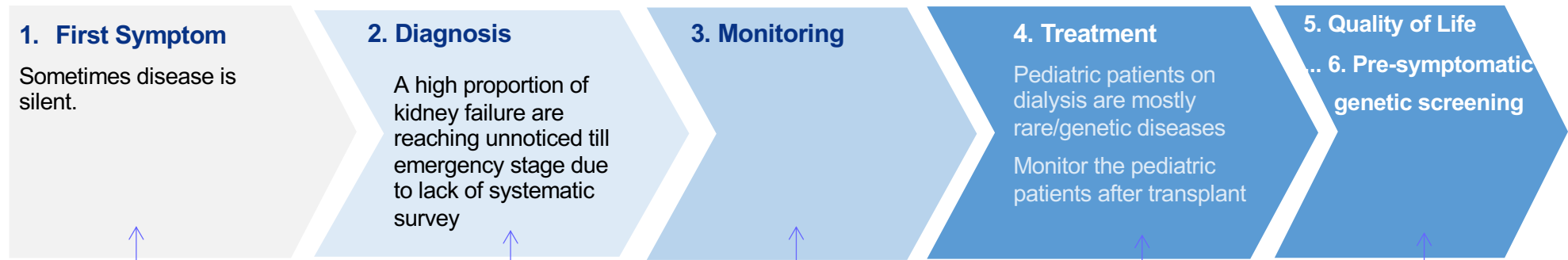


Rare Renal Diseases – Common Patient Journey



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**