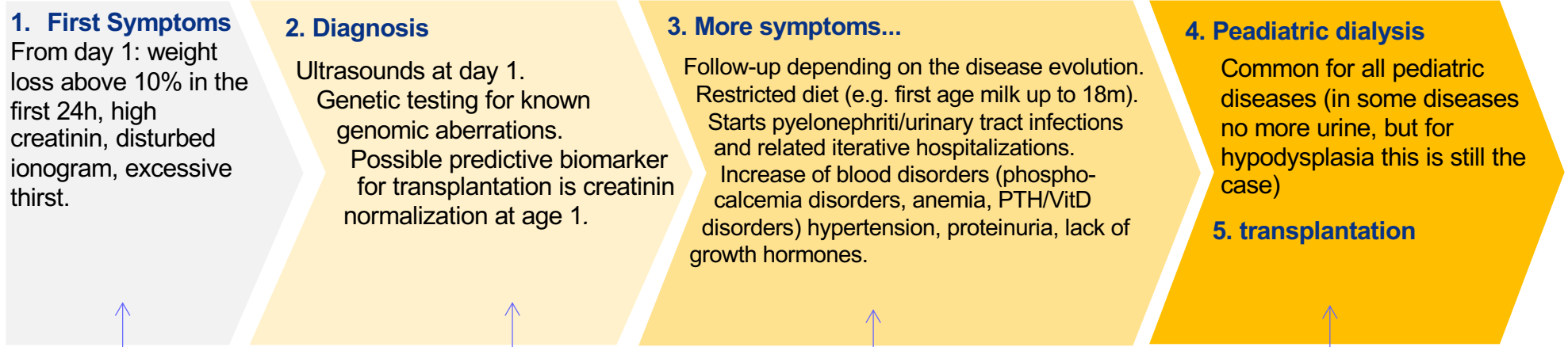
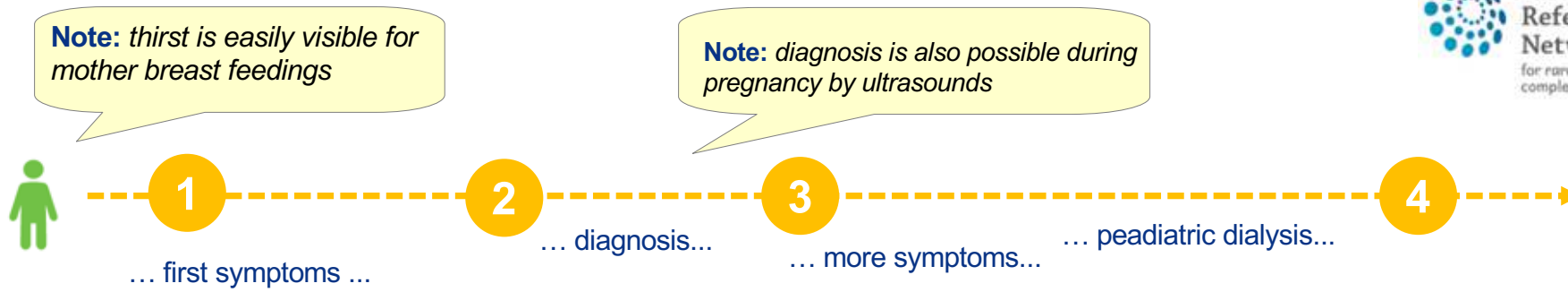


# CAKUT Patient Journey



**1. First Symptoms**  
From day 1: weight loss above 10% in the first 24h, high creatinin, disturbed ionogram, excessive thirst.

**2. Diagnosis**  
Ultrasounds at day 1.  
Genetic testing for known genomic aberrations.  
Possible predictive biomarker for transplantation is creatinin normalization at age 1.

**3. More symptoms...**  
Follow-up depending on the disease evolution.  
Restricted diet (e.g. first age milk up to 18m).  
Starts pyelonephriti/urinary tract infections and related iterative hospitalizations.  
Increase of blood disorders (phospho-calcemia disorders, anemia, PTH/VitD disorders) hypertension, proteinuria, lack of growth hormones.

**4. Paediatric dialysis**  
Common for all pediatric diseases (in some diseases no more urine, but for hypodysplasia this is still the case)  
**5. transplantation**

**Needs:** Train obstetricians and pediatricians from maternity services on renal rare diseases leading to natal CKD and on tracking family history.

**Ideally:** Early diagnosis is crucial to ensure the correct diet and to start medication when needed.

**Needs:** Ensure the ultrasounds is performed by an experimented radiologist. Do not wait to perform genetic testing, if care-givers agreed on it. For ultrasounds follow-up during pregnancy, it would worth to develop a more relevant diagnostic tool, allowing to identify more precisely monitoring. In case of future pregnancy, explain the risk level of having a child with a severe renal disease before conception when possible.

**Ideally:** Psychological support will be needed to parents/family

**Needs:** Minimize as much as possible the follow up visits in reference centers, liaise as much as possible with the local pediatrician and allow patients to perform blood samplings in local laboratories. Ensure as much as possible hospitalization at home and/or in a close hospital from home with a direct connection with reference center. For specific surgery perform it in a reference center. Offer psychological support for the children. Provide educational material on rare kidney diseases to the teacher.

**Ideally:** Accept parents requesting a second opinion/advice on the disease. Free access via prescription to psychologists, to dietiticians, physiotherapies.

**Needs:** Explain the different dialysis

**Ideally:** Maintain as much as possible QoL. Psychological support mandatory.