# WHEN TO CONSIDER LYSINURIC PROTEIN INTOLERANCE (LPI)

Association and severity of symptoms vary greatly, depending on patients and intra-family variability. Secondary urea cycle deficit



#### FIRST EARLY SYMPTOMS

Often occurs when the baby is weaned and when a range of foods is introduced



# **Digestive impairment**

Mainly due to secondary urea cycle deficit

**Recurrent vomiting** 

Chronic diarrhoea

Anorexia, feeding problems

Aversion to high-protein foods

Sometimes antenatal hyperechogenic colon



Rupture in the growth curves for weight and height

Hepatosplenomegaly

**Hypotonia** 



# **EPISODES OF ACUTE/SUBACUTE DEGRADATION**

Can occur at any age, aggravated by catabolism and high-protein meals

# Hyperammonaemia<sup>1</sup>

**Emergency treatment** 

Exacerbation of vomiting, anorexia, nausea

#### Acute neurological disorders:

impaired vigilance, confusion, sleepiness, impaired balance, behavioural issues, tremors, abnormal movements, etc.

Risk of degradation to coma +/- convulsions and risk of death and neurological sequelae



# PROGRESSIVE ONSET OF **SYMPTOMS / COMPLICATIONS**

Sometimes, early onset when diagnosed, or only in adults



### **Growth issues and bone damage**

Failure to thrive, severe osteoporosis (pathological fractur

### Lung damage

Progressive interstitial changes, sometimes severe pulmonary alveolar proteinosis (life-threatening), pulmonary fibrosis



Hepatosplenomegaly, cytopaenia, biological markers of macrophage activation

Predisposition to autoimmune diseases (ANF, anti-DNA antibodies, etc.)

## Kidney disease (adolescents/adults)

Progressive proximal glomerular and/or tubular disease, kidney failure

#### Other

Acute pancreatitis

Psychomotor delay (possible consequence of episodes of hyperammonaemia)

Laboratory: Sometimes high ammonia levels1 (particularly after meals or during decompensation), fluctuating liver cytolysis, possible: cytopaenia (anaemia, thrombocytopaenia), signs of macrophage activation syndrome (hyperferritinaemia, hypertriglyceridaemia, elevated LDH, low fibrin, etc.), coagulation disorders, signs of tubulopathy, microalbuminuria, proteinuria

Thorax X-ray / scan: possible reticular interstitial syndrome



sometimes progressing to kidney failure (frequent in adulthood)



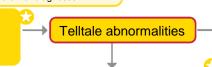
# Lysinuric protein intolerance?

Specialist workup in collaboration with Centre of Excellence

At the same time as looking for other potential differential diagnoses2

Plasma and urinary amino acid chromatography

Urinary orotic acid determination



Confirmatory genetic analysis to be carried out subsequently by a specialist centre

# **Urgent specialist advice from a Centre of Excellence:**

Rare Disease Centre of Reference / Competence

https://www.filiere-g2m.fr/annuaire/

Start the parallel treatment, urgently depending on type of presentation

Refer to the emergency protocols for each symptom and/or disease: https://www.filiere-g2m.fr/urgences

Specialist treatment coordinated by a Centre of Excellence Genetic counselling, family screening in a specialist centre

For more information: PNDS French National Authority for Health - Urea Cycle Disorders (has-sante.fr)



Specialist medical opinion and reference laboratory



