WHEN TO CONSIDER GALACTOSAEMIA

Clinical signs

MOST OFTEN PRESENTS IN THE NEONATAL PERIOD
(in breastfed newborns or those fed formula containing galactose)

Hepatic involvement ★
(progressing to severe acute liver failure)

- Feeding difficulties
- Insufficient weight gain
- Growth retardation
- Vomiting
- Jaundice
- Hepatomegaly
- Lethargy

+/- Neonatal E. coli infection
(more common)

+/- Ophthalmic involvement

- Cataract

+/- Renal involvement

- Proximal tubular involvement

OTHER LATER MANIFESTATIONS/COMPLICATIONS

Gonadal insufficiency due to premature ovarian failure[1]

- Delayed puberty
- Subfertility

Skeletal involvement

- Decreased bone density

Neurological involvement[2]

- Global neurodevelopmental disorder
- Language delay
- Attention deficit/ hyperactivity disorder (ADHD)
- Autism spectrum disorder
- Anxiety
- Depression

Additional tests

Laboratory tests:
Possible hypoglycaemia

↘ Prothrombin time (PT), coagulation factors and albumin

↗ Total and conjugated bilirubin transaminases

Laboratory tests:
Signs of tubulopathy

Specialist assessment

Galactosaemia due to galactose-1-phosphate uridylyltransferase (GALT) deficiency ?

Specialist assessment in collaboration with a Centre for Rare Diseases ★
in parallel with investigation of other possible differential diagnoses[3]

Spot test [4] (screening for congenital galactosaemia using blood on filter paper): Decreased GALT activity
(+ increased intra-erythrocyte galactose-1-phosphate)

Follow-up confirmatory genetic testing by a specialist centre

Urgent referral to a hepatologist and specialist centre for Inherited Metabolic Disorders
Reference and Expert Centre for Rare Diseases:
https://www.filiere-g2m.fr/annuaire/

Simultaneously begin emergency management:
Stop breastfeeding (or galactose-containing formula) as soon as galactosaemia is suspected, without waiting for the results of the spot test

Specialist management coordinated by an Expert Centre

Genetic counselling, family screening in a specialist centre

Further information:
emergency protocols by symptom and/or disease:
https://filiere-g2m.fr/urgences

★ Specialist medical opinion and reference laboratory

[1]Rare form of presentation. No gonadal insufficiency in males.
[2]Common but not systematic; may be limited to psychological aspects.
[3]Differential diagnoses for liver failure in neonates/infants: neonatal haemochromatosis, infectious, metabolic, toxic, immune-mediated, neoplastic or vascular causes
[4] To be performed immediately if no obvious cause is found, in parallel with other investigations.
Do not wait for test results before discontinuing breastfeeding or galactose-containing formula if galactosaemia is suspected.

Maladies rares Héréditaires du Métabolisme
Filière nationale de santé