

## Classic congenital galactosaemia (GALT deficiency)

Label

### 1 PATIENT PRESENTING IN A&E

#### No risk of acute decompensation.

Patients being followed up for galactosaemia should be managed in the same way as for other patients. This disease does not affect patient triage in A&E.

Risk for a non-compliant patient (diet not being followed): liver failure, cirrhosis (if late diagnosis), cataracts.

### 2 IF ADMITTED TO HOSPITAL

Adhere to the patient or parents known diet, strictly avoiding lactose / galactose (including all dairy products). For children, use a specific milk free of lactose / galactose (e.g. Diargal®).

Usual treatment apart from diet: calcium and vitamin D supplements.

**Drugs:** No contraindications for emergency treatment. No contraindication for hepatotoxic drugs.

Read the list of excipients to avoid formulations containing lactose / galactose if given as long-term treatment.

No increased risk in the case of a one-off dietary error or accidental intake of lactose / galactose.

#### PATHOPHYSIOLOGY:

An inherited metabolic disease due to deficiency of galactose-1-phosphate uridylyltransferase (GALT). Patients with galactosaemia cannot metabolise galactose derived from foodstuffs, such as dairy products (lactose is broken down to glucose + galactose). The diagnosis is often made neonatally as a result of progressive toxicity from galactose-1-phosphate, which manifests itself as liver failure, classically under the age of 1 month old. Treatment consists of lifelong avoidance of lactose / galactose. Despite the diet, other complications may include cataracts, cognitive impairment, low fertility in women (hypergonadotropic hypogonadism) and reduced bone density. Patients with a late diagnosis may develop cirrhosis.

As adults, patients are able to tolerate an individually specific amount of exogenous galactose, enabling their diet to be gradually broadened.

Look up the  
emergency

### NUMBERS AND MEDICAL SPECIALISTS

On-call telephone numbers for metabolic emergencies:

At night, only medical teams can call in emergency situations, and only if the emergency certificate has not been understood or if the clinical state or test results are worrying. As far as possible, make calls before nighttime.

Secretarial issues must be dealt with via the medical secretariat during the week, or by email addressed to the patient's metabolic medicine specialist.

Certificate issued on

Dr