Broad clinical spectrum, continuum of more or less severe forms<sup>2</sup> Infant form: early onset in first few months of life, juvenile form: onset > 1 year and up to end of adolescence and adult form



## **PROGRESSIVE** NEUROMUSCULAR IMPAIRMENT



#### Infantile form

Major hypotonia Hypomobility Hypomimia **Tonque** fasciculations Areflexia or Hyporeflexia Awareness is fine



**Delayed motor skill** acquisition

forms

Possible hyporeflexia (juvenile form)

Mainly proximal damage: progressive myopathy of pectoral, particularly pelvic, as well as scapular girdles

Axial damage:

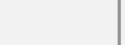
hyperlordosis. camptocormia

Intolerance of exercise and/or myalgia are possibilities but are rarely the most prominent sign

**Blood workup** 

Elevated CPK 3 (<5N) most often

Possible increase in muscle transaminases



## All forms

**Restrictive respiratory** I failure (severe in the infantile form, possible isolated diaphragm damage. occasionally acute, which is a telltale sign in adults): dyspnoea, orthopnoea, recurrent infection, daytime drowsiness, sleep apnoea

Difficulty swallowing

Oropharyngeal dysphagia



#### CARDIAC IMPAIRMENT



#### Infantile form

**Hypertrophic** cardiomyopathy (HCM) severe, may be obstructive (lifethreatening)



#### Juvenile and adult forms

Rare cardiac impairment Moderate HCM or arrhythmia



### Infantile and sometimes iuvenile form

**Hepatomegaly** Feeding problems, growth retardation Language delay. joint impairment

**Hearing loss** 





# All forms

Macroglossia Fatique, asthenia Possible cognitive impairments

Thoracic x-ray: cardiomegaly ECG: Short PR interval, high voltage

QRS, repolarisation abnormalities



Specialist medical opinion and reference laboratory



# Glycogen Storage Disease type II (Pompe disease)?

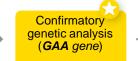
Seek specialist neuro-metabolic advice

## Specialist workup to guide the diagnosis

In collaboration with the centre of excellence, and at the same time as looking for other potential differential diagnoses<sup>4</sup>

Lysosomal acid alpha-glucosidase ( = acid maltase) activity assay

(spot of blood on blotting paper or venous blood): a deficiency indicates the disease5





Rare Disease Centre of Reference / Competence: G2M network: https://www.filiere-G2M.fr/annuaire/ and Filnemus network: https://www.filnemus.fr/

Initial assessment, specialist care, specific treatment (indication, initiation) to be coordinated by a Centre of Excellence. Advantage of early initiation of treatment in infantile form

Genetic counselling, family screening in a specialist centre

For more information:

PNDS French National Authority for Health -Glycogen Storage Disease type II (has-sante.fr), CETL website(Lysosomal disease treatment assessment committee: www.cetl.net), protocol for monitored patients (https://www.filiere-q2m.fr/urgences)





<sup>1=</sup> Glycogen storage disease type II or acid alpha-glucosidase deficiency (GAA).
2Age of onset and prognosis are related to residual enzyme activity. Very low activity correlates to an earlier age of onset and more rapid progression. In the infantile form with no treatment, death normally occurs within the first year of life

<sup>3</sup> Sometimes discovered by chance and can be the telltale sign in juvenile and adult forms. CPK is sometimes normal in adults 4 Other causes of muscular damage and/or damage to the anterior horn of the spinal cord.

<sup>&</sup>lt;sup>5</sup> Enzyme activity may be reduced in heterozygotes