Enhancing POMPE DISEASE Recognition to Prompt Diagnosis

Progressive muscle weakness leads to loss of independent ambulation and respiratory function

Respiratory muscle dysfunction can be the first symptom of Pompe disease (PD)

One-third of adults with PD will present with respiratory dysfunction

IOPD

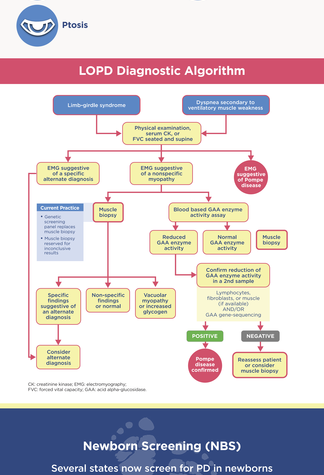
Infantile-Onset Pompe Disease

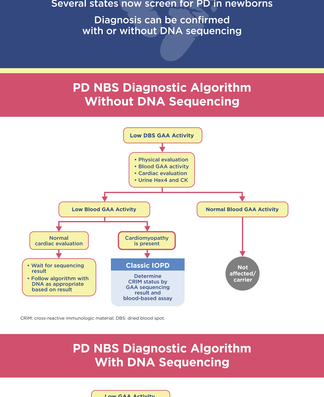
- Signs and symptoms present within first year of life
- Cardiomyopathy is an identifying feature of the classical form of IOPD

LOPD

Late-Onset Pompe Disease

- Variable age of onset
- There is marked variability in the specific pattern of involvement between patients
- Differential involvement of musculoskeletal and respiratory system
  - 25%-30% of symptomatic adults will have a normal muscle biopsy
- Cardiomyopathy typically absent, yet patients can have cardiac involvement





LOPD Evolving Phenotypes and Presentations

Cardiac
- Wolff–Parkinson–White syndrome
- Left ventricular hypertrophy

Vascular
- Basal artery aneurysm
- Aneurysmal dilation of the thoracic aorta

Lingual weakness

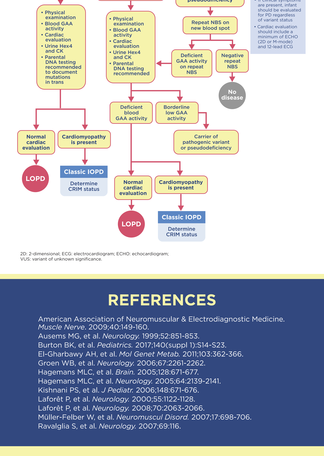
Ptosis

Bladder and bowel incontinence

Gastrointestinal
- Dysphagia
- Gastroesophageal reflux

Skeletal
- Scoliosis
- Rigid spine

LOPD Diagnostic Algorithm



CK: creatinine kinase; EMG: electromyography; FVC: forced vital capacity; GAA: acid alpha-glucosidase.

Newborn Screening (NBS)

Several states now screen for PD in newborns

Diagnosis can be confirmed with or without DNA sequencing

PD NBS Diagnostic Algorithm Without DNA Sequencing

CRIM: cross-reactive immunologic material; DBS: dried blood spot.

PD NBS Diagnostic Algorithm With DNA Sequencing

2D: 2-dimensional; ECG: electrocardiogram; ECHO: echocardiogram; VUS: variant of unknown significance.

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