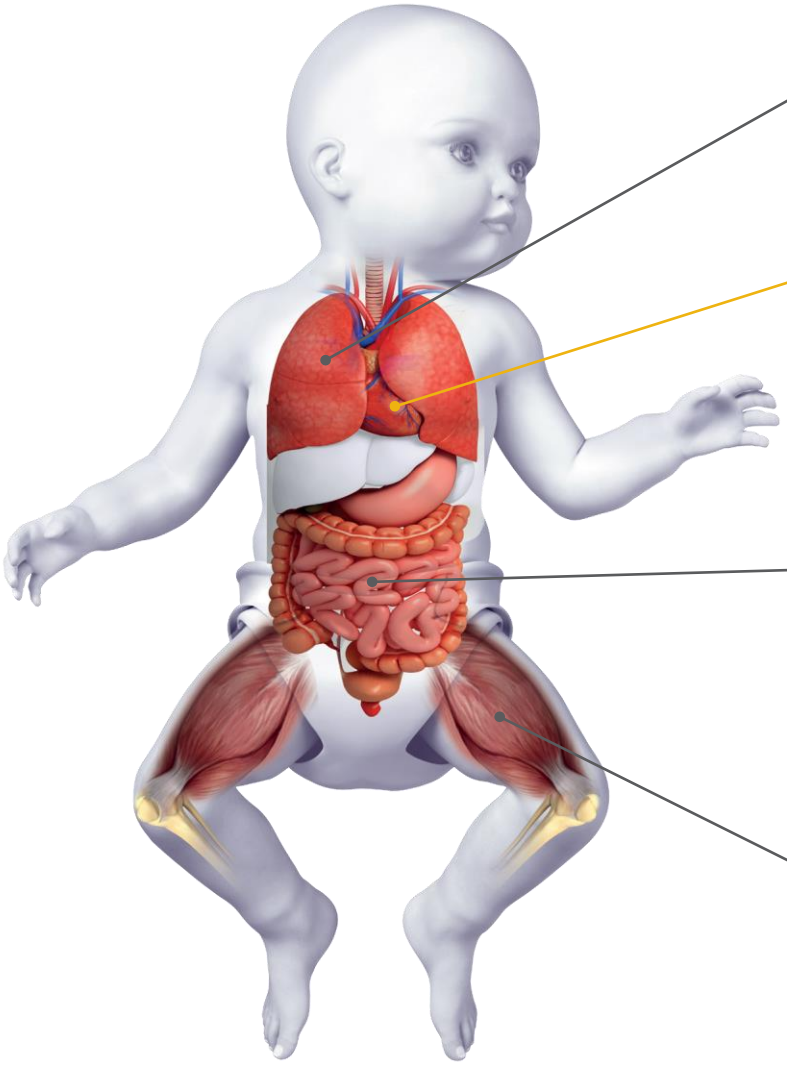
THINK OF **INFANTILE ONSET POMPE DISEASE** IN YOUR **DIFFERENTIAL DIAGNOSIS**

It is a treatable disease

Common signs and symptoms[1]



LUNG

Frequent infections[4]
Respiratory failure/insufficiency
Sleep-disordered breathing

CARDIAC

Cardiomegaly
Cardiomyopathy
Heart failure
Arrhythmia[4]
>specific for infantile form

GASTROINTESTINAL

Macroglossia
Feeding & swallowing difficulties
Hepatomegaly[4]
Gastroesophageal reflux
Failure to thrive

MUSCOSKELETAL

Hypotonia/"Floppy baby"
Muscle weakness
Loss of early motor milestones

Pompe disease is classified as follows[2]:

Infantile Pompe disease (IOPD):

- Patients who exhibit **rapidly progressive disease** characterized by prominent **cardiomegaly**, hepatomegaly, weakness and hypotonia and death due to cardiorespiratory failure in the first year.This represents the most severe end of the disease spectrum and is often referred to as **classic infantile Pompe disease**. Little to no detectable enzyme activity (<1%)[3].
- Patients with the infantile variant form with slower progression and less severe cardiomyopathy but still presenting in the first year of life are classified as **nonclassic infantile pompe disease**.

Late-onset Pompe disease (LOPD)

- Childhood, juvenile or muscular variant that is a heterogeneous group usually **presenting later** than infancy and typically not including severe cardiomyopathy.
- Adult-onset form characterized by a **slowly progressive myopathy** predominantly involving skeletal muscle that can present as late as the second to sixth decade of life.

1. Kishnani PS, et al. J Pediatr 2006;148:671-676., 2. Kishnani et al. Genet Med 2006;8:267-88 , 3. AANEM. Muscle Nerve. 2009;40(1):149-160, 4. Hirschhorn et al. OMMBID New York, NY: McGraw-Hill; 2001. http://ommbid.mhmedical.com/content.aspx?bookid=971&sectionid=62641992. Accessed July 28,2017.

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POMPE disease

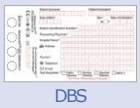
TEST THE PATIENT & THEIR SIBLINGS, TREAT EARLY

When you have diagnosed one patient, it is quite possible that their brother(s) and/or sister(s) may have Pompe disease as well. They may or may not present with signs or symptoms or may present signs and symptoms differently to the diagnosed patient. However, Pompe disease is progressive in nature and can lead to significant and irreversible muscle damage.[2] Early diagnosis and intervention are critical.[2]

1

ENZYME ACTIVITY TESTING

GAA

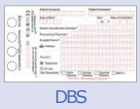


DBS

2

GENOTYPING

GAA



DBS

or

Whole blood sample (EDTA)

3

CONFIRMATION OF ENZYME ACTIVITY ON CULTIVATED FIBROBLASTS

Cultivated fibroblasts

Confirmation of a deficiency of acid α-glucosidase on a culture of skin fibroblasts is required for refund in Belgium.

To confirm Pompe disease and subsequently initiate therapy, refer the patient to specialized centers for neuromuscular or metabolic disorders.

! A lysosomal glycogen accumulation is to be detected in a muscle biopsy (vacuolar myopathy) if only one GAA-PATHOLOGICAL MUTATION of the gene is found.

V Please contact your Sanofi Product Specialist V

Contact person in your lab:.....

Tel.:.....

CRIM ANALYSIS[1]

Cross-reactive immunological material (CRIM) status refers to the presence or absence of endogenous GAA protein with or without enzymatic activity. CRIM-negative patients produce no functional GAA protein of their own and have been reported to have a poorer prognosis than CRIM-positive patients.

1. Kishnani PS, et al. Mol Genet Metab. Jan 2010;99(1):26-33, 2. Toscano et al. Acta Myol. 2013; 32(2): 78-81, 2. AANEM. *Muscle Nerve*. 2009;40(1):149-160.

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A rare commitment to the Pompe community

Sanofi focuses on developing specialty treatments for debilitating diseases that are often difficult to diagnose and treat, providing hope to patients and their families.