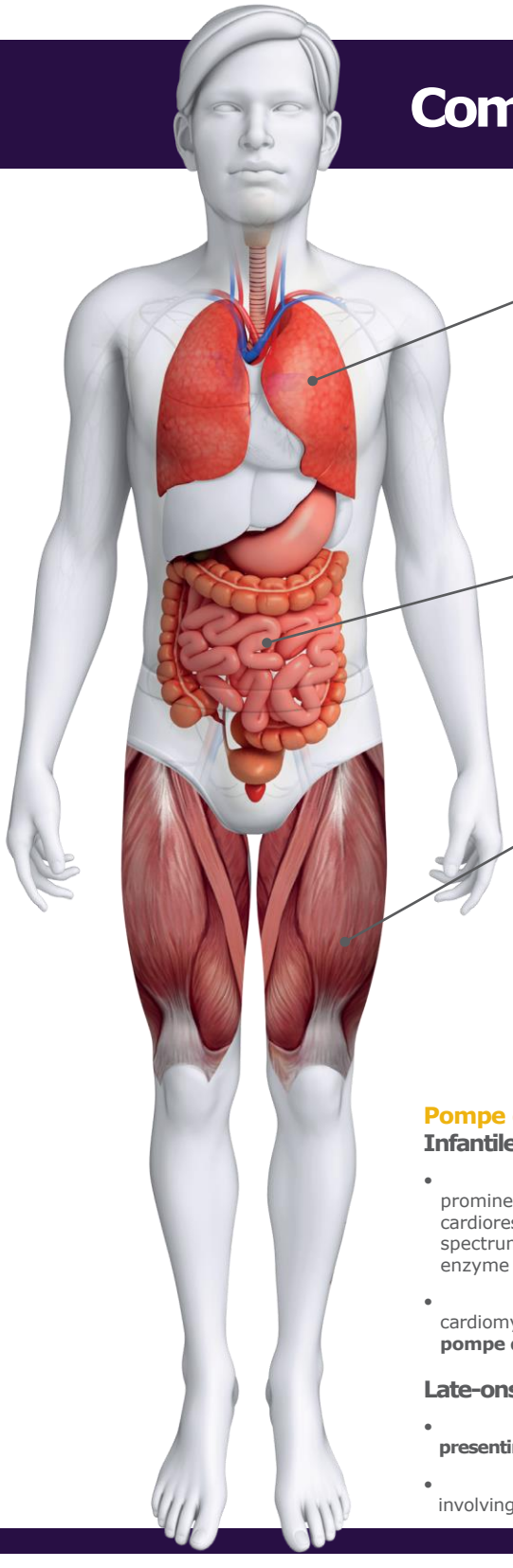
THINK OF **LATE ONSET POMPE DISEASE (LOPD)** IN YOUR **DIFFERENTIAL DIAGNOSIS**

It is a treatable disease



Common signs and symptoms[1,2,3]

LUNG

- Respiratory failure/insufficiency
- Diaphragm weakness
- Sleep-disordered breathing
- Orthopnea
- Dyspnea

GASTROINTESTINAL

- Feeding and swallowing difficulties
- Poor weight gain/maintenance
- Gastroesophageal reflux

MUSCOSKELETAL

- Limb-girdle weakness
- Persisent, mild-to-moderate raised CK* level
- Scoliosis/scapular wings
- Gait abnormalities
- Elevated liver enzymes: aspartate aminotransferase (AST) and alanine aminotransferase (ALT)

Pompe disease is classified as follows[3]:

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%)[1].
- 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. AANEM. Muscle Nerve. 2009;40(1):149-160. 2 Hirschhorn et al. OMMBID New York, NY: McGraw-Hill; 2001. http://ommbid.mhmedical.com/content.aspx?bookid=971&sectionid=62641992. Accessed July 28,2017., 3. Kishnani et al. Genet Med 2006;8:267-88,
*CK =creatine kinase

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POMPE disease

SUSPECT **LATE ONSET POMPE DISEASE** IN CASE OF...

1. Proximal/ axial weakness (LGMD*-like)[1]

AND /OR

2. Restrictive respiratory insufficiency[1]

AND /OR

3. Asymptomatic hyperCKemia[1]

*Limb-Girdle Muscular Dystrophy

10 EARLY WARNING SIGNS OF POMPE DISEASE[2]

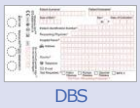
- Trouble climbing stairs
- Leg weakness
- Abnormal walk
- Difficulty getting out of a chair
- Difficulty reaching overhead
- Fall easily
- Morning headaches
- Daytime sleepiness
- Trouble breathing
- Excessive fatigue

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.[1] **Early diagnosis and intervention are critical.**[1]

1

ENZYME ACTIVITY TESTING

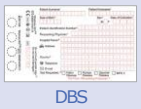


GAA

DBS

2

GENOTYPING



DBS

GAA

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.:.....

1.Toscano et al. Acta Myol. 2013; 32(2): 78-81, 2. AANEM. Muscle Nerve. 2009;40(1):149-160.

Sanofi Belgium, MAT-BE-2001062-2.0-02/24

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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.