

Plenary Abstract

Early Recognition of Late-Onset Pompe Disease by Skeletal Muscle Signs and Symptoms

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In the last decades, both the natural history and clinical spectrum of late-onset Pompe disease (LOPD) have been extensively reviewed, with the identification of a continuum, ranging from severe childhood cases to very late overt manifestations, occurring in the sixth or even seventh decade. Some of these patients may complain of non-specific symptoms and signs (hyperCKemia, myalgia, fatigability) for years; therefore they may be misdiagnosed, due to the great number of other neuromuscular conditions presenting with similar patterns. It has been observed that enzyme replacement therapy is mostly effective when the treatment is started at an early stage of the disease, before advanced damage has occurred in muscle fibres; consequently, the timing of diagnosis has taken on an increasingly important role, both in children and in adults. Although differential diagnosis is challenging, some clues may be helpful to early suspect LOPD at any age, such as history of poor athletic or

physical performance; disproportionate fatigue and exertional dyspnoea; persistently moderately elevated serum CK; long-standing high serum ALT and AST with negative hepatological examinations. The most common clinical presentation, however, is the limb girdle muscular weakness syndrome (LGMW), characterised by subacute onset of chronic weakness involving the shoulder and/or pelvifemoral muscular regions, with difficulties in raising arms or getting up from the ground or sitting. All these patients are usually first seen in non-specialist settings, such as emergency rooms, general medical offices or general neurological offices. For a relatively quick screening in all these patients, the dosage of GAA enzyme in a small amount of blood collected on absorbent paper (DBS or dried blood spot, used for the neonatal screening enzyme) can be easily performed in dedicated laboratories, thereby allowing for early identification of LOPD.

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