Journal of Neuromuscular Diseases 1 (2015) S8
DOI 10.3233/JND-159008
IOS Press

Plenary Abstract

A Troublesome Debate: When to Start Treatment in Adult Pompe Patients?

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BACKGROUND

As Pompe disease became a treatable hereditary disease, often clinicians are questioned by their patients and colleagues when to start treatment in asymptomatic or symptomatic patients.

MATERIALS AND METHODS

A literature review of established guidelines regarding this topic from several countries around the world was performed.

RESULTS

Based on those guidelines, distinct scenarios will be discussed. Scenario 1: When shall we start treatment in an asymptomatic Pompe patient with hyperCKemia only? Here, most Pompe experts would recommend a wait-and-see strategy with close clinical follow-up every 3 months with functional measures, such as six-minute walking test, lung function tests, and a muscle MRI investigation at least every 6 months, and conceivably a sleep lab night. Scenario 2: In any case of sole or combined hypoventilation as a sign of diaphragmatic insufficiency, a clear recommendation for starting ERT is given. Scenario 3: If axial weakness and proximal weakness e.g. combined with Trendelenburg´s sign is already present, once more ERT should be started immediately. Scenario 4: However, what is the situation and decision maker in Pompe patients with already massive disease burden? E.g. in Pompe patients with a long-lasting disease course over many years and a rigid spine syndrome? Here, in line with the international guidelines, most experts would recommend at least a 1-year ERT course with 3-monthly clinical follow-up and functional scoring of the disease burden. Here the chief intention of treatment is stabilisation of this highly chronic disease with specific lung function issues. The same decision line can be commended for Pompe patients with already the need of non-invasive ventilation or in elderly patients with a moderate Pompe disease burden.

CONCLUSIONS

The major hurdle to tackle is our incomplete and not fully validated disease-specific measurement toolbox, which has to be developed to better reflect the disease burden changes under long-term ERT in a clinical setting.

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ISSN 2214-3599/15/$27.50