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. 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.