Frequently Asked Questions

How do you speak with patients about the choices they have with second-generation enzyme replacement therapies (ERTs)?

Take a step back, one of the most challenging questions for a treating physician is "Are patients really stable on first-generation ERT? Currently, there is an expectation that patients will continue first-generation ERT unless they show a serious health decline.

In patients who are deteriorating, the discussion about which of the two new ERTs is very complex. However, it comes down to patient choice. Some patients will be very interested in the idea of a dual approach with an enzyme and stabilizer. Other patents may be put off by the fact that have to fast before taking the stabilizer and would prefer to have a single infusion protocol. It is a personal choice, and it is the role of the physician to be extremely balanced in that discussion to enable patients to come to their own conclusion.

Physicians should also be clear with patients that if there is a lack of response with one second-generation ERT there is another second-generation ERT. Physicians and patients should keep in mind that the observed benefit of a second-generation ERT may not be immediate. One might consider 2 years as a potential observational window for treatment before considering a switch to another second-generation ERT. Importantly, more real-world data on patients receiving second-generation ERTs is needed. We need to as the patient community to engage in monitoring process as this will help further our understanding of how to use second-generation ERTs in practice.

How do you monitor the central nervous system (CNS) in patients with late-onset Pompe disease (LOPD)? How do you monitor the basilar artery?

The best monitoring tools are magnetic resonance imaging (MRI) and angiography. Transcranial ultrasound is now used in many places, which is especially helpful for monitoring the flow conditions of the basilar artery. Unfortunately, there are not adequate recommendations on how to manage CNS manifestations, such as stroke risk, in LOPD. In the case of patients who are at risk of stroke, physicians must use the same management protocol as any other patient at risk for stroke (eg, monitoring cholesterol levels).

Older-aged patients with LOPD may have develop comorbidities unrelated to Pompe disease (eg, cancer). Patients should be regularly monitored. If there an unexpected change is observed, then the physician should investigate whether another disease process is responsible for that change.

Is "late-onset" Pompe disease an accurate description of this disease process?

The nomenclature of "late-onset" Pompe disease is confusing because it suggests disease onset in older children. However, newborn screening provides the opportunity to identify patients with LOPD at the earliest stages. In the past, early manifestations of LOPD have been overlooked. In hindsight, many patients have spoken about early symptoms, such as tiring more easily, not being able to keep up when playing sports, not being able to climb stairs. Often these patients were being labelled as "lazy children". However, newborn screening and muscle biopsies of patients at earlier stages have broadened our understanding of the pathophysiology of LOPD. Onset of LOPD can occur at very young ages and with appropriate monitoring physicians can intervene earlier in the disease process.