

## Poster Abstract: Diagnostic

### Epidemiology and Screening for Pompe Disease in Sweden

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#### BACKGROUND

Pompe disease (PD; acid maltase deficiency or glycogen storage disease type II) is an autosomal recessive inherited, potentially treatable metabolic myopathy with heterogeneous clinical presentations and with considerable overlap of signs and symptoms found in other neuromuscular disorders. According to previous reports, patients with PD have been incorrectly diagnosed for several years as limb–girdle muscular dystrophies (LGMDs). To diagnose both entities is challenging, and a diagnostic delay of several years seems to be common. The frequency of misdiagnosis is unknown. No epidemiologic studies have been carried out on PD in the nordic countries.

#### METHOD

This is the first nationwide, registry-based study on the epidemiology of PD in Sweden. First, we identified all patients with PD by retrospectively validating

the diagnosis using the medical records of individuals with the unspecified diagnosis code E74.0 (glycogen storage diseases) according to the International Classification of Diagnosis (ICD). Beyond this, we selected, from the Swedish National Patient Registry, all inpatients in Sweden with unclassified LGMD and limb–girdle muscle weakness, and then screened for PD by using the dried blood spot test. We hereby report the preliminary results of the screening study and data on the prevalence of PD in Sweden.

#### RESULTS AND CONCLUSIONS

PD and van Gierke disease are the most common glycogen storage diseases in Sweden. The occurrence of PD among unclassified LGMDs is only 1.5–3%, suggesting that the risk of misdiagnosis of PD is low in the Swedish population. The overall prevalence of PD in Sweden is 0.25/100,000 (1:400,000), which is much lower than that described in other European countries, except for Norway and Finland.

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