Pediatric Rheumatology



Poster presentation



# The Pompe Registry: tracking Pompe disease symptoms in a broad patient population

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## Introduction

Pompe disease is a rare, progressive, often fatal metabolic myopathy, which manifests as a clinical spectrum that varies with respect to age at onset, rate of disease progression, and extent of organ involvement. The underlying pathology is deficiency of acid alpha-glucosidase (GAA). To gain a better understanding of Pompe disease, a global, voluntary, observational Registry was developed to collect anonymous, longitudinal data.

## Preliminary data overview

As of March 2008, 494 patients from 23 countries were enrolled; the majority (72%) Caucasian. Europe and North America enroll 87% of patients. Median age of infants at symptom onset was 2.0 months (n = 94) and at diagnosis was 4.0 months (n = 93). Median age of adults at symptom onset was 27.7 years (n = 293) and at diagnosis was 35.3 years (n = 289). Symptoms most frequently reported by patients ≥ 18 years old (n = 321) include: muscle weakness [lower extremities (80%), upper extremities (69%), trunk (53%)]; shortness of breath after exercise (61%) and at rest (31%); dependence on respiratory support (38%); sleep disturbance/apnea (35%); orthopnea (32%); and scapular winging (31%). Approximately half of patients genotyped expressed the IVS1-13T>>G mutation.

## Summary

These results show significant delay from symptom onset to diagnosis in adult patients, highlighting the need for greater disease awareness. Registry data on prevalence and age at onset of symptoms may allow earlier patient identification, enabling intervention before irreversible muscle damage occurs. Analysis of registry data over time may increase understanding of the evolution of, and interaction between, impairments and function under varying conditions and interventions, allowing improved clinical management.