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Rate of disease progression during long-term follow-up of patients with late-onset Pompe disease

N.A.M.E. Van der Beek[ab], M.L.C. Hagemans[b], A.J.J. Reuser[c], W.C.J. Hop[d], A.T. Van der Ploeg[b], P.A. Van Doorn[a], J.H.J. Wokke[e]

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Abstract

To determine the rate of disease progression in patients with late-onset Pompe disease, we collected longitudinal data on pulmonary function and skeletal muscle strength in 16 patients whose symptoms had started in childhood or adulthood. The mean duration of follow-up was 16 years (range 4–29 years). During the follow-up period, eight patients (50%) became wheelchair bound and three (19%) became ventilator dependent. At a group level, pulmonary function deteriorated by 1.6% per year, and proximal muscle weakness progressed gradually. At the individual level, however, the rate and extent of progression varied highly between patients. In two thirds of patients, pulmonary function and muscle strength declined simultaneously and to the same extent. The remaining one third of patients showed a variable, sometimes rapidly progressive course, leading to early respirator or wheelchair dependency. These individual differences, especially in pulmonary dysfunction, indicate the need for regular monitoring every 6–12 months depending on the rate of disease progression.

Keywords: Pompe disease, Glycogen storage disease type II, Acid α-glucosidase, Late-onset, Disease progression

[a] Erasmus MC, University Medical Center, Department of Neurology, 's-Gravendijkwal 230, 3015 CE Rotterdam, The Netherlands

[b] Erasmus MC – Sophia Children's Hospital, Department of Pediatrics, Division of Metabolic Diseases, Rotterdam, The Netherlands

[c] Erasmus MC, University Medical Center, Department of Clinical Genetics, Rotterdam, The Netherlands

[d] Erasmus MC, University Medical Center, Department of Epidemiology and Biostatistics, Rotterdam, The Netherlands

[e] Rudolf Magnus Institute of Neurosciences, University Medical Center Utrecht, Department of Neurology, Utrecht, The Netherlands

Corresponding author. Address: Erasmus MC, University Medical Center, Department of Neurology, 's-Gravendijkwal 230, 3015 CE Rotterdam, The Netherlands. Tel.: +31 10 7043821; fax: +31 10 7044727.

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