

Poster Abstract: Clinical

Prevalence of Asymptomatic Vertebral Fractures in Late-Onset Pompe Disease

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CONTEXT

Bone fragility and low bone mass have been reported in small case series of patients with Pompe disease with severely reduced muscle strength or immobilization.

OBJECTIVE

To determine the prevalence of morphometric vertebral fractures and to evaluate bone mass in adults with late-onset Pompe disease.

DESIGN

A multicenter cross-sectional observational study. The study was performed from August 2012 to December 2013.

STUDY SETTING

All subjects were outpatients referred to University Referral Centers.

PATIENTS

Twenty-two late-onset Pompe disease patients with progressive proximal myopathy and minimal respira-

tory involvement without other diseases affecting bone mass.

MAIN OUTCOME MEASURES

The prevalence of morphometric vertebral fractures was systematically assessed by semiquantitative analysis of the lateral spine X-ray (T4–L5).

RESULTS

A high prevalence of morphometric vertebral fractures was found. At least one vertebral fracture was present in 17 out of 22 patients (77%). All vertebral fractures were asymptomatic. Bone mineral density was normal in 36.5% of the patients, while 36.5% were osteopenic and 27% were osteoporotic at at least one site. Fracture prevalence was independent of muscular and respiratory functional parameters and of genotype.

CONCLUSIONS

Our data show, for the first time, that asymptomatic and atraumatic vertebral fractures occur frequently in late-onset Pompe disease patients without significant impairment of bone mass. Screening for asymptomatic vertebral fractures should be routinely performed in Pompe disease, irrespective of the disease severity. Fracture risk should be confirmed in longitudinal studies.

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