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Effects of enzyme replacement therapy on bone density in late onset Pompe disease

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Pompe Disease is an inherited rare autosomal recessive disorder caused by the deficiency of enzyme acid α-glucosidase (GAA). The disease results in the accumulation of glycogen in smooth, cardiac, and skeletal muscles. Late Onset Pompe Disease (LOPD) is mainly characterized by skeletal muscle dysfunction, proximal muscle weakness, and early respiratory insufficiency. Although many patients exhibit decreased bone density and fractures, there is currently no official protocol for surveillance and management of osteoporosis and osteopenia in Pompe disease. Results of studies of the effect of ERT on bone mineralization in LOPD patients are inconclusive because of the small number of studies, and small size of some of the cohorts. Our cohort includes 15 Pompe patients (13 males, 2 females) ranging in age from 21 to 74 years, 14 of whom were on routine enzyme replacement therapy (ERT) for variable durations. Progressive impact of ERT on osteopenia or osteoporosis was tested by studying bone mineral density of hips and spine using DEXA scans. This study showed a significant positive correlation with the increase of Z-score = 0.04–0.09 in association with ERT treatment. We found that for each one-year increase in age, there was a 0.09 unit score significant increase in lumbar Z-scores ($p = .01$) and a 0.042 ($p = .04$) increase in the femur Z-score. This study emphasizes the importance of early management and ERT to prevent and lower the risk of fractures resulting from osteoporosis. A larger study of Pompe subjects will determine other variables that influence bone density.

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