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

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Pompe Disease is a rare autosomal recessive disorder lysosomal storage disorder caused by the deficiency of α-glucosidase (GAA). The disease results in the accumulation of glycogen, a polysaccharide of glucose, in the smooth, cardiac, and skeletal muscles. Late Onset Pompe is mainlycharacterized by skeletal muscle glycogen accumulation, proximal muscle weakness, and early respiratory insufficiency. Many patients have also exhibited decreased bone density and fractures as symptoms of Pompe Disease Our cohort included 15 Pompe patients ranging from 21- 74 years on ERT for variable durations. We tested the progressive impact of ERT on osteopenia or osteoporosis by studying bone mineral by comparing the z and t-scores of hips and spine using DXA scans. Our results demonstrated that females had a 2.17 units lower average z-score of the lumbar spine and 0.563 units lower average z-score of their femurs compared to males. Increasing age also changed the bone dynamics, as a one-year increase in age of the patients led to a 0.09 unit score significant increase in lumbar z-scores ($p=0.01$) and a 0.042 unit score significant increase in femur z-scores ($p=0.04$). Additionally every one year duration of ERT treatment resulted in a 0.15 unit score increase in z-scores ($p=0.01$) of the lumbar spine. However the femur z-score only showed a 0.08 unit score increase after one year of treatment ($p=0.10$) Results overall, denote that ERT treatment shows a positive correlation with a lower risk of fractures resulting from osteoporosis. ERT was associated with an increase of the z-scores of .09 of the lumbar spine. Females were at a higher risk of developing osteoporosis compared to males. This study emphasizes the importance of early management and ERT to prevent osteopenia and bone fractures. A genotype-phenotype correlation and a larger study of Pompe subjects is needed to determine other variables

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