Factors affecting bone mineral density in adult patients with Glycogen Storage Diseases - one centre experience.

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BACKGROUND

The clinical presentation of Glycogen storage diseases (GSD) varies and includes liver and muscle phenotypes. Reduced bone mineral density (BMD) is one complication of which the aetiology is unclear. Based on by dual-energy X-ray absorptiometry (DXA) it is classified into two stages based the number of standard deviations away from age- and gender-matched controls:
• Osteopenia (Z-score <-1.0 and >-2.5)
• Osteoporosis (Z-score ≤-2.5)

METHODS

A retrospective analysis was performed of DXA reports in adults with GSDs type Ia, Ib, III, VI and IX, who attend Adult Metabolic Clinics in one tertiary centre. DXA scan results were available for 19 patients (10M/9F; mean age 30.5; range 21 - 46 at the time of DXA scan): 2 GSD Ia, 3 GSD Ib, 11 GSD III, 1 GSD VI and 2 GSD IX.

RESULTS

BMD within the Osteopenia range was documented in 10/19 cases (53%) and osteoporosis in 1/19 case (5%). In 8/19 cases (42%) the DXA scan showed normal range BMD.

In all cases vitamin D was insufficiently replaced (serum vitamin D <50 nmol/L). None of patients had received bisphosphonates. In one GSD III patient, chronic immobilization likely contributed to a reduced BMD. Their fasting tolerance and metabolic control varied with no clear correlation with their BMD.

CONCLUSION

This small cohort of GSD patients highlights the common occurrence of low BMD in this group of disorders. Likely to be multifactorial in origin, it highlights the importance of regular assessment with DXA. Correcting factors like low vitamin D may help improve BMD although more robust treatment strategies are needed to reduce future morbidity which results from bone fragility.

Conflicts of interest: None

REFERENCES