THE EFFECT OF VITAMIN D RECEPTOR POLYMORPHISM ON BONE MINERAL DENSITY IN EGYPTIAN PATIENTS WITH BETA THALASSEMIA MAJOR

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Background

- Beta thalassemia major (BTM) is considered a major health problem. Despite optimal conventional treatment, bone disease comprising of low bone mineral density (BMD), bone pain, and fractures is still a characteristic feature of thalassemia. The etiology of bone disease in thalassemia is multifactorial. Vitamin D receptor (VDR) mediates the action of 1,25(OH)2D, The VDR polymorphism may be responsible for modifying the activity of VDR protein.

Objective

- To study the effect of Vitamin D status and VDR polymorphism on bone mineral density in Egyptian patients with BTM.

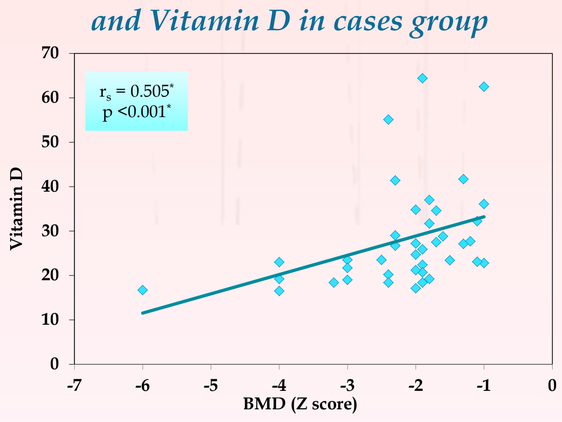
Subjects & Methods

- The Study included eighty children with BTM and eighty age & sex- matched children as control group. Patients with any hepatic or renal impairment, hyper-parathyroidism or using medications affecting bone mineral metabolism (as glucocorticoids or anticonvulsant drugs) were excluded. Serum calcium, phosphorus, ALP, ferritin and 25OHD levels were measured. VDR genotyping regarding BsmI, TaqI, FokI single nucleotide polymorphisms was carried out. Every patient underwent dual-energy X-ray absorption (DEXA) scan of the lumbar spine.

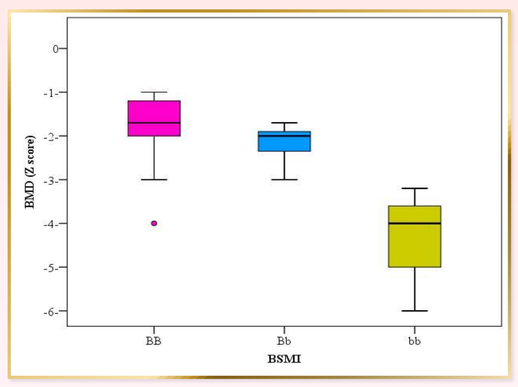
Results

- The serum levels of phosphorus, ALP and ferritin were significantly higher in patients with βTM than the control group. The serum calcium levels was significantly lower in the patients with βTM than the control group.
- We reported a significant positive correlation between serum 25OHD levels and the of BMD Z score at the lumbar spine.
- Serum 25OHD levels and BMD Z score were significantly lower in patients with *BsmI* bb genotype compared to Bb and BB genotypes
- Serum 25OHD levels and BMD Z score were significantly lower in patients with *FokI* ff genotype compared to Ff and FF genotypes.
- No significant association was observed between serum 25OHD levels and the BMD Z score with *TaqI* polymorphism.

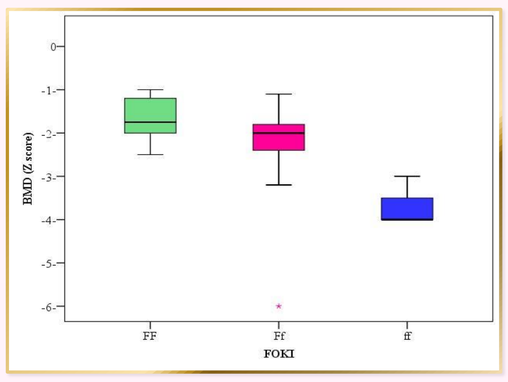
Correlation between BMD (Z score) and Vitamin D in cases group



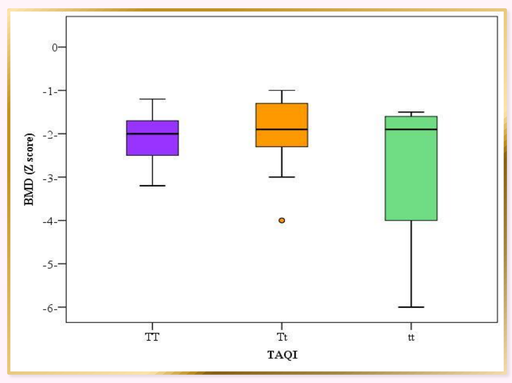
Box Plot showing the relation between BsmI with BMD (Z score) in cases group



Box Plot showing the relation between FokI with BMD (Z score) in cases group



Box Plot showing the relation between TaqI with BMD (Z score) in cases group



Conclusion

- The VDR genotyping can be used as an additional test in children who are vulnerable to osteoporosis so that early preventive can be taken.

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