Osteopetrosis and its effects on the elbow

Health & Medicine

Osteopetrosis al Affiliation According to Michael Whyte (2006), osteopetrosis or the marble bone disease was first described by Albers-Schonberg in 1940, (p. 398). After it was described, there were two forms of the disease with distinct clinical manifestations that were discovered. The first was the autosomal recessive infantile which was termed as malignant and the benign autosomal dominant adult type, (p. 398). Gene defects have been associated with the predisposition of the victims to the disorder. The pathogenesis of the disorder as has been described by various scientists involves the failure of osteoclast-mediated resorption of the skeleton leading to the disorder. In disease diagnosis, radiographic images are taken. The radiographic findings in the case of osteopetrosis show symmetrical increase in the mass of bone in the affected region e. g. the elbow (p. 399). This accumulation of bone mass in the affected region leads to disturbance of bone formation processes which include bone growth, bone modelling and bone remodeling.

Using radiographic images, the elbow and joint will show accumulated mass of bone and hence joint movement will be hampered (Arun, Rysavy, & Wozniak, 2007). Laboratory tests are also applied to clear and explain the findings after the radiographic analysis. In most cases hypocalcemia is evident due to low levels of calcium in the body culminating from the disease. Rachitic changes in growth plates is seen in cases where hypocalcemia is severe (p. 400). Treatment is complex and may involve bone marrow transplant from human leukocyte antigen identical donors. This is used to avert the situation as the disease could be hereditary. Hormonal and dietary therapy using calcium deficient diet has also been used in treatment of the disease.

References

Arun, K. P., Rysavy, M. & Wozniak, A. (2007). Fracture Treatment in Intermediate Autosomal Recessive Osteopetrosis. Orthopedics Journal. 30(7), 2007.

Whyte, P. M. (2006). Genetic, Development, and Dysplastic Skeletal Disorders. American Society for Bone and Mineral Research. p. 398-401.