



كلية الطب – جامعة الفيوم
قسم الأطفال

البحث السابع

عنوان البحث :

Evaluation of Early Musculoskeletal Response After Enzyme Replacement Therapy In Gaucher Disease Using MRI-Based Bone Marrow Burden Score.

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مكان وتاريخ النشر :

Abstract

Introduction: Gaucher disease (GD) is a rare hereditary disease caused by lack of lysosomal enzyme that result in accumulation of glucocerebroids in the cells of reticulo-endothelial system (RES) including bone marrow, which leads to variable presentations of variable onsets that varies from early during childhood to late during adulthood with mild to severe clinical complications that may end with death. Musculoskeletal involvement is common in type 1 and type 3 Gaucher disease and may cause marked disability to untreated patients. New modalities of treatment as enzyme replacement therapy (ERT) aim to replace the defective enzyme to decrease disease progression and improve life expectancy with good quality. **Patient and Methods:** The study was done among 10 cases (5 males, and 5 females) of Gaucher disease to assess early musculoskeletal response after Enzyme Replacement therapy (ERT). All cases were diagnosed by molecular techniques. Severity of the disease was evaluated using MRI-based bone marrow burden (BMB) scoring system. **Results:** The mean age of diagnosis was 7.6 years \pm 4.22. The most common gene mutation was Homozygous D409H which was present in 5 cases. Two cases showed fractures prior to ERT, and one case presented with dorsolumbar scoliosis. However, both were not significantly related to GD. All cases started ERT since 2017 and no cases showed Fractures, or bone crises, only one case complained from non-significantly related hip problem that improved within one month (Irritable hip). Bone marrow infiltration was found in all patients. The mean total BMB score (femora and lumbar spine) was 11, ranging from 7 to 13 points. **Conclusion:** ERT is an effective treatment in preventing severe skeletal complications and improve the quality of life in patients with GD. Follow up of patients using BMB scoring system is a simplified method for evaluation of bone marrow infiltration in GD.