MONITORING BONE MARROW INVOLVEMENT IN GAUCHER DISEASE USING BONE MARROW BURDEN SCORE

Introduction: Semiquantitative MRI is a useful tool to study bone marrow involvement and complications in Gaucher disease (GD). Bone marrow burden (BMB) score has been used to evaluate the severity and monitoring the bone involvement in GD patients. Aim: Report the experience of the use of BMB score in the cohort of patients from the Reference Center for Gaucher Disease of Rio Grande do Sul State, Brazil. Methods: Thirteen GD patients on treatment (12 GD type 1 and one GD type 3) were submitted to a lumbar spine and femur MRI at point 1 and at 12 months after (mean; point 2). BMB score was classified in four categories according to the severity: from 0 to 2 (no bone involvement), 3 to 7 (mild), 8 to 12 (moderate) and 13 to 16 (severe). Results: Mean BMB score was 6.6 and 5.7, at points 1 and 2, respectively. At point 1, two patients had no bone involvement and 5 had mild bone disease, and there were no differences on the scores found at point 2. Three patients had moderate bone involvement; one of them had a significant decrease of BMB score at point 2 (from 12 points to 4 points). This patient started ERT one month before the basal MRI. Two patients had severe bone disease, and one of them had a decrease on BMB score on the follow up MRI (from 13 points to 10 points). Conclusion: The use of BMB score is a useful tool to evaluate the bone marrow involvement in GD patients and their response to ERT. Our data suggest patients with a more severe bone marrow involvement have a faster response to ERT regarding the bone disease than patients with a mild bone marrow involvement. Support: Genzyme, Actelion. Palavra-chave: Gaucher; Reossonância; BMB.