Mutational Analysis in Gaucher Disease: Implications in Genetic Counseling and Management

Abstract:

Gaucher disease (GD) is the most common LSD worldwide. The disease is caused due to mutations in β-glucocerobrosidase (GBA) gene located on chromosome 1. The mutations results in the deficient activity of acid β-glucosidase (glucocerebrosidase) enzyme. It is inherited in an autosomal recessive fashion and both men and women are affected equally. We report here two families wherein the mutation analysis for the disease was performed as the clinical features of the children were suggestive of GD. In the first family the enzyme analysis reports of the children were normal but GD was confirmed upon mutation analysis. In another family who had come for prenatal diagnosis, the parents were confirmed to be heterozygote of normal mutation whereas the foetus was found to be of carrier status. The family had already lost two children who had clinical features suggestive of Gaucher. We conclude that in some cases the enzyme analysis report may not be conclusive and mutation analysis has to be carried out to confirm the disorder. Prenatal diagnosis for lysosomal storage disorders like GD is also recommended among high risk couples.