World Academy of Science, Engineering and Technology
International Journal of Bioengineering and Life Sciences
Vol:8, No:10, 2014

Phenotypic and Genotypic Diagnosis of Gaucher Disease in Algeria

Authors : S. Hallal, Z. Chami, A. Hadji-Lehtihet, S. Sokhal-Boudella, A. Berhoune, L. Yargui

Abstract : Gaucher disease is the most common lysosomal storage in our population, it is due to a deficiency of β –glucosidase acid. The enzyme deficiency causes a pathological accumulation of undegraded substrate in lysosomes. This metabolic overload is responsible for a multisystemic disease with hepatosplenomegaly, anemia, thrombocytopenia, and bone involvement. Neurological involvement is rare. The laboratory diagnosis of Gaucher disease consists of phenotypic diagnosis by determining the enzymatic activity of β - glucosidase by fluorimetric method, a study by genotypic diagnosis in the GBA gene, limiting the search recurrent mutations (N370S, L444P, 84 GG); PCR followed by an enzymatic digestion. Abnormal profiles were verified by sequencing. Monitoring of treated patients is provided by the determination of chitotriosidase. Our experience spaning a period of 6 years (2007-2014) has enabled us to diagnose 78 patients out of a total of 328 requests from the various departments of pediatrics, internal medicine, neurology. Genotypic diagnosis focused on the entire family of 9 children treated at pediatric CHU Mustapha, which help define the clinical form; or 5 of them had type III disease, carrying the L444P mutation in the homozygous state. Three others were composite (N370/L444P) (N370S/other unintended mutation in our study), and only in one family no recurrent mutation has been found. This molecular study permits screening of heterozygous essential for genetic counseling.

Keywords : Gaucher disease, mutations, N370S, L444P
Conference Title : ICGMB 2014 : International Conference on Genetics and Molecular Biology
Conference Location : Paris, France
Conference Dates : October 30-31, 2014