Prenatal Genotyping of Gaucher Disease in Egypt

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Abstract

Objective: to use chorionic villi sampling (CVS) and amniocentesis to determine the genotyping of Gaucher Disease (GD) of fetuses of pregnant mothers who had a previous child affected by GD.

Methods: The study was conducted between January 2009 and December 2012. It included 42 pregnant women that gave informed written consent. Thirty mothers presented early so they underwent CVS at 10-12 weeks of pregnancy while 12 mothers presented later and underwent amniocentesis at 14-16 weeks. Strip assay for the identification of Glucocerebrosidase (GBA) gene mutations in the samples of chrorionic villi and amniotic fluid was based on polymerase chain reaction (PCR) and reverse hybridization.

Results: The age of the studied pregnant women ranged from 19 to 26 years. Consanguinity was present in 38 cases. Eighteen women were pregnant in affected fetuses. The results of genotyping revealed 15 cases were homozygous L444P / L444P and one case homozygous (N370s/N370s) while 2 cases were heterogeneous (L444P/D409H). Twenty four pregnant women had carrier fetuses which were all heterozygous L444P.

Conclusion: This study highlights the findings of an extended gene mutation examination for prenatal diagnosis of Guacher Disease. The study found out that the most common mutation was L444P/L444P.

Key words: Gaucher diseases, Prenatal diagnosis, Egypt, gene, mutation