

25th world NUTRITION AND PEDIATRICS HEALTHCARE CONFERENCE

May 03, 2021 | Webinar

A Novel Mutations in the glucose-6-phosphatase gene in heterozygous state that cause glycogen storage disease type 1a**Priyanka Udawat***Institute of Digestive & Liver Care, S L Raheja Hospital, Mumbai, India*

Glycogen storage disease (GSD) type 1a (von Gierke disease) (OMIM 232200) is an inborn error of metabolism caused by the deficiency of glucose-6-phosphatase-a in liver and kidney. Affected children exhibit failure to thrive hepatomegaly, growth retardation, delayed puberty, symptoms of early morning hypoglycemia such as drowsiness & seizures. Biochemical features include fasting hypoglycemia, lactic acidosis, hypertriglyceridemia and hyperuricemia.

Confirmatory diagnosis is based on enzyme assay on liver biopsy. The G6PC gene comprises of five exons and sequencing the gene for molecular genetic analysis is a reliable and convenient alternative to enzyme assay in fresh liver biopsy specimens for the diagnosis of GSD 1a with 100% sensitivity and specificity [2]. We sequenced this gene in case with clinical, biochemical and histopathology features of GSD type 1a. The DNA sequencing of G6PC1 exons revealed the missense mutation in exon 5; a substitutive point mutation at 992nd base position of exon 5, showing substitution of T in the place of C. The patient and mother are heterozygous for this missense mutation and father is normal. Thus, we report a rare case of GSD type 1a with novel mutation which causing disease in heterozygous state.

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