Glycogen Storage Disease VII

Alternative Names
GSD7
GSD VII
Muscle Phosphofructokinase Deficiency
PFKM Deficiency
Tarui Disease

Record Category
Disease phenotype

WHO-ICD
Endocrine, nutritional and metabolic diseases > Metabolic disorders

Incidence per 100,000 Live Births
Unknown

OMIM Number
232800

Mode of Inheritance
Autosomal recessive

Gene Map Locus
12q13.11

Description
Glycogen storage disease type VII (GSDVII) is an autosomal recessive disorder caused by a mutation in the PFKM gene. It occurs due to the inability to break down glycogen in muscle cells. There are four different types of GSDVII. The classical form of the disease is the most common one and is characterized by muscle pain and cramps, exercise intolerance, renal failure caused by myoglobin accumulation in the kidney, and jaundice. Affected patients have high level of bilirubin and creatine kinase in blood. Patients with the infantile form have severe hypotonia and cardiomyopathy. The only manifestation for the late-onset form of GSDVII is myopathy. The hemolytic form is characterized by hemolytic anemia and does not show signs of muscle problems. More than 100 cases of GSD VII have been described in the literature.

Molecular Genetics

Mutations in the Muscle Phosphofructokinase (PFKM) gene, which is located on the long arm of chromosome 12, cause GSD VII. Among the genes coding the three phosphofructokinase (PFK) isozymes present in humans, this gene encodes the muscle type isoform. This enzyme catalyzes the breakdown of glycogen in muscles, providing these cells with energy. Defects in the enzyme, therefore, do not allow muscle cells to get enough energy from the breakdown of glycogen, resulting in weakness and cramps.

Epidemiology in the Arab World

Saudi Arabia
Al-Hassnan et al. (2007) described a 2-year-old boy born to consanguineous parents who presented at the age of 3-days with subtle seizures in the form of facial twitching and eye blinking. He was treated with anti-seizure medications. All investigations including brain computed tomography (CT) and EEG were unremarkable. He went on to develop hypotonia and mild developmental delay. Electron microscopic examination showed excessive accumulation of free glycogen in the subsarcolemmal location. Muscle phosphofructokinase was found to be deficient with a level of 0.2 U (normal, 25.12 ± 10.3 U). The patient had five other siblings, two of them having died with a similar illness. Al-Hassnan et al., (2007) suggested considering GSDVII in the differential diagnosis of neonatal seizures and early infantile nonprogressive muscle weakness.

References

Related CTGA Records

External Links
Contributors