Geneticheck - Genetic Report

Gene

PFKM

Associated Diseases

Glycogen Storage Disease Vii
Glycogen Storage Disease Due To Muscle Phosphofructokinase Deficiency

Phenotype

Increased total bilirubin
Increased concentration of total (conjugated and unconjugated) bilirubin in the blood.

Jaundice
Yellow pigmentation of the skin due to bilirubin, which in turn is the result of increased bilirubin concentration in the bloodstream.

Exercise intolerance
A functional motor deficit where individuals whose responses to the challenges of exercise fail to achieve levels considered normal for their age and gender.

Exercise-induced muscle cramps
Sudden and involuntary contractions of one or more muscles brought on by physical exertion.

Hyperuricemia
An abnormally high level of uric acid in the blood.

Muscle weakness
Reduced strength of muscles.

Reticulocytosis
An elevation in the number of reticulocytes (immature erythrocytes) in the peripheral blood circulation.

Exercise-induced myoglobinuria
Presence of myoglobin in the urine following exercise.

Gout
Recurrent attacks of acute inflammatory arthritis of a joint or set of joints caused by elevated levels of uric acid in the blood which crystallize and are deposited in joints, tendons, and surrounding tissues.

Reduced erythrocyte 2,3-diphosphoglycerate concentration
This term refers to an inappropriate low 2,3-DPG concentration in erythrocytes. 2,3-diphosphoglycerate (2,3-DPG) controls the movement of oxygen from red blood cells to tissues. Anemia is usually accompanied by an increased level of 2,3-DPG in order to promote tissue oxygenation.

Anemia
A reduction in erythrocytes volume or hemoglobin concentration.

Skeletal muscle atrophy
The presence of skeletal muscular atrophy (which is also known as amyotrophy).

Cholelithiasis
Hard, pebble-like deposits that form within the gallbladder.

Hemolytic anemia
A type of anemia caused by premature destruction of red blood cells (hemolysis).

Autosomal recessive inheritance
A mode of inheritance that is observed for traits related to a gene encoded on one of the autosomes (i.e., the human chromosomes 1-22) in which a trait manifests in individuals with two pathogenic alleles, either homozygotes (two copies of the same mutant allele) or compound heterozygotes (whereby each copy of a gene has a distinct mutant allele).

Increased muscle glycogen content
An increased amount of glycogen in muscle tissue.

Myotonia
An involuntary and painless delay in the relaxation of skeletal muscle following contraction or electrical stimulation.