Alpha-galactosidase A

Fabry disease is caused by a mutation in the enzyme alpha-galactosidase-A. A mutation in the alpha-galactosidase A gene that controls this enzyme causes insufficient breakdown of lipids, which build up to harmful levels in the eyes, kidneys, autonomic nervous system and cardiovascular system. Lipid storage may lead to impaired arterial circulation and increased risk of heart attack or stroke. The heart may also become enlarged and the kidneys may also be affected.

AMP-activated protein kinase (AMPK)

AMP-activated protein kinase (AMPK) is a widely distributed cellular kinase that is activated during times of metabolic stress. It has been termed a cellular "fuel gauge", and primarily functions to turn off energy consuming pathways and turn on energy producing pathways during metabolic stress.

AMPK-activated protein kinase gamma subunit (PRKAG2)

The AMPK enzyme is composed of three subunits, an alpha, beta and gamma subunit. These subunits also exist in a number of different isoforms. PRKAG2 is the gene that encodes the gamma2 subunit of AMP-activated protein kinase (AMPK).

Amylo-1,6-glucosidase

Amylo-1,6-glucosidase is an enzyme that catalyzes the hydrolysis of glycogen at specific branch points in its glucose residue chains. It is sometimes called a "debrancher enzyme". Mutations in this gene cause glycogen storage disease. Forbes disease is an inherited mutation that results in an amylo-1,6-glucosidase deficiency. This enzyme deficiency causes excess amounts of an abnormal glycogen to be deposited in the liver, muscles and, in some cases, the heart.

Carnitine palmitoyltransferase

Carnitine palmitoyltransferase (CPT) is an important enzyme involved in the transport of fatty acids across membranes, particularly the mitochondrial membrane. CPT 1 is one isofrom of CPT that is a key enzyme transporting fatty acids into the mitochondria. Mutations in this enzyme can result in serious energetic deficiencies in muscle.

Globotriaosylceramide (Gb3)

Globotriaosylceramide (Gb3) is a glycosphingolipid with glucosylceramide as its base cerebroside. Fabry disease is an inherited deficiency of the enzyme, alpha-galactosidase A, which is normally responsible for the breakdown of globotriaosylceramide (Gb3). The subsequent abnormal level of Gb3 causes the symptoms of Fabry disease described above.

Glycogen Phosphorylase

Glycogen phosphorylase is one of the enzymes that break up glycogen into glucose subunits. Glycogen is left with one less glucose molecule. McCardle's disease is a condition caused by an inborn glycogen phosphorylase deficiency. Symptoms include muscular pain, fatigability, and muscle cramping following exercise. Unlike other types of glycogenosis the disease is not fatal and the missing enzyme does not impair the functioning of other body systems.

Glycosphingolipid

Glycosphingolipids are a subtype of glycerol containing lipids (glycolipids) that contain the amino alcohol sphingosine. The class of glycosphingolipids include a group of specialized glycolipids including cerebrosides, gangliosides and globosides.

Long Chain Acyl CoA Dehydrogenase

Long chain acyl CoA dehydrogenase (LCAD) is the first enzyme involved in the ß-oxidation of fatty acids in mitochondria. ß-oxidation of fatty acid oxidation is a major source of energy for the heart. Mutations in the LCAD gene can lead to serious cardiomyopathies due to inadequate production of energy.
Lysophospholipid

Phospholipids are important components of all cellular membranes in organisms. They consist of a glycerol backbone, a polar head group and two fatty acid moieties linked to the glycerol backbone. If one of these fatty acids is removed, a lysophospholipid is formed. These lysophospholipids can disrupt the normal membrane and the normal function of the proteins contained in the membrane.

Lysosomal acid glucosidase

Lysosomal acid glucosidase is a glycogen-degrading lysosomal enzyme. Pompe disease is a hereditary metabolic disorder caused by the complete or partial deficiency of lysosomal acid glucosidase. This enzyme deficiency causes excess amounts of glycogen to accumulate in the lysosomes of many cell types but predominantly in muscle cells. The resulting cellular damage manifests as muscle weakness and/or respiratory difficulty.

Lysosome associated membrane protein 2

Danon disease, a lysosomal glycogen storage disease, occurs as a result of a lysosome associated membrane protein 2 (LAMP 2) deficiency. Danon disease is associated with a severe cardiomyopathy and variable skeletal muscle weakness are constant features and mental retardation is very frequently associated.

Mutations in the AMPK-activated protein kinase gamma subunit (PRKAG2)

Mutations in PRKAG2 have recently been shown to cause cardiac hypertrophy, cardiac glycogen accumulation, Wolff-Parkinson White syndrome and conduction system disease causing pre-excitation. Dominant mutations in PRKAG2 have recently been shown to result in massive myocardial thickening, AV conduction system disease and ventricular pre-excitation.

Tafazzin

Tafazzin is a protein highly expressed in cardiac and skeletal muscle that is involved in the metabolism of specialized lipids. Mutations of the tafazzin gene are associated with a number of clinical disorders including dilated cardiomyopathy.