Climb™

Supporting those affected by Inherited Metabolic Disorders

Aldolase A Deficiency

Aldolase A Deficiency is a form of Glycogen Storage Disease. Glucose is stored in the body as glycogen which is used as fuel providing energy for the body. Glycogen Storage Diseases are caused by a deficiency or absence of an enzyme involved in the process of producing or breaking down glycogen. The Aldolase A enzyme is found mostly in muscles and red blood cells. A deficiency of the enzyme causes haemolytic anaemia which occurs when the bone marrow isn't making enough red cells to replace the ones that are being destroyed. Aldolase A Deficiency also causes muscle disease (myopathy) resulting in rhabdomyolysis; the break down of damaged skeletal muscle tissue releasing their contents into the bloodstream. There are reports of a case of Aldolase A Deficiency where haemolytic anaemia is not present.

Synonyms

Alternative names for this condition are:

- Aldolase Deficiency Red Cell
- Glycogen Storage Disease type 12
- Glycogen Storage Disease 12
- Glycogen Storage Disease due to Aldolase A Deficiency
- GSD12
- Red Cell Aldolase Deficiency

Further information about this condition is available from Climb

Disclaimer

Updated 12/05/17

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