Triosephosphate Isomerase Deficiency

Other names that may be used for this disorder are:
- Enzymopathic Hemolytic Anemia
- TPI
- TPI Deficiency

Triosephosphate Isomerase Deficiency is a very rare disorder that is characterised by a low amount of red blood cells. This occurs when the bone marrow activity is unable to compensate for the increased loss of red blood cells (haemolytic anaemia). Triosephosphate Isomerase is an enzyme that is present in all tissues. The gene coding the enzyme has been located on the short arm of chromosome 12 (12p13).

Treatment for individuals with this disease aims to provide relief for any symptoms and support in the care of the individual. Blood transfusions may be needed during episodes of haemolysis or anaemia and assisted ventilation may be required for diaphragm paralysis.

This information is fully sourced and referenced, for more detailed information and references please contact CLIMB by email, letter or telephone.

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