

Open Peer Review on Qeios

Triosephosphate Isomerase Deficiency

National Cancer Institute

Source

National Cancer Institute. <u>Triosephosphate Isomerase Deficiency</u>. NCI Thesaurus. Code C131652.

A rare, autosomal recessive, inherited disorder caused by mutation of the TPI1 gene. It is characterized by hemolytic anemia and severe, progressive neuromuscular dysfunction beginning in early childhood.

Qeios ID: 75FQN1 · https://doi.org/10.32388/75FQN1