Non-spherocytic hemolytic anemia due to hexokinase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Non-spherocytic hemolytic anemia due to hexokinase deficiency. ORPHA:90031

Nonspherocytic haemolytic anaemia due to hexokinase deficiency is characterised by severe hemolysis, appearing in infancy. Seventeen affected families have been reported so far. Transmission is autosomal recessive. Mutations have been described in HK1, the gene that encodes red blood cell-specific hexokinase-R.