Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency. ORPHA:35120

Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency is a rare, hereditary, hemolytic anemia due to an erythrocyte nucleotide metabolism disorder characterized by mild to moderate hemolytic anemia associated with basophilic stippling and the accumulation of high concentrations of pyrimidine nucleotides within the erythrocyte. Patients present with variable features of jaundice, splenomegaly, hepatomegaly, gallstones, and sometimes require transfusions. Rare cases of mild development delay and learning difficulties are reported.