Glutathione synthetase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Glutathione synthetase deficiency. ORPHA:32

Glutathione synthetase deficiency is characterised by hemolytic anemia, associated with metabolic acidosis and 5-oxoprolinuria in moderate forms, and with progressive neurological symptoms and recurrent bacterial infections in the most severe forms.