Crigler-Najjar syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Crigler-Najjar syndrome. ORPHA:205

Crigler-Najjar syndrome (CNS) is a hereditary disorder of bilirubin metabolism characterized by unconjugated hyperbilirubinemia due to a hepatic deficit of bilirubin glucuronosyltransferase (GT) activity. Two types have been described, CNS types 1 and 2 (see these terms). CNS1 is characterized by a complete deficit of the enzyme and is unaffected by phenobarbital induction therapy, whereas the enzymatic deficit is partial and responds to phenobarbital in CNS2.