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Joubert syndrome with hepatic defect

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Joubert syndrome with hepatic defect. ORPHA:1454*

Joubert syndrome with hepatic defect is a very rare subtype of Joubert syndrome and related disorders (JSRD, see this term) characterized by the neurological features of JS associated with congenital hepatic fibrosis (CHF).