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Joubert syndrome with renal defect

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. [Joubert syndrome with renal defect](#). ORPHA:220497*

Joubert syndrome with renal defect is a rare subtype of Joubert syndrome and related disorders (JSRD, see this term) characterized by the neurological features of JS associated with renal disease, in the absence of retinopathy.