Joubert syndrome and related disorders

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Joubert syndrome and related disorders. ORPHA:140874

Joubert syndrome (JS) and related disorders (JSRD) are a group of developmental delay/multiple congenital anomaly syndromes in which the mandatory feature is the ``molar tooth sign'' (MTS), a complex midbrain-hindbrain malformation recognizable on brain imaging. The MTS is characterized by cerebellar vermis hypodysplasia, thickening and malorientation of the superior cerebellar peduncles and abnormally deep interpeduncular fossa.