

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Kategoria

Synonimy

JSRD

JSRD

Kod ORPHA

140874

Kod OMIM

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Kod ICD10

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Kod ICD11

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*Źródło

orphanet