Opis choroby *

Definicja

A rare genetic developmental defect during embryogenesis characterized by the association of the classic features of Joubert syndrome (congenital midbrain-hindbrain malformations causing hypotonia, abnormal breathing and eye movements, ataxia and cognitive impairment) together with the skeletal anomalies of Jeune asphyxiating thoracic dystrophy (short ribs, long and narrow thorax causing respiratory failure, short-limbs, short stature, and polydactyly). Additional variable manifestations include cystic kidneys, liver fibrosis, and retinal dystrophy.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
JBTS with JATD
JBTS z JATD
Zespół Joubert z JATD
Joubert syndrome with JATD

Kod ORPHA
397715

Kod OMIM
616546

Kod ICD10
Q04.3

Kod ICD11
-

---

*Źródło

orphanet