Opis choroby *

Definicja

A rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by developmental delay, intellectual disability and mild to moderate facial dysmorphism in association with variable brain malformations (including abnormal gyration patterns, ventriculomegaly, white matter abnormalities, hypoplasia of the corpus callosum and cerebellar hemispheres), musculoskeletal abnormalities (including hemivertebrae, scoliosis or kyphosis, contractures, and joint laxity), ocular involvement (strabismus, hypermetropia and cortical visual impairment) and hypotonia. Additional clinical manifestations may include seizures, short stature urogenital malformations, heart defects and gastrointestinal malformations.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
ZTTK syndrome
Zespół ZTTK
Zespół Zhu,Tokita, Takenouchi i Kima
Zhu-Tokita-Takenouchi-Kim syndrome

Kod ORPHA
500150

Kod OMIM
617140

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet