

## 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 ZTTK syndrome

#### Synonimy

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