

Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome with intellectual disability characterized by global developmental delay, intellectual disability, macrothrombocytopenia, lymphedema, and dysmorphic facial features (like synophrys, ptosis, eversion of the lateral portion of the lower eyelid, and thin upper lip, among others). Additional reported manifestations include cardiac and genitourinary anomalies, sensorineural hearing loss, ophthalmologic abnormalities, skeletal anomalies, and immunodeficiency. Brain imaging may show enlarged ventricles, cerebellar atrophy, or white matter changes.

Dane

Klasyfikacja

Synonimy

Zespół wad wrodzonych Takenouchi-Kosaki syndrome

Zespół Takenouchiego i Kosakiego

Kod ORPHA

487796

Kod OMIM

616737

Kod ICD10

Q87.8

Kod ICD11

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*[Źródło](#)

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