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
Zespół wad wrodzonych

Synonimy
Takenouchi-Kosaki syndrome
Zespół Takenouchiego i Kosakiego

Kod ORPHA
487796

Kod OMIM
616737

Kod ICD10
Q87.8

Kod ICD11
-

---

*Źródło

orphanet