Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay and intellectual disability, infantile hypotonia, microcephaly, movement disorder, and impaired balance. More variable manifestations are hearing loss, cortical visual impairment, abnormalities of fingers and/or toes, congenital cardiac anomalies, kyphoscoliosis, dysmorphic facial features, abnormal sleep pattern, and seizures, among others.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
You-Hoover-Fong syndrome
Zespół You'a i Hoover-Fong

Kod ORPHA
488642

Kod OMIM
616954

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet