Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by intellectual disability, developmental delay, delayed bone age, short stature, generalized muscle weakness, and dysmorphic facial features (such as high arched eyebrows, downslanting palpebral fissures, prominent nose, and narrow palate and mouth). Additional reported manifestations include blue sclerae, ophthalmoplegia, and intention tremor. Brain imaging may show white matter abnormalities.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
457365

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet