Opis choroby *

Definicja

A rare genetic multisystemic neurodevelopmental disorder characterized by a distinct facial appearance, cardiac anomalies (most frequently supravalvular aortic stenosis), cognitive and developmental abnormalities, and connective tissue abnormalities (e.g., joint laxity). Facial dysmorphism is characterized by a broad forehead, bitemporal narrowing, periorbital fullness, stellate and/or lacy iris pattern, short upturned nose with bulbous tip, long philtrum, wide mouth, full lips and mild micrognathia.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Deletion 7q11.23
Delecja 7q11.23
Monosomia 7q11.23
Zespół Williamsa i Beurena
Monosomy 7q11.23
Williams-Beuren syndrome

Kod ORPHA
904

Kod OMIM
194050

Kod ICD10
Q93.8

Kod ICD11
LD44.70

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*Źródło

orphanet