Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by facial dysmorphism (brachycephaly, long, narrow, triangular face, prominent forehead, hypertelorism, flat philtrum, microstomia, thin lips, hypoplastic maxilla), marfanoid habitus with arachnodactyly, and moderate to severe intellectual disability. Additional features may include clinodactyly, triphalangeal thumbs, hammer-shaped toes, hyperextensible joints, hypotonia, hyperreflexia and underdeveloped musculature. Delayed external genitalia development, as well as seizures and mitral regurgitation have been reported in some cases. There have been no further descriptions in the literature since 1995.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
De Die-Smulders-Vles-Fryns syndrome
Zespół De Die, Smuldersa, Vlesa i Frynsa

Kod ORPHA
1130

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet