

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

Kod ORPHA	Kod OMIM	Kod ICD10
1130	-	Q87.8

Kod ICD11
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*Źródło

orphanet