

## 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 De Die-Smulders-Vles-Fryns syndrome

#### Synonimy

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