

Opis choroby *

Definicja

Stüve-Wiedemann syndrome (SWS) is a rare autosomal recessive congenital primary skeletal dysplasia, characterized by small stature, bowing of the long bones, camptodactyly, hyperthermic episodes, respiratory distress/apneic episodes and feeding difficulties that usually lead to early mortality.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Neonatal Schwartz-Jampel syndrome Dysplazja Stüve'a i Wiedemanna SJS2 Schwartz-Jampel syndrome type 2 Stüve-Wiedemann dysplasia

Kod ORPHA
3206

Kod OMIM
601559

Kod ICD10
Q78.8

Kod ICD11
LD24.C

[*Źródło](#)

orphanet