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
Zespół wad wrodzonych

Synonimy
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

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*Źródło

orphanet