Zespół Germana
Kod Orpha: 2077 Kod OMIM: 231080

Opis choroby *

Definicja
German syndrome is an autosomal recessive arthrogryposis syndrome, described in 5 cases. Three of the four known families with affected children were Ashkenazi Jews. German syndrome is characterized by arthrogryposis, hypotonia-hypokinesia sequence, and lymphedema. Patients present distinct craniofacial appearance (tall forehead and ''carp''-shaped mouth, cleft palate), contractures, severe hypotonia manifesting as motor delay, and swallowing difficulties. The disease has a severe morbidity and mortality rate and survivors present a small stature, hypotonia, frequent upper respiratory infections, and psychomotor delay. There have been no further descriptions in the literature since 1987.
Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Hypotonia-arthrogryposis-facial dysmorphism-lymphedema syndrome

Kod ORPHA
2077

Kod OMIM
231080

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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