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
-

---

*Źródło

orphanet