

Opis choroby *

Definicja

An extremely rare arthrogryposis syndrome, described in only two pairs of siblings from two unrelated families to date, and characterized by the association of arthrogryposis, congenital torticollis, dysmorphic facial features (i.e. asymmetry of the face, myopathic facial movements, ptosis, posteriorly rotated ears, cleft palate), progressive scoliosis and episodes of malignant hyperthermia. There have been no further descriptions in the literature since 1988.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Froster-Iskenius-Waterson-Hall syndrome

Zespół Frostera, Iskeniusa i Watersona

Zespół hipertermii złośliwej, artrogrypozy i kręczu szyi

Malignant hyperthermia-arthrogryposis-torticollis syndrome

Kod ORPHA

2215

Kod OMIM

217150

Kod ICD10

G71.8

Kod ICD11

-

*Źródło

orphanet