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	Synonimy
Zespół wad wrodzonych	Froster-Iskenius-Waterson-Hall syndrome
	Zespół Frostera, Iskeniusa i Watersona
	Zespół hipertermii złośliwej, artrogrypozy i kręczu szyi
	Malignant hyperthermia-arthrogryposis-torticollis syndrome

Kod ORPHA	**Kod OMIM**	**Kod ICD10**
2215	217150	G71.8

Kod ICD11
-

---

*Źródło

orphanet