Opis choroby *

Definicja

An extremely rare type of arthrogryposis multiplex congenita characterized by the combination of multiple joint contractures with movement limitation, microstomia with a whistling appearance of the mouth that may cause feeding, swallowing, and speech difficulties, a distinctive expressionless facies, severe developmental delay, central and autonomous nervous system dysfunction (excessive salivation, temperature instability, myoclonic epileptic fits, bradycardia), occasionally Pierre-Robin sequence, and lethality generally occurring during the first months of life. Arthrogryposis multiplex congenita-whistling face syndrome has been suggested to be a fetal akinesia deformation sequence.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Illum syndrome
Zespół Illum

Kod ORPHA
1150

Kod OMIM
208155

Kod ICD10
Q87.8

Kod ICD11
LD26.41

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*Źródło

orphanet