Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome with intellectual disability characterized by severe congenital contractures of the limbs and face, hypotonia, neonatal respiratory distress, and global developmental delay. Dysmorphic facial features include downslanting palpebral fissures, broad nasal bridge, large nares, long philtrum, and deep nasolabial folds, among others. Limb deformities (camptodactyly, clubfoot), short neck, scoliosis, as well as seizures have also been reported. Brain MRI may show cerebral and cerebellar atrophy in some cases.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
CLIFAHDD syndrome
Zespół CLIFAHDD

Kod ORPHA
562528

Kod OMIM
616266

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet