

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	Synonimy
Zespół wad wrodzonych	CLIFAHDD syndrome
	Zespół CLIFAHDD

Kod ORPHA	Kod OMIM	Kod ICD10
562528	616266	Q87.8

Kod ICD11

-

*Źródło

orphanet