Opis choroby *

Definicja

A rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by craniofacial dysmorphism (including an abnormal skull shape, hypertelorism, downslanting palpebral fissures, epicanthal folds, low-set ears, depressed nasal bridge, micrognathia), short stature, ectodermal anomalies (such as sparse eyebrows, eyelashes, and scalp hair, hypolastic toenails), developmental delay, and intellectual disability. Additional features may include cerebral/cerebellar malformations and mild renal involvement.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Developmental delay-short stature-dysmorphic features-sparse hair syndrome
Zespół opóźnienia rozwoju, niskiego wzrostu, cech dysmorficznych i rzadkich włosów
Loucks-Innes syndrome

Kod ORPHA
459061

Kod OMIM
616901

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet