

Opis choroby *

Definicja

A rare, fatal multiple congenital anomalies/dysmorphic syndrome characterized by facial dysmorphism (incl. dolichocephaly/scaphocephaly, high frontal hairline, laterally overlapping upper eyelids, hypertelorism, prominent eyelashes, deep-set eyes, macrocornea, nystagmus, dysplastic ears, abnormal auricles, prominent nasal bridge, dental dysplasia), visual impairment, deafness, seizures, generalized skeletal dysplasia, high fingerprint ridge count, cryptorchidism, hypospadias, spasticity and severe intellectual disability. An increased chromosome breakage and a fatal lymphoid malignancy have been reported. There has been no further description in the literature since 1974.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

2608

Kod OMIM

310465

Kod ICD10

Q87.8

Kod ICD11

LD2H.Y

*Źródło

orphanet