Opis choroby *

Definicja

A rare, genetic, developmental defect during embryogenesis syndrome characterized by generalized keratosis follicularis, severe proportionate dwarfism and cerebral atrophy. Alopecia (of scalp, eyebrows and eyelashes) and microcephaly are additionally observed features. Intellectual disability, inguinal hernia and epilepsy may also be associated. There have been no further descriptions in the literature since 1974.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2339

Kod OMIM
308830

Kod ICD10
Q87.1

Kod ICD11
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*Źródło

orphanet