Opis choroby *

Definicja

A rare genetic syndromic intellectual disability that is characterized by congenital permanent alopecia universalis, intellectual disability, psychomotor epilepsy and periodontitis (pyorrhea). Total permanent alopecia and pyorrhea are invariably concomitant while intellectual disability and psychomotor epilepsy are observed in most patients. No other abnormality of nails or skin (apart from absence of hair) has been reported. Transmission is autosomal dominant.

Dane

Klasyfikacja
Choroba

Synonimy
Shokeir syndrome
Zespół Shokeira

Kod ORPHA
1008

Kod OMIM
104130

Kod ICD10
Q87.8

Kod ICD11
LD90.Y

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*Źródło

orphanet