

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

*Źródło

orphanet