

Opis choroby *

Definicja

Tricho-retino-dento-digital syndrome is an autosomal dominant ectodermal dysplasia syndrome, characterized by uncombable hair syndrome (see this term), congenital hypotrichosis and dental abnormalities such as oligodontia (see this term) or hyperdontia, and associated with early-onset cataract, retinal pigmentary dystrophy, and brachydactyly with brachymetacarpia. Furthermore, hyperactivity and a mild intellectual deficit have been reported in affected patients.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Bork syndrome

Niedające się uczesać włosy - dystrofia

barwnikowa siatkówki - wady zębów -

brachydaktylia

Zespół Bork

Uncombable hair-retinal pigmentary dystrophy-

dental anomalies-brachydactyly syndrome

Kod ORPHA

1264

Kod OMIM

191482

Kod ICD10

Q82.4

Kod ICD11

LD27.0Y

[*Źródło](#)

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