

Opis choroby *

Definicja

A rare ectodermal dysplasia syndrome with bone abnormalities characterized by onychodystrophy; anomalies of the lower jaw, oral vestibule and dentition; post-axial polydactyly; moderately restricted growth with short limbs; and normal intelligence. Although it closely resembles Ellis-van Creveld syndrome (see this term), an allelic disorder and another type of ciliopathy, WAD is usually a milder disease without the presence of heart abnormalities and is inherited in an autosomal dominant manner.

Dane

Klasyfikacja

Zespół wad wrodzonych Curry-Hall syndrome

Synonimy

Dyzostoza kończynowo-zębowa typu Weyersa

Dyzostoza kończynowo-zębowa Weyersa

Zespół Curry i Halla

Weyers acrofacial dysostosis

Weyers acrofacial dysostosis

Kod ORPHA

952

Kod OMIM

193530

Kod ICD10

Q75.4

Kod ICD11

LD25.2

[*Źródło](#)

orphanet