Opis choroby *

Definicja

A rare ectodermal dysplasia syndrome with bone abnormalities characterized by onychodystrophy; anomalies of the lower jaw, oral vestibule and dentition; post-axialpolydactyly; 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

Synonimy
Curry-Hall syndrome
Dyzostoza kończynowo-zębowa typu Weyersa
Dyzostoza kończynowo-zębowa Weyersa
Zespół Curry i Halla
Weyers acrodental dysostosis
Weyers acrofacial dysostosis

Kod ORPHA
952

Kod OMIM
193530

Kod ICD10
Q75.4

Kod ICD11
LD25.2

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*Źródło

orphanet