

Opis choroby *

Definicja

A rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by hypoplasia, aplasia or atresia of the lacrimal system, anomalies of the ears with sensorineural or mixed hearing loss, hypoplasia, aplasia or atresia of the salivary glands, dental anomalies, and digital malformations. Patients present obstruction of the nasal lacrimal ducts that can lead to epiphora, and chronic conjunctivitis due to alacrimia. Aplasia or hypoplasia of the salivary glands lead to dry mouth and early onset of severe dental caries. Dental features include late tooth eruption, small and peg-shaped lateral maxillary incisors and mild enamel dysplasia. The digital features are variable and include fifth finger clinodactyly, duplication of the distal phalanx of the thumb, triphalangeal thumb, and/or syndactyly. Unilateral radial aplasia and radial-ulnar synostosis have also been reported in association.

Dane

Klasyfikacja

Zespół wad wrodzonych LADD syndrome

Synonimy

Zespół LADD

Zespół LARD

Zespół Levy'ego i Hollistera

Zespół łzowo-uszno-promieniowo-zębowy

LARD syndrome

Lacrimoauricularodental syndrome

Levy-Hollister syndrome

Kod ORPHA

2363

Kod OMIM

620192

Kod ICD10

Q87.8

Kod ICD11

LD27.0Y

*Źródło

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