Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by aplasia cutis congenita of the scalp, breast anomalies ranging from hypothelia or athelia to amastia, and anomalies of the external ears. Variable clinical characteristics include nail and dental anomalies, syndactyly and camptodactyly of fingers and/or toes, sparse or absent secondary sexual hair, renal malformations, and facial dysmorphism. Cases with severe hypotonia and developmental delay have been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Finlay-Marks syndrome
Zespół Finlay'a i Marksa

Kod ORPHA
2036

Kod OMIM
181270

Kod ICD10
Q87.8

Kod ICD11
LD27.0Y

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*Źródło

orphanet