Opis choroby *

Definicja

A rare, dominantly inherited multiple congenital anomalies syndrome characterized by highly variable clinical phenotype involving the three main affected systems: branchial (cutaneous) defects, ophthalmic malformations and facial anomalies. Additional features can be present.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
BOFS
BOFS

Kod ORPHA
1297

Kod OMIM
113620

Kod ICD10
Q18.8

Kod ICD11
LD2F.1Y

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*Źródło

orphanet