

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	Synonimy
Zespół wad wrodzonych BOFS	BOFS

Kod ORPHA	Kod OMIM	Kod ICD10
1297	113620	Q18.8

Kod ICD11
LD2F.1Y

*Źródło

orphanet