Opis choroby *

Definicja

A rare, multiple congenital anomalies syndrome characterized by craniofacial dysmorphology, congenital heart disease, dermatological abnormalities (most commonly hyperkeratotic skin and sparse, curly hair), neurological manifestations (hypotonia, seizures), failure to thrive and intellectual disability.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
CFC syndrome
Zespół CFC

Kod ORPHA
1340

Kod OMIM
615280

Kod ICD10
Q87.8

Kod ICD11
LD27.0Y

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*Źródło

orphanet