

Opis choroby *

Definicja

A rare genetic multiple congenital anomalies syndrome characterized by abnormal bone maturation with skeletal anomalies, airway obstructions, failure to thrive, developmental delay, moderate to severe intellectual disability and characteristic facial features with macrocephaly, prominent forehead, shallow orbits, proptosis and blue sclerae.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Accelerated skeletal maturation-facial dysmorphism-failure to thrive syndrome
Zespół zaawansowanego wieku kostnego, dysmorfii twarzy i niedoboru wzrostu i masy ciała

Kod ORPHA

561

Kod OMIM

602535

Kod ICD10

Q87.3

Kod ICD11

LD2C

*Źródło

orphanet