

Opis choroby *

Definicja

A rare developmental defect during embryogenesis characterized by premature closure of metopic sutures and/or other sutures, short stature, and developmental delay. Dysmorphic features include trigonocephaly, metopic ridge, narrow forehead, bitemporal narrowing, arched eyebrows, hypotelorism, deep-set eyes, epicanthal folds, strabismus, wide nasal bridge, small pointed nose, anteverted nostrils, long philtrum, low-set ears, malar flattening, narrow mouth, thin lips, high-arched palate, crowded teeth, and micrognathia. Variable additional manifestations may include conductive hearing loss, cerebral (mainly involving the white matter), skeletal (e.g. brachymesophalangy of the fifth fingers), cardiovascular and renal anomalies, inguinal hernia, hypospadias, and seizures.

Dane

Klasyfikacja

Zespół wad wrodzonych Say-Meyer syndrome
Zespół Say'a i Meyera

Synonimy

Kod ORPHA

3369

Kod OMIM

314320

Kod ICD10

Q87.0

Kod ICD11

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*Źródło

orphanet