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

Synonimy
Say-Meyer syndrome
Zespół Say'a i Meyera

Kod ORPHA
3369

Kod OMIM
314320

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet