Opis choroby *

Definicja

Delayed membranous cranial ossification is a rare, genetic primary bone dysplasia characterized by absent ossification of calvarial bones at birth and characteristic facial dysmorphisms (frontal bossing, hypertelorism, downward-slanting palpebral fissures, proptosis, flat nasal bridge, low-set ears, midface retrusion). Patients present a soft skull at birth which, over time, progressively ossifies and in adulthood typically results in a deformed skull (with brachycephaly and prominent occiput). No other skeletal abnormalities are associated and patients have normal cognitive and motor development.

Dane

Klasyfikacja Synonimy

Zespół wad wrodzonych Gonzales-del Angel syndrome Zespół Gonzales del Angela

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 3034
 155980
 075.8

Kod ICD11 LB70.Y

*Źródło

orphanet