

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	Q75.8

Kod ICD11
LB70.Y

*Źródło

orphanet