Opis choroby *

Definicja

A rare cranial malformation syndrome characterized by the premature closure of both lambdoid sutures and the posterior sagittal suture, resulting in abnormal skull contour (frontal bossing, anterior turricephaly with mild brachycephaly, biparietal narrowing, occipital concavity) and dysmorphic facial features (low-set ears, midfacial hypoplasia). Short stature, developmental delay, epilepsy, and oculomotor dyspraxia have also been reported. Associated anomalies include enlargement of the cerebral ventricles, agenesis of the corpus callosum, Arnold-Chiari malformation type I, venous anomalies of skull, and hydrocephalus.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
BLSS
Niesyndromiczna kraniosynostoza obejmująca oba szwy węgłowe i szew strzałkowy
Bilateral lambdoid and sagittal synostosis
Isolated sagittal and bilambdoid craniosynostosis
Non-syndromic sagittal and bilateral lambdoid synostosis

Kod ORPHA
1516

Kod OMIM
218350

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet