Opis choroby *

Definicja

Pseudoaminopterin syndrome is a developmental anomalies syndrome that resembles the aminopterin embryopathy (see this term) without history of fetal exposure to aminopterin. It is characterized by skull (craniosynostosis and poorly mineralized cranial vault), dysmorphic (ocular hypertelorism, palpebral fissure anomalies, micrognathia cleft lip and/or high arched palate and small and low set/rotated ears) and limb (brachydactyly, syndactyly and clinodactyly) anomalies, associated with mild-to-moderate intellectual deficit and short stature.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
ASSA
ASSA
Zespół podobny do zespołu aminopterynowego bez aminopteryny
Aminopterin syndrome-like sine aminopterin

Kod ORPHA
221120

Kod OMIM
600325

Kod ICD10
Q82.0

Kod ICD11
LD24.GY

---

*Źródło

orphanet