

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

Kod ORPHA	Kod OMIM	Kod ICD10
221120	600325	Q82.0

Kod ICD11
LD24.GY

*Źródło

orphanet