

Opis choroby *

Definicja

A rare syndromic craniosynostosis with variable phenotypic expression characterized by craniosynostosis, intellectual disability, distinctive facies, abnormalities of the fingers and toes (brachydactyly, polydactyly and syndactyly), short stature, congenital heart disease, skeletal defects, obesity, genital abnormalities and umbilical hernia.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych ACPS2	ACPS2
	Akrocefalopolisyndaktylia typu 2
	Acrocephalopolysyndactyly type 2

Kod ORPHA	Kod OMIM	Kod ICD10
65759	201000	Q87.0

Kod ICD11
LD24.GY

*Źródło

orphanet