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
Zespół wad wrodzonych

Synonimy
ACPS2
ACPS2
Akrocefalopolisyndaktylia typu 2
Acrocephalopolysyndactyly type 2

Kod ORPHA
65759

Kod OMIM
201000

Kod ICD10
Q87.0

Kod ICD11
LD24.GY

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*Źródło

orphanet