

Opis choroby *

Definicja

Craniolenticulosutural dysplasia (CLSD), also known as Boyadjiev-Jabs syndrome, is characterized by the specific association of large and late-closing fontanels, hypertelorism, early-onset cataract and mild generalized skeletal dysplasia.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Boyadjiev-Jabs syndrome
	Zespół Boyadjiev i Jabsa

Kod ORPHA	Kod OMIM	Kod ICD10
50814	607812	Q75.8

Kod ICD11
LB70.Y

*Źródło

orphanet