

Opis choroby *

Definicja

Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome is a developmental anomalies syndrome characterized by coloboma of the iris and optic nerve, facial dysmorphism (high forehead, microretrognathia, low-set ears), intellectual deficit, agenesis of the corpus callosum (ACC), sensorineural hearing loss, skeletal anomalies and short stature.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Graham-Cox syndrome
	Zespół Grahama i Coxa

Kod ORPHA	Kod OMIM	Kod ICD10
52055	300472	Q87.8

Kod ICD11

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*Źródło

orphanet