Opis choroby *

Definicja

A rare, genetic dysostosis malformation syndrome characterized by skeletal dysplasia (rabbit ear-shaped iliac alae, delayed bone age, abnormalities of the vertebral bodies and schisis of the vertebral arches), seizures, short stature, cerebral atrophy and moderate to severe intellectual disability. Additional variable manifestations include corneal and retinal abnormalities, cataract, prognathism, dental malocclusion, brachydactyly, clinodactily, slight generalized hypotonia and hyper extensible joints.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Gurrieri-Sammito-Bellussi syndrome
Zespół Gurrieri, Sammito i Bellussi

Kod ORPHA
1858

Kod OMIM
601187

Kod ICD10
Q87.5

Kod ICD11
LD24.8Y

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*Źródło

orphanet