

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

Kod ORPHA	Kod OMIM	Kod ICD10
1858	601187	Q87.5

Kod ICD11
LD24.8Y

*Źródło

orphanet