

Opis choroby *

Definicja

A total autosomal trisomy that is caused by the presence of a third (partial or total) copy of chromosome 21 and that is characterized by variable intellectual disability, muscular hypotonia, and joint laxity, often associated with a characteristic facial dysmorphism and various anomalies such as cardiac, gastrointestinal, neurosensorial or endocrine defects.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Trisomy 21
	Trisomia 21

Kod ORPHA	Kod OMIM	Kod ICD10
870	190685	Q90.9

Kod ICD11
LD40.0

*Źródło

orphanet