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
Zespół wad wrodzonych

Synonimy
Trisomy 21
Trisomia 21

Kod ORPHA
870

Kod OMIM
190685

Kod ICD10
Q90.9, Q90.0

Kod ICD11
LD40.0

---

*Źródło

orphanet