Tetrasomia 21
Kod Orpha: 96055 Kod OMIM:

Opis choroby *

Definicja
Tetrasomy 21 is an extremely rare autosomal anomaly resulting from the presence of 4 copies of chromosome 21, characterized by features of trisomy 21 including developmental delay/intellectual disability, muscular hypotonia, short neck with redundant skin, brachycephaly, microcephaly, flat face, epicanthus, upslanted palpebral fissures, small ears, protruding tongue, single transverse palmar crease, brachydactyly, hypoplastic iliac wings, together with additional features such as prematurity, intrauterine growth retardation, high and broad forehead, hypertelorism. Haematological malignancies are also associated and may occur earlier than in trisomy 21.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Isochromosome 21
Izochromosom 21

Kod ORPHA
96055

Kod OMIM
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Kod ICD10
Q99.8

Kod ICD11
LD7Y

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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