Opis choroby *

Definicja

Paternal uniparental disomy of chromosome 21 is an uniparental disomy of paternal origin that most likely does not have any phenotypic expression except from cases of homozygosity for a recessive disease mutation for which only father is a carrier.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
UPD(21)pat
UPD(21)pat

Kod ORPHA
96195

Kod OMIM
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Kod ICD10
Q99.8

Kod ICD11
LD45.1

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*Źródło

orphanet