Opis choroby *

Definicja

A uniparental disomy of maternal origin that does not seem to have an adverse impact on the phenotype of an individual. There is a possibility of homozygosity for a recessive disease mutation for which the mother is a carrier and specific phenotype depends on the inherited disorder.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
UPD(X)mat
UPD(X)mat

Kod ORPHA
261519

Kod OMIM
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Kod ICD10
Q99.8

Kod ICD11
LD45.0

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*Źródło

orphanet