

Opis choroby *

Definicja

Paternal uniparental disomy of chromosome 1 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	Synonimy
Zespół wad wrodzonych UPD(1)pat	UPD(1)pat

Kod ORPHA	Kod OMIM	Kod ICD10
251004	-	Q99.8

Kod ICD11
LD45.1

*Źródło

orphanet