Opis choroby *

Definicja

Ring chromosome Y syndrome is a rare chromosome Y structural anomaly, with a highly variable phenotype, mostly characterized by short stature, partial to total gonadal failure, sexual infantilism, genital anomalies (e.g. ambiguous genitalia, hypospadias, cryptorchidism), and azoospermia or oligozoospermia. Additional reported features include speech delay, obesity, and acanthosis nigricans. Gender dysphoria and comorbid bipolar disorder have also been observed.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Ring chromosome Y
r(Y)
r(Y)

Kod ORPHA
261529

Kod OMIM
-

Kod ICD10
Q98.6

Kod ICD11
-

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*Źródło

orphanet