Ring chromosome Y syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ring chromosome Y syndrome. ORPHA:261529

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.