49,XXXYY syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 49,XXXYY syndrome. ORPHA:261534

49,XXXYY syndrome is a rare gonosome anomaly syndrome characterized by a eunuchoid habitus with gynecoid fat distribution and shape, normal to tall stature, moderate to severe intellectual disability, distinctive facial features (e.g. prominent forehead, epicanthic folds, broad nasal bridge, prognathism), gynecomastia, hypogonadism, cryptorchidism, small penis and behavioral abnormalities (incl. solitary, passive disposition but prone to aggressive outbursts, autistic). Skeletal malformations, such as delayed bone age, fifth finger clinodactyly, elbow malformations and slow molar development, may also be associated.