16p13.3 microduplication syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 16p13.3 microduplication syndrome. ORPHA:96078

16p13.3 microduplication syndrome is a rare chromosomal anomaly syndrome resulting from a partial duplication of the short arm of chromosome 16 and manifesting with a variable phenotype which is mostly characterized by: mild to moderate intellectual deficit and developmental delay (particularly speech), normal growth, short, proximally implanted thumbs and other hand and feet malformations (such as camptodactyly, syndactyly, club feet), mild arthrogryposis and characteristic facies (upslanting, narrow palpebral fissures, hypertelorism, mid face hypoplasia, bulbous nasal tip and low set ears). Other reported manifestations include cryptorchidism, inguinal hernia and behavioral problems.