
CHROMOSOME 1p PARTIAL TRISOMY

Chromosome 1p partial trisomy is a rare chromosomal abnormality with a limited number of patients reported. Although a distinctive phenotype is not apparent, the reported patients do share some frequent features.

MAIN FEATURES: Craniofacial anomalies, growth delay, malformed ears

ABNORMALITIES

Growth: Growth delay

Performance: Developmental delay, motor delay, hypotonicity, mental retardation, seizures

Eyes: Ptosis, hypertelorism, long eyelashes

Craniofacies: Microcephaly, anteverted nares, micrognathia, cleft lip/palate, flat nasal bridge, high-arched palate, malformed ears, epicanthal folds

Other: Syndactyly, thoracic stenosis, adrenal hyperplasia, genital ambiguity, cryptorchidism

CLINICAL COURSE: Early demise has been reported in several patients.

CYTOGENETICS: Inherited rearrangements are unusual.

REFERENCE

Mohammed FM, Farag TI, Gunawardana SS, al-Digashim DD, al-Awadi SA, al-Othman SA, Sundareshan TS. 1989. Direct duplication of chromosome 1, dir dup(1)(p21.2-p32) in a Bedouin with multiple congenital anomalies. *Am. J. Med. Genet.* 32:353-355.



Note microcephaly, sloping forehead, hypertelorism, long eye lashes, anteverted nares, long philtrum, long, malformed pinnae, and micrognathia. (From Mohammed et al., 1989. *Am. J. Med. Genet.* 32:353–355. Copyright © 1989 John Wiley & Sons. Reprinted by permission of Wiley-Liss, Inc.)