Partial Trisomy 5p12-Q11.2 Resulting From a Marker Chromosome: A New Case Report with Attention Deficit Hyperactivity Disorder

H.B. Erdem, I. Sahin, S. Tasdemir And A. Tatar

GENETIC COUNSELING, Vol. 27, No 3, 2016, pp 295-303 H.B.

Summary:

Partial trisomy 5p12-q11.2 resulting from marker chromosome: a new case report with attention deficit hyperactivity disorder. Partial trisomy of chromosome 5 was first described by Lejeune et al. in 1964 on the short arm (12). The vast majority of the partial trisomy 5 cases include 5p duplications; however we reported a small supernumerary marker chromosome. General symptoms include developmental delay, mental retardation, seizures, respiratory difficulties, congenital heart defects, abdominal muscle hypoplasia and dysmorphic features such as macrocephaly, enlarged anterior fontanelle, dolichocephaly, upslanting palpebral fissures, epicanthal folds, hypertelorism, abnormal ears, midface hypoplasia, short nose, broad nasal bridge and microretrognathia. Arachnodactyly and club foot may be seen as cytoskeletal abnormalities and hypotonia may be determined in neurological exam. Here we reported a case with developmental delay, attention deficit hyperactivity disorder, mild mental retardation and dysmorphic features, caused by a new small supernumerary marker chromosome, generating partial trisomy 5p12-q11.2. To our knowledge, this small supernumerary marker chromosome has not been reported before. Severe type of partial trisomy 5 includes seizures, congenital heart defects, hypotonia and failure to thrive. Previously reported partial trisomy 5 cases, who showed severe phenotype, had usually duplicated 5p13 region. Therefore, patients, who do not have duplicated 5p13, showed mild phenotype. Also, duplication of the long arm of chromosome 5, may contribute to the milder phenotype and the longer survival in partial trisomy 5 patients. Attention deficit hyperactivity disorder, which we described in the present case, may be a result of partial trisomy 5, because it includes ADHD4 gene. This case may help better understanding the karyotype/phenotype correlation related to partial trisomy 5.