Further delineation of the phenotype maps for partial trisomy 16q24 and Jacobsen syndrome by a subtle familial translocation t(11;16)(q24.2;q24.1)

Abstract

We report on two cases of distal monosomy 11q and partial trisomy 16q due to a familial subtle translocation detected by FISH subtelomere screening. Exact breakpoint analyses by FISH with panels of BAC probes demonstrated a 9.3-9.5 megabase partial monosomy of 11q24.2-qter and a 4.9-5.4 megabase partial trisomy of 16q24.1-qter. The index patient displayed craniofacial dysmorphisms, mild mental retardation and postnatal growth retardation, muscular hypotonia, mild periventricular leukodystrophy, patent ductus arteriosus, thrombocytopenia, recurrent infections, inguinal hernia, cryptorchidism, pes equinovarus, and hearing deficiencies. In his mother's cousin who bears the identical unbalanced translocation, mild mental retardation, patent ductus arteriosus, hypogammaglobulinemia, recurrent infections, unilateral kidney hypoplasia, pes equinovarus, and hearing deficiencies were reported. Since only four descriptions of cryptic or subtle partial trisomies 16q have been published to date, our patients contribute greatly to the delineation of the phenotype of this genomic imbalance. In contrast to this, terminal deletions of the long arm of chromosome 11 cause a haploinsufficiency disorder (Jacobsen syndrome) in which karyotype-phenotype correlations are already being established. Here, our findings contribute to the refinement of a phenotype map for several Jacobsen syndrome features including

abnormal brain imaging, renal malformations, thrombocytopenia/pancytopenia, inguinal hernia, testicular ectopy, pes equinovarus, and hearing deficiency.

Zahn S, Ehrbrecht A, Bosse K, Kalscheuer V, Propping P, Schwanitz G, Albrecht B, Engels H. Further delineation of the phenotype maps for partial trisomy 16q24 and Jacobsen syndrome by a subtle familial translocation t(11;16)(q24.2;q24.1). Am J Med Genet A. 2005 Nov 15;139(1):19-24. doi: 10.1002/ajmg.a.30995. PMID: 16222663.