

Subtelomeric monosomy 11q and trisomy 16q in siblings and an unrelated child: molecular characterization of two der(11)t(11;16)

Abstract

We report here three children with a der(11)t(11;16), two sibs (patients 1 and 2) having inherited a recombinant chromosome from a maternal t(11;16)(q24.3;q23.2) and a third unrelated child with a de novo der(11)t(11;16)(q25;q22.1), leading to partial monosomy 11q and trisomy 16q. Fluorescent in situ hybridization (FISH) using bacterial artificial chromosome (BAC) clones and array-CGH were performed to determine the breakpoints involved in the familial and the de novo rearrangements. The partial 11 monosomy extended from 11q24.3 to 11qter and measured 6.17-6.21 Mb in Patients 1 and 2 while the size of the partial 11q25->qter monosomy was estimated at 1.97-2.11 Mb for Patient 3. The partial 16 trisomy extended from 16q23.2 to 16qter and measured 8.93-8.95 Mb in Patients 1 and 2 while the size of the partial 16q22.1->qter trisomy was 20.82 Mb for Patient 3. Intraventricular hemorrhage and transitional thrombocytopenia were found in both sibs but not in the third patient. The FLI1 gene, which is the most relevant gene for thrombocytopenia in Jacobsen syndrome, was neither deleted in family A nor in Patient 3. We suggest that a positional effect could affect the FLI1 expression for these two sibs. Deafness of our three patients confirmed the association of this anomaly to 11q monosomy and tended to confirm the hypothetic role of DFNB20 in Jacobsen syndrome hearing loss. Both sibs shared most of the features commonly observed in Jacobsen syndrome, but not the third patient. This confirmed that terminal

11q trisomy spanning 1 to 1.97-2.11 Mb is not associated with a typical Jacobsen syndrome.

Basinko A, Audebert-Bellanger S, Douet-Guilbert N, Le Franc J, Parent P, Quemener S, La Selve P, Bovo C, Morel F, Le Bris MJ, De Braekeleer M. Subtelomeric monosomy 11q and trisomy 16q in siblings and an unrelated child: molecular characterization of two der(11)t(11;16). *Am J Med Genet A*. 2011 Sep;155A(9):2281-7. doi: 10.1002/ajmg.a.34162. Epub 2011 Aug 10. PMID: 21834034.