Interstitial 11q deletion derived from a maternal ins(4;11)(p14;q24.2q25): a patient report and review

Abstract

We present a family with multiple cytogenetic abnormalities, identified through a girl with several dysmorphic features and cardiac problems, suspected for Jacobsen syndrome. Cytogenetic analysis showed a 46,XX,del(11)(qter) karyotype, which was confirmed by fluorescence in situ hybridization (FISH). Cytogenetic investigation of the parents showed a chromosome aberration in both: the father had a t(11;12)(p13;q22) translocation and the mother was carrier of an ins(4;11)(p14;q24q25). FISH analysis with an 11q-subtelomeric probe from the second-generation telomere clone set and BACs from 11q24-q25 suggested a complex maternal rearrangement. However, subsequent array analysis showed a single interstitial deletion in the proband, derived from the maternal insertion. The aberrant karyotypes in both parents implicated an increased risk of unbalanced fetal chromosome composition, thus high risk for a child with multiple congenital abnormalities. Therefore, during the next pregnancy, the couple opted for prenatal diagnosis by means of amniocentesis. An interphase FISH strategy for uncultured amniotic fluid cells predicted two possible unbalanced fetal chromosome constitutions. Karyotyping of cultured amniotic cells confirmed one of the predicted unbalanced cytogenetic options, demonstrating the value of a fast interphase strategy for parents who both are carriers of a chromosomal abnormality. In addition, we present an overview of patients with Jacobsen syndrome and an interstitial 11q deletion reported thus far in literature.

Van Zutven LJ, van Bever Y, Van Nieuwland CC, Huijbregts GC, Van Opstal D, von Bergh AR, Corel LJ, Tibboel D, Wouters CH, Poddighe PJ. Interstitial 11q deletion derived from a maternal ins(4;11)(p14;q24.2q25): a patient report and review. Am J Med Genet A. 2009 Jul;149A(7):1468-75. doi: 10.1002/ajmg.a.32714. PMID: 19449434.