Familial balanced translocation leading to an offspring with phenotypic manifestations of 9p syndrome

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ABSTRACT. We report two similarly affected cousins (children of monozygotic twin sisters) with phenotypic features consistent with 9p deletion syndrome, including dysmorphic craniofacial features (trigonocephaly, midface hypoplasia, upward-slanting palpebral fissures and long philtrum), intellectual disability and disorders of sex development. Initial cytogenetic examination showed normal karyotypes in the probands and their respective parents, though multiplex ligation probe amplification revealed a 1q terminal duplication and a 9p terminal deletion in both affected children. Further analysis by fluorescence in situ hybridization, identified a familial balanced cryptic translocation t(1;9)(q44;p23) in the mothers, showing the importance of the association of molecular cytogenetic techniques in clinical genetics, given the implications for the care of
patients and for genetic counseling.

**Key words:** Multiplex ligation prole amplification subtelomeric; Trigonocephaly; 9p deletion; Fluorescence *in situ* hibridization; Familial rearrangement; Genetic counseling