ISSN 0972-3757
INTERNATIONAL JOURNALOF HUMAN GENETICS

PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 8(4): 317-323 (2008)
DOI: 10.31901/24566330.2008/08.04.01

Mosaic Trisomy 1q Due to a *de novo* Translocation in a Foetus with Early Developmental Abnormalities (Karyotype 46,XY,der(14),t(1;14)(p11;p11.2)/46,XY) Delineation of Parent and Cell Stage of Origin

Thomas Eggermann[1], Ulrike Gamerdinger[2], Gisela Knöpfle[3], Ursula Pätzold[4], Ulrich Gembruch[5], Manfred Hansmann[6], Gesa Schwanitz[4] and Dagmar Hansmann[7]

[1] Institute of Human Genetics, University Hospital Aachen, Germany
[2] Institute of Pathology, University Medical Center of Giessen/Marburg, Germany
[3] Department of Pathology, University Hospital Bonn, Germany
[4] Institute of Human Genetics, University Hospital Bonn, Germany
[5] Department of Obstetrics and Prenatal Medicine, University Hospital Bonn, Germany
[6] Prenatal Medicine and Genetics, Bonn, Germany
[7] Institute of Prenatal Medicine and Genetics, Meckenheim/Bonn, Germany

KEYWORDS Mosaic trisomy 1q; de-novo translocation 1/14; paternal origin of structural aberration; combination of meiotic and postzygotic aberration

ABSTRACT Pure trisomies of the whole long arm of chromosome 1 are extremely rare and have been reported only once in association with mosaicism. We report on a malformed foetus with mosaic trisomy 1p11 to 1qter whose clinical features were partially in accordance with those of previously described trisomy 1q patients. An additional long arm of chromosome 1 was translocated onto 14p11.2 (karyotype: mos46,XY,der(14)t(1;14)(p11;p11.2)/46,XY). Mosaic formation of the partial trisomy 1 was investigated in seven different somatic tissues of first and second trimester pregnancy. The distribution of the pathologic cells was unequal, ranging from 4 to 93%. The duplicated region was paternal in origin. We were able to delineate two possible complex formation mechanisms involving paternal meiosis and postzygotic mitoses.