ISSN 0972-3757
International Journal of
HUMAN GENETICS

PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 14(2): 83-90 (2014)
DOI: 10.31901/24566330.2014/14.02.04

Prenatal Diagnosis of Fetus with Short Limbs Caused by Three Abnormal Chromosomes Inherited from Parents

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KEYWORDS Karyotypic Abnormality. Parental Transmission. inv(9). t(15;16). Skeletal Dysplasia

ABSTRACT A 29 year old woman with history of G2P0+1 and shortening of limbs in past pregnancy referred herself to undergo a fetal anomaly scan at 18th week of gestation. The ultrasonographic imaging of the present pregnancy admeasuring the growth of 12-13 weeks at 18th week detected hydrocephalous condition with short limbs and kyphotic spine. This report aimed at looking for chromosomal aberrations in association with the imaging results and discussed the case in light of available literature. Amniocentesis and conventional cytogenetic analysis was performed following the standard protocol of tissue culture and chromosome banding. Karyotypic analysis of 50 metaphases detected an abnormal female karyotype with 46,XX,inv(9)(p11q13),t(15;16)(q15;q22) pattern. The karyotype revealed two constitutive abnormalities involving four break-points on three different chromosomes in a female genome. Upon counseling, the parents decided to terminate the pregnancy. However, at delivery the external genitalia of the female fetus was found to be of male phenotype and ambiguous. Parental karyotyping revealed transmission of inversion from mother and the balanced translocation from father. Finally the fetal karyotype was expressed as 46,XX,inv(9)(p11q13)mat,t(15;16)(q15;q22)pat.