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Partial Trisomy 2q(2q37.3 → qter) and Monosomy 7q(7q34 → qter) Due to Paternal Reciprocal Translocation 2;7
: A Case Report

We report an unbalanced translocation involving chromosome 2 and 7 due to a balanced reciprocal translocation 2;7 in the father. The female fetus had a partial trisomy of the long arm of chromosome 2 with a partial monosomy of distal 7q. Ultrasound at the first trimester had indicated normal fetal anatomy, including normal intracranial structures. Parental karyotypes showed a paternal balanced translocation: 46,XY,t(2;7)(q37.3;q34). The unbalanced translocation in the fetus resulted in trisomy for 2q37.3 →qter and monosomy for 7q34→qter. Postnatal examination showed that the female abortus had a cleft lip and palate, and mild dysmorphic features. The clinical phenotype was in agreement with previous descriptions and allowed us to propose a fetal phenotype for this chromosomal abnormality.

Key Words: *Prenatal Diagnosis; Chromosomes, Human, Pair 2; Trisomy 2q; Monosomy 7q; Translocation 2;7*

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INTRODUCTION

Partial trisomy 2q is a rare chromosomal aberration with only 9 cases reported in the literature (1, 2) since the first report by Ricci et al. (3). Most cases were the result of parental translocation or insertion and isolated partial trisomies for the long arm of chromosome 2 were represented in only 6 cases (4). We report a case of abnormal fetus that revealed an unusual karyotype of 46,XX,der(7)t(2;7)(q37.3;q34), resulting from paternal t(2;7) reciprocal translocation.

CASE REPORT

A 43-yr-old woman was referred for genetic counseling at 6 gestational weeks because of frequent miscarriages of 3 times. Detailed history revealed that her husband's younger sister gave birth to an anomalous child. The patient gave birth to a normal male child following a missed abortion and experienced two more missed abortions after a live birth. Analysis of the father's blood showed a balanced reciprocal translocation between the long arm of chromosome 2 and the long arm of chromosome 7,46,XY,t(2;7)(q37.3;q34). Karyotypes of the patient and her live male child were normal. Prenatal ultrasonogram at 13 gestational weeks showed no abnormalities in fetal structure, especially normal nuchal translucency without decrease or increase of amniotic fluid. Amniocentesis was thus performed at 16 weeks of gestation. Cytogenetic analysis using GTG banding revealed partial trisomy 2q and distal 7q monosomy, 46,XX,der(7)t(2;7)(q37.3;q34), resulting from paternal reciprocal translocation, t(2;7). The parents opted to terminate the pregnancy. At 18 weeks of gestation, a female abortus measuring 20 cm in length and 370 g in weight was delivered. The pathologic analysis of placenta was apparently normal. Physical examination of this proband showed dysmorphic features of a high forehead, a flattened face, narrow and upward slanting palpebral fissures, fine eyebrows, flat nasal bridge, low-set and posterioly rotated ears, cleft lip and palate, a mouth with down-turned corners, and retrognathia (Fig. 1). Other internal organs were normal.

DISCUSSION

There have been case reports on trisomy 2q but not identical with the present case (3-5). To our knowledge, this is the first case report in Korea. Many reported cases of partial trisomy 2q resulted from unbalanced product of reciprocal translocations and therefore were partial trisomic for the terminal part of 2q, often accompanied by partial monosomy elsewhere. Partial 2q trisomy is a well-known syndrome characterized by the rare occurrence of internal malformations (6)

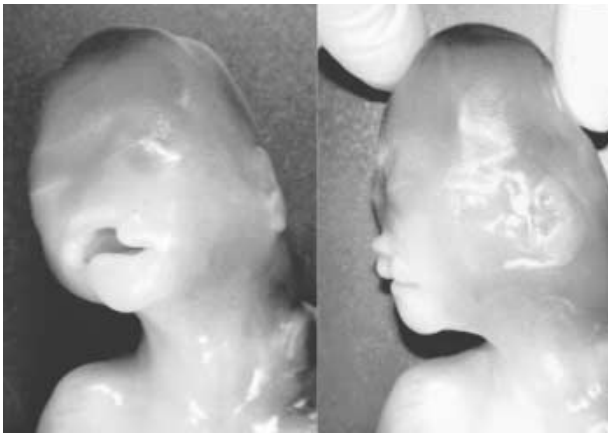

Fig. 1. Facial appearance of the 2q37.3 →qter trisomic fetus.

and a distinctive facial phenotype.

The findings of facial anomalies in the present case were similar to the previous reports except cleft lip and palate, and the present case lacked major internal malformations as in the case described by Moller et al. (5). SHH (Sonic hedgehog) is the gene that provokes the cleft lip and premaxillary agenesis and is located on 7q36, so monosomy 7q34→qter in this case is thought to be associated with cleft lip and palate. There seems to be some relationship between the extent of the trisomic region and the severity of the clinical manifestations, with the cases trisomic for a more extensive part of 2q being associated with internal malformations (7). The present case had a partial trisomic region on the terminal part of 2q, which could explain the absence of internal malformations of the fetus.

The findings of the present case confirm the phenotype of a partial 2q trisomy and emphasize the importance of parental chromosome studies in cases of familial structural abnormalities and frequent miscarriages.

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