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A Case of Partial Trisomy 13 Findings with 46,XX,der(7)t(7;13)(p22;q31)mat Karyotype

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Authors have shown that the trisomy of the distal part of chromosome 13 is related to different clinic findings than cases with classic trisomy 13 (1,2). They reported that different trisomic segments of the long arm of chromosome 13 might be either translocated or inserted in different chromosomes (1,3-17). According to the literature, this is the first report of a case where the trisomic segment of chromosome 13 is translocated to chromosome 7.

Case Report

Second degree related parents had three daughters and two sons. Our case was the fourth child of this family. She was born at term following a normal pregnancy. At the age of 6 years, her height was 112 cm (25-50 %), weight was 20 kg (50-75 %) and head circumference was 54.4 cm (97 %).

Typical craniofacial characteristics were as follows: trigonocephaly, high forehead, large and low-set ears, a hemangioma on the middle of the forehead, synophrys, bushy eyebrows, long and curled eyelashes, downslanting palpebral fissures, long philtrum, thin upper lip and high arc palate (Figure 1). She had minimal umbilical hernia,

additionally, the palms and the soles were erythematous and there were flexion contractures in the distal interphalangeal joints. There were hemangiomas of varied size on the occipital, thoracic and lumbar regions of her body. The liver was 3 cm palpable. The patient was observed to have a 2/6 systolic murmur in the mesocardiac and aortic loci upon auscultation. Neurological examination revealed severe mental retardation and that her deep tendon reflexes were hypoactive. She started to walk at the age of five, and was not able to walk easily. She could speak a few words only and could not even make a sentence.

Radiographic examination indicated that her bone age was concordant with calendar age and cranial tomography was normal. Abdominal ultrasonography confirmed the clinically evident hepatomegaly. Moreover, an increase in the paranchymal echo and in the wall echos of intrahepatic paths of the gall bladder was observed. It was seen by echocardiography that the patient had ASD and VSD.

In the hemoglobin electrophoresis HbA was 97.8 % and HbA₂ was 2.2 %. Hematologic analysis also revealed an increased frequency of nuclear projections in the neutrophils.



Figure 1. Facial Appearance of the Patient

We observed an ulnar loop in the 1st and 5th fingers, a radial loop in the 2nd finger and a arch pattern in the 3rd and 4th fingers in the right hand. There was an ulnar loop pattern in the 1st and 5th fingers, an arch pattern in the 2nd and 4th fingers and a whorl pattern in the 3rd finger of the left hand. Her right foot had a distal loop pattern and her left foot an S fibular arch pattern.

In the analysis carried out using Tripsin Giemsa G banding (GTG), the karyotype 46,XX,der(7)t(7;13)(p22;q31) was determined in our patient (Figure 2). Her mother and siblings were investigated while paternal evaluation was impossible due to his death three years previously. Although her siblings had normal karyotypes, abnormality in our patient was seen to result from the fact that the mother with the karyotype 46,XX,t(7;13)(p22;q32) was a balanced carrier (Figure 3). FISH analysis using the specific telomeric probe for chromosome 7p (Cat No: PCT 007, Cytocell) confirmed translocation and the signal was not observed on the derivative chromosome 7p end (data not shown). Maternal cytogenetic examination also confirmed the translocation between chromosomes 7 and 13 and the same 7p telomeric probe gave a clear signal at the deleted chromosome 13q end (data not shown). When scrutinized, the mother's parents were seen to have normal karyotypes.

Discussion

Partial trisomy of the distal segment of chromosome 13 (13q14→qter), has a characteristic phenotype

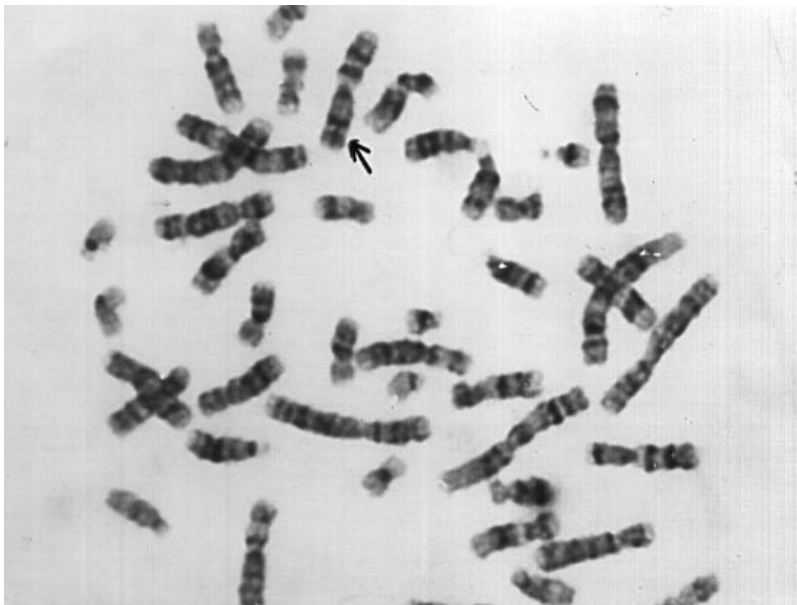


Figure 2. The Karyotype of the Patient (GTG banding)
(The arrow shows the derivative chromosome 7)



Figure 3. The Karyotype of the Mother (GTG banding)

(The large arrow shows the translocate chromosome 7 and the small arrow the deleted 13th chromosome)

associated with severe mental deficiency, frontal capillary hemangiomata, a short nose with upturned tip and elongated philtrum, synophrys, bushy eyebrows and long, incurved lashes and a prominent antihelix. Trigenocephaly and arinencephaly are seen occasionally. One fourth of the patients are generally lost in the early postnatal period (2).

Clinical findings of the case made us believe that the excessive segment in chromosome 7 belongs to the distal part of chromosome 13. FISH analysis confirmed that this cytogenetic event was a translocation, not an insertion. The reciprocal translocation between the distal short arm of chromosome 7 and chromosome 13q31 led to both partial monosomy 7 and partial trisomy 13. However, loss of this chromosomal segment on chromosome 7 did not cause any additional abnormal phenotype suggesting that there is no remarkable gene or genes in that region.

Bonioli et al. classified the clinic findings of a total of 33 cases who had partial trisomy of distal part of the chromosome 13, which had been presented up until 1981, as to where trisomic segment is located (1). Their classification includes 1st, 2nd and 3rd groups whose trisomic segments are q12 or q13→qter (6 cases), q14 or q21→qter (18 cases) and q22 or q31→qter (9 cases), respectively.

The characteristic clinical findings of the latter group having the same localization as our case were summed up

as follows where relevant information could be obtained: 3000 g or above birth weight (5 of 6 cases), normal development (7 of 7 cases), psycho-motor retardation (8 of 8 cases), microcephaly (3 of 8 cases), prominent forehead (8 of 9 cases), long and curled eyelashes (7 of 7 cases), short and broad nose (8 of 8 cases), long philtrum (7 of 7 cases), high palate (8 of 9 cases), prominent antihelix (9 of 9 cases), postaxial hexadactyly (7 of 9 cases), capillary hemangioma (6 of 6 cases).

Except for microcephaly and postaxial hexadactyly, all these findings were observed in our patient. According to Bonioli et al., the ratio of microcephaly in the first group (15/19) is much higher than that of the third group. This led us to think that the genes causing microcephaly are localized at the proximal rather than q31. The frequency of potential hexadactyly is reported to be the same in the three groups. The third group comprises the cases having q22 excluding trisomic segment in our subject, which strengthens our former idea that the genes belonging to postaxial hexadactyly are more proximal than q31, as in microcephaly.

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