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A CASE OF SPONDYLOCOSTAL DYSOSTOSIS WITH A FRA (5) (q32)*

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SUMMARY: Satar M, Temoçin AK, Atıcı A, Demirhan O. (Division of Neonatology, Department of Pediatrics, Çukurova University Faculty of Medicine, Adana, Turkey). A case of spondylocostal dysostosis with a fra (5) (q32). Turk J Pediatr 1997; 39: 547-549.

Spondylocostal dysostosis is a rare hereditary syndrome with various costal and vertebral deformities. No chromosomal abnormalities in connection with this syndrome were previously reported in literature. We report a case with spondylocostal dysostosis and chromosomal abnormality [fra (5) (q32)]. *Key words: spondylocostal dysostosis, chromosomal abnormality.*

Spondylocostal dysostosis is an uncommon heritable disease with multiple chondral and vertebral deformities (including cleft vertebrae, hemivertebrae and butterfly vertebrae) and bizarre costal features, such as variations in the size and thickness of the ribs and in the widths of the intercostal spaces, and partial rib fusion or missing ribs[1].

"Syndrome of bizarre vertebral anomalies", "syndrome of costovertebral segmentation anomalies" "spondylothoracic dysplasia" are some of the names given to this condition. There are two major subtypes of dysostoses with different survival rates, associated malformations, and inheritance patterns[1,2]. Chromosomal abnormalities in spondylocostal dysostosis and spondylothoracic dysostosis were not previously reported.

In this paper, a boy is presented with spondylocostal dysostosis and a fragile site at 5q32.

Case Report

The propositus was born in Balcalı Hospital, Obstetric and Gynecology Departments of Çukurova University. The male infant was the product of a full-term pregnancy with an uncomplicated delivery. He was admitted to the Neonatal Intensive Care Unit of the same hospital due to respiratory difficulty. His weight was 3510 g (50^{th}-90^{th} percentile), length was 46 cm (25^{th}-50^{th} percentile) and head circumference was 36 cm (50^{th}-90^{th} percentile). Clinical examination

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revealed mild cyanosis, a short neck, a markedly short and wide chest, and a protruding abdomen (Fig. 1). Radiological findings included multiple vertebral deformities (hemivertebrae, cleft vertebrae and butterfly vertebrae) and marked costal anomalies (different widths of intercostal spaces and the absence of the 10^{th}, 11^{th} and 12^{th} ribs) (Fig. 2). Cranial and abdominal ultrasonographies were normal. Serum concentrations of amino acids and urinary excretion of mucopolysaccharides were normal. Chromosomal analysis of peripheral blood cells with G-banding and Giemsa stain revealed 46, XY, fra (5) (q32) (Fig. 3). This fragile site was seen in 20 metaphases. During the clinical course, the baby's respiratory difficulty gradually diminished and he was discharged on the 13^{th} day. At 14 months, the baby was healthy, but his length was 70 cm below the 3^{rd} percentile).

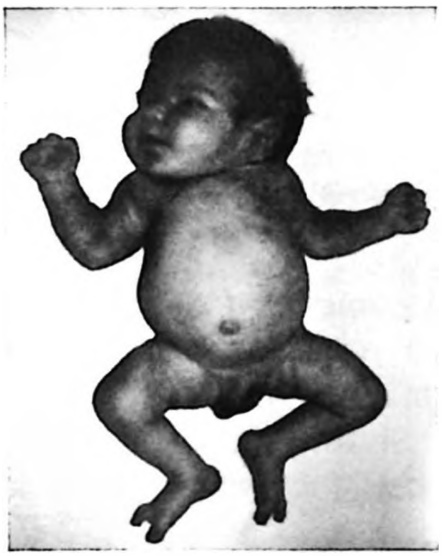

Fig. 1: General appearance of the case.

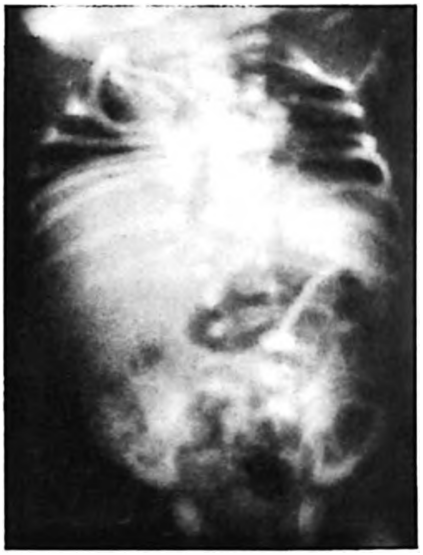

Fig. 2: Trunk anteroposterior roentgenogram of the case.

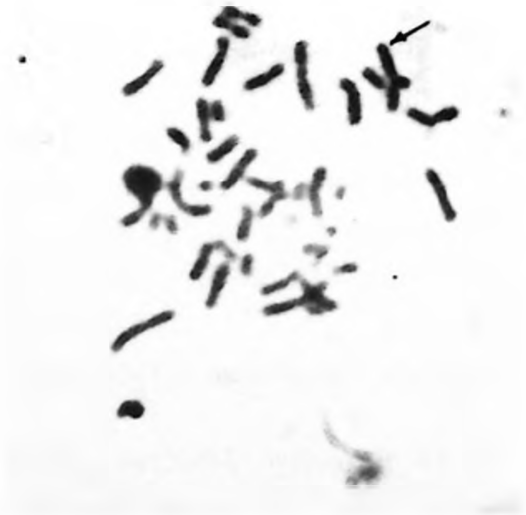

Fig. 3: Karyotype of the case (arrow indicates fragile site).

The father (33 years old) and the mother (28 years old), who were second cousins, were both healthy and their chromosomes were normal. The baby's two elder sisters were without deformities. However, the pedigree study did reveal a maternal uncle with chest deformities who died at 65 years of age.

Discussion

Chromosomal abnormalities in spondylocostal dysostosis were not previously reported. Some abnormalities on chromosome 5q were reported in certain skeletal dysplasia syndromes. Haastbacka et al.[3] concluded that the locus is probably in the 5q31-q34 area in diastrophic dysplasia. Cooke et al.[4] reported that a related gene may be located at 5q33.1 or 8q21.4 in camptomelic dysplasia. However, Maria et al.[5] observed a de novo paracentric inversion of 17q in an infant with camptomelic dysplasia. Chromosomal analysis revealed a fra (5) (q32) in this infant. We conclude that the gene related to spondylocostal dysostosis syndrome may be located on 5q32 or close to this region.

REFERENCES

1. Hall JG. Dysostoses. In: Emery AE, Rimoin DL (eds). Principles and Practice of Medical Genetics (2nd ed). London: Churchill Livingstone; 1990: 967-988.
2. Satar M, Kozanoğlu MN, Atilla E. Identical twins with an autosomal recessive form of spondylocostal dysostosis. Clin Genet 1992; 41: 290-292.
3. Hästbacka J, Sistonen P, Kaitila I, Weiffenbach B, Kidd KK, de la Chapella A. A linkage map spanning the locus for diastrophic dysplasia (DTD). Genomics 1991; 11: 968-973.
4. Cooke CT, Mulcahy MT, Cullity GJ, Watson M, Sragué P. Camptomelic dysplasia with sex reversal: morphological and cytogenetic studies of a case. Pathology 1985; 17: 526-529.
5. Maraia R, Saal HM, Wangsa D. A chromosome 17q de novo paracentric inversion in a patient with camptomelic dysplasia, case report and etiologic hypothesis. Clin Genet 1991; 39: 401-408.