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Case Report

Bilateral Palmar Single Transverse Crease in an Infant Girl with Ring Chromosome 13 and Multiple Facial Anomalies: A Case Report

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ABSTRACT

Background: Ring chromosome 13 is a rare cytogenetic disorder resulting from breakage and reunion of the distal ends of the chromosomal arms. The incidence of this disorder is about 1 in 58,000 live births. This syndrome usually presents with particular clinical features, including developmental delay, microcephaly, genital malformation in males, and dysmorphism, such as hypertelorism, broad nasal bridge, thin lips, up-slanting palpebral fissure, and ear anomalies.

Case Report: We report a 2-month-old girl who was referred to a clinical geneticist because of growth retardation and distinctive facial features. Her parents were consanguineous (second-cousins). The patient had a history of IUGR and was the product of a normal delivery at 39 weeks of gestation. At presentation, hypotonia, microcephaly, micrognathia, low-set ears, bilateral palmar single transverse creases (simian creases), broad nasal bridge, and thin lips were observed.

Conclusion: The karyotype revealed the presence of mosaic ring chromosome 13. This is the first report of a case of ring chromosome 13 with bilateral palmar single transverse creases.

Keywords: Cytogenetic, Microcephaly, Palmar single transverse crease, Ring chromosome 13, Simian crease

Introduction

Ring chromosome 13 is a rare cytogenetic disorder which was first reported by J Lejeune et al. in 1968 (1). The incidence of this disorder is approximately 1 in 58,000 live births (2). Similar to other ring chromosomes, ring misshaping of chromosome 13 is the result of breakage and reunion of the distal ends of the chromosome, which is associated with deletion or duplication of chromosomal segments (3).

The phenotype of patients with ring chromosome 13 is variable and seems to be dependent on the amount of deleted segments of chromosomal arms (4). The most common phenotypic features reported in this syndrome are growth retardation and developmental delay, microcephaly, unusual formation and position of

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feet or toes, abnormal facial features including broad nasal bridge, low-set ears with unusual shape, small jaw or chin, hypertelorism, and slanting eyes. Other rare anomalies have also been reported in hair, skin, brain structures, heart, kidneys, and GI system (5). We present a case of ring chromosome 13 in a 2-month-old infant girl with typical facial anomalies and single transverse creases of both hands (formerly known as simian creases). To the best of our knowledge, palmar single transverse creases have not yet been reported in the list of clinical manifestations of cases with ring chromosome 13.

Case report

A 2-month-old girl was referred to a genetic clinic because of abnormal facial features and growth retardation. She was the first child of a 19-year-old mother and a 32-year-old father. Both parents were healthy and consanguineous (second-cousins). Routine prenatal screening tests had been performed (NT, PAPP-A, beta hCG, inhibin A, AFP, and free estriol) and showed no serious problems. However, ultrasound imaging in the third trimester showed a decrease in the head circumference diameter and IUGR. She was born at full term (39 weeks) by vaginal delivery with a low birth weight (2,100 g), length of 49 cm, and small head circumference (29 cm). The brain ultrasonography and cardiac echocardiography findings were normal.

She was visited by a clinical geneticist at 2 months of age because of facial anomalies. At the time of the visit, her weight, length, and head circumference were 2,600 g, 51 cm, and 31 cm, respectively. Physical examination revealed the following anomalies: microcephaly, hypotonia, micrognathia, hypertelorism, broad nasal bridge, low-set ears, thin lips, bilateral palmar single transverse creases, and an increased space between the first and second toes in both feet. The external genitalia were also normal (Figure 1).

Karyotyping with GTG banding of the cultured peripheral blood lymphocytes was performed. Findings exhibited ring chromosome 13 as follows: 46,XX,r(13)(p11.2q34)[19]/46,XX,dic(13;13)(p11.2q34;p11.2q34)[1] (Figure 2).

Two different clones were observed with the dominant simple ring chromosome 13 with a breakpoint at p11.2q34. The second clone contained a dicentric ring chromosome 13. Chromosome 13 deletion was not observed in any metaphases. The parents refused to participate in further karyotype analyses.

Ethical Approval

This report was in compliance with the Ethics Committee of TUMS (registration number: IR.TUMS.CHMC.REC.1403.104).

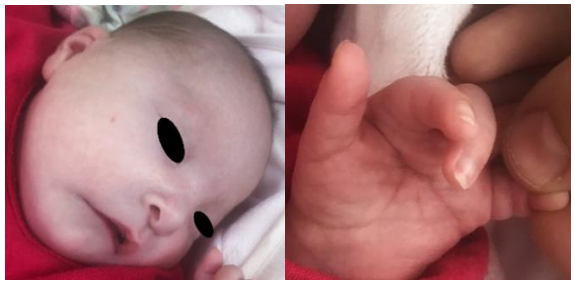

Figure 1. phenotypic features including microcephaly, broad nasal bridge, low set ear, small chin, hypertelorism, and slanting eyes. (left) palmar single transverse crease(right)

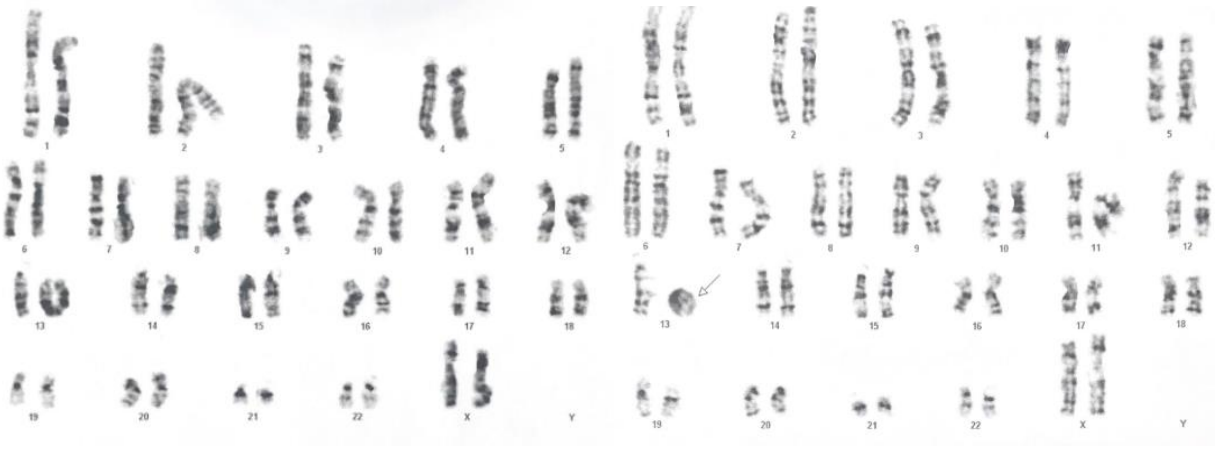

Figure 2. chromosome 13 ring formation, 46, XX, r (13) (p11.2q34) (left) and dicentric ring of chromosome 13, 46, XX, dic(13;13) (p11.2q34;p11.2q34) (right)

Discussion

Ring chromosome 13 is a rare cytogenetic abnormality and is the most common ring chromosome syndrome (6). The clinical features of this syndrome are variable, and it seems to be dependent on the extent of deleted segments of chromosomal arms, which is seen in karyotype but is best appreciated using array techniques (7).

Niebuhr and Ottosen classified these patients into three groups according to three different breakpoints. The first group, with a breakpoint at 13q33 or 13q34, showed severe mental retardation, microcephaly, hypertelorism, ear anomalies, and a broad nasal bridge. The second group had a breakpoint at 13q31 or 13q32, which presented all the aforementioned features of the first group as well as thumb aplasia or hypoplasia, genital and eye malformations, foot or toe anomalies, and anal atresia. The last group, with a breakpoint at 13q21, had a predilection for the development of retinoblastoma (8, 9). Some other studies do not support the above association between clinical features and breakpoints (7).

The clinical features of our case were similar to those of the first group of Niebuhr and Ottosen's descriptions, such as microcephaly, hypotonia, micrognathia, hypertelorism, broad nasal bridge, low-set ears, and thin lips. In addition, our case showed bilateral palmar single transverse creases, which had not been reported in previous cases of ring chromosome 13.

The palmar single transverse crease is a full-length transverse line formed by the fusion of the proximal transverse crease (PTC) and distal transverse crease (DTC) (10). Multiple genetic disorders and environmental factors can affect the pattern of palmar single transverse creases (11).

Some genetic diseases, such as trisomy 13/18/21, 4p, 18q chromosomes, and monosomy 21, are associated with a palmar single transverse crease. It has also been reported to be associated with cri-du-chat syndrome and triploidy (12). The prevalence of this sign differs in these disorders. It has been noted in 60% and 30% of trisomies 13 and 18, respectively (13). In Down syndrome, palmar single transverse crease (unilateral or bilateral) has been reported in 33.2% to 60% of cases (14, 15).

The prevalence of palmar single transverse crease in normal populations varies in different populations, both bilateral and unilateral. The highest prevalence was observed in African pygmies (34.7%) and the lowest prevalence was observed in the Swiss population (1.2%). In the Iranian population, this is approximately 2.5% (16). Bilateral palmar single transverse creases are very rare but do not have a preference for unilateral ones, indicating an increased possibility of associated genetic disorders (17-19).

Due to the higher association of palmar single transverse crease with genetic disorders, and with attention to the simple and painless examination of this sign, physicians should be informed about the importance of careful palmar examination in genetically suspicious patients, especially those with facial and structural anomalies.

Most previously reported cases of ring chromosome 13 were diagnosed during the first year of life, and some of them were detected in prenatal evaluation, which shows the importance of cytogenetic studies in infants with structural anomalies and unusual prenatal screening results during pregnancy (3, 4, 7, 20-22).

Our knowledge about the pathogenesis and molecular abnormalities of this syndrome is insufficient, and more precise methods (such as CGH array) to determine deleted and duplicated genes and their associated clinical features are required.

The majority (99%) of ring chromosomes 13 are sporadic due to accidental events in gametogenesis; however, rare cases are inherited from a parent, mostly the mother (23). Therefore, evaluation of parents' karyotype before the second conception, as well as prenatal diagnosis, could be helpful to prevent repeat anomalies in these families.

This highlights the interplay between genetics and clinical practice, emphasizing preventive approaches for managing future pregnancies in affected families.

Conclusion

Palmar single transverse crease is associated with various type of chromosomal anomaly. So palmar examination as a permanent part of infant physical examination could be helpful in diagnosis of these anomalies.

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Conflicts of interest

None declared.

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