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Case Report

Apert Syndrome: A Case Report

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ABSTRACT

***Background*:** Primary craniosynostosis is a form of premature fusion of the cranial sutures, which commonly occurs prenatally. The condition appears in both syndromic and nonsyndromic forms.

Case report: The cause of most cases of primary craniosynostosis are unknown, with genetic syndromes explaining 10%–20% of cases. The most prevalent syndromes associated with primary craniosynostosis are Crouzon, Apert, and Pfeiffer. Scaphocephaly is the most typical form of craniosynostosis that occurs due to premature closure of the sagittal suture. Frontal plagiocephaly is another form of this condition that is caused by the premature fusion of a sphenofrontal or coronal suture. The suture line palpation at birth usually exhibits a bony ridge. In these case, head CT or skull radiograph may be prescribed. Some genetic types of craniosynostosis are triggered by FGFR1, TWIST, FGFR2, or FGFR3 mutations.

Conclusion: A rare congenital condition, Apert syndrome is associated with craniosynostosis and severe symmetrical syndactyly of the feet and hands. In this case study, the goal has been to present a newborn with all characteristics of a classical Apert syndrome.

Keywords: Apert, Craniosynostosis, Midface hypoplasia, Syndactyly

Introduction

Head circumference (H.C.) measurement is important parameter in the routine physical examination and may be an early indicator of intracranial pathology and dysmorphic syndromes (1).

A rare congenital disorder, Apert syndrome, is associated with midface hypoplasia, craniosynostosis, proptosis, hypertelorism, down-slanting, prominent forehead, flattened nasal bridge, palpebral fissures high-arched palate, ocular manifestations, and severe symmetrical syndactyly of the feet and hands (1-2). Eugene Apert, a French physician, discovered the acrocephalosyndactylia syndrome in 1906 (1). The Apert syndrome incidence is 1 per 65,000 to 200,000 population, and this syndrome is passed between generations in an autosomal dominant manner (3-4).

In this case study, we aimed to present a newborn diagnosed with Apert syndrome, based on dysmorphic facial manifestations and severe syndactyly of the feet and hands, in the light of the existing literature.

Case report

A full-term male newborn was presented with hyperbilirubinemia, head deformities, and symmetric syndactyly of the feet and hands. The baby was born through normal vaginal delivery. The parents had no medical condition and were in the their 30s. The birth weight, height, and head circumference of the infant were reported to be 3,115 grams, 56 cm, and 37 cm, respectively. Apgar score was reported to be 9 and 9 at minutes one and 5 after birth, respectively. All vital signs were within normal limits. This was the second baby of a young Torkman couple who was not relatives and had no family history of a similar condition. The first baby of this couple was a normal female child. The mother had a normal vaginal delivery and no history of infection, trauma and drug use during her pregnancy. The baby was born in Bojnord city of

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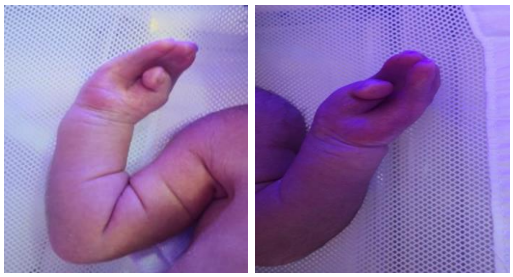
Figure 1. An infant with the cone-shaped calvarium, midface hypoplasia, hypertelorism, and down slanting palpebral fissures

North Khorasan province, Iran. He was kept under phototherapy for two days, and his total bilirubin decreased from 17.09 mg/dL to 7.14mg/dL.

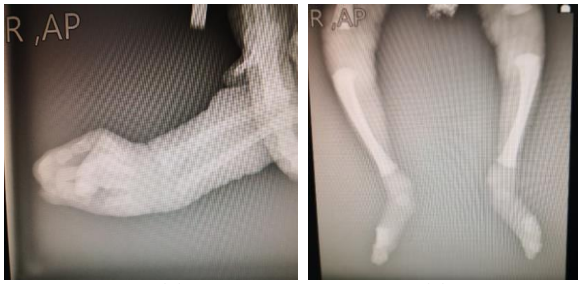
Physical examination of the infant revealed cone-shaped calvarium, midface hypoplasia, down slanting, hypertelorism, depressed nasal bridge, palpebral fissures, ocular proptosis, low set ears, and orbit shallow (Figure 1).

The severe symmetrical syndactyly of all fingers and toes and radial deviation of thumbs was observed as well (Figures 2, A, and B). The cardiovascular system and respiratory system of the newborn were normal, based on the physical examination. Both hands and feet radiographs exhibited soft tissue syndactyly of all the digits (Figures 3, A, and B). A three-dimensional computerized tomography image showed a midline defect that stretched from glabella to posterior fontanelle and bilateral symmetric synostosis of coronal suture (Figures 4A and B). All findings were diagnostic of Apert syndrome.



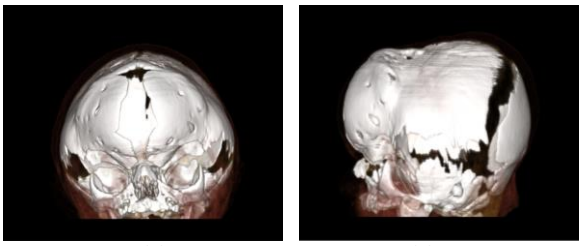
(A) (B)

Figure 2. Bilateral symmetrical syndactyly characterized with an inwardly placed thumb as well as complete fusion of all five digits of both hands.



(A) (B)

Figure 3. Radiograph manifesting soft tissue syndactyly of all the digits as well as synostosis that involve phalanges of the second, third, and fourth digits, as well as metacarpals of hands and feet



(A) (B)

Figure 4. 3D computerized tomography indicating a midline defect that stretches from glabella to posterior fontanelle with an abnormally wide posterior and anterior fontanelle

Discussion

An autosomal dominant disorder, Apert syndrome is induced by the mutation of fibroblast growth factor receptor-2 (FGFR-2) on chromosome 10q. Most cases are sporadic, arising from novel mutations with a paternal age impact (5). Apert syndrome Patients suffer from craniosynostosis, midline hypoplasia, palatal abnormalities, ocular disorder, down slanting palpebral fissures, keratoconus, and strabismus (6). Syndactyly or webbing leads to the immobility of fingers, which is caused by the interphalangeal joints' ossification. The first or fifth digits could be involved to varying degrees in this bony mass. There is an identical deformity that affects foot called mitten hand and sock foot (7). There are also neurologic, cardiovascular, gastrointestinal, genitourinary, and cutaneous issues (8-12).

The two common amino acid missense substitutions (Ser252Trp and Pro253Arg) on FGFR2 are responsible for over 98% of cases of Apert syndrome (13).

The incidence is reported to be approximately one case per 65,000 live birth. Apert syndrome does not depend on gender, and its prevalence is equal in both genders (14).

Craniosynostosis is also observed in Beare-Stevenson, Antley-Bixler, Jackson-Weiss, Crouzon Pfeiffer, and Saethre-Chotzen syndromes (15). The Apert syndrome treatment starts from the birth

requiring a multidisciplinary approach to come up with a collaborative remedial plan for the defects. Future advancements promise the non-surgical correction of Apert syndrome (e.g., by using selective inhibitors of the FGFR-kinase domain). Genetic counseling is of paramount importance as the risk of recurrence in the children of patients is 50% (16, 17).

Conclusion

A rare congenital condition, Apert syndrome is associated with craniosynostosis and severe symmetrical syndactyly of the feet and hands. In this case study, the goal has been to present a newborn with all characteristics of a classical Apert syndrome.

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Conflicts of interest

None declared.

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