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Apert's Syndrome: A Rare Case Report

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ABSTRACT

Background: Apert syndrome is a genetic disorder inherited in an autosomal dominant manner, with an occurrence rate of about 15 cases per 100,000 live births. It is caused by a mutation in the fibroblast growth factor receptor-2 (FGFR-2) gene located on chromosome 10q26. The condition mainly impacts the first and second branchial arches, leading to the early closure of cranial sutures (craniosynostosis) with fusion of fingers and toes of the hands and feet. Apert syndrome is rare in India, and a case report is presented.

Clinical Description: 21 months female toddler presented with global developmental delay along with distinctive craniofacial features. Clinically toddler exhibited an abnormal head shape and contour, characterized by turribrachycephaly, a depressed nasal bridge, frontal bossing, midface hypoplasia, and a characteristic "crossbow" appearance of the upper lip. Limb examination revealed symmetrical soft tissue syndactyly affecting all digits.

Management & Outcome: This case is notable for its rarity and the similarity of its features to other craniosynostosis syndromes, such as Crouzon and Pfeiffer syndromes, posing a diagnostic challenge. Therefore, genetic counselling for the family was recommended, along with early intervention for the child, including plastic surgery for the affected limbs.

Conclusion: Acrocephalosyndactyly is an autosomal dominant condition seen rarely, marked by craniosynostosis, craniofacial deformities, and pronounced symmetrical clubbing of fingers and toes of the hands and feet. In the majority of Apert syndrome cases, the condition occurs sporadically, often due to new mutations in the relevant gene.

Keywords: *Acrocephalosyndactyly, Craniosynostosis, Midface hypoplasia.*

1. INTRODUCTION

Apert syndrome (AS) was first described in 1894 by Wheaton, with the name later derived from a series of works by French physician Eugène Apert, who reported nine such cases. In 1906, Apert formally described the syndrome as acrocephalosyndactyly.

The incidence is approximately 15 cases per 1,000,000 live births[1].

AS is a rare autosomal dominant disorder caused by a mutation in the fibroblast growth factor receptor-2 (FGFR-2) gene on chromosome 10q26. This mutation affects the FGFR-2 expression by suture progenitor cells, leading to a disruption in signaling that impairs the production of the fibrous material needed for normal cranial suture formation [2].

Classified as a branchial arch syndrome, AS af- fects the first and second branchial arches. The hall- mark of the condition is the premature closure of cranial sutures (craniosynostosis), which restricts cranial growth and results in craniofacial abnormalities. Common features include frontal bossing, a narrow high-arched palate, midfacial hypoplasia, and symmetrical clubbing of fingers and toes of the hands and feet, with short, broad fused nails. While intellectual disability is often present, the exact in- cidence remains unclear[2,3].

AS is seen less commonly in India and one such case is reported.

Clinical Description:

A 21 months old female toddler presented with global development delay with distinctive facial features. Toddler had unusual craniofacial appearance, prompting a more detailed evaluation which led to a clinical diagnosis of Apert syndrome. A thorough examination was subsequently conducted.

The toddler was the first Caesarean section born daughter to non-consanguineous parents. Her birth history was uneventful, with no known exposure to infections, drugs, or radiation during her mother's pregnancy. There was no family history of similar malformations on either side.

On examination (Fig. 1), the child had atypical skull shape and contour, consistent with turribrachycephaly, along with a depressed nasal bridge, frontal bossing, midfacial hypoplasia, and a characteristic "crossbow" shape of the upper lip. The mouth was trapezoidal in shape, with a cephalometric dolichofacial pattern and proptosis.

Examination of the upper limbs (Fig. 2) revealed symmetric soft tissue clubbing of all digits, which almost fused into a single unit, with a single fused nail looking like a spoon-like deformity. The lower limbs also exhibited symmetrical clubbing of all toes, with a single broad fused nail. There was delayed developmental with Developmental Quotient (DQ) of 60%. No other systemic abnormalities were observed clinically.

Given the characteristic features, a clinical possibility of Apert syndrome was considered. Although AS usually has an autosomal dominant (AD) inheritance pattern, both parents were unaffected, suggesting a possibility of a sporadic mutation or gonadal mosaicism. The parents were advised to undergo further genetic testing for confirmation.

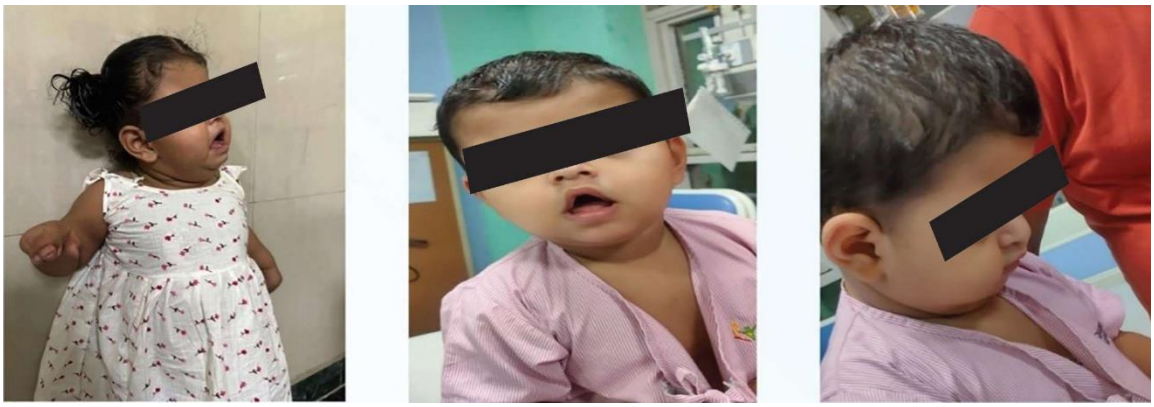

Figure 1: Abnormal shape and contour of the head.

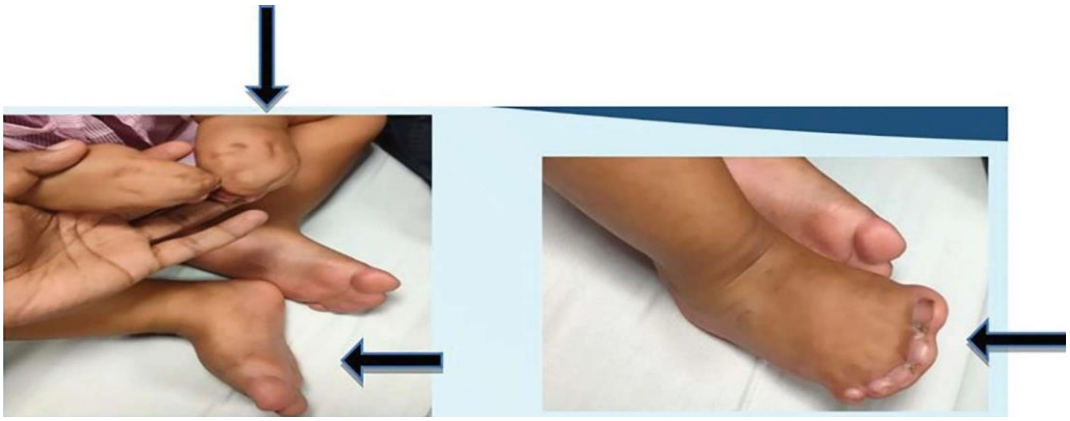

Figure 2: Limbs showing symmetric soft tissue syndactyly of all digits.

Management and Outcome:

Apart from being seen less commonly, AS has resemblance of its features to other craniosynostosis syndromes, such as Crouzon syndrome and Pfeiffer syndrome, thus pose a diagnostic challenge [2].

Management typically involves conventional surgical procedures, such as Le-Fort III osteotomy for midface advancement. Early intervention includes optimizing hearing, potentially with hearing aids, airway management, psychological counselling, and speech therapy[3,4].

Genetic counselling was recommended for the family, along with early intervention for the child, which includes plastic surgery to address the limb abnormalities[5].

2. DISCUSSION

Apert syndrome (AS) is a form of craniosynostosis and is characterized by premature craniosynostosis, hypertelorism, clubbing of hands and feet, Inheritance is generally autosomal dominant but may develops due to a mutation of fibroblast growth factor receptor -2 gene (FGFR2) located on 10q26 gene locus. FGFR2 gene enables coding of a protein required for suture closure. Two different types of mutations have been demonstrated in the binding site. Most of the patients have a normal karyotype [1].

Paternal mutations may be seen though most of the cases are sporadic, and develop because of new mutations. Incidence varies from 9.9 to 15.5 per one million live births with no sex variation.[1]

Phenotypic manifestations can be explained by premature closure of cranial sutures. Premature fusion of coronal sutures causes shorter anteroposterior diameter, high, and prominent forehead. The most prominent symptoms of this syndrome are clubbing of hands, and feet.[1,2]

This toddler had the clinical features of atypical skull shape namely acrocephaly, clubbing of

fingers, and toes with Mid-face hypoplasia. Eye

examination revealed hypertelorism, proptosis, and down slanting palpebral fissures. Nasal root was short and widened. This patient had all specific facial characteristics.[1]

AS children can have increased occurrence of upper respiratory tract infections, sleep apnoea and malnutrition. Respiratory difficulty can be severe requiring endotracheal intubation or tracheostomy. In patients with AS, mental disorders are rarely seen[1,2].

3. CONCLUSION

Acrocephalosyndactyly, is a less common inherited (autosomal dominant) disorder, characterized by craniosynostosis, craniofacial anomalies, and severe, symmetrical clubbing of the hands and feet. While many cases of AS have sporadic origin and result from de novo mutations in the FGFR2 gene, however the genetic basis remains crucial for diagnosis [1,2].

Genetic testing and counselling are essential for each diagnosed case. Advances in prenatal diagnostic techniques have made it possible to detect the condition early, enabling timely, multidisciplinary intervention. This early approach significantly improves the quality of life for affected individuals[5].

This case report aims to throw light on this rare and complex syndrome.

Lessons learnt

1. Apert syndrome is a rare inherited (autosomal dominant) disorder marked by craniosynostosis, craniofacial deformities, and severe, symmetrical clubbing of both the hands and feet.
2. The majority of Apert syndrome cases are sporadic, often arising from de novo mutations in the relevant gene.
3. Genetic counselling for the family was recommended, along with early intervention for the child, which includes plastic surgery for the facial and limb abnormalities.

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Conict of Interest: Nil

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