

# Apert Syndrome: A Case Report

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Date of Submission : 03.06.2022  
Date of Acceptance : 26.06.2022

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## Abstract

**Background :** Apert syndrome is a rare autosomal dominant disorder characterized by craniosynostosis, facial dysmorphism and syndactyly of hands and feet. Eugene Apert in 1906 describe the syndrome acrocephalosyndactyly.

**Case Presentation :** The reporting case was a 3 months old female baby presented with early fusion of all cranial bone, facial dimorphism like hypertelorism, cleft palate and syndactyly of both hands and feet. There was some bony defect also found in radiology.

**Conclusion :** It is a noncurable disease but treat with multidisciplinary approach can reduce the complication and prevention can be done by genetic counselling and antenatal ultrasonography.

**Key words:** Apert syndrome; Acrocephalosyndactyly; Midfacial hypoplasia; Craniosynostosis; Syndactyly.

## INTRODUCTION

Apert syndrome is a rare type acrocephalosyndactyly syndrome characterized by craniosynostosis, severe syndactyly of the hands and feet and dysmorphic facial features.<sup>1</sup> It was first described by Wheaton<sup>1</sup> in 1894 and subsequently further cases were reported by Apert. Apert syndrome, named after this French physician "Eugene Apert" who first described it in 1906 and is a relatively uncommon cranio-facial anomaly.<sup>2</sup> Apert syndrome presents autosomal dominant inheritance assigned to mutations in the fibroblast growth factor receptors (FGFR-2) gene at locus 10q26.<sup>1</sup> Males and females may be affected with equal severity.<sup>1</sup> Due to the high infant mortality rate, the incidence in general population is lower, that is, 1:1,60,000.<sup>3</sup>

## CASE PRESENTATION

On 1st March 2022 patient 3 months old female child 1<sup>st</sup> issue of consanguineous healthy parent presenting with dysmorphic faces and syndactyly of both hand and both foot at Chattogram Maa Shishu-O-General Hospital. Antenatal and perinatal period was uneventful. There was no history of trauma, infection and taking any offending drug by her mother during pregnancy. There was no history of similar or any other type of congenital anomaly in her family. Examination revealed apparently small size head, flat occiput, closure of sagittal and coronal suture, anterior fontanel measuring 1.8 cm x 1.4 cm. There was hypertelorism, depressed nasal bridge, thick nose with bulbous tip and cleft palate also present. Bilateral symmetrical syndactyly with fusion of fourth and fifth finger of both hand. There was syndactyly present in both feet, where one foot shows syndactyly of all five toes and another one shows lateral 3 toes. Anthropometry shows weight-2.8 Kg, length- 52 cm and OFC- 32 cm. There was no other systemic or congenital abnormality was found.

Radiology of skull shows premature closure of all the suture with deformed skull and protrusion of mandible. X-ray hand shows polydactyly and X-ray foot shows number and size of bones are abnormal in both foot and focal gigantism in left great toe and 2<sup>nd</sup> toe. Echocardiography and abdominal sonography shows no abnormality.



Figure 1 Craniofacial dysmorphism



Figure 2 Bilateral syndactyly of hands

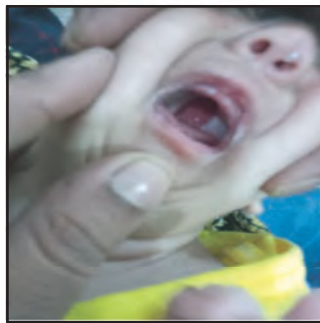


Figure 3 Cleft palate



Figure 4 Bilateral severe syndactyly

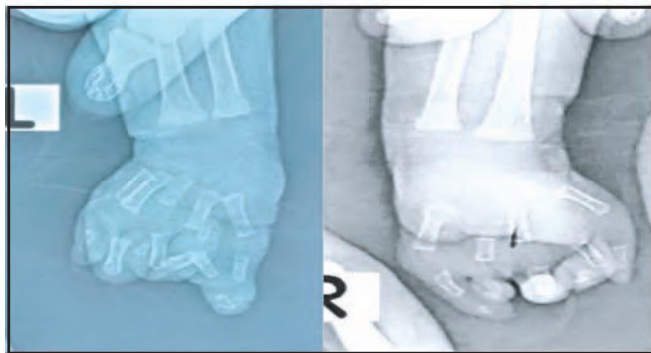


Figure 5 Bony abnormality of hands

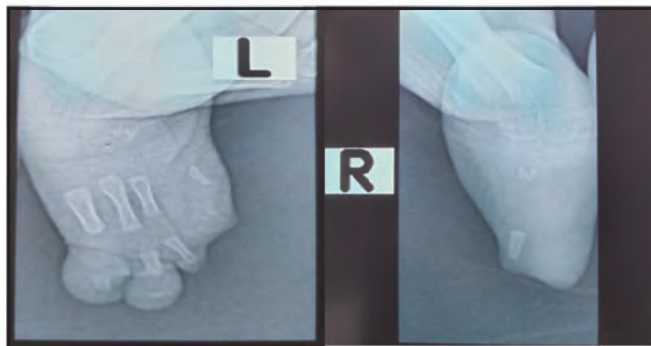


Figure 6 Bony abnormality of both feet

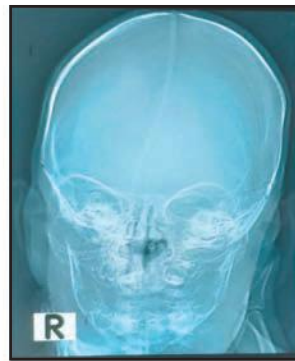


Figure 7 Craniosynostosis

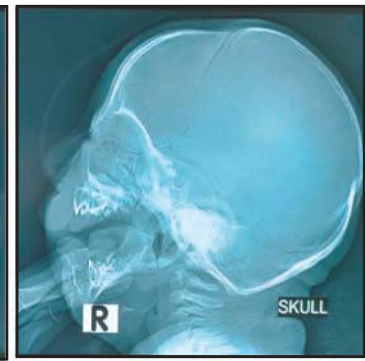


Figure 8 Craniosynostosis with mandibular protrusion

## DISCUSSION

Apert syndrome is a genetic disorder inherited in an autosomal dominant pattern. Almost all cases of Apert syndrome result from a sporadic or spontaneous mutation in the gene, and occur in people with no history of the disorder in their family.<sup>4</sup> People suffering with Apert syndrome, however, can pass along the condition to the next generation. Mutations in the Fibroblast Growth Receptor-2 (FGFR2) gene which is on chromosome number 10 cause Apert syndrome.<sup>2</sup>

In our case both parent and other family members have no such type of illness or other congenital anomaly.

Apert, in 1906, described the triad craniosynostosis, severe syndactyly of the hands and feet, and dysmorphic facial features, characterizing the syndrome.<sup>1</sup> In the reporting case all three criteria was present - craniosynostosis of all suture, small size head, brachycephaly, facial dysmorphism include hypertelorism, bulbous nose but turricephaly or exophthalmos was not present may be due to early age. Syndactyly of both hand and both foot was present.

Other skeletal defects include congenital cervical spine fusion, especially C5-C6 (68% cases). Cardiovascular anomalies are seen in 10% cases which include ASD, VSD, PDA, PS, TOF, COA etc. Genitourinary anomalies are seen in 9.6% cases ranging from polycystic kidney, hydronephrosis, duplication of renal pelvis etc. Gastrointestinal and respiratory system anomalies are uncommon seen in 1.5% cases.<sup>5</sup>

In the index case there was cleft palate and radiology of hand showed polydactyly and feet shows number and size of bones are abnormal in both foot and focal gigantism in left great toe and 2<sup>nd</sup> toe. No cardiovascular and genitourinary anomalies was found.

The differential diagnosis includes several such genetic disorders such as Crouzon syndrome, Carpenter syndrome, and Pfeiffer syndrome.<sup>6</sup> When compared to the Apert syndrome, in the Crouzon syndrome, extremities are unaffected, and craniofacial deformities with a milder course are noted, in the Pfeiffer syndrome, enlarged thumb and toes are typical, however, whereas in the Carpenter syndrome, the cloverleaf skull is a typical manifestation along with facial paralysis.<sup>7</sup>

Mortality and morbidity in children with Apert syndrome is due to upper airway as well as lower airway compromise causing early death, obstructive sleep apnoea and cor-pulmonale. Elevated ICP due to craniosynostosis is another cause of mortality. Many patients exhibit mental retardation but patients with normal intelligence have been reported.<sup>4</sup>

The treatment of apert syndrome begins at birth and a multidisciplinary approach is required arrive at a collaborative corrective plan for the deficiencies. Craniectomy is often performed during 6 months of age to treat the craniosynostosis. Corrective surgery for syndactyly is done in first year of life and completed by 3 to 4 years of age. Cosmetic correction for midface deficiency and pseudocleft is at 4 to 6 years age. Orthodontic and orthognathic surgery is performed after eruption of permanent dentition and completion of growth. Nonsurgical manipulation of Apert syndrome may be a possibility in the future, for example by using selective inhibitors of the FGFR-kinase domain.<sup>7</sup> Genetic counselling is an important factor as recurrence risk for an affected individual to have an affected offspring is 50%.<sup>8</sup>

Medical management of Apert syndrome includes corneal protection by instilling lubricating eye ointments and artificial tear drops. Management of upper airway obstruction by suctioning humidified oxygen and topical nasal decongestants and Sleep apnoea management by polysomnography and continuous positive pressure. Antibiotic treatment is required for chronic middle ear effusion.<sup>9</sup>

#### CONCLUSION

There is no specific cure treatment for Apert syndrome but multidisciplinary approach can minimize the complication. Though most of the cases are sporadic but prevention can be done by genetic counselling if there is family history or early diagnosis by prenatal ultrasonography and termination of pregnancy.

#### DISCLOSURE

All the authors declared no competing interest.

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