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Case Report

A Case Report on Apert Syndrome

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ABSTRACT

Apert syndrome (Acrocephalosyndactyly) is a rare congenital disorder characterised by craniosynostosis, midfacial malformation, and symmetric syndactyly of the hands and feet. The prodromal characteristics for the typical cranio-facial appearance are early craniosynostosis of the coronal suture, cranial base, and agenesis of the sagittal suture. The reporting case presented with several cranio-facial deformities, including brachycephaly, midface hypoplasia, flat face, hypertelorism, ocular proptosis, down-slanting palpebral fissures, and syndactyly with osseous fusion of the hands and feet. Intraoral findings included a high arched palate with pseudocleft in the posterior one-third.

Keywords: *Apert syndrome, Craniosynostosis, Syndactyly.*

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INTRODUCTION

Apert syndrome or acrocephalosyndactyly type-I is a rare genetic disease that is present from birth. It is one of the most severe craniofacial synostoses. In addition to craniosynostoses (Premature closure of coronal sutures), there is also craniofacial dysmorphism and malformation of fingers and toes[1,2]. This syndrome was first described by Baumgertner in 1842 and by Wheaton in 1894; later, it was extensively reviewed by French paediatrician Eugene Apert in 1906, hence its name[3]. Transmission occurs in an autosomal dominant mode; however, most cases are sporadic or can be associated with advanced paternal age, maternal infections, drug use during pregnancy, as well as a cranial inflammatory process[4]. With a prevalence of 1/50,000 to 1/65,000, this syndrome affects both sexes equally[2].

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CASE REPORT

An 8 months old boy presented with complaints of fever, cough, breathing difficulty, an abnormal head shape, and deformities in both hands and feet. Both parents were normal and in their third decade of life. He was the second child from a non-consanguineous marriage and had one sibling who was normal, and his mother had a caesarean delivery with no history of trauma, infection, or drug use during pregnancy. No family history of similar complaints or any other congenital abnormality was reported. He had age-appropriate milestones of development. On examination, his heart rate was 136 beats per minute, respiratory rate was 56 breaths per minute, and crepitations were present over both lung fields. He had a turribrachycephalic head contour, a flat occiput, and a protuberant frontal region. Ocular proptosis, strabism, hypertelorism, and down-slanting lateral palpebral

fissures were present. He had a depressed nasal bridge, a thick nose with a bulbous tip and a cross-bow-shaped lips. He had midfacial hypoplasia with a retruded maxilla (Figures-1 and 2). There were bilateral symmetric syndactylies with a complete fusion of all five digits of the hands and inwardly placed great toes. The fused fingers and toes had separate nails (Figures-3 and 4). Intraorally, there was absence of teeth, a v-shaped maxillary arch, and a narrow cleft palate. There was no other apparent congenital malformation. His chest x-ray showed bilateral patchy opacities, and complete blood count revealed neutrophilic leucocytosis. Radiograph of both hands and feet showed soft tissue syndactyly of all the digits and synostosis involving the phalanges and the metacarpals and metatarsals of both hands and feet with deformed great toes (Figure-5). Karyotyping could not be performed due to its unavailability. The baby was clinically diagnosed as bronchopneumonia with Apert syndrome. The patient was treated with antibiotics and other supportive measures for bronchopneumonia. After improvement, the patient was referred to a higher centre for reconstructive and orthodontic surgery.

Figure-1: *Facial features of Apert syndrome*

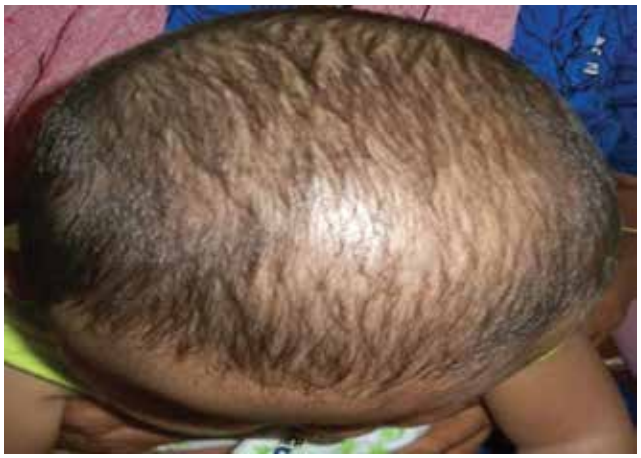

Figure-2: *Head findings of Apert syndrome*

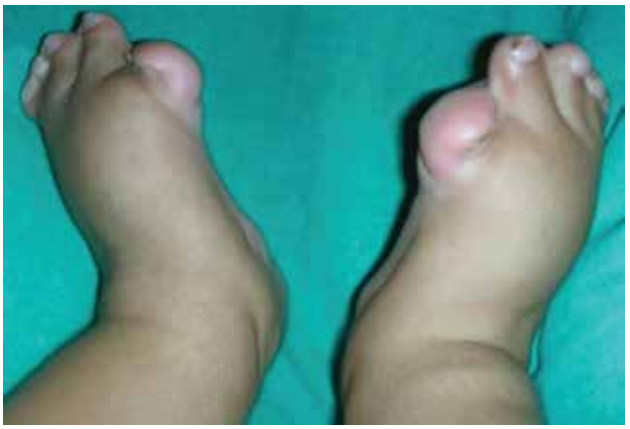

Figure-3: *Toe findings of Apert syndrome*

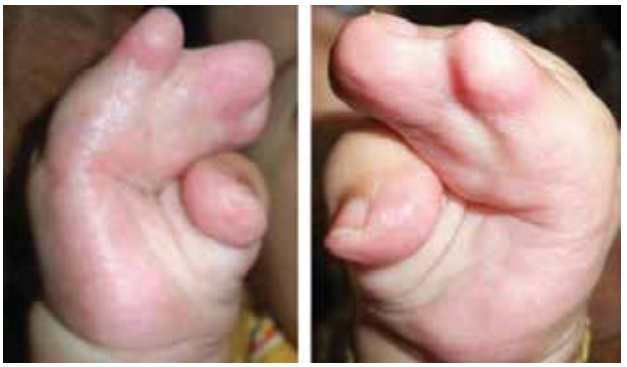

Figure-4: *Hand findings of Apert Syndrome*

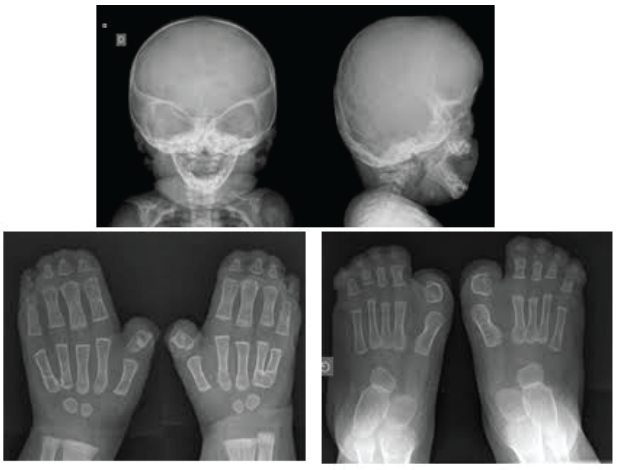

Figure-5: *Radiographic findings of Apert syndrome.*

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 families[5]. People suffering from Apert syndrome, however, can pass along the condition to the next generation. Mutation in the fibroblast growth factor receptor 2 (FGFR2) genes, which is located on chromosome 10, causes Apert syndrome[6].

Apert, in 1906, described the triad of craniosynostosis, syndactyly of hands and feet, and facial dysmorphism, characterising the syndrome[7]. In the reporting case, all three criteria were present: craniosynostosis of all sutures, small head, brachycephaly, facial dysmorphism including hypertelorism, bulbous nose, turricephaly, proptosis, and syndactyly of both hands and feet. Other skeletal defects include congenital cervical spine fusion, especially C5-C6 (68% cases). Cardiovascular anomalies are seen in 10% of cases, which include ASD, VSD, PDA, PS, TOF, COA, etc. Genitourinary anomalies are seen in 9.6% of cases, such as polycystic kidney, hydronephrosis, duplication of the renal pelvis, etc. Gastrointestinal and respiratory system anomalies are uncommon, seen in 1.5% cases[8]. In the index case, no cardiovascular, genitourinary, or GIT anomalies were found.

The differential diagnosis includes Crouzon syndrome, Carpenter syndrome, and Pfeiffer syndrome. When compared to the Apert syndrome, in Crouzon syndrome, extremities are unaffected and cranio-facial deformities with a milder course are noted. In the Pfeiffer syndrome, enlarged thumbs and toes are typical, whereas in the Carpenter syndrome, the cloverleaf skull is a typical manifestation along with facial paralysis[9].

Mortality and morbidity in children with Apert syndrome are due to upper airway as well as lower airway compromise, causing early death, obstructive sleep apnoea, and cor-pulmonale. Elevated intracranial pressure (ICP) due to craniosynostosis is another cause of mortality. Many patients exhibit mental retardation, but patients with normal intelligence have been reported[5]. The treatment of Apert syndrome begins at birth, and a multidisciplinary approach is required to achieve a collaborative corrective plan for complications. Craniectomy is often performed at 6 months of age to treat craniosynostosis. Corrective surgery for syndactyly is done in the first year of life and is completed by 3 to 4 years of age. Cosmetic correction for midface hypoplasia and pseudocleft is done at 4 to 6 years of age. Orthodontic and orthognathic surgery is performed after the eruption of permanent dentition and the completion of growth[10]. Medical management of Apert syndrome includes corneal protection by instilling lubricating eye ointments and artificial tear drops. Management of upper airway obstruction includes repeated nasopharyngeal suction, humidified oxygen therapy, and topical nasal decongestants. Sleep apnoea management by polysomnography and CPAP. Antibiotics are required for middle ear infection[11]. Our patient presented with pneumonia, was treated with antibiotics, and was then referred for surgical management.

Nonsurgical manipulation of Apert syndrome may be a possibility in the future, for example, by using selective inhibitors of the FGFR-kinase domain[11]. Genetic counselling is an important part, as the recurrence risk for an affected individual to have an affected offspring is 50%[12].

CONCLUSION

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

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