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Apert Syndrome (Acrocephalosyndactyly): A Rare Case Report

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ABSTRACT

Apert syndrome is the rare acrocephalosyndactyly syndrome, characterized by a classic triad of craniosynostosis, syndactyly of hands and feet, and midfacial hypoplasia. It demonstrates autosomal dominant inheritance attributed to mutations in the fibroblast growth factor receptor gene. Presently described is case of an 18-year-old female patient diagnosed on physical examination with Apert syndrome based on acrocephaly, ocular hypertelorism, frontal bossing, short and broad nose, dental crowding and ectopia, maxillary hypoplasia and bilateral syndactyly of hands and feet. The multiple phenotypic signs of Apert syndrome make multidisciplinary team, including dentist, neurosurgeon, plastic surgeon, physiatrist, ophthalmologist, perinatalogist and geneticist, essential for successful management.

KEYWORDS: Apert syndrome, Craniosynostosis, Syndactyly, Midfacial hypoplasia

INTRODUCTION

Apert syndrome is also known as acrocephalosyndactyly, characterized by a classic triad of craniosynostosis, hypoplastic middle third of face, and syndactyly of the hands and feet [(1,2)]. Cohen states that there are 15 cases of Apert syndrome per 10,000 live births [(3)]. Craniofacial deformities specific to Apert syndrome (AS) include acrocephaly (cone-shaped calvarium) or brachycephaly, prominent forehead, flat occiput, ocular proptosis, downward slanting palpebral fissures, exophthalmos and hypertelorism [(4)]. Nasal bridge is often depressed with a bulbous nasal tip resulting in a characteristic parrot-beaked appearance [(5)]. Oral signs may be enumerated as cleft palate, bifid uvula, high-arched palate, drooping angle of the mouth, hypotonic lips, dental crowding, delay in dentition, ectopic teeth, shovel shaped incisors and supernumerary teeth can be enumerated [(6)]. Presence of hypoplastic maxilla leads to mid faces deficiency and a more prominent mandible [(5)]. Rarely symptoms related to central nervous system, cardiac, gastrointestinal, and urogenital system, and vertebral anomalies have been reported [(4)]

Herein, we have aimed to present a 18-year-old female patient diagnosed as AS based on dysmorphic facial manifestations (Frontal bossing, Ocular hypertelorism, Proptosis, Absence of malar prominence, Incompetent lips, Mandibular prognathism, Depressed nasal bridge, Hypoplastic maxilla), Severe syndactyly of hands, and feet, in the light of the literature information.

CASE REPORT

An 18-year-old female patient presented with the complaints of irregularly placed teeth in the upper front teeth region since childhood. Her past medical history reveals history of epilepsy at the age of 1 year and not under medication, history of frontal advancement at the age of 1.5 years, history of frontoorbital advancement at the age of 5 years, history of syndactyly release of 2^{rd},3^{rd},4^{th} and 5^{th} bilateral fingers. Her personal history reveals history of delayed milestones of the child. Both the parents were normal and in fourth decade of life. She was the single child from a nonconsanguineous marriage and her mother had a normal delivery with no history of trauma, infection, drug use during the term. No family history of similar complaints or any other congenital abnormality was reported.

On General examination, short stature (Height - 112 cm tall and weighed - 65 kgs), dysmorphic facial features noted, deformities in fingers and toes noted. Evidence of short hands with broad and thick fingers on both hands. Evidence of complete fusion of middle finger and the ring finger was noted in her left hand and also evidence of partial fusion of middle and ring finger was noted in her right hand. Evidence of short and broad big toes on both legs with fusion of pointer and middle toe on both legs (syndactyly) (Figure 1)

Figure 1: Syndactyly of hands and feet

On Extraoral examination, ocular proptosis noted, depressed nasal bridge, midfacial hypoplasia, hypertelorism, increased lower facial height, mandibular prognathism, absence of malar prominence, frontal bossing, incompetent lips (Figure 2).

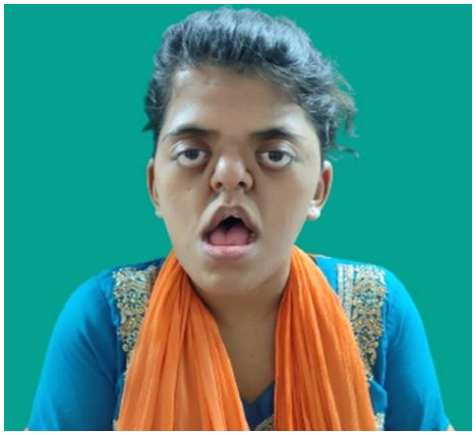

Figure 2: Profile photo

On Intraoral examination: Upper right and left canine were missing, V shaped maxillary arch, malpositioned : 15,18 , palatally placed 25,27. Root stumps was there in 47. Upper and lower arch crowding, Anterior open bite, Reverse overjet, palatal deep groove was noted in the midpalate region, Bifid uvula, Macroglossia, deep fissure noted in the dorsum of tongue was noted (Figure 3)

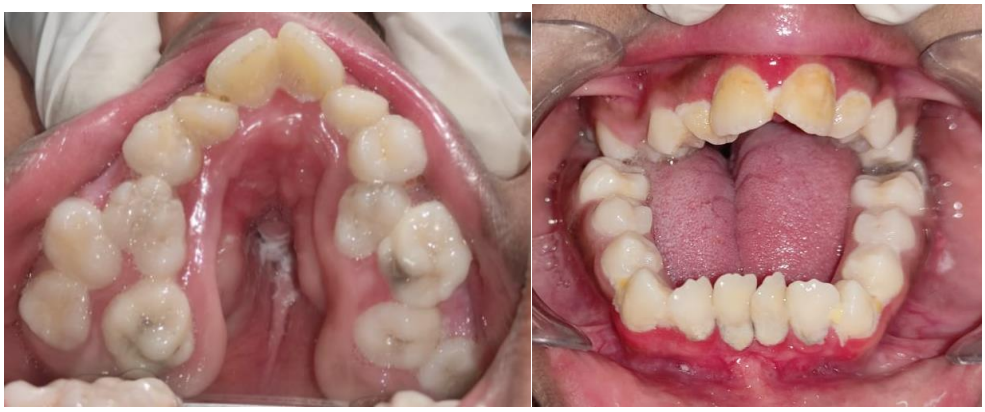

Figure 3: Intraoral Pictures

By correlating the patient's history and clinical findings, we arrived at a provisional diagnosis of Apert syndrome.

INVESTIGATIONS:

Patient was subjected to radiological examination where Orthopantomogram (Figure 4) revealed in zone 1: Total number of teeth present: 30, Missing: 13, 23. In 37, evidence of coronal radiolucency involving enamel, dentin and pulp with well-defined periapical radiolucency surrounded by radio opaque rim like structure. Suggestive of Dental caries with periapical cyst in 37. In 47, evidence of coronal radiolucency involving enamel, dentin and pulp with ill-defined periapical radiolucency. Suggestive of Dental caries with chronic periapical abscess 47. Altered root morphology with single canal in 37,38,47,48. In zone 2, deviated nasal septum to right side was noted. In zone 5, prominent antegonial notch was noted.

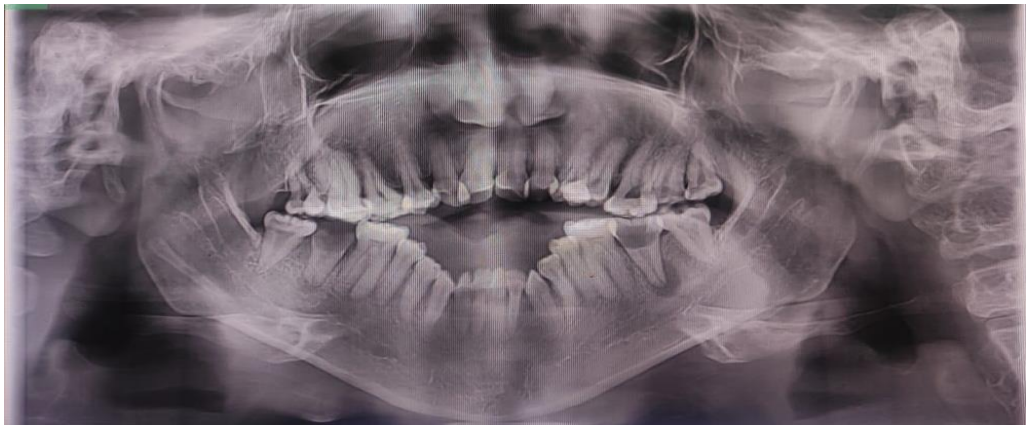

Figure 4: Orthopantomogram

Patient was subjected to Hand wrist radiograph (Figure 5). It shows short hands with fused distal and middle phalanx in the third and fourth digits with lateral bending of second digit in her right hand whereas fused distal and middle phalanx in the third, fourth and fifth digits with forward bending of second digit in her left hand. Evidence of broad phalanges in the first digits of both hands.

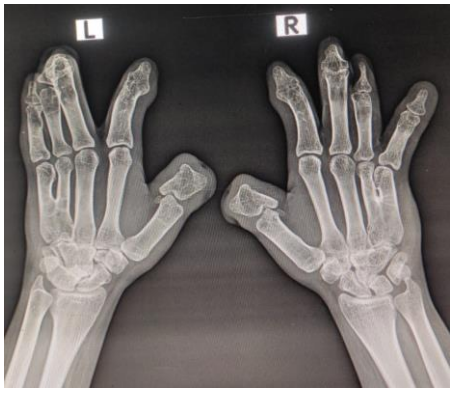

Figure 5: Hand wrist radiograph

Later patient was subjected to X-ray of foot (Figure 6) which revealed evidence of short and broad great toe in both the legs.

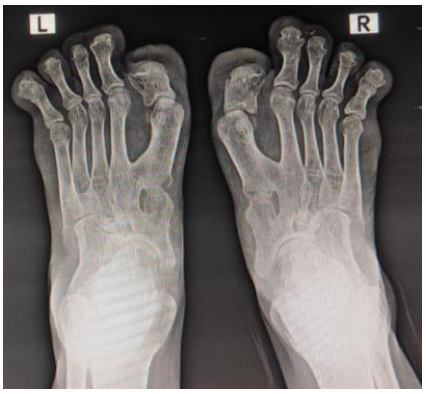

Figure 6: X-ray of foot

Lateral skull (Figure 7) revealed evidence of hazy radiolucent line on the cranial suture suggestive of premature closure of coronal suture. Open anterior fontanelle, midfacial deformity, multiple surgical radioopaque plates, pins and screws were noted in the cranial vault and also evidence of vertical disharmony of the posterior teeth leading to anterior open bite.

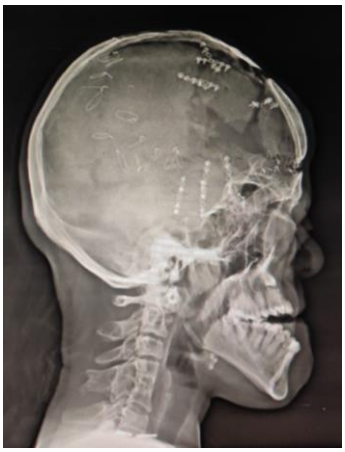

Figure 7: Lateral skull

Posteroanterior view of skull (Figure 8) was taken. It revealed facial asymmetry noted on right side and also evidence of multiple surgical radioopaque plates, pins and screws were noted in the cranial vault.

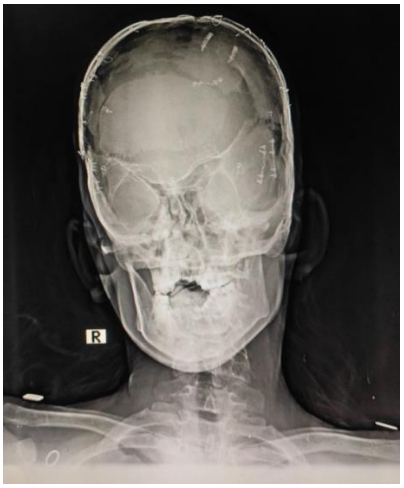

Figure 8: Posteroanterior view of skull

By correlating patient's history, clinical and radiographic findings, we arrived at a final diagnosis of Apert syndrome. Patient was advised for orthodontic treatment for correction of malocclusion and she is kept under regular follow-up.

DISCUSSION

Craniosynostosis are a heterogenous group of syndromes characterized by a premature sutural fusion that may occur alone or together with other anomalies [(7)]. More than 70 craniosynostosis syndromes are described, which amongst many others, include AS, Crouzon syndrome and Pfeiffer syndrome [(8)]. AS is classified as branchial arch syndrome, affecting the first branchial arch [(9)]. Apert syndrome is named after the French pediatrician Eugene Apert in 1906 as "acrocephalosyndactylia" characterized by a classic triad of craniosynostosis, hypoplastic middle third of face, and syndactyly of the hands and feet [(5)]. It is an autosomal dominant disorder and it is due to mutations in the gene coding for FGFR2 in chromosome locus 10q25. Such mutations lead to increase in the osteogenic precursor cells resulting in increased subperiosteal bone formation and premature ossification of calvaria during foetal development [(10)]. Cohen (2005) states that sutural agenesis in the midline region is characteristic of AS. This midline defect normally obliterates during 2^{nd}-4^{th} year of life [(11)].

Premature fusion of cranial sutures explains the disease's phenotypic features. Acrocephalic (cone-shaped) heads are associated with a high, prominent forehead and shorter anteroposterior diameter due to premature closure of coronal sutures before three months of age. The most prominent symptoms of this syndrome are syndactyly of hands, and feet [12,13]. Our patient also had the most marked symptoms namely acrocephaly, syndactyly of fingers, and toes [10].

The patient can have a flat occiput and a high, prominent forehead. Low-set ears with sporadic conductive hearing loss are observed. Downward slanting palpebral fissures, hypertelorism, shallow orbits, ocular proptosis, and exophthalmos may be present. Nasal bridge is often depressed with a bulbous nasal tip resulting in a characteristic parrot-beaked appearance. Syndactyly involves the hands and feet, with partial-to-complete fusion of the digits, often involving second, third, and fourth digits with a common nail, termed mitten hands and sock feet.

It has been reported that elbow, shoulder, vertebral column (vertebral malsegmentation, fusion, hemivertebra) deformities, lysencephaly, hypogenesis or agenesis of corpus callosum, ventriculomegaly, and infrequently, internal organ (renal, cardiac, gastrointestinal, genitourinary) involvement can all lead to mental disorders. Malnutrition, sleep apnea, and upper respiratory tract infections are observed in people with Apert syndrome [2,4,12,13,14].

By identifying a heterozygous pathogenic variant in FGFR2 through molecular genetic testing and phenotypic features consistent with Apert syndrome, the diagnosis of Apert syndrome is established in a proband with classic clinical characteristics (multisuture craniosynostosis, midface retrusion, and syndactyly) [15].

Various complications arising due to late diagnosis, include defective brain development, mental retardation, increase in facial deformity, prognathic mandible, etc. [16] Though functional improvement is generally small, surgical digit separation (mitten glove syndactyly) yields cosmetic results. [16,17] Planning the procedure will help define the digits and promote unhindered skeletal growth. Inconsistencies in finger length and growth, if surgery is delayed, typically result in more deformities to an already malformed hand [18].

CONCLUSION

The rarity of the AS, the typical craniofacial and dental features, the genetic transmission makes it necessary to carry out genotyping and genetic counseling of each diagnosed case. Prenatal diagnostic methods have advanced to the point where early detection and prompt interdisciplinary management are now possible, improving the quality of life for those who are affected. This case report attempts to throw some light on this rare syndrome.

Declaration by Authors

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