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# Apert syndrome in South Asian (Bangladeshi) male child: A rare case report

Maliha Nawar

## ABSTRACT

**Introduction:** To report a case of Apert syndrome (AS) in a 9-year-old South Asian Bangladeshi male child. This case is novel due to its rare combination of classical AS features with notable ophthalmic findings and early multidisciplinary intervention, which is seldom reported in similar settings.

**Case Report:** We presently describe the case of a 9-year-old male patient with prominent forehead, proptosis, short and broad nose, pseudoprognathism, crowding of teeth and ectopia, and severe bilateral syndactyly of hands and feet. Examination of the patient revealed the findings of abnormal contour of the head, symmetric syndactyly of hands and toes, mental retardation, delayed milestones, mid-face hypoplasia, exophthalmos, exorbitism, high arched palate, and crowded teeth. *Results:* From the clinical characteristic and radiology evaluation, the patient concluded with suspicion of AS.

**Conclusion:** We reported the case of a 9-year-old male patient with AS, characterized by craniosynostosis and acrocephalosyndactyly. The characteristics of clinical manifestation of AS should be aware by physicians to be able to diagnose the disease.

**Keywords:** Acrocephalosyndactyly, Apert syndrome, Craniosynostosis

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## INTRODUCTION

Cranial stenosis is premature healing of one or more sutures. Decreased or asymmetrical growth of the skull occurs and the calvaria or base is deformed. In 1851, Virchow found that growth stopped in the direction perpendicular to the affected suture line and continued in the direction parallel to it [1]. Common features include suture fusion, mid-face hypoplasia, and facial and limb abnormalities. Apert syndrome (AS) is one of the disorders defined by craniosynostosis, mid-facial hypoplasia, and symmetrical hands and ankles [2].

Apert syndrome, or acrocephalosyndactyly, is a rare, congenital anomaly with an incidence of 1 in 160,000 live births [3]. This syndrome was described in detail in 1906 by Eugene Apert, hence the name. Baumgartner first mentioned the syndrome in 1842, and Wheaton did so again in 1894 [4]. Apert syndrome is distinguished by craniosynostosis, exorbitism, mid-face hypoplasia, and symmetric syndactyly of both hands and feet. We report one such case.

## CASE REPORT

A 9-year-old South Asian boy hailing from Dhaka, Bangladesh, came to Bangladesh Medical University (BMU) Dhaka, outpatient department (OPD) with the complaints of conjunctival chemosis, with lagophthalmos for seven months along with symmetric syndactyly of both hands and feet, abnormal shape of the head and mental impairment, as demonstrated by a lack of speech development since infancy. He was born out of a nonconsanguineous marriage. The patient is his parents' first child and has one female sibling. No illness or medication use during pregnancy was mentioned. The birth went smoothly and on time. The patient's physical examination revealed aberrant contours of the

head (turribrachycephaly) and symmetric syndactyly of the second, third, fourth, and fifth digits of the hands (Figure 1) and all the toes of the feet (Figure 2), mental retardation, delayed milestones, mid-face hypoplasia, proptosis, hypertelorism, exorbitism (Figure 3), funnel chest (Figure 4), a high arched palate, and crowded teeth (Figure 5). The patient's ocular examination revealed exophthalmos and a visual acuity of 6/6 in both eyes. The patient additionally complained of frequent redness of the eyes due to keratitis. Radiographic examination revealed on both hands, syndactyly of the skin flexion, deformity of all fingers, enlargement of the distal phalanx of the thumb, and occasional subluxation of the distal phalanx (Figure 6). Radiographs of both feet revealed soft tissue syndactyly of all toes, as well as synostosis of the metatarsals. Phalanges of the great toe were deformed (Figure 7). A computed tomography (CT) scan of the brain indicated total loss of interdigitation in the coronal and partial loss on the left lambdoid suture, protrusion of both eyeballs, abnormal head shape, decreased nasal bridge, and a smaller orbital cavity (Figure 8). From the clinical characteristic and radiology evaluation, the patient concluded with suspicion of AS.



Figure 1: Symmetric syndactyly of second, third, fourth, and fifth digits of the hands.



Figure 2: Syndactyly of foot.



Figure 3: Clinical manifestation of patient.



Figure 4: Funnel chest.



Figure 5: High arched palate and crowded teeth.




Figure 6: Syndactyly of the skin flexion, deformity of all fingers, enlargement of the distal phalanx of the thumb, and occasional subluxation of the distal phalanx.




Figure 7: Soft tissue syndactyly of all the toes with synostosis involving metatarsals with deformed phalanges of great toe.

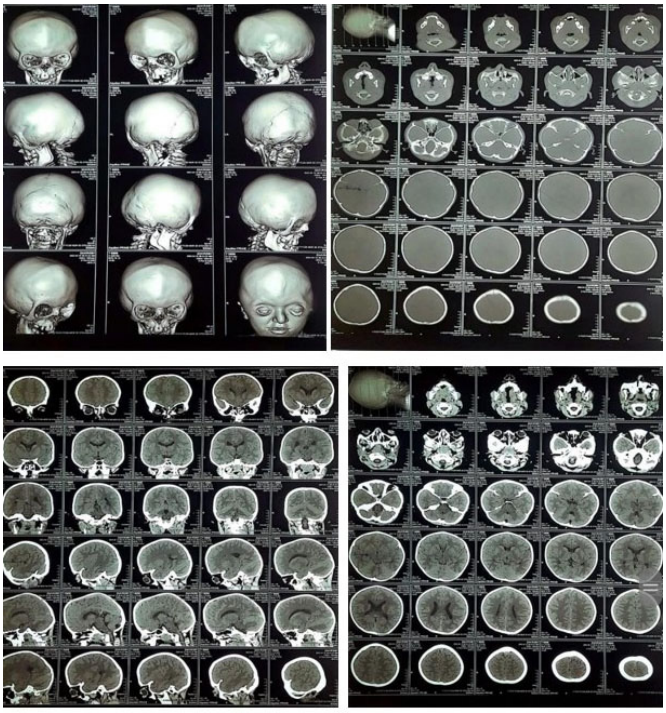


Figure 8: Complete loss of interdigitation in coronal and partial loss on left lambdoid suture, protrusion of both eyeballs, abnormal shape of head, decreased nasal bridge, and smaller orbital cavity.

## DISCUSSION

Acrocephalosyndactyly, a kind of cranial syndactyly, is another name for AS. The main features of AS are early cranial synostosis, hypertension, limb synostosis, and variable phenotypic manifestations. It is an autosomal dominant characteristic, sometimes may be sporadic and occurs as a mutation in the fibroblast growth factor receptor 2 (FGFR2) gene at the 10q26 locus. The FGFR2 gene encodes a protein named the fibroblast growth factor receptor 2. This protein is one of four FGFRs. The fibroblast growth factor receptor binds fibroblast growth factors with higher affinity and plays an important role in the signaling pathways active during craniofacial fusion [5]. It can be inherited from a parent who has Apert, or may be a fresh mutation. This mutation usually occurs in a sperm and AS is one of the few genetic conditions linked to older fathers, particularly men over the age of 50 [6]. The clinical features are distinctive. The coronal suture fuses early (within three months), resulting in an acrocephalic (cone-shaped) head with a decreased anteroposterior diameter and a high prominent forehead. The middle face is hypoplastic. Hypertelorism, proptosis, and palpebral fissures with a downward slant are examples of ocular abnormalities. The nose is short and broad, with a depression on the nasal bridge. Previous research has identified affected individuals with malformations of the viscera, elbows and shoulders, bones, and central nervous system, which frequently result in decreased mental function. The oral cavity of Apert patients is likewise distinctive. Clinical findings include a reduction in maxilla size, particularly in the anteroposterior direction. This reduction may cause dental crowding and an anterior open bite in the maxilla. The mandible is within normal size and shape, and simulates a pseudoprognathism. Common dental malformations include impacted teeth, delayed eruption, ectopic eruption, supernumerary teeth, and thick gingiva. Abnormalities of the upper and lower respiratory tracts include cleft soft palate, bifid uvula,

Byzantine-arch palate, choanal stenosis, and tracheal cartilage defects. Mouth breathing, reported in most cases of Apert's disease, is associated with changes in facial growth. Syndactyly involving partial to complete fusion of the digits results in a spoon-like deformity of the hands and feet [7]. Our patient had abnormal contour of the head (turribrachycephaly), symmetric syndactyly hands and feet, mental retardation, delayed milestones, mid-face hypoplasia, proptosis, hypertelorism, exorbitism, funnel chest, high arched palate, and crowded teeth, with occasional keratitis which resemble with the case of Cahyana [8]. Our patient was born of nonconsanguineous marriage which is dissimilar with case report of Ileri and Goyenc as they report patient born of consanguineous marriage [9].

Lastly, AS is a rare autosomal inheritance disorder that affects various body components. Ophthalmologists, neurosurgeons, plastic surgeons, dentists, and geneticists ought to adopt a multidisciplinary approach to the effective planning and treatment of such patients as an integral component of healthcare delivery.

## CONCLUSION

We reported a 9-year-old male patient with clinical manifestation of AS with craniosynostosis and acrocephalosyndactyly. This case underscores the importance of early recognition of AS based on clinical features and imaging, enabling timely multidisciplinary management. Prompt diagnosis can significantly improve functional outcomes and quality of life. The case also highlights the need for awareness among clinicians, especially in resource-limited settings, to facilitate appropriate referral and genetic counseling.

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### Author Contributions
Maliha Nawar – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

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Written informed consent was obtained from the patient for publication of this article.

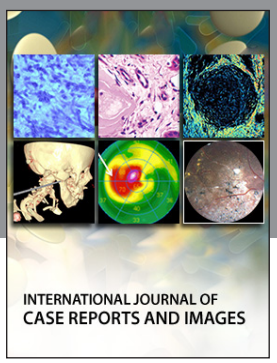
### Conflict of Interest
Author declares no conflict of interest.

### Data Availability
All relevant data are within the paper and its Supporting Information files.

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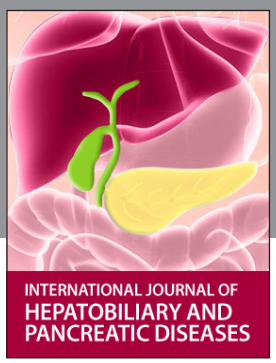
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