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Case Report

Atypical Apert syndrome with mitten-hand deformity and mild neurodevelopmental delay: a case report highlighting phenotypic variability in a resource-limited setting

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ABSTRACT

Apert syndrome (AS) is a congenital disorder that is inherited in an autosomal dominant manner. It accounts for approximately 4.5% of all cases of craniosynostosis. This report describes a rare case of a three-year-old male who presented with classical features of AS, including complex syndactyly of the hands and feet, midface hypoplasia, and congenital speech difficulties. The child had macrocephaly at birth that resolved without surgical intervention. Despite the craniofacial abnormalities, the child exhibited normal hearing and vision. This case report highlights an atypical presentation of AS with spontaneous correction of cranial morphology and mild neurodevelopmental involvement. This underscores the clinical heterogeneity of AS and emphasizes the importance of functional assessment and genetic counselling, especially in sporadic cases. The phenotypic overlap with other craniosynostosis syndromes, such as Carpenter, Crouzon, Pfeiffer, and Saethre-Chotzen, remains a diagnostic challenge.

Keywords: Acrocephalosyndactyly, FGFR2, Macrocephaly, Sporadic mutation, Syndactyly

INTRODUCTION

Apert syndrome (AS), or type I acrocephalosyndactyly, is the most common form of severe craniosynostosis. It is often accompanied by anomalies of other organs and appears in approximately 1 in 65,000-200,000 live births, irrespective of gender.[1,2] Baumgartner first reported AS in 1842, followed by Wheaton in 1894. The name originated from the French pediatrician Dr. Eugene Charles Apert, who documented nine cases in 1906.[3] The presence of pathogenic fibroblast growth factor receptor 2 (FGFR2) gene variants across multiple craniosynostosis syndromes may complicate diagnosis, especially for non-genetic specialists managing AS.[4]

Individuals with AS encounter a variety of medical, developmental, and psychosocial challenges.[5] The management of AS requires a team-based approach involving geneticists, neurologists, neurosurgeons, perinatologists, pediatricians, physiatrists, ophthalmologists, psychiatrists, orthopaedics and plastic surgeons.[4,6] Treatment includes established surgical interventions such as, Le Fort III osteotomy, for correcting midface hypoplasia. Timely multidisciplinary measures include hearing aids, airway stabilization, psychological support, speech therapy, and reconstructive procedures for limb abnormalities.[7] The need for this study is to report a case of AS and highlight its phenotypic features because AS is relatively rare in India, and such cases are infrequently documented. This case report intends to enhance clinical awareness and contribute to the existing literature by facilitating prompt diagnosis and collaborative care.

CASE REPORT

A three-year-old male, born to non-consanguineous parents as their second child, presented to the Burns and Plastic Surgery department with congenital bilateral complex syndactyly of the hands (mitten-hand configuration) and feet involving the central toes. The

child was diagnosed with AS at birth. The mother was 24, and the father was 27 when he was born. He was admitted for the release of syndactyly of the second and fourth web spaces in both upper limbs with full-thickness skin grafting from the bilateral groins. The child was delivered by cesarean section because of a history of nuchal cord. Birth weight was three kilograms. The parents noted proptosis, rhinorrhea, and an unusual increase in head circumference immediately after birth. Although the size of the head gradually reduced with growth, symmetric syndactyly of both limbs persisted.

The child had an older male sibling, aged five years, and was healthy, with no congenital anomalies. There was no history of visual or hearing impairment, cleft palate, nasal regurgitation of food, nasal obstruction, feeding difficulties, or neonatal intensive care unit (NICU) admission. He could sit without support at 2 years and began walking at 2.2 years of age, but still required assistance while getting dressed. At the time of assessment, according to Denver development screening test (DDST) 1967, child showed delay in developmental milestones, particularly motor and speech development, whereas social milestones were appropriate for age. He also had a history of recurrent upper respiratory tract infections, that necessitated medical treatment.

On examination, the child was active and playful. Extraoral examination revealed an abnormal contour of the head (brachycephalic skull), frontal bossing, wide palpebral fissures, hypertelorism, proptosis, flat nasal bridge, and sparsely distributed hair. Intraoral examination revealed an anterior open bite and a high-arched palate. On examination of the hands, his right hand showed a mitten-hand deformity. The thumb was partially separated, but short and broad with radial deviation (Type II Apert hand). The second, third, fourth, and fifth digits were completely fused, forming a single soft tissue mass. There was no digital web separation, and synonychia was observed in the third, fourth and fifth digits. The dorsum was smooth; the skin appeared stretched but not compromised, and the palmar crease was absent. The consistency was firm, with synostosis observed in the distal phalanges of the third, fourth, and fifth digits. Passive palpation revealed a bony mass on the distal phalanx. No motion was observed at the interphalangeal or metacarpophalangeal joints. Grasp function was present but markedly reduced.

His left hand also showed a classic mitten-hand deformity, consistent with complex complete syndactyly involving the second, third, fourth, and fifth digits, with synonychia of the third, fourth, and fifth digits. The left thumb was dystrophic, partially free, radially deviated, short and broad, with hypoplastic features. The palmar aspect appeared flat, with loss of the normal skin crease, suggestive of deep tissue fusion. The dorsal skin was intact with no evidence of erythema, ulceration, scarring, contractures, or skin redundancy, and the passive range of motion was limited to approximately 10-20º. The thumb exhibited restricted and palpable movement at the metacarpal joints across all planes. Gross grasp was limited, with only partial opposition of the thumb to the palm. Radial and ulnar pulses were palpable in both hands. Overall, the skin appeared healthy with normal turgor and no evidence of maceration, ischemia, infection, masses, vascular lesions, hypertrophic scarring, or callus formation.

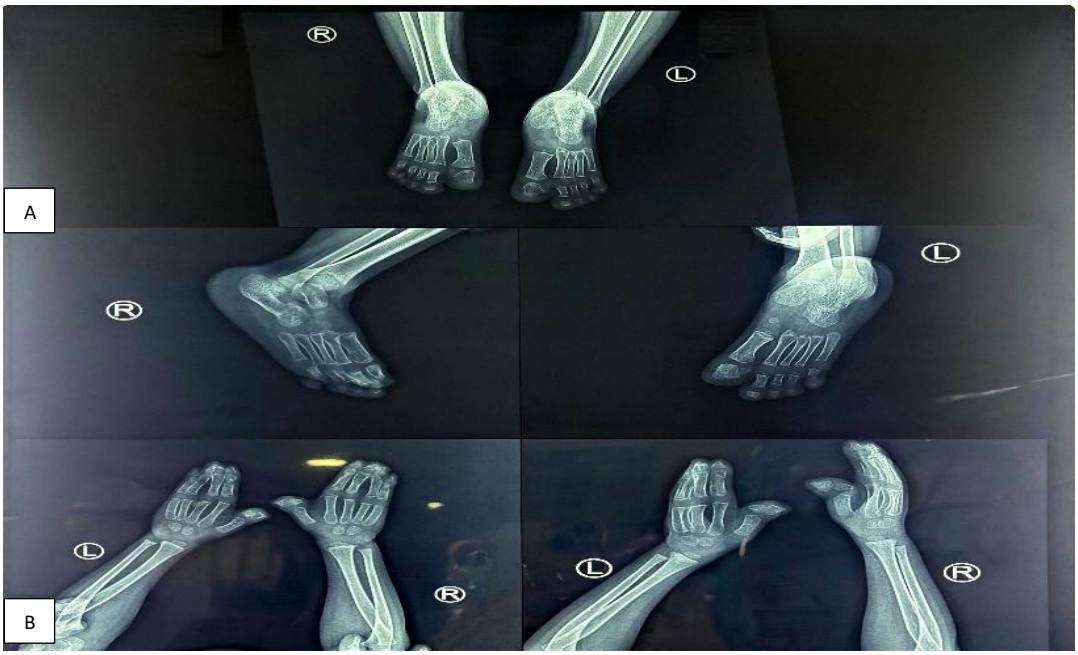

Figure 1 (A and B): Figure 1: X-ray of right (R) and left (L) feet showed soft tissue syndactyly and bony fusion of the toes and X-ray of right (R) and left (L) hand showed soft tissue syndactyly of second, third, fourth and fifth digits and synostosis of third, fourth and fifth digits with deformed phalanges of the first digit.

Upon examination of the feet, bilateral complete simple acrosyndactyly involving the second, third, and fourth toes was noted, along with incomplete simple syndactyly of the great and little toes. Passive movement of the great toe was possible but restricted, with a normal capillary refill. The child was ambulatory, with an altered weight distribution. A small healing scar was observed on the lateral aspect of the right ankle. Radiographic investigations of both feet showed soft-tissue syndactyly in the second, third, and fourth toes. Both hands showed soft-tissue syndactyly in the second, third, fourth and fifth digits, synostosis of the phalanges of the third, fourth and fifth digits, and deformed phalanges of the first digit (Figure 1). No other systemic abnormalities were detected on clinical examination. Echocardiography and abdominal ultrasonography revealed normal findings, and bilateral hearing sensitivity tests were within normal limits.

As per the maternal history, the mother received regular antenatal care, was appropriately immunized, and underwent routine ultrasonography, during which no anomalies were reported. There was no significant medical or surgical history, including gestational diabetes mellitus, pregnancy-induced hypertension, TORCH infection, trauma, or radiation exposure. The mother had not consumed any medications other than iron or folic acid supplements. There was no family history of major congenital malformation.

DISCUSSION

AS is associated with factors such as advanced age among fathers, maternal infections, medication exposure during pregnancy, and inflammatory changes in the cranium. Since the mother provided antenatal history without supporting medical documentation, the information may be subject to recall bias.[4] The father was 27 years old when the child was born, which contrasts with existing literature identifying advanced paternal age as a risk factor for AS.[4,6,8]

Phenotypic variability

This case report highlights a unique presentation of AS in a three-year-old male, which differs from previously documented cases owing to the presence of macrocephaly at birth. The child exhibited hallmark clinical signs of AS, such as midface hypoplasia associated with an anterior open bite and high-arched palate, and symmetric interdigital fusion of the hands and feet.[2,4] A case report by Baditela et al in a twenty one-month-old male demonstrate classic AS features with genetic and clinical similarities. Both cases emphasize the genetic complexity of the syndrome, suggesting the involvement of sporadic mutations despite the typical inheritance pattern.[7]

Abdatam et al reported a similar case in a twenty-month-old Tanzanian child with complete digital fusion and turribrachycephaly. The child had an isolated upper-limb fine-motor deficit, with preserved intellect and no systemic complications. This divergence suggests a distinct underlying genetic and molecular mechanism. In the present case, multisystem involvement including limb anomalies and midface hypoplasia affecting speech and respiration, created a cumulative developmental burden. This aligns with the documented correlations between adverse outcomes and systemic complications.[1]

However, in a case report by Abdatam et al cardiovascular or respiratory symptoms were absent. This demonstrates that osseous fusion alone does not necessitate global developmental delay. Both cases maintained age-appropriate cognitive potential, suggesting that the developmental impacts are secondary to mechanical and physiological effects, rather than intrinsic cognitive disabilities related to the syndrome. Critical diagnostic gaps include the absence of genetic testing and the failure of prenatal ultrasonography to detect obvious anomalies. Nonetheless, bilateral complex syndactyly provides strong phenotypic specificity for AS and other acrocephalosyndactyly syndromes.[1]

Another case report by Ourrai et al presented three infants diagnosed with AS based on clinical assessment and medical imaging findings. These cases depict the clinical features of AS, such as craniofacial dysmorphia, bilateral syndactyly of the hands and feet, and premature coronal suture fusion, as observed in the present case.[8] In their case-based analysis, Khatri et al reported a four-year-old female with AS who was born to phenotypically normal parents, suggesting a sporadic mutation. This case demonstrated variable phenotypic expression of AS despite identical genetic aetiologies, underscoring the clinical heterogeneity of the condition.[9]

Genetic implications

The clinical presentation of the child aligns with a diagnosis of AS. While AS typically follows an autosomal dominant inheritance pattern, the absence of characteristic features in both parents indicates the possibility of sporadic mutation or gonadal mosaicism. AS can result from a pathogenic FGFR2 variant located at 10q26. FGFR2 encodes a protein that is essential for angiogenesis, tissue repair, embryogenesis, and the regulation of cellular proliferation, differentiation, and maturation. FGFR2 plays a critical role in the signaling pathways necessary for cranial suture fusion.[6] Two specific mutations, Ser252Trp and Pro253Arg, have been identified in the receptor-binding region.[4]

The karyotype is normal in children with AS. Phenotypic changes may result in premature fusion of cranial sutures, leading to a reduced anteroposterior diameter and a prominent forehead, as observed in acrocephaly. AS rarely, affects the internal organs, elbows, shoulders, vertebral columns, and central nervous system (CNS). The prevalence of intellectual disability is low. Upper respiratory tract infections and obstructive sleep apnoea

are frequent complications of AS, that result in respiratory distress. In the present case, the child also had recurrent upper respiratory tract infections.[6]

Diagnostic challenges

AS can be diagnosed through a combination of clinical assessment, imaging modalities, and genetic analysis. In the present case, the diagnosis was made based on the findings of the clinical evaluation and imaging studies. Fetal morphological abnormalities can be prenatally identified using ultrasound in conjunction with fetal magnetic resonance imaging. Cranial developmental anomalies can be detected at around 19th week of pregnancy. Furthermore, advancements in 3D prenatal ultrasonography have demonstrated its efficacy in delineating characteristic features during the third trimester.[10]

Clinical management and multidisciplinary care

Owing to the multifaceted nature of AS, a multidisciplinary approach is essential for the diagnosis and management of affected children. Inter-professional collaboration has the potential to enhance a child's overall well-being and minimize the risk of long-term complications. Although a definitive cure remains elusive, corrective surgery offers a viable solution for anatomical deformities.[6] The child exhibited profound limitations in gross grasp and prehension of both hands, with absent thumb-index opposition and an inability to perform digital manipulation or fine motor tasks. Consequently, the decision to pursue surgical intervention was made by the parents. The child underwent successful release of syndactyly in the second and fourth web spaces of the hands bilaterally, within two weeks of admission after addressing upper respiratory tract infections. This procedure is expected to markedly improve the manual function.

Resource constraints in low- and middle-income countries

Molecular genetic testing is now available in developed countries to identify a heterozygous pathogenic variant of FGFR2, which can be used to differentiate AS from other acrocephalosyndactyly syndromes. However, developing countries continue to face significant challenges due to its high costs.[11] A comprehensive review by Li et al concluded that combining advanced prenatal imaging to identify the diagnostic triad of AS, that is, brachycephalic skull, midface hypoplasia, and symmetric syndactyly, with rapid FGFR2 molecular testing enables early, comprehensive care planning.[12]

Differential diagnosis

Craniosynostosis syndromes such as Crouzon, Pfeiffer, Carpenter, and Saethre-Chotzen bear a close resemblance to AS, yet, they remain clinically and genetically distinct entities. AS and Crouzon syndrome (CS) share a significant phenotypic overlap, particularly in craniosynostosis and facial dysmorphia. However, CS is distinguished by the absence of limb anomalies, which is a hallmark of AS. Given the prominent involvement of the extremities, CS was initially excluded from differential diagnosis. Pfeiffer syndrome (PS), also referred to as type V acrocephalosyndactyly, is characterized by craniofacial abnormalities. Unlike PS, AS lacks cloverleaf skull deformity, which is a typical feature of PS. Limb abnormalities, particularly broad, medially deviated thumbs and great toes, are most commonly observed in individuals with PS. Interestingly, the presence of bilateral thumb enlargement in the present case resembled the digital phenotype reported in the PS.[6,11,13]

Carpenter syndrome, an acrocephalopolysyndactyly type II disorder, follows an autosomal recessive inheritance pattern and results from biallelic mutations in the RAB23 (Ras-associated protein) gene. While both AS and Carpenter syndrome share features, such as craniosynostosis and limb malformations, they have distinct genetic origins. Carpenter syndrome is distinguished by features including preaxial polydactyly of the toes, congenital heart disease, obesity, and renal abnormalities-none of which were present in the child. In contrast, AS may exhibit preaxial polydactyly, but generally lacks the broader systemic involvement of Carpenter syndrome.[6,14] Saethre-Chotzen syndrome (SCS), another autosomal dominant disorder resulting from TWIST1 haploinsufficiency, manifests as variable craniosynostosis, milder limb abnormalities, drooping of eyelids, and ear anomalies. Distinguishing between these syndromes requires comprehensive clinical assessment and molecular genetic analysis.[15]

CONCLUSION

This case report highlights the substantial phenotypic variability of AS, particularly in resource-limited setting where access to advanced genetic diagnostic modalities remains restricted. The atypical presentation of the child emphasizes the need for comprehensive clinical evaluation beyond classical phenotypic expectations. In the Indian context, where many orphan diseases remain underdiagnosed and underreported, strengthening early diagnosis through prenatal imaging, improving access to specialized surgical and genetic services, and establishing structured referral networks are essential. Policies that promote rare disease registries, affordable genetic testing, and collaboration among medical specialists can greatly enhance the long-term health of children with rare conditions and reduce the challenges faced by their families.

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