CASE REPORT

APERT SYNDROME (ACROCEPHALO SYNDACTYLY): A RARE BUT RECOGNIZABLE HUMAN MALFORMATION SYNDROME

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ABSTRACT: BACKGROUND: Apert syndrome is a rare autosomal dominant Mendelian disorder characterized by a set of recognizable patterns of human malformations, having paediatric, plastic surgical, Orthopaedic and Dental implications requiring early recognition & effective management strategies for good Cosmetic and functional out-come.

KEYWORDS: Apert Syndrome, Acrocephalosyndactyly, Craniosynostosis.

Case Characteristics: A 2 Years and 4 months – female child admitted for simple febrile seizures and found to be dysmorphic.

OBSERVATIONS: The Child was first born to nonconsanguineous parents, (mother age 25 Years and father age 28 Years) brought for Fever, Cold, Cough of one day duration and 1 episode of seizure (simple febrile seizures) following upper respiratory tract infection and found to have mental retardation, Brachycephaly, Craniosynostosis, hypertelorism, midfacial hypoplasia, shallow orbits, mitten hands, syndactyly of both feet and palms (osseous and cutaneous)

DISCUSSION: Apert Syndrome (Acrocephalosyndactyly) was reported by Wheaton in 1894.¹ In 1906, Apert summarized nine cases.² By 1960, Blank noted the recording of 150 cases.³ The incidence was expected to vary greatly with estimates of 1:60,000 live births to 1: in 2,00,000 live births as suggested by different studies. Males and females are affected equally. The Syndrome is characterized by more frequent abnormalities includes mental deficiency may be present (but normal intelligence has been observed) Craniofacial abnormalities (like Craniosynostosis, Flat facies, shallow orbits, hypertelorism, strabismus, maxillary hypoplasia, Small nose, Narrow palate with or without cleft palate, Pseudomandibular prognathism and crowding of teeth). Limb defects (like Osseous and/ or cutaneous syndactyly, varying from total fusion to partial fusion most commonly with complete fusion of second, third, and fourth fingers. Distal phalanges of the thumbs are often broad and in valgus position. Fingers may be short. Cutaneous syndactyly of all toes with or without osseous syndactyly. Distal hallux may be broad and malformed) and Skin changes (like a Moderate to severe acne, including the forearms at adolescence).

Occasional Abnormalities (like short humerus, Pyloric stenosis, ventricular septal defect, polycystic kidney and bicornuate uterus).

DIFFERENTIAL DIAGNOSIS: Includes all other syndromes characterized by Acrocephalosyndactyly such as 1) crouzon syndrome 2) carpenter syndrome 3) Pfeiffer syndrome.

The hands in patients with Apert syndrome always show four common features⁴

1. A short thumb with radial deviation.

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2. Complex syndactyly of the index, long and ring finger.
3. Symbrachyphalangism.
4. Simple syndactyly of the fourth webspace.

Based on the first webspace, we can differentiate three different types of hand deformation. The typical hand deformities in patients with Apert syndrome distinguish it from the other syndromes.⁵

	Type I ("spade")	Type II ("mitten")	Type III ("rosebud")
First webspace	Simple Syndactyly	Simple Syndactyly	Complex Syndactyly
Middle three fingers	Side –to-side fusion with flat palm	Fusion of fingertops forming a concave palm	Tight fusion of all digits with one conjoined nail
Fourth webspace	Simple and incomplete syndactyly	Simple and complete syndactyly	Simple and complete syndactyly

Natural History Etiology: Autosomal dominant, with the vast majority of cases representing a fresh mutation.⁶ One factor in the sporadic cases has been older paternal age. The recurrence risk for the unaffected parents of a child with Apert syndrome is negligible, whereas the recurrence risk for the offspring of the affected individual is 50 per cent.

There are no adequate data on the long term follow up of patients. Early surgery for craniosynostosis, however, would seem indicated when the condition is of sufficient magnitude to give rise to increased intracranial pressure. Though there can be mental deficiency in patients who have no evidence of increased intracranial pressure, it is an irregular occurrence. There should be vigorous early management of the syndrome. When the thumb is immobilized, early surgery to allow for a pincer grasp is indicated, with later attempts at further improvement of hand function. Newer Plastic oral and maxillofacial (OMS) re- constructive surgical techniques may allow for improved facial cosmetic reconstruction as well as functional outcome of hand performance.

Therefore, every neonate suspected of having Apert syndrome deserves a complete evaluation for other malformations as well as referred to appropriate surgical faculties for improved cosmetic and functional outcome.

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