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Apert Syndrome : a Case Report in Sncu

KEYWORDS: Acrocephalosyndactyly, Craniosynostosis, Syndactyly, Hypertelorism

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ABSTRACT Apert Syndrome is a rare acrocephalosyndactyly characterised by dysmorphic facial features, craniosynostosis and severe symmetrical syndactyly of hands and feet (cutaneous and bony fusion). Inherited as a autosomal dominant trait, but most cases are sporadic. It includes prominent clinical features i.e. turricephaly,anterior open-bite maxilla, impacted and crowded teeth, cleft palate, bifid uvula,thick gingiva and mandible simulating pseudoprognathism. Clinical, genetic and biochemical approach have recently allowed tremendous scientific advances in understanding the molecular basis of Apert Syndrome. In the present case report we report a 3 day old male child with all the clinical features in agreement with Apert Syndrome like hypertelorism, exophthalmos, Depressed nasal bridge, cleft palate, mitten hand. The pt. was referred to specialized centre for clinical care with special needs.

INTRODUCTION :-

Craniosynostosis encompasses many congenital diseases like Apert Syndrome, Crouzon Syndrome, Carpenter Syndrome, Pfeiffer Syndrome. Out of all these , Apert Syndrome is the most common and most widely recognised craniosynostosis. Though this syndrome was mentioned as early as 1842 by Baumgartner, the eponymic credit was given to a French paediatrician Eugene Apert for his presentation in 1906. Apert summarised 9 cases and in 1920, Park and Powers published an exceptional essay on this entity. Apert Syndrome has typical clinical features but the relative rarity having prevalence of birth 1:65000 live births) still poses diagnostic dilemma[1,2].

CASE REPORT:-

A 3 day old ,term, 2nd order male infant born to nonconsanguinous married couple (Age of father-32, age of mother-21) was brought to SNCU of our Institute with chief complaint of poor feeding, abnormal facies and fused fingers and toes. He was born at 39 wks gestation,by LSCS following complain of PROM and leaking of amniotic fluid for 2 days measuring 3045 grams. During pregnancy, the mother was registered and immunised and undergone blood tests without adequate antenatal check up and antenatal USG. There was no significant past medical or surgical history (GDM, PIH, Viral exanthem, Trauma or Exposure to radiation. No family history of major congenital anomalies. Also no history of drug intake apart from Iron and folic acid. The first child, 2 ½ yrs.old,female was doing well with no congenital anomalies. On examination the baby was alert, with stable vitals. Anthropometry revealed head circumference to be 34.5cm, Length-50cm. Cry, reflex, activity and tone were good. Capillary refill time was normal. Moro and other reflexes present. He had a tower shaped calvarium (Turricephaly), flat occiput, prominent frontal region, prominent midface hypoplasia, Hypertelorism, prominent ocular proptosis, downslanting of lateral canthi, depressed nasal bridge,small nose, low set ears, flat face, trapezoid-like mouth, prognathism, Syndactyly of both fingers and toes (both simple and complex) with complete fusion of digits (spoon like deformity), concave palm (fig-1,2). Also on examination of oral cavity High arched palate and post.cleft palate was found. In the skull coronal sutures were fused, AP diameter was short, but anterior fontanelle and posterior fontanelle were large and wide open. All other systemic examinations were normal. Further investigations and imaging studies were performed. Radiographs of both hands and feet showed soft tissue syndactyly of 2nd,3rd,4th,5th digits. Phalanges of great toe were deformed in both feet (fig-3). TFUSG and NCCT Scan was normal. ECHO scan showed normal study.USG KUB and Abdomen was normal. Complete blood count including Hb%,total and differential count,platelet count and CRP were within normal limits. LFT and RFT were normal. Thyroid function tests were normal.

(fig-1: galaxy of facial deformities along with craniosyn-

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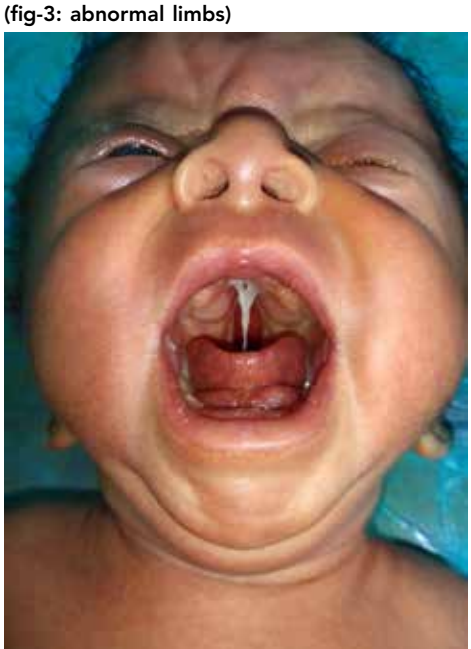
ostosis)

(fig-2: abnormal limbs- trident finger)

(fig-3: abnormal limbs)



(fig-4: cleft palate)

DISCUSSION:-

Apert Syndrome is a rare *Type Acrocephalosyndactyly* characterised by the triad of craniosynostosis, facial dysmorphism and severe syndactyly of hands and feet[1]. Craniosynostosis refers to premature fusion of one or more cranial sutures that normally separate the bony plates of the cranium. Reduced or asymmetrical skull growth ensures deformity of skull vault or base. Virchow (1851) noted that there is cessation of growth in a direction perpendicular to that of the affected suture while growth proceeds in a parallel direction. Syndromic Craniosynostosis involve single/multiple fused sutures and additional anomalies (limb,cardiac,CNS)[3]. Apert Syndrome is one such syndrome having typical clinical features. The incidence at birth is 1:65000 -1:80000 live births[1]. It has been rarely reported in India. Although inherited as autosomal dominant but most cases are sporadic in nature. It appears to originate exclusively in paternal germline. Most of(98%) the molecularly characterised cases of Apert Syndrome result from missense substitution mutation in FGFR2 gene(Fibroblast Growth Factor Receptor2),located on Chr.10q26[4]. Apert Syndrome is characterised by a galaxy of clinical features.

Among them 1) Craniofacial features i.e. short AP diameter, high prominent forehead, flat occiput,irregular cranisynostosis esp.of coronal suture,fontanelles large and close late,flat or hypoplastic facies,supraorbital horizontalgroove, shalloworbits, proptosis,hypertelorism, strabismus, downslanted palpebral fissures, small wide nose with bulbous tip, depressed nasal nasal bridge, low set ears, maxillary hypoplasia, cleft palate or narrow palate, thick gingiva, trapezoid mouth, bifid uvula, delayed or ectopic or malocclusion of teeth, shovel shaped incisors. 2) Limb features i.e. osseus or cutaneous syndactyly (Mitten hand, Sock foot) ranging from complete fusion to partial fusion. M.C.with complete fusion of 2nd ,3rd and 4th fingers,concave palm,contiguous nail bed (Synonychia), radial deviation of short and broad thumb (Hitchhiker posture) . 3) CNS features i.e. Agenesis of corpus callosum (ACC),Nonprogressive ventriculomegaly, progressive hydrocephalus, absent or defective septum pellucidum, gyral and hippocampal abnormalities, megalencephaly, mental deficiency (57% have IQ <70). Growth deceleration becomes more prominent after adolescence. 4) Orthopedic abnormalities include – short humerus,synostosis of radius and humerus, joint mobility limitation, multiple epiphyseal dysplasia, short or absent neck of scapula, small capitulum,flat radial head,genu valgum,fusion of C5→C6[5,67]. The recurrence risk for unaffected parents of a child with Apert's is negligible. But for affected parents recurrence risk is 50%. Various etiological hypothesis have been proposed for Apert Syndrome – a) High paternal age b) Antenatal drug consumption by mother c) Maternal infection. High paternal age is thought to cause Apert syndrome due to androgen end organ hyper response affecting the epiphyses and sebaceous gland causing early suture closure, selection and high no. of mutant sperms. Vigorous early surgical intervention is vital for survival and normal development[8].

CONCLUSION:-

The following case report demonstrates the typical c/f of Apert's after ruling out the other similar syndromes like Crouzon's, Carpenter, Pfeiffer, Beare-Stevenson Syndromes.

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REFERENCE

1. Verma, Shyam, and Michelle Draznin. "Apert syndrome." Dermatology online journal 11.1 (2005). | 2. PARK, EDWARDS A., and GROVER F. POWERS. "Acrocephaly and scaphocephaly with symmetrically distributed malformations of the extremities: A study of the so-called acrocephalosyndactylism." American Journal of Diseases of Children 20.4 (1920): 235-315. | 3. Buchanan, R. C. "Acrocephalosyndactyly, or Apert's syndrome." British journal of plastic surgery 21.4 (1968): 406-418. | 4. Wilkie, Andrew OM, et al. "Apert syndrome results from localized mutations of FGFR2 and is allelic with Crouzon syndrome." Nature genetics 9.2 (1995): 165-172. | 5. Batra P, Duggal R, Hari P. Dentofacial characteristic in Apert syndrome: A case report. J Indian Soc Pedo PrevDent. 2002;20:118–123. [PubMed] | 6. Ferraro NF. Dental, orthodontic, and oral/maxillofacial evalution and treatment in Apert syndrome. Clin PlastSurg. 1991;18:291–307. [PubMed] | 7. Upton J. Apert Syndrome. Classification and pathologic anatomy of limb anomalies. Clin Plast Surg 1991;18:32155. | 8. Glaser RL, Broman KW, Schulman RL, et al. The paternalageeffect in Apert syndrome is due, in part, to the increased frequency of mutations in sperm. Am J Hum Genet 2003;73:93947