International Journal of Medical and Health Sciences

Journal Home Page: http://www.ijmhs.net

Case Report

Apert syndrome- a case report

Muddu Gopi Krishna*[1], Saritha Purushothaman[2]

[1]Senior Resident, [2]Senior Consultant Department of Pediatrics, Sri Sathya Sai General Hospital, Prashanthi Nilayam, Andhra Pradesh, India.

ABSTRACT:

Apert syndrome is a rare congenital disorder which is a part of Acrocephalosyndactyly syndromes. It is characterized by craniosynostosis, symmetrical syndactyly of hands and feet. We present a case of Apert syndrome with clinical features of brachycephaly, proptosis, flat forehead, low set ears with syndactyly of hands and feet.

KEY WORDS: Apert syndrome, Brachycephaly, Syndactyly.

INTRODUCTION:

Apert syndrome is a rare Autosomal Dominant disorder, but in the majority of the cases it is sporadic in inheritance [1]. It was named after a French Physician "Eugene Apert", who first described it in 1906 in a 9 case series [2]. Its incidence is 1 in 160,000 live births and it comprises approximately 4.5% of total craniosynostosis. There is no sex predilection [3]. Apert syndrome is rarely reported from India [4].

CASE REPORT:

At 10 months old male child, product of non-consanguineous marriage brought to the outpatient departmentt with the complaints of hand and foot deformities since birth.

The antenatal period was uneventful, with no history of unknown drugs or irradiation exposure. Born full term, normal vaginal delivery. Child was able to sit with support, making babbling sounds and identify the mother. The weight of the child was 6 kgs (<3rd percentile), length was 68 cm (<3rd percentile), head circumference was 44 cms (within 2 SD). Anterio posterior diameter of head was 12 cms and transverse diameter was 14 cms.

On physical examination there was Turri brachycephaly. The transverse diameter was more than the anterio posterior diameter. Anterior fontanella was wide open. Child had a flat fore head with horizontal groove above the supra orbital ridge. There were sparse eyebrows bilaterally with hypertelorism. The palpebral

Corresponding author: Dr. Muddu Gopi Krishna
Email: gopikrishna.dr@gmail.com

fissure was down slanting with ocular proptosis. The nasal bridge was depressed and "parrot beak nose" was noted (Figure 1). Eruption of primary teeth was delayed.

There was high arched palate with low set ears.

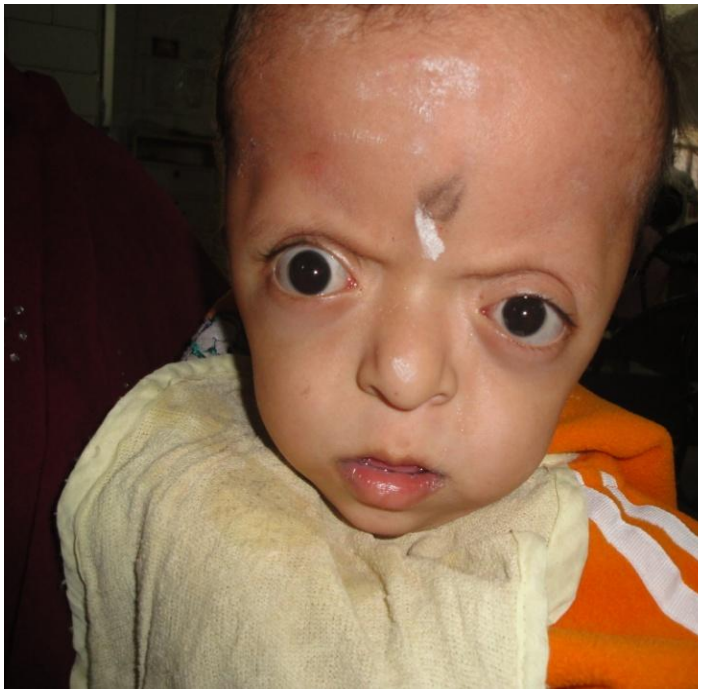

Figure 1: Depressed nasal bridge, hypertelorism, proptosis, horizontal supra orbital groove, flat forehead and turribrachycephaly.

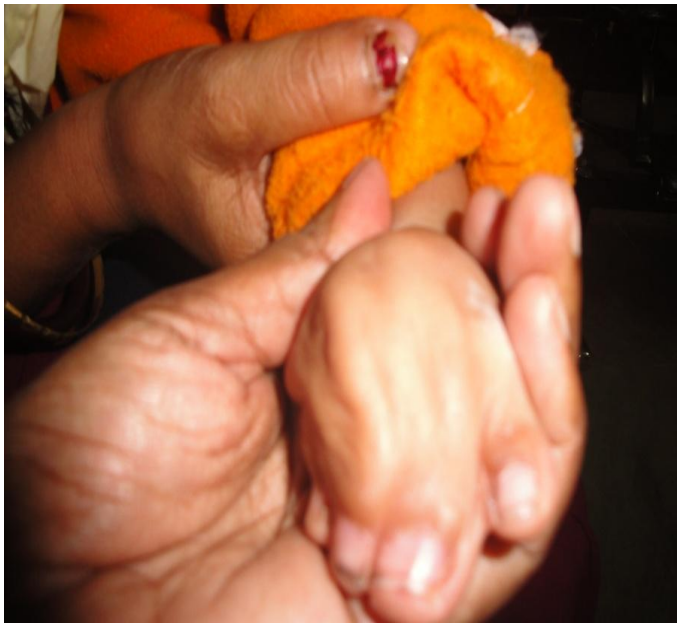

Figure 2: Syndactyly of hand "mitten hand appearance".

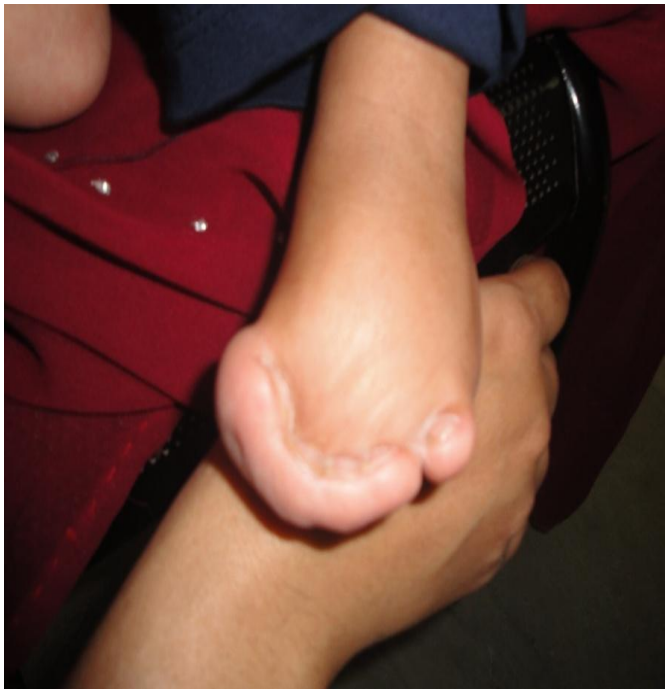

Figure 3: Syndactyly of foot "sock foot deformity".

There was bilateral syndactyly in the hands. The 2^{nd}, 3^{rd}, 4th & 5^{th} digits were fused. Thumb was fused to the index finger by incomplete syndactyly, giving "mitten hand" appearance to the hand (Figure 2). The nails of 2^{nd} and 3^{rd} digits were fused (synocychia). Thumb had separate nail. Palm was concave. At the foot, there was a complete fusion of toes symmetrically. 1^{st} to 4^{th} toes were fused, with incomplete syndactyly of the 5^{th} toe, giving a "sock foot deformity" appearance (Figure 3).

DISCUSSION:

Apert syndrome is an Autosomal Dominant disorder. It can occur in families where there is no previous similar condition, due to sporadic incidences. It occurs as a result of mutation in Fibroblast growth factor receptor 2 (FGFR2) on chromosome 10q25.3-26 [5-6]. Advanced paternal age appears to be a risk factor [7]. Apert syndrome occurs as a result of androgen end organ hyper response affecting the epiphysis and sebaceous glands, which results in early

epiphysial fusion leading to short stature, craniosynostosis and fused digits. Our patient had the triad of brachycephaly, mid face hypoplasia and syndactyly of hands and feet.

There are other acrocephalopolysyndactyly syndromes like Carpenter's syndrome, Crouzen's syndrome, Pfeiffer syndrome which shows some similarities in facial features but Apert is clinically distinguished by a complete distal fusion of the soft tissues of 2^{nd} to 4^{th} digits with variable degree of bony fusions in both hands and foot [8].

This gives characteristic "mitten hand" and "sock foot" deformities in the hands and foot respectively which is seen in our case. The severity of syndactyly of hand in Apert was divided into 3 types by Wilkie [9]. Type I is less severe where thumb and part of fifth finger are separated called "spoon hand".

Type II where the little finger is not separated and variable simple complete or incomplete syndactyly of thumb with radial deviation is seen called "mitten hand". Type III is severe form where all 5 digits are fused called "rose bud". There can be cervical vertebral fusion at C5-C6 level [10].

Orbital proptosis may lead to exposure keratopathy. Keratoconus, congenital glaucoma and optic atrophy, can also occur [11]. Intelligence can be subnormal with an average I.Q. of 70 [12]. There can be progressively increased intra cranial pressure with other anomalies, as a defect in corpus callosum [13]. Dental problems, cardiac defects, conductive hearing loss, chronic otitis media, trachea-esophageal anomalies were also reported with Apert syndrome [14, 15].

Treatment is usually multidisciplinary, involving plastic and reconstructive surgery. Craniotomy is done to reduce the increased intra cranial pressure. It is usually done during infancy. Correction of dental malocclusion is done by orthognathic surgery. Mid face hypoplasia is corrected by Ilizarov procedure, which opens up the facial bones and helps them to grow. Surgical separation of the fingers and toes may help in better cosmetic appearance but not in better functioning [16].

CONTRIBUTORS:

MGK and SP were involved in clinical evaluation, research, and conceptualization of work and drafting of the manuscript. The final work was approved by both the authors. MGK will act as guarantor.

COMPETING INTEREST: Nil.

REFERENCES:

1) Eastman JR, Escobar V, Bixler D: Linkage analysis in dominant acrocephalosyndactyly. J Med Genet 1978; 15:292.

2) Apert E: Del'acrocephalosyndactylie. Bull Mem Soc Med Hop Paris 1906; 243:1310.

3) Blank CE: Apert's syndrome (a type ofaerocephalosyndactyly): Observations on a British series of thirty-nine cases. Ann Hum Genet 1960; 24:151.

4) Sohi BK, Sohi AS. Apert's syndrome. Indian J Dermatol Venereol Leprol 1980; 46:169 -72.

5) Park WJ, Theda C, Maestri Ne et al: Analysis of phenotypic features and FGFR2 mutations in Apert syndrome. Am J Hum Genet 1995; 57:321.

6) Wilkie AO, Starey SF, Oldridge M et al: Apert syndrome results from localized mutations of FGFR2 and is allelic with Crouzon syndrome. Nat Genet1995; 9:165.

7) Erickson JD, Cohen MM Jr: A study of parental age effects on the occurrence of fresh mutations for the Apert syndrome. Ann Hum Genet 1974; 38:89.

8) Kaplan, L C). "Clinical assessment and multispecialty management of Apert syndrome". *Clinics in plastic surgery* 1991-04; 18 (2): 217–25.

9) Wilkie AO, Slaney SF, Oldridge M, Poole MO. Apert syndrome results from localized mutation of FGFR2 and is allelic with Crouzon syndrome. Nature Genet 1995;9:165-72

10) Cohen MM Jr, Kreiborg S. Visceral anomalies in the apert syndrome. Am J Med Genet1993; 45:758-60.

11) Mathur JS, Nema HV, Metra KS et al: Apert's anomaly a transition. Acta Ophthalmol1968; 46:1041.

12) Murcovic JA, Posnick JC, Drake JM et al: Hydrocephalus in the Apert syndrome: A retrospective review. Pediatr Neurosurg1993; 1963:151.

13) Cohen MM, Kreigorg S: Unusual cranial aspects of Apert syndrome. J Craniofac Genet1994; 14:48.

14) Cohen MM Jr: Cardiovascular anomalies in Apert type acrocephalosyndactyly. Birth Defects1982; 8:239.

15) McGill T: Otolaryngologic aspects of Apert syndrome. Clin Plast Surg 1991; 18:309.

16) Gleicy VS, Carnelro Farias JG, Fred AP, Patrica L. Apert syndrome: Review and case report. Indian J Dermatol Venereol Leprol 2008; 74:640.