APERT SYNDROME (ACROCEPHALO SYNDACTYLY)

(A Case Report)

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Introduction

Apert, E. (1906) described a rare developmental deformity syndrome characterised by (*a*) Cranio-synostosis leading to turribrachycephaly and (*b*) Syndactyly of hands and feet, often alongwith (*c*) various ankyloses and (*d*) progressive synostoses of the hands, feet and cervical spine. Blank (1960) divided the cases of acrocephalo-syndactyly into typical (Apert type) and atypical forms. Grabb, W.C. (1973) has enumerated the following characteristics of a typical Apert's syndrome (*a*) Deformity of skull which is typically towershaped with a broad forehead, (*b*) Exophthalmos and hypertelorism, (*c*) Maxillary hypoplasia and (*d*) Hand anomalies. Gorlin, R. J. et al (1976) could find the reports of only 200 case s of this syndrome upto date.

Case Report

Dha·mendr Singh, 1½ years old, Hindu, male, chi.' reported for treatment of multiple congenital deformities involving craniofacial region and limbs. The baby was the first child born of a father aged 26 years and mother aged 16½ years. Father was reported to be mentally deranged since last 7 years, though he was a good student initially. No family history of occurrence of any congenital deformity could be had either on maternal or paternal side. Also, mother had neither suffered from any illness worth the name or was on any drugs during the period of gestation.

On examination the general health of the child was found to be satisfactory and his intelligence appeared to be normal. Local examination of the skull revealed that it was a tower shaped skull (oxycephaly) with a broad forehead. Its maximum anteroposterior diameter was 27 cm. and lateral diameter 17 cm. with cephalic index of 63% (Dolictocephaly). X-ray of the skull showed that sutures had fused. The eyes were having increased distance between the pupils as compared to normal (hypertelorism) and the distance between the two medial canthi was 3.5 cm., whereas the distance of lateral to medial canthus of one eye was 2 cm. The maxillary bones appeared to be slightly less developed as compared to mandible. The tip of the nose was traversed by a 1″ long longitudinal skinlined groove separating the two normally formed nostrils to either sides. The groove in its turn was transversely traversed by a semilunar septum of cartilage covered by skin.

The right hand had normal thumb and index with small webbed 3rd, 4th and 5th

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digits. On the other hand, left hand had only 1st digit (thumb) normal and the rest of the digits were small and webbed.

So far the inferior extremities were concerned both the right and left legs were having congenital constriction rings. The right foot, though developed, was having equinovarus deformity whereas the left foot was not developed at all.

Discussion

To be born normal, the new born must successfully overcome the possible obstacles associated with unfavourable heredity and hostile environmental factors.

Park and Power (1920) suggested that acrocephalo-syndactyly was caused by a hereditary defect in the tissues which separated the various bone analgen from one another before the fifth and sixth weeks of intrauterine life.

Blank (1960) and Cohen (1971) are of the opinion that most cases are sporadic. Only on a few occasions, females with the Apert Syndrome have given birth to an affected child, or an increased paternal age has been detected.

Dodson (1970) could find chromosomal translocation in a few cases of acrocephalo syndactyly.

The Apert syndrome occurs once in 160,000 births and because of high mortality rate in neonatal period only one case of this syndrome is seen per 2,00,000 population.

It is no wonder that upto 1976 only 200 cases were reported in the Anglo-American literature and this happens to be the first case report from this part of the country.

The embryo-pathogenesis of these anamolies and their interrelationship is not fully understood. The embryological development of the face and limbs takes place between fourth and eighth weeks of intrauterine life. Hence, it can be conceived that during a short period of four weeks an extreme demand is placed upon the coordination of cell separation, migration and interaction. The proper amount of tissue must be present at an exact time in the correct three dimensional relationship and their precise movements and timings are critical. It is no wonder, any mishap in this intricate programme can lead to a bizarre group of anomalies. The mid part of the face develops immediately anterior to the forebrain by the differentiation of the broad midline fronto nasal process which becomes divided into medial and lateral nasal process by the nasal placodes. The failure of fusion of globular process of the medial nasal process could lead to the formation of a bifid nose, lip, frenulum, alveolus or even anterior palate. Frontonasal dysplasia of varying degree may be responsible for the more lateral placing of the eyes. Simultaneously the limbs also suffer from various degrees of arrested and abberrent development resulting into the group of deformities of limbs seen in this syndrome.

A plastic surgeon has to tax his ingenuity to deal with these deformities and has often to take assistance of neurosurgeons and orthopaecic surgeonsin dealing with craniotosis and osteoskeletal problems. The mid-line dehiscence of the face has to be suitably closed. Correction of syndactyly, deepening of webs, stabilisation of loose joints etc. may greatly improve the function of the maimed hands. Provision of prosthetic hand or fingers may take care of the appearance. The construction rings require excision alongwith "Z" plasty.

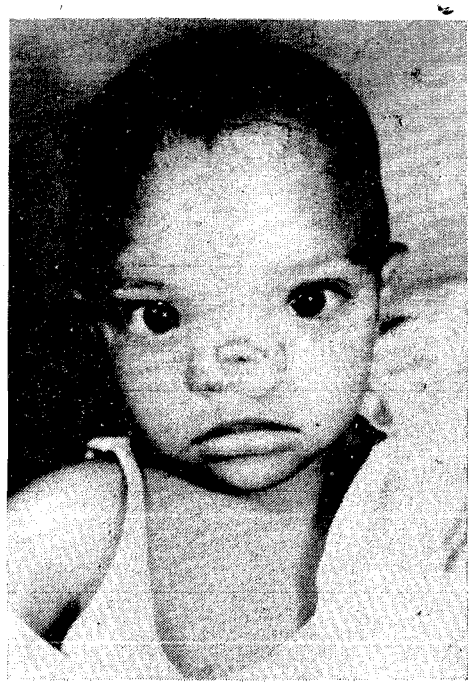

Fig. 1

Fig. 2

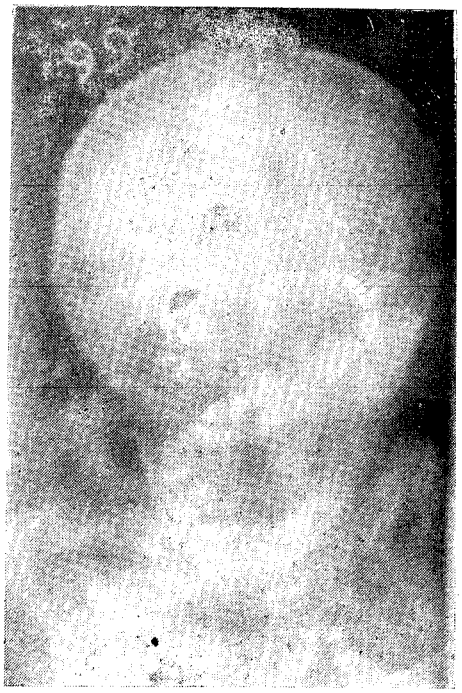

Fig. 3

The talepes deformities of the foot have to be corrected and provision of suitable prosthetic lower limbs may enable the child to walk, Apart from the above, the hypertelorism may require before III type of advancement osteotomy and only bone grafting may have to be done for hypoplastic jaws. Thus, the life of these unfortunate patients can be made to a great extent, comfortable, bearable and livable.

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