APERT'S SYNDROME
(Case report and review of literature)

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Summary

A case of Apert's syndrome in a one year old female child is described and literature reviewed. She was the first born of a young couple. She had congenital syndactyly of toes and fingers, acrocephalic skull, flat facies, exophthalmos, hypertelorism and greasy skin. In addition to the typical radiological features of this syndrome which the patient showed, thickened first metacarpals forked at the base were also seen. There were two phalanges for each toe. Calcification was seen intracranially. These radiological features have not been mentioned so far in the literature reviewed.

Apert's syndrome is an uncommon disorder producing craniofacial malformations along with syndactyly of hands and feet. The first person to describe this entity was Troquart in 1886[1]. By 1960 Blank[2] noted recording of 150 cases. Very few cases have been reported from India. We are presenting a one year old child having features of this syndrome.

Case Report

One year old female child was brought for treatment of congenital syndactyly of toes and fingers. She was the first born; her mother being twenty and father twenty four years old at the time of her birth. Mother remained healthy throughout the pregnancy. The child's milestones were slightly delayed. She started holding up her head at the age of five months, sitting up at seven months and standing with support at ten months. She started speaking monosyllables at the age of eleven months. The left lower incisor tooth erupted at the same age.

On examination, child was found to have acrocephalic skull with flattened occiput. Facies was flat with supraorbital horizontal groove, exophthaloms and hypertelorism. There was no squint. Nose was wide and beaked. Palate was high arched and narrow. Mandible was normal (Fig 1). The skin was rather 'greasy' and fontanelle were closed. There was complete syndactylism in both hands and feet; but all nails were present. X-Ray skull (Fig 2), showed acrocephaly and fusion of sutures with hypoplastic maxillae and intracranial calcification. X-Ray of hands (Fig. 3 A) showed synphalangism, (osseous syndactyly) in the first, second, third and fourth fingers of one hand and between third and fourth fingers of the other hand. X-Ray of feet (Fig. 3 B) revealed thickened first metatarsals which were forked at the base, two phalanges for each big toe and crowding of phalanges of other toes, (perhaps due to cutaneous synostosis). There were no other congenital anomalies or defects in any other system. Fundi were normal.

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Received for publication on 22—6—1979

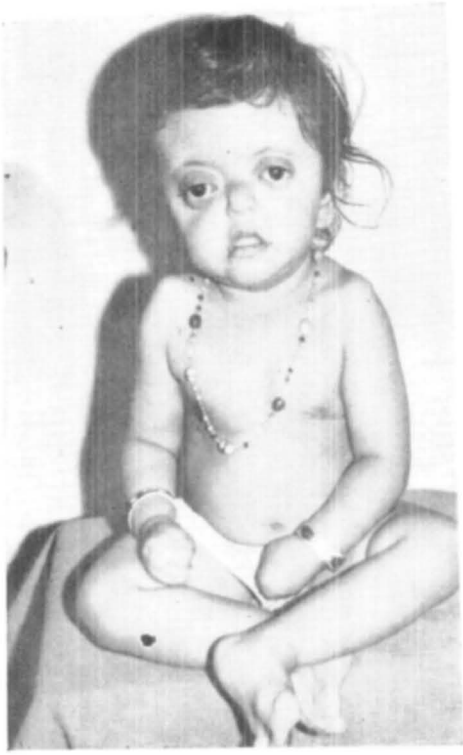

Fig 1 Photograph of the child showing flat facies, hypertelorism, wide beaked nose and synphalangism in both hands and feet.

Discussion

Though the original description of this condition was given by Troquart in 1866[1], Apert in 1906[3] reviewed nine cases, including the one reported by Troquart and the syndrome was named after him. In this syndrome children are acrocephalic with flat occiput. There is irregular craniostenosis especially of four coronal sutures and the closure of fontanelle may be delayed[4]. Facies is flat with supraorbital horizontal groove. The maxilla is hypoplastic and recessed with small maxillary sinuses. The nose is wide and sometimes beaked. There is hypertelorism and divergent squint[5]. Other ocular abnormalities include optic atrophy, keratoconus and hyperopic eyes[6]. The palate is high arched, narrow and occasionally cleft. Usually the mandible is normal but prognathia may be present and because of size disparity and displacement of alveolar ridges, malocclusion is frequent. All these features except ocular changes and hypomandibulosis were seen in our patient.

Deformities of limbs are symmetrical and more pronounced in distal portions. Osseous synphalangism or cutaneous syndactyly or both may be present in varying severity, from total fusion to partial fusion. Apert's syndrome is the only entity with complete syndactyly and bone fusion. Commonest form of syndactyly is seen as fusion of second, third and fourth fingers. Distal phalanges of thumbs are often malformed[4]. There may be cutaneous syndactyly of all toes as in our case. Fusion of metacarpals gives rise to so called "lobster claw" deformity, with varying degrees of

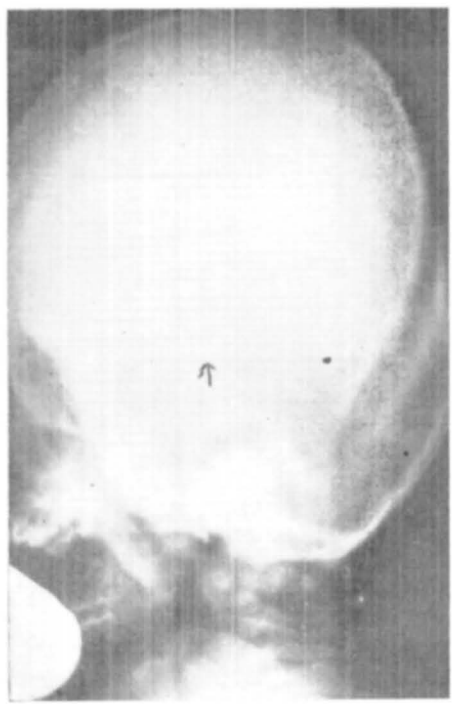

Fig. 2 X-Ray skull lateral view Showing acrocephaly, fusion of all sutures, hypoplastic maxillae and calcification at the level of corpus callosum.

syndactylism[8]. Fusion of vertebrae, deformities of hip joints, diastasis of sympysis pubis, short humerus, synostosis of radius and humerus and limitation of joint mobility may be present[4].

Skin may appear 'greasy' and thickened[2], as it was seen in the case under discussion. Other occasional abnormalities besides those of the skeletal system are oesophageal atresia, pyloric stenosis, ectopic anus, pulmonary aplasia, atrophy of pulmonary arteries, anomalies of tracheal cartilage, pulmonary stenosis, overriding aorta, ventricular septal defect, endocardial fibroelastosis, polycystic kidney, hypernephrosis and bicornuate uterus[4]. None of these were present in our case.

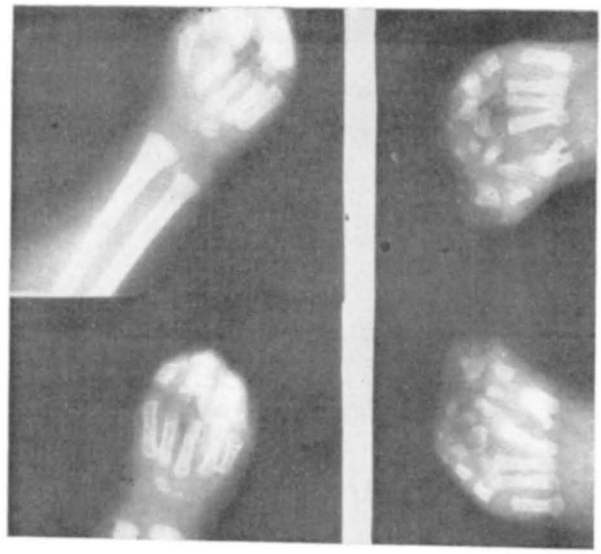

Fig. 3 A. X-Ray hands. Shows osseous syndactyly between first second and between third and fourth finger phalnages in one hand; between thumb and second finger and between third, fourth and fifth finger phalanges in the second hand.

B. X-Ray feet. Thickened first metatarsals, forked at the base. Two phalanges for each big toe.

Some patients have normal intelligence[9] but subnormal faculty is commonly noted[2]. Antisocial behaviour has been observed in some patients[10]. Musallam et al[6] have reported one case with agenesis of corpus callosum. Most of the patients have cyanotic spells after birth which he postulated could contribute to psychomotor retardation. Our patient being just one year old, it is too early to comment on mental development and social adjustment. According to Blank[2], many affected children die in early infancy and survivors into adulthood are not reported to have married due to their grotesque appearance, even though social adjustment apparently has been accomplished by some.

As regards etiology, autosomal dominant multigenic inheritance is indicated in majority of the cases. The risk of occurence in offspring of the affected individual is 50% Some have represented fresh mutation. No chromosomal abnormalities have been described[15]. Critical teratogenic effects are produced sometime about the end of first month of embryonic life[16]. In cases occuring sporadically, one of the deciding factors may be disparity on parental age, as fathers of these children are usually much older than their mothers[9]. In our case both the parents were of young age group and there was not much disparity in their age. Incidence of Apert's syndrome is similar in males and females and there is no definite racial predilection[5].

There may be difficulty in differentiating Apert's syndrome from Carpenter's syndrome, which is characterized by acrocephaly, peculiar facies, brachydactyly of fingers, polydactyly and preaxial syndactyly of toes, obesity and mental retardation[13]. It is inherited as

an autosomal trait. Moreover Apert's syndrome is the only entity with complete syndactyly and bone fusion and there is no hypogenitalism or obesity in this Chotzen's syndrome[14], another acrocephalic syndactyly syndrome can be differentiated from Apert's syndrome in which several generations with affected members have been reported.

As for management, corrective surgery on skull and extremities can be carried out. Craniectomy should be done early in life, partly to improve the appearance but mainly to lower the intracranial pressure[11]. This will help in the growth of cerebral cortex and decrease incidence of damage to the brain and optic nerves. Follow up during period of active brain growth is necessary as fusion can occur at previous craniectomy sites. Plastic surgery on the hands and feet should be delayed till kindergarten age, if fingers are growing straight and there is no synphalangism[12]. Synphalangism will necessitate early surgery. Contractures are apt to develop during periods of rapid growth particularly on volar surfaces. Attainment of function should take precedence over appearance in determining type and time of surgery. When function is adequate, treatment should be delayed until ossification centres have developed[12]. Coexisting defects of muscles and tendons should be ascertained. Remarkable functional and perceptual adaptation of deformed hands can occur usually with impairment of stereognosis[8].

Keratoconus, proptosis, errors of refraction, exposure keratitis and strabismus should be treated by orbital decompression or corneal transplants[9]. Speech and articulation defects resulting from abnormal facial configuration, malocclusion and cleft palate require reconstructive surgery and speech therapy.

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