Apert Syndrome: A Rare Presentation

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Abstract

Apert syndrome (acrocephalosyndactyly) is a rare congenital disorder characterised by craniosynostosis, mid-facial malformation and symmetrical syndactyly. We present a 7-year-old female having all the features of classical Apert syndrome.

Keywords: Apert, Syndactyly, Craniosynostosis.

Introduction

Apert syndrome (acrocephalosyndactyly) is a congenital disorder characterised primarily by craniosynostosis, mid-face hypoplasia, and syndactyly of the hands and feet with a tendency for fusion of bony structures. Craniosynostosis (or craniostenosis) is defined as premature closure of the cranial sutures producing deformity of the skull. It may be primary, originating from a sutural pathology, or secondary resulting from dysgenesis of the underlying brain. It has been described in various syndromes like Apert, Crouzon, Pfeiffer and Jackson-Weiss, and is associated with specific systemic anomalies. We present a 7-year-old girl with all the features of Apert syndrome.

Case report

A seven-year-old girl presented with the complaints of abnormal shape of the head, webbed fingers, and developmental delay. On examination, the baby was found to have flattened occiput with frontal prominence, abnormal contour of the head (brachycephaly), shallow orbits with bilateral proptosis, hypertelorism, depressed nasal bridge, hypoplastic maxillae, low set ears, and dental anomalies (Fig. 1). She had symmetrical syndactyly with complete fusion of all the five digits of both hands and both feet (Fig. 2). The fused fingers and toes had separate nails. There was no other apparent congenital malformation, and systemic examination revealed no other abnormality. On investigation, X-ray of the spine, abdominal ultrasonography, and echocardiography were normal. Radiographs of both hands and feet showed soft tissue syndactyly of all the digits and toes. Skull radiographs revealed fused coronal sutures, brachycephalic skull contour, elongated flat forehead with bitemporal widening, increased convolutional markings suggestive of increased intracranial pressure and hypertelorism. All findings were diagnostic of Acrocephalosyndactyly or Apert syndrome.

Fig. 1: Characteristic features of Apert syndrome showing ocular hypertelorism, proptotic eyes, depressed nasal bridge, and short wide nose with bulbous tip are seen in this seven-year-old girl.

Fig. 2: Same child with mitten appearance of the hands with syndactyly and sock-like appearance of the feet with syndactyly.
**Discussion**

Apert syndrome was first described by Eugene Apert in the year 1906. He described a triad of craniosynostosis, syndactyly, and maxillary hypoplasia. The incidence of Apert syndrome is approximately one in 50,000 births. More than 98% of cases with Apert syndrome are caused by specific missense substitution mutations, involving adjacent amino acids (i.e., Ser252Trp, Ser252Phe, Pro253Arg) in the linker between the second and third extracellular immunoglobulin domains of FGFR2, which maps to chromosome bands 10q25 - q26. The remaining cases are due to Alu-element insertion mutations in or near exon 9 of FGFR2. The majority of cases are sporadic, resulting from new mutations with a paternal age effect.

Apert syndrome is thought to occur as a result of androgen end-organ hyper-response affecting the epiphyses and sebaceous glands. This results in early epiphyseal fusion resulting in short stature, short and fused digits, and acrocephaly.

In Apert syndrome, the cranial vault deformity is variable, but most often presents as a short anteroposterior dimension with craniosynostosis involving the coronal sutures resulting in a turribrachycephalic skull. The typical craniofacial appearance includes a flat, elongated forehead with bitemporal widening and occipital flattening. There is also mid-face hypoplasia accompanied by orbital proptosis, downslanting palpebral fissures and hypertelorism. High arched palate, clefts of the secondary palate, and crowding of the dental arch can also be seen. The nose is downturned at the tip, the bridge is depressed, and the septum deviated. Other central nervous system abnormalities include malformations of the corpus callosum, the limbic structures, or both, megalencephaly, gyral abnormalities, encephalocoele, pyramidal tract abnormalities, hypoplasia of cerebral white matter and heterotopic gray matter. There is also an increased incidence of delayed mental development in these children, but many of them develop normal intelligence. The usual hand abnormality in Apert syndrome consists of a bony fusion of the second, third, and fourth fingers, with a single common nail. Involvement of the first or fifth digits in this bony mass is variable. There can be a similar deformity involving the foot (mitten hand and sock foot). Other skeletal abnormalities have been described in Apert syndrome. These include limited mobility at glenohumeral joint and elbow joint, multiple epiphyseal dysplasia, very short or absent neck of scapula, small capitulum, and flat radial head. Commonly associated systemic features include cardiac anomalies, visual and hearing defects, cleft palate and varying degrees of acne. Acne is usually severe, extensive, and resistant to treatment. Skin, eyes, and hair may show pigmented dilution. Other cutaneous abnormalities reported are hyperhidrosis and oculocutaneous albinism. Psychological counselling should include attachment and interaction with peers. Genetic counselling is an important factor. Recurrence risk for an affected individual to have an affected offspring is 50%. Treatment involves multidisciplinary teamwork including craniofacial surgeon, neurosurgeon, pediatrician, speech pathologist, and an orthodontist. Surgical care involves early release of the coronal suture and fronto-orbital advancement and reshaping to reduce dysmorphic and unwanted skull growth changes. Craniosynostosis requires multistaged operative procedures. Surgical separation of digits (mitten-glove syndactyly) provides relatively little functional improvement.

**Reference**