Apert syndrome: a case report

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Abstract

Apert syndrome was described as a triad of craniosynostosis, syndactyly and maxillary hypoplasia. The incidence of Apert syndrome is approximately one in 50,000 births. A three year old boy was brought with a history of facial, hand and feet deformities to the Pediatrics out patient department. On examination, he had symmetric syndactyly of the hands and feet. He also had craniosynostosis with deformed skull. This patient also exhibited midface hypoplasia, exophthalmia, ocular hypertelorism and high arch palate. Crowding of the teeth, malocclusion with anterior open bite is also found. The X-ray of the hand and feet showed skeletal fusion of phalanges (complex syndactyly). The case represents a rare condition where there is a mutation in the FGFR2 gene causing Apert syndrome.

Key words: craniosynostosis, acrocephalosyndactyly, spade hand, rose bud deformity

Introduction

Apert syndrome is a congenital disorder characterized by malformations of the skull, face, hands and feet. It is a form of acrocephalosyndactyly; also classified as a branchial arch syndrome, affecting the first branchial (or pharyngeal) arch, the precursor of the maxilla and mandible. Disturbances in the development of the branchial arches results in malformations of the skull, face, hands and feet. Apert’s syndrome was first described by Eugene Apert in 1906. He described a triad of craniosynostosis, syndactyly and maxillary hypoplasia. It is known to be inherited as an autosomal dominant, but most cases are sporadic. The sporadic cases are postulated to be associated with advanced paternal age. The incidence of Apert syndrome is approximately one in 50,000 births. Some investigators state that 4.5 percent of all craniosynostosis represent Apert syndrome.

Case report

A three year-old boy presented in paediatrics out patient department, Silchar Medical College & Hospital with deformities of hands and feet along with facial deformities. He was first in birth order and his mother’s age was 33 and father’s age was 36 yrs. There was no history of consanguinity or history of drug intake or radiation exposure at the time of pregnancy. In her family, there was no known history of congenital anomalies. On examination, he had symmetric syndactyly of the hands and feet. The syndactyly was complex with the fingers fused together at the tip (Type III Apert hand or rose bud deformity)

He also had craniosynostosis with deformed skull. This patient also exhibited midface hypoplasia, exophthalmia, ocular hypertelorism, and high arch palate. Crowding of the teeth and malocclusion with anterior open bite were also found. The X-ray of the hand and feet showed skeletal fusion of phalanges (complex syndactyly). On CT scan, patients had skeletal deformities of skull with premature fusion of sutures. No other systemic anomalies were found. Karyotyping of the patient showed no abnormalities.

Discussion

In the Apert syndrome, skull is deformed due to premature fusion of the sutures. Many of those patients may have agenesia of the corpus colossum, progressive hydrocephalus, and hippocampal abnormalities. They have shallow ocular orbits and the accompanying
Fig. 1: showing the case of the Apert syndrome

Fig. 2: showing syndactyly of both the hands

Fig. 3: showing syndactyly of both the feet

Fig. 4: showing high arch palate

Fig. 5: X-rays showing complex syndactyly with fusion of bones

Fig. 6: CT showing craniosynostosis and hypertelorism
exophthalmia may lead to blindness. The ocular manifestations, hypertelorism and exophthalmia are present in most of the cases. Increased intracranial pressure results from premature fusion of sutures with continued brain growth. There is hypoplastic midface and a vertically accentuated craniofacial complex. Ocular proptosis, down slanting of lateral canthus and palpebral fissures (antimongoloid slant), hypertelorism are present due to shortening of the bony orbit. Nasal bridge may be depressed with deviated nasal septum. There may be hypoplastic and retropositioned maxilla. The lips are bow shaped and often unable to form a lip seal.

Syndactyly or webbing of fingers causes immobility of fingers due to ossification of interphalangeal joints due to segmentation of embryonic phalanges. There can be a similar deformity involving the foot (sock foot). The deformity of the space between the index finger and the thumb may be variable. Based on this first web space, we can differentiate three different types of hand deformity.

Type I: Also called a "spade hand". The most common and least severe type of deformation. The thumb shows radial deviation but is separated from the index finger. The index, long and ring finger are fused together in the distal interphalangeal joints and form a flat palm. In the fourth web space, we always see a simple syndactyly, either complete or incomplete.

Type II: Also called a "spoon" or "mitten" hand. Here the thumb is fused to the index finger by simple complete or incomplete syndactyly. Only the distal phalanx of the thumb is not joined in the osseous union with the index finger and has a separate nail. Because the fusion of the digits is at the level of the distal interphalangeal joints, a concave palm is formed.

Type III: Also called the "hoof" or "rosebud" hand. This is the most uncommon but also most severe form of hand deformity in Apert syndrome. There is a solid osseous or cartilaginous fusion of all digits with one long, conjoined nail.

The oral cavity of Apert patients is also characteristic. Intraorally, there may be high arched and sagittally narrow palate seen with lateral palatal swellings with prominent central fissure. The maxillary arch is V shaped and sagittally narrow. Posterior slanting maxilla can give rise to class III malocclusion.

The findings include a reduction in the size of the maxilla, particularly in the anteroposterior direction. This reduction may result in tooth crowding. Cleft palate or bifid uvula is found in approximately 75 percent of those affected. Dental anomalies such as impacted teeth, delayed eruption, ectopic eruption, supernumerary teeth, and thick gingiva are also common.

Children with Apert syndrome may have mental deficiencies, short stature, hearing impairment, frequent ear infections, prominent and/or bulging eyes, a large or late-closing soft spot on the skull, other skeletal and congenital abnormalities.

According to the literature, Apert's and Crouzon's syndrome seem to be the same syndrome, with the exception of syndactyly of hands and feet in Apert's syndrome. Cleft or pseudocleft palate is a frequent finding in Apert's syndrome, whereas these traits are extremely rare in Crouzon's syndrome.

Apert syndrome may be an autosomal dominant disorder. Males and females are affected equally; however research is yet to determine an exact cause. Nonetheless, almost all cases are sporadic, signifying fresh mutations or environmental insult to the genome. It is inherited with mutations of either Ser 252 Trp or Pro253Arg in fibroblast growth factor receptor 2 (FGFR2) on chromosome 10q25. The FGFR2 is active at the metaphysis; diaphysis and also in the interdigital mesenchyme. Mutations in the FGFR2 gene cause Apert syndrome and alters the protein and causes prolonged signalling, which can promote the premature fusion of bones in the skull, hands, and feet.
Conclusion

Multimodality treatment is required at multiple settings, aimed at correction and rehabilitation of the patient with Apert syndrome. There is also scope of genetic diagnosis and research for this condition.

References