

Apert's Syndrome – Unusual cause of Syndactyly

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Abstract: Apert syndrome is a set of complex malformations of the first brachial arch, with manifestations on the skull, face, hands and feet. . It is a rare disease with an incidence of around 1 per 160,000 live births. This case report outlines rare occurrence of syndactyly associated with Apert's syndrome.

Keywords: Apert syndrome, syndactyly.

INTRODUCTION

Apert Syndrome was named after the French paediatrician Eugene Apert, who first described the collection of signs in 1906. It is a congenital disease that is a form of acrocephalosyndactyly, and is characterized by malformations of the skull, hands, feet and face. It is a rare disease with an incidence of around 1 per 160,000 live births ^[1]. Increased paternal age is a risk factor. It is thought that the affected chromosome is chromosome 10.

This syndrome complex involves syndactyly of the second, third and fourth digits with distal bone fusion; simple syndactyly of the fifth digit; foreshortened thumb with radial clinodactyly and symphalangism excluding the fifth digit. Unreleased syndactyly can significantly impair finger and hand function. The impairment is worse when the syndactyly is complete, complex, or involves the border digits with fingers of uneven lengths, such as the ring and small fingers or the thumb and index.

Complications arising due to late diagnosis, include defective brain development, mental retardation, increase in facial deformity, prognathic mandible, etc. Surgical separation of digits (mitten glove syndactyly) provides cosmetic results, and relatively good functional improvement. If surgery is postponed, incongruities of length and growth between fingers usually produce additional deformities in an already deformed hand. The typical craniofacial and dental features, the genetic transmission and its rare occurrence make it necessary to screen and carry out genotyping and genetic counseling of each diagnosed case. This case report attempts to throw some light on this rare syndrome.

CASE REPORT

A 14-year-old boy presented in our plastic surgery department with the complaints of recurrent ear discharge, delayed milestones, complex syndactyly right hand, mild bifid tongue, mild bat ears and facial deformity. His chest radiograph revealed no abnormality. His parents were non consanguineously married in their 3rd decade. No other family members were affected by same features. The child had a delay in psychomotor development, language disorders, academic delay, and recurrent ear infections. He had symmetrical syndactyly with complete fusion of all digits of hands (except thumb and little finger) [Figure-1].



Figure 1

Radiographs of right hand showed soft tissue syndactyly of second, third and fourth fingers) [Figure-2].



Figure 2

Pre anesthetic checkup revealed no other abnormality. Biochemical parameters were normal.

Corrective hand surgery in the form of syndactyly release and split skin grafting was performed for right hand. Post operative course was uneventful.

DISCUSSION

Apert's syndrome (AS) was described by Wheaton in 1894. In 1906, Apert published a summary on nine cases. Apert's syndrome makes up approximately 4% of all cases of craniosynostosis. The incidence is reported to be 1/160,000 live births . Amar T and others have reported a high incidence of delayed mental development in these children ^[2].

Apert's and Crouzon syndrome seem to be the same syndrome, with the exception of syndactyly of hands and feet in AS; when fusions are present, C5–C6 involvement in the Apert's syndrome and C2–C3 involvement in Crouzon syndrome separate the two conditions in most cases. Cleft or pseudocleft palate is a frequent finding in Aperts; whereas these traits are extremely rare in Crouzon syndrome. These features favor the diagnosis of Apert's syndrome over Crouzon syndrome in the current case. There is also an increased incidence of delayed mental development in these children, but many of them develop normal intelligence ^[3].

Chang, Ladd, Danton and Hertz outlined that digital separation of an Apert hand should begin at 9 months of age and should be completed by 2 to 4 years of age. They advocated early bilateral surgery on border digits followed by unilateral separation of middle syndactyly combined with thumb and digit osteotomies and bone grafts ^[4].

Holten and Smith concluded that Apert is a genetic anomaly causing variable and uncoordinated differentiation of the mesenchyme at the time of embryologic separation into its various skeletal components, particularly in the distal limb bud and craniofacial skeleton ^[5].

There is syndactyly or webbing involving second, third, and fourth fingers with the expression ranging from partial fusion of the skin to a true osseous syndactyly of fingers and toes^[6,7]. Prenatal detection of specific FGFR mutations now allows definitive antenatal diagnosis of Aperts.

Roje Z and others reported seven patients with Apert syndrome who underwent bilateral separation of border digits, which started between 1 and 2 years of age. The unilateral middle syndactyly mass division was done with osteotomy of the thumb and other digits. Bone grafting was carried out in later surgeries, which are usually completed by 4 years of age^[8]. The preoperative CT angiography imaging identified abnormalities in vascular anatomy that were incorporated into surgical planning for complete release of their syndactyly.

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