

Case Report

Apert Syndrome with CCHD: A Rare Association

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ABSTRACT

An 8months female child came to our Dept. of Paediatrics, M.K.C.G. Medical College, Berhampur, Odisha, India for evaluation of motor delay with history of cyanotic spell with features of Apert Syndrome. Apert syndrome is a rare type 1 acrocephalosyndactyly syndrome characterized by dysmorphic facial features, craniosynostosis and severe syndactyly of hands and feet. It represents an autosomal dominant inheritance and occurs due to the gene mutations in the receptors of the fibroblast growth factor 2(FGFR2).

Key Words: Acrocephalosyndactyly, Apert Syndrome

CASE REPORT

An 8months female child came to our OPD for evaluation of motor delay. There was no history of cough and cold or convulsion. There was history of repeated attacks of cyanosis for which she was hospitalized two times and Echocardiography report revealed D Transposition of Great Arteries with Unrestricted Arterial Septal Defect (L to R Shunt). That patient has low set ear, depressed nasal bridge, mitten hand. There was no similar illness in the family. There was no pallor, icterus, edema. Cyanosis and Clubbing was present. Pulse Rate was 136/min, Respiratory Rate was 34/min, BP was 93/68mm Hg. S1 and S2 normally heard without murmur, Bilateral Air entry equal without any added sound, conscious and oriented. Altogether Diagnosis of Apert Syndrome was made and sent for CTVS consultation. On follow up at the age of 14m she has undergone Modified Senning's Arterial Switch Operation at Fortis Hospital, New Delhi.



Figure 1: Image of mitten hand of our patient



Figure 2: Image of syndactyly of Toes



Figure 3: Post operative image of D TGA

DISCUSSION

Apert, in 1906, described the triad syndactyly of the hands and feet, dysmorphic facial features, and craniosynostosis characterizing the syndrome. ⁽¹⁻³⁾ With the mutations in the fibroblast growth factor receptors (FGFR-2) gene at locus 10q26, ^(2,4) a rare autosomal dominant heritage was linked to the syndrome. Apert syndrome have well established clinical features and are in agreement with the case described in the present report. Clinically the syndrome is characterized by premature fusion of the coronal suture and hypoplastic mid face. ^(1,2,5,7)

Short nose with depression of the nasal bridge and ocular anomalies, could also be observed. Tooth crowding and an anterior open-bite of the maxilla are the characteristics oral cavity findings. ^(3,5,7,8)

Ambylopia and Strabismus is more common in patients with FGFR2 Ser252Trp mutation and optic disc pallor is more frequent with FGFR2Pro253Arg mutation. Patients with FGFR2 Ser252Trp mutation have a significantly greater prevalence of visual impairment compared with patients with the FGFR2 Pro253Arg mutation. ⁽⁹⁾

Anomalies of the elbows and shoulders, viscera, skeleton and central nervous system ^(5,6) or abnormalities of the upper and lower respiratory tracts ^(1,3) have

been reported in some affected individuals. However in literature Apert Syndrome is not associated with occurrence of Congenital Cyanotic Heart Disease (CCHD). Thus, our case with D-TGA makes this a rare entity.

CONCLUSION

Apert syndrome is a rare autosomal dominant inheritance with multiple affects in various parts of body. For the effective planning and treatment of such patients, a multidisciplinary approach provided by dentists, plastic surgeons, neurosurgeons, ophthalmologists and geneticists should be included in the integral healthcare delivery system.

A Syndromic Approach must be made in each and every case of CHD along with Limb Defect. Few Syndromes like Holt-Oram Syndrome, Di George Syndrome, Ellis-van Creveld syndrome are examples of them. As mitten hand is not a feature of any of the above related syndrome most likely possibility of Apert Syndrome made considering the total phenotype. Our case is rare as it is associated with CHD (D-TGA).

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