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Case Report

A Rare Case Report: Acrocephalosyndactyly-Apert Syndrome

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Abstract

Apert syndrome is a rare genetic disorder involving mutation of Fibroblast growth factor receptor 2 (FGFR2) gene. FGFR2 gene is essential for growth of bones. Its deficiency results in abnormal premature fusion of bones of cranium, hands and feet. This results in abnormal size and shape of skull and syndactyly of hands and feet with its associated consequences and complications.

Keywords: Apert Syndrome; Case Report; Image

Abbreviations

ACS: Acrocephalosyndactyly; FGFR2: Fibroblast Growth Factor Receptor2 Gene

Case Report

A female infant brought by her parents for clinical opinion on her dysmorphic face and upper extremity skeletal deformities. Her pregnancy was uneventful, and she had regular antenatal check-ups. Antenatal ultrasound anomaly scan revealed abnormal skull. Baby was born to non-consanguineous young couple and delivered by spontaneous vaginal delivery at term to primigravida mother at a health care facility. Immediate postnatal period was uneventful. Baby was on mixed feeds (breast feeds cum top feeds). Baby vaccinated as per schedule.

At our outpatient clinic, we examined this baby at one year of age and found abnormal skull, face and acral skeletal deformities in both upper limbs. Both eyeballs were protruding anteriorly, depressed nasal bridge, low set ears, down slant palpebral fissure, prominent frontal bossing, hypertelorism, anterior fontanel open, flat occipital area and mid facial hypoplasia noticed. All fingers of hand on both sides were fused with single confluent nail plate (Mitten hand). Both bony and cutaneous fusion of hands was noticed. Both lower extremities and feet were normal. No other skeletal abnormalities noticed.

Neurosonogram: Revealed dilated ventricles, intact carpus callosum and presence of all major cerebral arteries.

- **Hearing screen:** Baby passed Oto acoustic emission test for both ears.
- **ECHO:** Echocardiography revealed no cardiac congenital defects.
- **Follow up:** On follow up at 6 years, baby was thin built and her gross motor milestones were normal but lag in academic performance to her peers at school. She was attending lower standard to her age equal peer group. Child able to hear, understand, speak and communicate with parents and peers.

Discussion

Synonym: Acrocephalosyndactyly.

Abnormal acrocephaly skull with craniosynostosis involving both coronal sutures and its facial developmental consequences like frontal bossing with high and broad forehead and flat oc-

ciput, Mitten hands (syndactyly of all digits of hands and or feet) and raised intracranial tension secondary to craniosynostosis is a clinical syndrome called as Apert syndrome (synonym: acrocephalosyndactyly). First case was described by Eugene Charles Apert, French paediatrician in 1906 (Driessen C, 2017). Primary diagnosis is based on characteristic clinical features only. It is a rare anomaly with incidence of 1 in 50,000 to 80,000 live births (Pius S, 2016). Most cases are sporadic but autosomal dominant inheritance is possible with 50% penetrance equally affecting both sexes. It is caused by mutation involving fibroblast growth factor receptor 2 gene (FGFR2). FGFR2 gene is essential for fibroblast growth factors in skeletal development. Other associated abnormalities include ventriculomegaly, carpus callosum abnormalities, raised intracranial pressure, mental sub normality, seizural tendency, hearing and visual impairment, psychosocial maladjustment, cleft palate, acral skeletal deformities involving both upper and lower limbs and congenital heart defects. Child should be managed with multidisciplinary approach involving paediatrician (for regular vaccination, nutritional advice, monitoring growth and development), neurosurgeon (cranial suture releasing craniotomy to allow brain growth and treat intracranial tension), otolaryngologists (for taking care of ENT and hearing), ophthalmologist (for caring acuity of vision), cardiologist(for managing congenital cardiac defects and improving quality of life), orthopaedic surgeon (for surgical management of syndactyly, improve thumb movements and to restore pincer grasp for carrying out daily activities), developmental neurologist (for seizures and neurodevelopmental follow up) and psychologist (for emotional support to the family and child) [1,2].

Conflict of Interest

There are no conflicts of interest.

Sponsor

There are no sponsors to this study paper.

Consent

Consent was obtained from parents to publish the images for scientific and academic purposes.

Bibliography

1. Driessen C., *et al*. "Apert syndrome: the Paris and Rotterdam philosophy - REVIEW". *Expert Opinion on Orphan Drugs* 5.7 (2017): 599-605.

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