Case Report

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A Case Report Apert Syndrome

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Abstract

Apert syndrome (acrocephalosyndactyly) is autosomal dominant disorder characterized by craniosynostosis, syndactyly and midfacial malformation.

Keywords: Possible mechanism of injury and operative procedures are discussed.

Introduction

Apert syndrome is autosomal dominant inherited craniosynostosis syndrome. Apert syndrome is a type of acrocephalosyndactalia (Type 1) and is charecterized by craniosynostosis, a cone shaped calvarium, pharyngeal attenuation, midfacehypoplasia, ocular manifestations and syndactly of hands and feet.

Case Report

A 5 month old male child was brought with complaints of large head, symmetric syndactyly of both hands and feet, and proptosis. He was first born from a non-consanguinous marriage. No family history of foetal wastage or maternal exposure to radiation and use of unprescribed medications. No family history of similar complaints or any other congenital abnormality was reported.

Examination revealed abnormal cone shaped head (turribrachycephalic head) and AFpatent measuring 5 by 6 cm, and posterior fontanelle was also widely patent measuring 4 by 3 cm with sutural diasthesis involving saggital and mitopicsutures. Ocular manifestations proptosis, strabism, hypertelorism, down slidinglateral palpebral fissures were present. He had depressed nasal bridge, cross bow shaped lips. Bilateralsyndactyly was present, symmetricaly in voloving both hands and foot known as mitten hand. A similar

syndactyly was present in the foot.

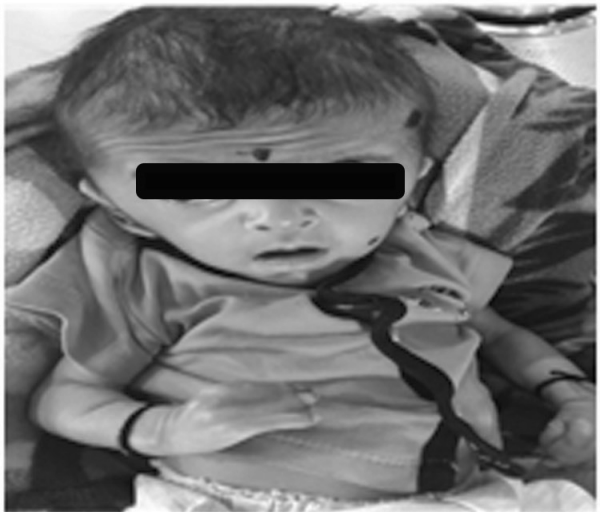

Fig. 1: Infant with a turribrachycephalicskull, frontal bossing, hypertelorism, depressed nasal bridge, antimongoloid slant of eyes

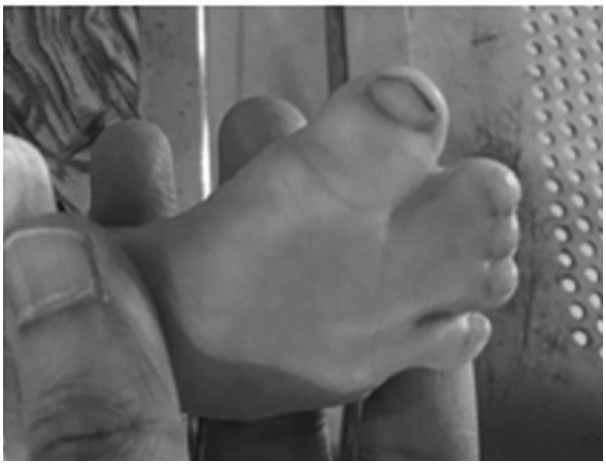

Fig. 2: Syndactyly with fusion of digits

Fig. 3: Syndactyly of feet

Discussion

Apert syndrome is an autosomal dominant disorder that occurs in 6 to 15.5 out 1 million livebirths.Most cases are sporadic. Higher mutation rates are seen in male than females because of higher germline mutations in males[2]. Increased paternal age increases the rate of mutations[3]. Most of the cases of Apert syndrome (around 98%) are caused by specific missense substitution mutations, involving adjacent amino acids in the linker between the second and third extra cellular immunoglobulin domains of FGFR2, which maps to chromosome bands 10q26. Alu-element insertion mutations in or near exon 9 of FGFR2 accounts for remaining cases of apert syndrome[4]. Increased paternal age accounts to most of the sporadic cases resulting from new mutations. Increased paternal age exponentially increases the incidence of FGFR2 mutations. Premature fusion of skull bones results in characteristic facial features of Apert syndrome. Bicoronal synostosis and maxillary hypoplasia that causes recessed forehead and flat midface are the two noticeable craniofacial defects. Proptosis of eyes, beaked nose and underdeveloped upper jaw leads to crowded teeth and other dental problems. Malformation of the corpus callosum and limbic system structures, gyral abnormalities, hypoplastic white matter, and heterotopic gray matteroccres due to early fusion of skull bones. Ventriculomegaly is seen in most patients of apert syndrome resulting from distortion imposed upon a large brain within a misshapen skull leading to (cone-shaped) head with flat occiput, short anterior-posterior diameter, prominent elongated forehead and a short broad nose and reduced nasolabial angle[5]. Syndactyly is characteristic feature of apert syndrome and the severity of fusion varies from three digits on each hand and foot is fused together.In addition, signs and symptoms of Apert syndrome may include hyperhidrosis, hearing impairment[6] resulting from persistent middle ear effusion,severe acne, patches of missing hair in the eyebrows, fusion of spinal bones in the (cervical vertebrae), may be associated with opening in the roof of the mouth (a cleft palate), this case did not have cleft palate. Shoulders, elbow, hips, knees and ribs abnormalities can be seen in patients of Apert syndrome[7].

Treatment of apert syndrome, The modality is generally divided into two major approaches, the first one which include the medical management and the second one is the surgical management which is the main modality of the treatment. Measures should be taken to protect cornea, by instillation of lubricating ointment in the eyes at

bedtime to protect corneas from desiccation and also artificial tear drops during the day.Upper airway infection should be treated and excessive secretions should be removed. Humidification with added oxygen and judicious use of topical nasal decongestants is helpful.There may be a need to employ continuous positive pressure ventilation[1,4]. Antibiotic therapy is essential in the presence of chronic middle ear effusion associated with bilateral conductive hearing deficit.Rehabilitative measures to prevent cognitive impairment and delay in speech and language developmentshould be undertaken.Aim of surgery in treatment of Apert syndrome is to release the cranial sutures in order to permit a proper brain development, to prevent hydrocephalus due to raised intracranial pressure, repair the cleft in order to prevent nasal regurgitation as well as facilitate proper speech[8,9]. Correction of midfacial hypoplesia to facilitate proper breathing improves body image and self-esteem. Surgical release of the digits (syndactyly) is to provide better grasp.

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