A rare case of neonatal Apert syndrome

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ABSTRACT

Apert syndrome is a rare Type I acrocephalosyndactyly syndrome. It is a congenital disorder characterized by premature fusion of cranial sutures (craniosynostosis), malformation of skull, hands, face, and feet. Apert syndrome has an incidence of 1/50,000 to 1/80,000 live births. It is 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 branchial arches in the fetal development create lasting and widespread effects. It is inherited as an autosomal dominant and occurs due to the gene mutations in the receptors of the fibroblast growth factor. Management of Apert syndrome requires multidisciplinary approach. We, hereby, report a case of a 5-h-old neonate with Apert syndrome.

Key words: *Acrocephalosyndactyly, apert syndrome, autosomal dominant, fibroblast growth factor*

Apert syndrome was first described by French Physician Dr. Eugene Charles Apert in 1906. It is a rare congenital disorder [1], it is inherited as an autosomal dominant, assigned to mutations in the fibroblast growth factor receptors-2 (FGFR-2) gene at locus 10q26 [2,3]. FGFR-2 have a high affinity for fibroblast growth factors that play a role in signaling pathways with multiple biologic effects including cranial growth and development when bound to their specific receptor. Embryologically, the hands and feet have a selective cell death or apoptosis causing separation of digits; however, in acrocephalosyndactyly, apoptosis fails to occur and the skin and rarely bone between the fingers and toes fuse [4,5]. This rare clinical abnormality has to be differentiated from other craniofacial syndromes such as carpenter syndrome, Crouzon disease, Pfeiffer syndrome, and Saethre-Chotzen syndrome. The case of a 5-h-old neonate with Apert syndrome is reported here.

CASE REPORT

A 5-h-old neonate referred from peripheral secondary care hospital with complaints of respiratory distress since birth and multiple congenital malformations noticed at birth. The baby was born to a consanguineous parent of gravida four mothers of age 23 with three previous abortions with uneventful antenatal and natal history. The baby was delivered at term gestation by normal vaginal delivery, and the baby was said to have cried immediately after birth with Apgar score of 7 at 1 min and 8 at 5 min, respectively. While during postdelivery care, the baby was found to have breathing difficulty and congenital malformations; hence, the baby was referred to our hospital.

On examination, the baby had mild respiratory distress with acrocyanosis, baby's weight was 2.5 kgs, and length was 48 cm and found to have multiple congenital malformations. On examination, the child had dusky peripheries, brachycephaly with head circumference of 31 cm, flat occiput, and prominent forehead. The anterior fontanelle was wide open measuring about 3.5 by 3.5 cm with sutural diathesis and low set ears. There were pinched nose and trapezoid mouth with cleft palate (Figs. 1 and 2). In upper limbs, the baby has syndactyly characterized by fusion of all the fingers giving the appearance of spoon (Fig. 3). In lower limbs, all toes are fused giving it socks like appearance (Fig. 4). After initial stabilization, the baby was referred to higher center for multidisciplinary approach after giving supportive care.

DISCUSSION

Apert syndrome belongs to acrocephalosyndactyly group of disorders, and it is a genetic disorder characterized by craniosynostosis, midline facial hypoplasia, severe symmetrical cutaneous syndactyly of the hands and feet, as well as central nervous system, heart and kidney abnormalities. Craniofacial findings of the Apert syndrome include closed coronal suture during birth and the presence of a large fontanel glabella. The forehead is upfront, more prominent and elevated, while the superciliary arch is depressed due to the anterior dislocation of the sphenoid bone, temporal regions appear protruding, while the occiput is flattened. This arrangement negatively affects the development of maxillary bone, thus prevents the development of the nasopharyngeal cavity. Typical ophthalmologic findings

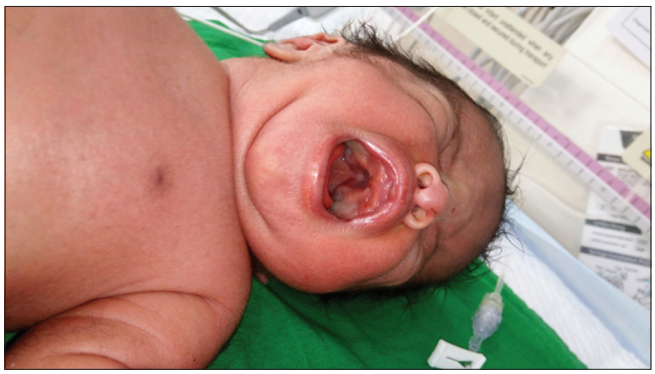

Figure 1: Cleft palate

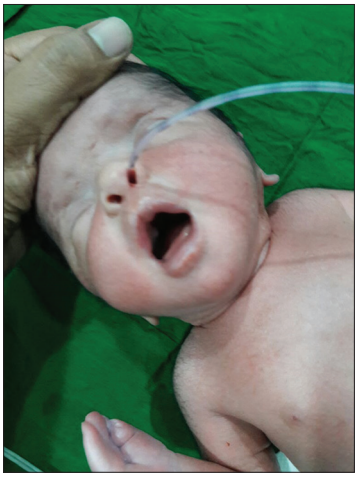

Figure 2: Pinched nose and trapezoid mouth

of the Apert syndrome include hypertelorism, papilla edema, and proptosis [6,7].

In Apert syndrome, syndactyly is characterized by progressive fusion of the bones of the hands and feet during skeletal development. Symmetrical syndactyly most frequently occurs between the second, third, and fourth fingers, while the first and fifth fingers are generally free. In addition to musculoskeletal abnormalities, abnormalities of the cardiovascular system, cleft palate, genital-urinary system, and central nervous system can also be encountered. Among these cases, the most common cardiovascular abnormalities are ventricular septal defect and dextra positioning of the aorta [8,9]. While the majority of cases are sporadic, some cases are associated with autosomal dominant inheritance. The rate of occurrence is equal between men and women.

Differential diagnosis should include evaluation of another genetic disorder associated with craniosynostosis. The most common genetic disorders accompanying craniosynostosis include Crouzon, Apert, Carpenter, Apert-Crouzon syndrome, Jackson-Weis syndrome, and Pfeifer syndrome. Apert syndrome can be differentiated by genetic analysis and typical appearance. Prenatal diagnosis can be made by establishing craniosynostosis and syndactyly in ultrasonography. The earliest gestational week to notice these findings varies between 16 and 32. In sporadic cases, molecular genetics support the diagnosis [10]. Treatment of patients with Apert syndrome requires a multidisciplinary approach, involving follow-up therapies provided by plastic and reconstructive surgery, neurosurgery, neurology, and psychiatry specialists.

Figure 4: Spoon like apperance of toes in upperlimbs

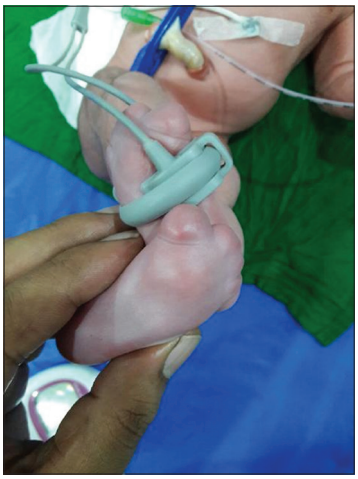

Figure 3: Socks like apperance of toes in lower limbs

CONCLUSION

Apert syndrome is a rare disorder. When a neonatologist is confronted with this condition, effort should be made to evaluate the patient so as to make a definitive diagnosis as early as possible and plan an effective multidisciplinary approach.

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