Apert’s Syndrome - A Rare Case Report

Dr Daggula Devi Bharathi¹, Dr. Santhi Sri Appireddy², Dr. M Chenchi Reddy³

¹(M.S), Post Graduate, Ophthalmology, Katuri Medical College and General Hospital, Chinakondrupadu, Guntur District, Andhra Pradesh – 522019, NTR University of Health Sciences

²(M.S), Post Graduate, Ophthalmology, Katuri Medical College and General Hospital, Chinakondrupadu, Guntur District, Andhra Pradesh – 522019, NTR University of Health Sciences

³M.S; Ophthalmology, Katuri Medical College And General Hospital, Chinakondrupadu, Guntur District, Andhra Pradesh – 522019, NTR University of Health Sciences

Abstract: Apert’s syndrome is the rare acrocephalosyndactyly syndrome type 1, characterized by craniosynostosis, dysmorphic facial features and severe syndactyly of hands and feet. It shows an autosomal dominant inheritance pattern assigned to mutations in fibroblast growth factor receptor gene. We present a case of a 8 year-old female patient diagnosed on physical examination with Apert’s syndrome based on acrocephaly, prominent forehead, ocular hypertelorism, proptosis, short and broad nose, pseudopognosthmus, dental crowding, webbed neck, and bilateral syndactyly of hands and feet. The multiple phenotypic signs of Apert’s syndrome make multidisciplinary team, including dentist, neurosurgeon, plastic surgeon, phystiatrist, ophthalmologist, perinatalologist and geneticist, essential for successful management.

Keywords: Acrocephalosyndactyly, Apert syndrome, craniosynostosis

1. Introduction

Apert syndrome was firstly described by a French physician, Eugene Apert. This syndrome is a form of acrocephalodactyly (Type 1). It is a rare congenital disorder characterized by an autosomal dominant inheritance which manifests itself with craniosynostosis, midface hypoplasia, and syndactyly of hands, and feet, with a tendency of fusion of boney structures. Males and females are equally affected. It has an autosomal dominant trait with the locus of mutation of FGFR2 on chromosome 10q26.

2. Case Report

An 8 year old female child was brought with a chief complaint of discharge from the right eye since 3 weeks. And also complaints of difficulty in chewing and facial deformity. She had no other past significant medical history. Her family history revealed no other family members were affected by same features.

On examination, the girl was found to have flattened occiput with frontal prominence, retruded midface, incompetent everted lips and prognathic mandible. She had symmetrical syndactyly with complete fusion of all digits of hands and feet. Intraoral examination showed normal mouth opening with high arched (V-shaped) palate. Maxillary alveolar ridges were thick with crowding of maxillary teeth.

On ocular examination she had shallow orbits with severe bilateral proptosis, hypertelorism and exposure keratopathy leading to keratitis and corneal ulceration resulting in discharge from the right eye.

The systemic examination revealed no other abnormality.

On Investigation

• Orthopantomogram showed deformity of maxilla with malaligned maxillary teeth and high arched palate,

• X-ray of hand-wrist showed that there was bony fusion of the phalanges.

• X-ray of spine, abdominal ultrasonography, and echocardiography were normal. Later CT scan of the face was done, which revealed that there was obvious abnormal growth of middle-third of the face with bilateral proptosis and deviation of nasal septum.

Due to the typical features and presence of the triad of craniosynostosis, syndactyly of hands and feet with maxillary hypoplasia, patient was diagnosed as Apert’s syndrome. Due to presence of syndactyly of hands and feet, Crouzon’s syndrome was ruled out.

Patient was advised orthodontic treatment with orthognathic surgery for correction of the facial dysmorphisms, and corrective plastic surgery for the syndactyly of hands and feet. Corneal ulcer with discharge was advised for a scraping and culture sensitivity. In the mean while the girl was started on topical moxifloxacin and lubricant eye drops, was advised dark goggles and lid taping to prevent exposure keratopathy. As patient was not ready for any interventional procedures, only oral prophylactic measures were taken and kept under regular follow-up.

Figure 1: A girl of age 8 years front view
3. Discussion

Apert’s syndrome is an autosomal dominant condition, but in many cases the inheritance is sporadic. A localized mutation of the gene FGFR2 with chromosomal localization at 10q26 is responsible. The incidence of Apert’s syndrome is approximately one in 50,000 births. Unique fibroblast growth factor receptor 2 (FGFR2) mutations lead to an
increase in the number of precursor cells that enter the osteogenic pathway. Ultimately, this leads to increased subperiosteal bone matrix formation and premature calvaria ossification during fetal development. Once a suture becomes fused, growth perpendicular to that suture becomes restricted, and the fused bones act as a single bony structure. Compensatory growth occurs at the remaining open sutures to allow continued brain growth. However, complex, multiple sutural synostosis frequently extends to premature fusion of the sutures at the base of the skull causing brachycephaly, midfacial hypoplasia, shallow orbits, hypertelorism, foreshortened nasal dorsum, maxillary hypoplasia and mandibular prognathism as seen in our case.

According to the literature, Apert’s and Crouzon’s syndrome has similar clinical presentation with the exception of syndactyly of hands and feet in Apert’s syndrome which is seen in our case.

Various complications arising due to late diagnosis, include defective brain development, mental retardation, increase in facial deformity, prognathic mandible, etc. It is very important to diagnose and treat this syndrome at an early age to prevent the late diagnosis effects. The treatment of patients presenting complex facial deformities is one of the most challenging multidisciplinary tasks.

4. Summary and Conclusion

The rarity of the Apert’s syndrome, its spectra heterogeneity, craniofacial anomalies and being a multifactor digenetic syndrome, it is necessary to carry out the genetic advising and detailed study in each individual affected by this syndrome. Therefore, the study has to be continuous to promote prenatal diagnosis and early multidisciplinary treatment approach to prevent the late diagnosis effects. So, our case is an addition to the literature of the clinical and radiographic features and treatment modalities of this rare syndrome.

References