Acrocephalosyndactyly Syndrome: A Rare Case Report

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Abstract

Acrocephalosyndactyly or Apert syndrome is a rare disorder in which the fingers and toes are fused, giving the appearance of webbed hand and feet. Also, there is craniosynostosis, leading to severe mid-face hypoplasia and characteristic facial deformities. Here there is a genetic mutation in the fibroblast growth factor receptor 2 (FGFR2) gene with autosomal dominant inheritance. A case of an 8-year-old male child is reported here with frontal bossing, acrocephaly, prominent ocular hypertelorism with divergent strabismus, saddle-shaped nose, mid-face hypoplasia, crowded anterior teeth, sparse hair and severe bilateral syndactyly of the feet and hands.

Keywords*: Craniosynostosis, Facial Deformity, Malocclusion, Syndactyly.*

Introduction

Eugene Apert, a French doctor first described Apert syndrome which has acrocephalosyndactyly. [1] This is a rare autosomal dominant developmental disorder in which the individual is affected from birth with craniosynostosis, midface hypoplasia, webbed, fused hands and feet.[2] Deformities like a cone-shaped skull, frontal bossing, proptosis, prominent hypertelorism with divergent strabismus, saddle-shaped nose and a hypoplastic maxilla are observed. Dental malocclusion, high arched palate, ectopic, malaligned crowded teeth are seen. The mandible may be normal but due to a hypoplastic maxilla, the mandible may appear prognathic. Other systemic diseases have been associated with the syndrome.[3]A case of an eight-year-old male child affected by acrosyndactyl syndrome with all the classic features is reported for documentation.

Case Report

An 8-year-old male child reported to the dental hospital for enhancement of facial aesthetic appearance. History revealed that the child was born with facial deformities and fused hands and feet. He was otherwise normal, and the medical and family history was non-contributory.

On clinical examination, the face was deformed, (figure 1 and 2) and the fingers and toes appeared web-like and fused. (figure 3 and 4) He had malocclusion, high arched palate, pseudo-class III malocclusion, cross-bite and delayed eruption of permanent teeth. The skull was cone-shaped or acrocephalous, hypoplastic maxilla, bilateral divergent strabismus, hypertelorism with propoptosis, scanty hairline, saddle-shaped nose and frontal bossing. Features were suggestive of Acrocephalosyndactyly syndrome.

The patient was referred to a paediatrician for further evaluation to exclude other systemic diseases. The patient's mental function was evaluated and found normal. He was found not to be suffering from any cardiac, gastrointestinal, renal or respiratory diseases. Full skeletal radiographs were taken and revealed syndactyly, acrocephaly and dental malocclusion.

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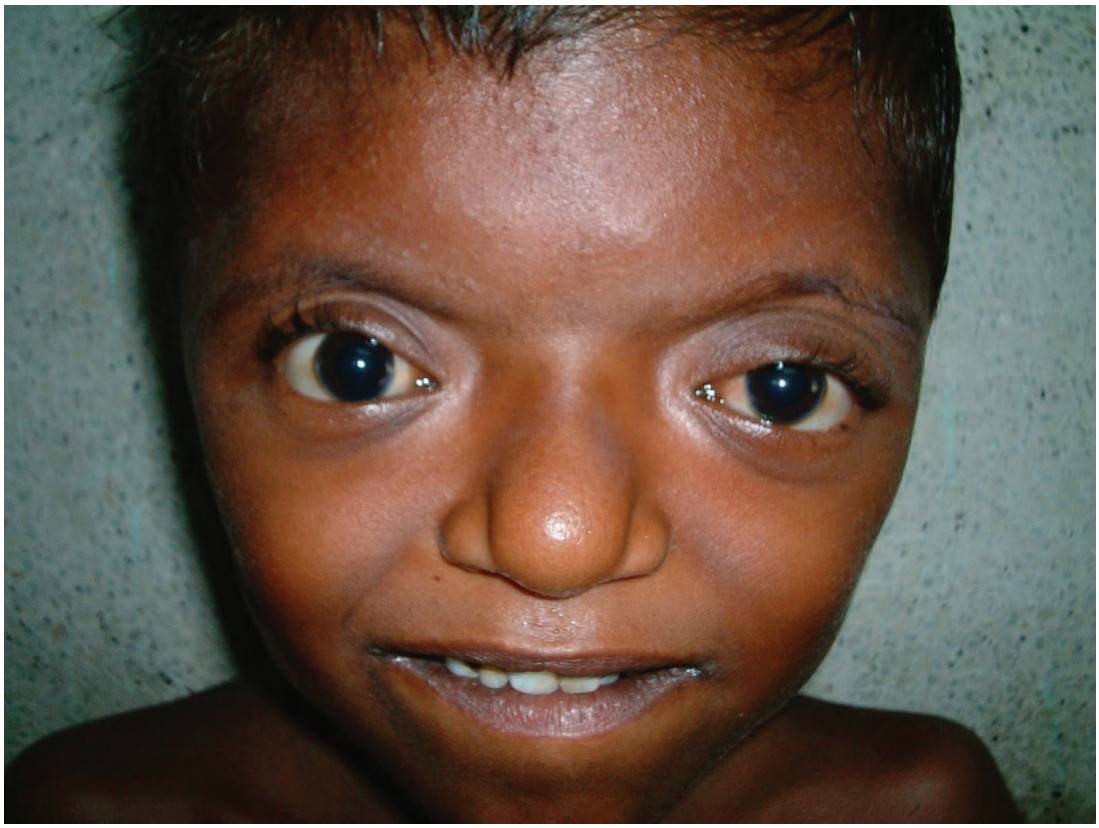

Figure 1: Frontal view showing divergent strabismus, hypertelorism, saddle shaped nose.

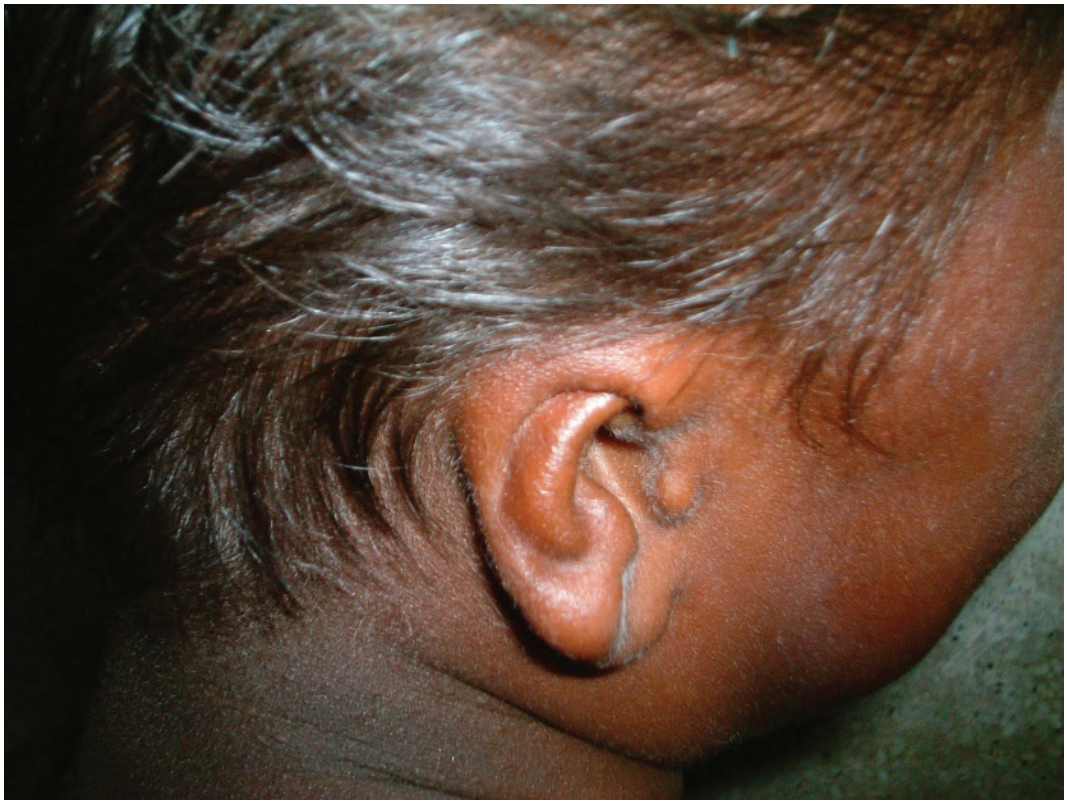

Figure 2: Profile picture showing ear tags and sparse hair.

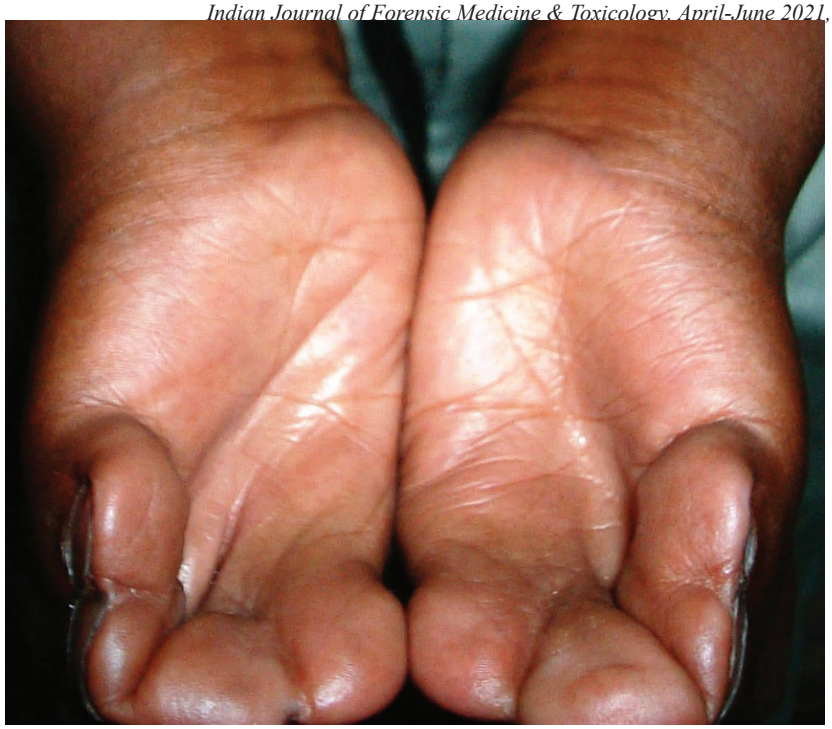

Figure 3:Syndactyly in hands appearing like web.

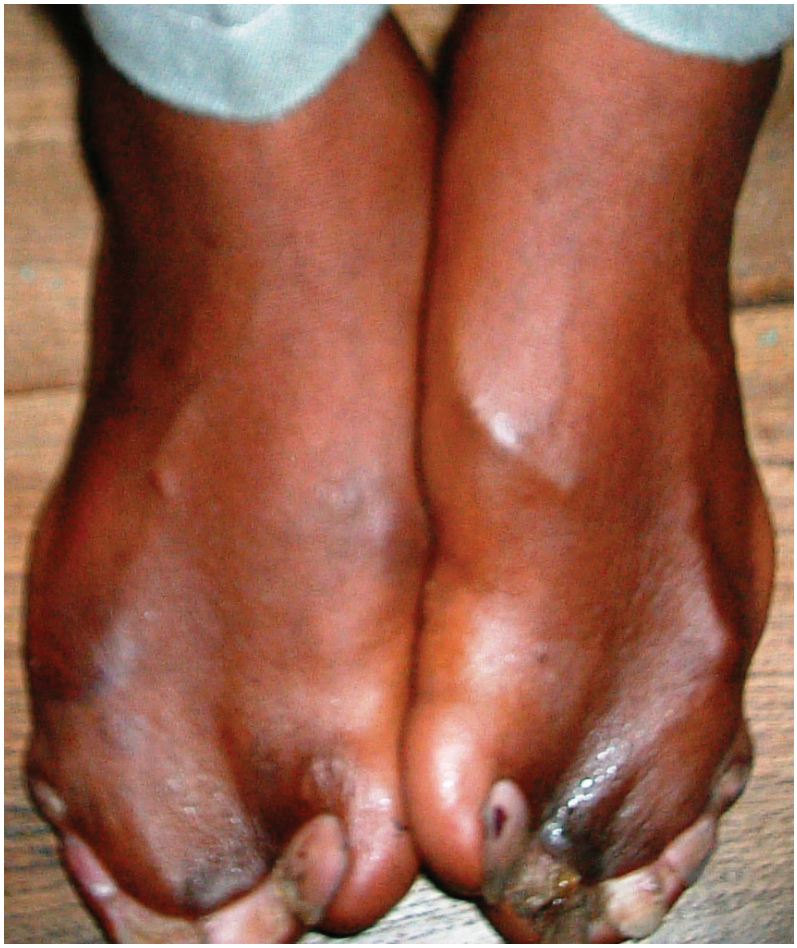

Figure 4: Syndactyly of the toes appearing like webbed feet.

Discussion

Acrocephalosyndactyly syndrome (AS) or Apert syndrome is one of the craniosynostosis syndromes and is associated with syndactyly. which is a form of craniosynostosis. [4] It has been reported in the literature that parenting at an advanced age leads to AS. [5] Though it is of autosomal dominant inheritance, sporadic mutations occur like in the present case without any family history of AS. There is no gender or racial predilection and the disorder is quite rare occurring about ten to fifteen in a million live births. [6]

The pathology is in the mutation of FGFR2 gene which codes for a protein known as fibroblast growth factor receptor and is responsible for forming vascular channels, healing of wounds, the evolution of an embryo, regulates the division of cells, cell growth and cell maturation. This mutated gene causes defective signalling in pathways that initiate the skull bone fusion, hence there is premature craniosynostosis.[7] There is also a defect in the epithelial-mesenchymal interaction leading to defective membranous and endochondral ossification. [8]

The facial appearance results due to premature fusion of the coronal sutures so there is a short anteroposterior diameter and enlarged forehead giving rise to the acrocephalic skull. Depressed nasal bridge and hypoplasia of maxilla, as well as dental malocclusion, are common findings. [7,8] Ocular anomalies like hypertelorism, strabismus, propoptosis are also frequent manifestations.[9] Syndactyly of the fingers and toes are the classic features and most important differentiating feature between other craniosynostosis syndromes. [8-10]

Extremely rare systemic manifestations like vertebral mal-segmentation, hypogenesis/ agenesis of the corpus callosum, defects of the cerebrum and cerebellum, enlarged ventricles have been reported. [11] Obstructive sleep apnoea, respiratory infections, mal-nourishment, etc have been rarely mentioned in the English literature. Mental retardation may also be rarely seen. [12]

Oral manifestations like malocclusion and crowded teeth are due to the decrease in the anteroposterior maxillary growth. [11]

Hand and foot deformities are characteristic and syndactyly may be associated with polydactyly also which was not seen in our patient. Since the quality of life of these patients gets severely affected due fused hands and toes, surgical correction of hands and feet is a priority. The patient's feet are unable to bear the weight like normal feet, and they cannot wear normal shoes also. Many a times patients are most uncomfortable wearing shoes. Other craniosynostosis syndromes like Crouzon syndrome, Carpenter syndrome, Chotzen syndrome and Pfeiffer syndrome must be considered in the differential diagnosis.[12]

Conclusion

The present reported case had the characteristic facial features and syndactyly, all suggestive of AS besides sparse scanty hair and short stature. It may be emphasized that when there is a family history of AS then foetal DNA analysis must be performed to diagnose any DNA mutations like the FNGR gene mutation Moreover pre-natal diagnosis can establish craniosynostosis as well as syndactyly on prenatal ultrasonographic images. AS is a rare disorder without a complete cure and the aesthetic issues associated with disease leads to depression and low confidence in the patients due to their facial appearance. Hence the aim is to diagnose such developmental anomalies early in the foetus and consider medical termination of pregnancy.

Conflicts of Interest: None

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Ethical Permission: Approved

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