A Case Report of Oral Findings in a Patient with Apert Syndrome

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ABSTRACT

Background: Apert syndrome, also called acrocephalosyndactyly, is a very rare genetic condition. It is characterized by craniosynostosis, midface hypoplasia and syndactyly with an autosomal dominant inheritance in most cases. Apert syndrome constitutes 4.5% of all cases of craniosynostosis, and it occurs in every 1 in 65,000 live births

Objective: To report a case of Apert syndrome in an 18-year-old with characteristic facial and oral features

Case presentation: An 18-year-old male with Apert syndrome presented with the characteristic craniofacial and oral features: Acrocephalic skull, exophthalmos, hypertelorism, downward slant of the palpebral fissures, depressed nasal bridge, syndactyly, retruded maxilla, prognathic mandible, bow-shaped lips, pseudocleft and malocclusion.

Conclusion: A multidisciplinary team of healthcare professionals should be involved in giving early diagnosis, early surgical intervention and comprehensive rehabilitation to improve the quality of life of patients with Apert syndrome.

Key words: Apert syndrome, craniosynostosis, syndactyly, midface hypoplasia, oral health

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INTRODUCTION

Apert syndrome, also called acrocephalosyndactyly, is a very rare genetic condition. It is characterized by craniosynostosis (early fusion of cranial sutures), mid-face hypoplasia, syndactyl, acrocephaly and often anomalies in other organs.[1,2]It has an autosomal dominant inherited pattern; however, some sporadic cases have been found.[3,4]Mutation in the fibroblast growth factor receptor 2 gene (FGFR2) occurs in 98% of patients with Apert syndrome.[5–7] S252W is the most common mutation, which causes severe craniofacial anomalies, and P253R mutation causes severe syndactyly.[6–8]

Apert syndrome is 4.5% of all cases of craniosynostosis, and it occurs in every 1 in 65,000 live births.[9] The first documented report of Apert syndrome in Nigeria was in 1982.[10]Features such as occipital flattening, bitemporal widening, shallow orbits, hypertelorism, ocular proptosis, depressed nasal bridge, down slanting of palpebral fissures (antimongoloid slant), syndactyl of hands and feet have been found to occur causing aesthetic and functional concerns in Apert syndrome.[1,3,5,11–14]

Individuals with Apert syndrome often present with features that affect oral health, such as midface hypoplasia, prognathic mandible, high arched palate with a midline pseudocleft, cleft of the soft palate, anterior open bite, delayed eruption, ectopic eruption, crowding of teeth, bilateral crossbite and bow-shaped lip which causes the inability to achieve an oral seal.[13,15–17].

Imaging studies (Skull, Spinal and Limb radiography, CT Scan and MRI) can be used for the investigation of Apert syndrome.[18,19]Molecular analysis in more than 98% of cases are caused by specific missense substitution mutations involving adjacent amino acids (Ser252Trp, Ser252Phe, or Pro253Arg) in exon 7 or FGFR2.[18,19] Diagnosis can be prenatal and often identified in the third-trimester based on acrocephaly, mitten-like hands and proximally placed and radially deviated thumbs using ultrasonograhy.[20]Non-invasive prenatal diagnosis of Apert syndrome using polymerase chain reaction and restriction enzyme digestion of cffDNA in maternal plasma has been reported.[21]

We report a case of an 18-year-old Nigerian male with Apert syndrome having characteristic facial and oral features.

CASE REPORT

An 18-year-old male SM with Apert syndrome presented to the paediatric dental clinic at Lagos University Teaching Hospital, Lagos State in Nigeria, with complaints of "disarranged teeth". His brother was the informant. There were also complaints of saliva drooling due to his inability to close his mouth. There is a positive history of mild intellectual disability, as he dropped out of school at JSS 2 due to poor performance. However, he was able to learn a skill and is currently a visual artist.

None of his siblings or relatives have Apert syndrome. His father was 51 years and his mother 44 years when he was born. There was no known medical condition at the time of presentation, and he was not on any routine medication. There was a positive history of surgical intervention for the syndactyly of his hands when he was seven years old. (Figure 4)

On general physical examination, there was the presence of acrocephalic skull, exophthalmos, hypertelorism, downward slant of the palpebral fissures and lateral canthus, depressed nasal bridge and syndactyly of both hands and feet. Extraoral examination revealed retruded maxilla, prognathic mandible, and inability to achieve oral seal due to bow-shaped incompetent lips. (Figure 1 and Figure 2) Intraorally, there was a high-arched palate and the palatine processes had bilateral swellings forming a pseudocleft (Figure 5 and Figure 6).

Patient had poor oral hygiene with a Simplified oral hygiene index (OHI-S) score of 3.1, according to Greene and Vermillion. Teeth present:18, 17, 16, 15, 14, 12, 11, 21, 22, 24, 25, 26, 27, 37, 36, 35, 34, 33, 32, 31, 41, 42, 43, 44, 45, 46, 47; Palatally displaced 15, 25. Maxillary canines were missing with the presence of anterior open bite. Orthopantomogram (OPG) showed erupted:18, 17, 16, 15, 14, 12, 11, 21, 22, 24, 25, 26, 27, 37, 36, 35, 34, 33, 32, 31, 41, 42, 43, 44, 45, 46, 47 and unerupted teeth are 13, 23, 28, 38, 48. (Figure 9)

Diagnosis of Angles class III malocclusion on a class 3 skeletal base pattern complicated by impacted maxillary canines, anterior open bite, palatally displaced maxillary premolars, impacted third molars and chronic gingivitis secondary to plaque and calculus accumulation in a patient with Apert syndrome.

Oral hygiene education, Ultrasonic Scaling and polishing were done. The patient was referred for orthodontic evaluation and treatment.

Figure 1: Extraoral clinical picture of SM (frontal view)

Figure 2: Extraoral clinical picture (profile view)

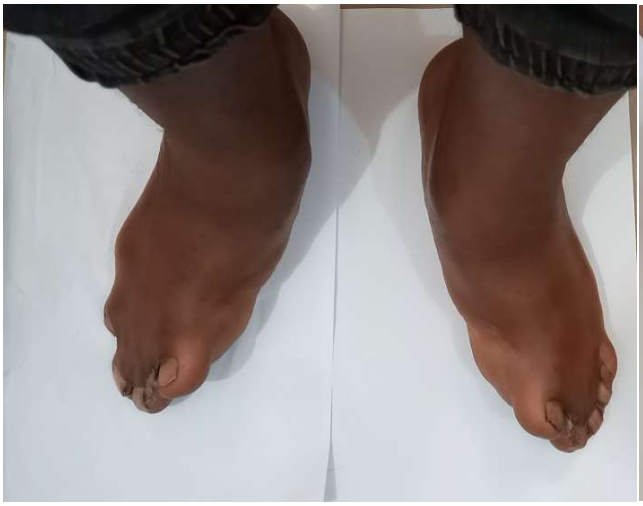

Figure 3: Syndactyly (feet)

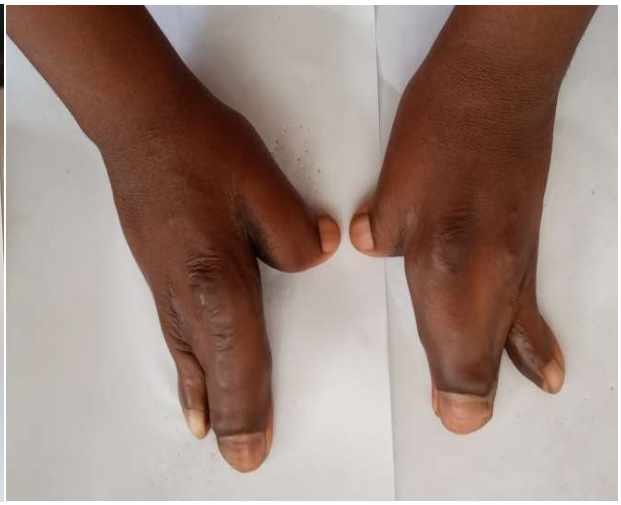

Figure 4: Syndactyly (hands)

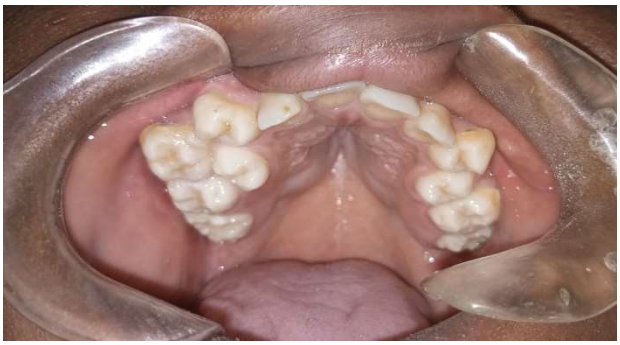

Figure 5: Intraoral picture showing high arched palate

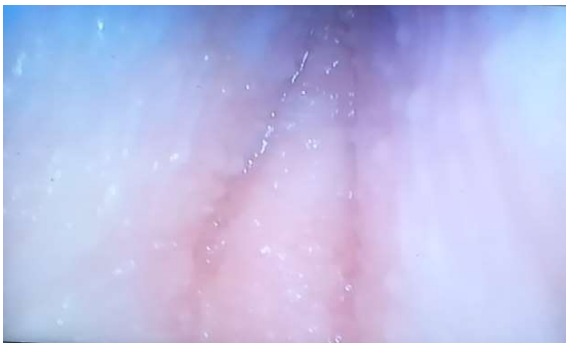

Figure 6: Intraoral pseudo cleft on palate

Figure 7: Intraoral clinical picture of teeth in occlusion

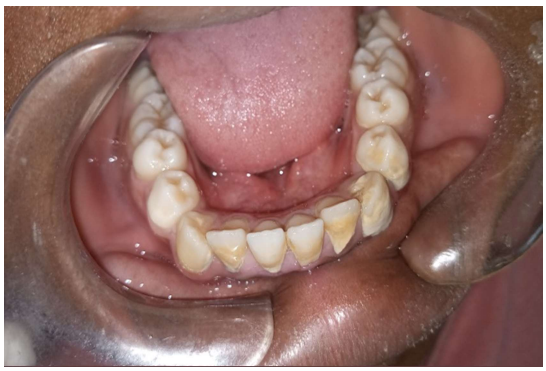

Figure 8: Intraoral clinical picture (mandibular arch

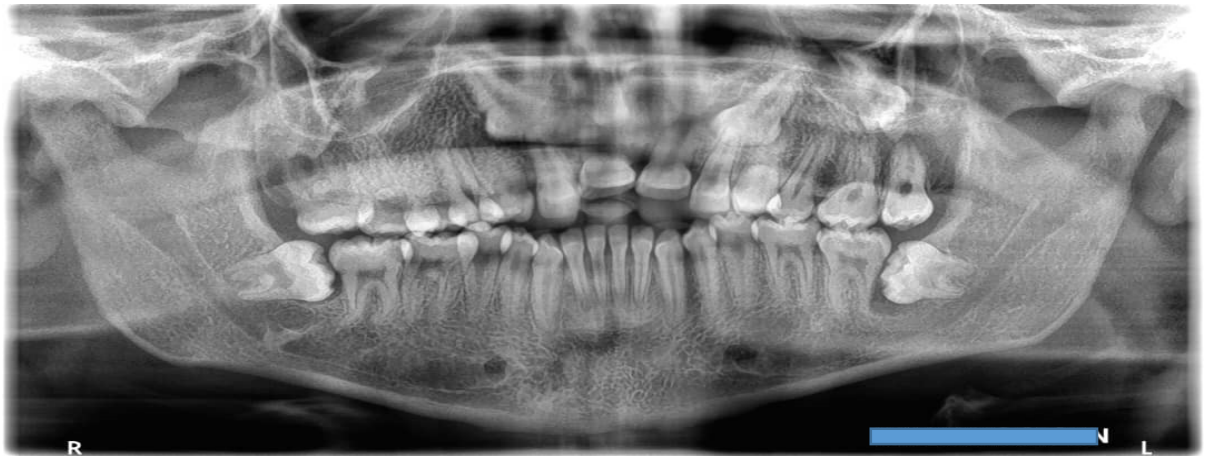

Figure 9: Orthopantomogram (OPG)

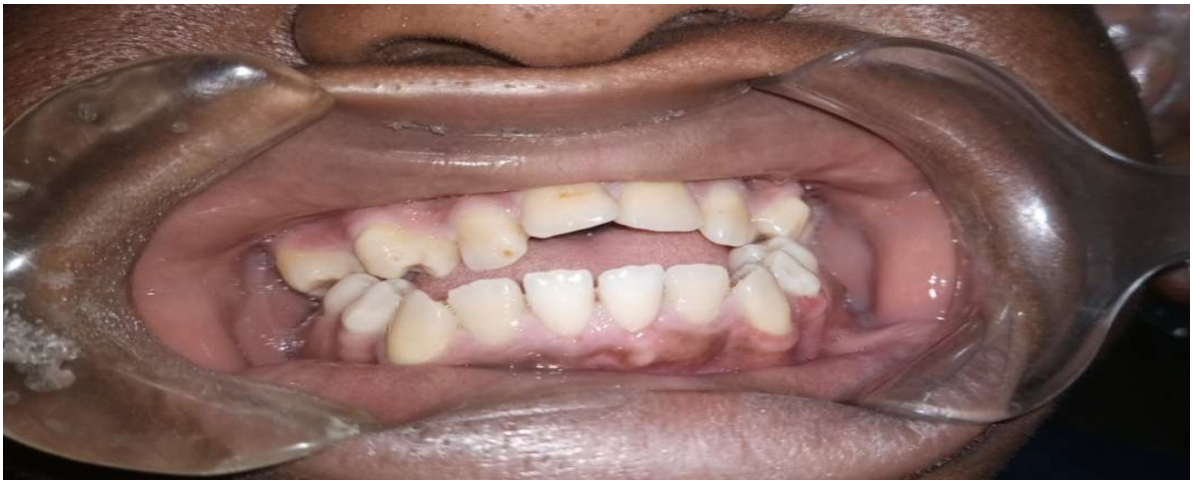

Figure 10: Intraoral clinical picture after scaling and polishing

DISCUSSION

A French paediatrician named Dr Eugene Charles Apert was the first to describe Apert syndrome in 1906.[1]Apert Syndrome is a very rare craniosynostosis syndrome that can be differentiated most times from other craniosynostosis syndromes such as Beare-Stevenson syndrome, Crouzon syndrome, Jackson-Weiss syndrome and Pfeiffer syndrome due to the presence of syndactyly.[5]

Apert syndrome affects males and females equally, and increased paternal age has been shown to be a risk factor, as seen in the case presented above, because the father was 51 years at his birth.[11,13,22] The midface in Apert syndrome is underdeveloped, and

retruded.[10,11,23]Underdeveloped midface causes shallow orbits, downward slant of the palpebral fissures, ocular proptosis, malocclusion and mandibular prognathism as seen in the clinical features of the case presentation above.[5,22]

Certain oral features such as impacted teeth, severe crowding, delayed eruption, posterior crossbite, class III molar relationship, midline deviation and anterior open bite are common in Apert syndrome and were also present in this case report.[11,24] The patient in this case also had the pathognomonic midline pseudo-cleft on the high-arched palate due to the bilateral swelling of the palatine process[23]. Kreiborg and Cohen[25] did a clinical study of patients with Apert syndrome and found the presence of pseudocleft or bifid uvula in 75% of their cases[25]. Cleft palate is also commonly seen in Apert syndrome.[1,5,8,13]

In individuals with Apert syndrome, intelligence ranges from normal intelligence to mild intellectual disability, as observed in the case presented above.[23] However, they are at increased risk for developing moderate to severe intellectual disability due to increased intracranial pressure in infancy and early childhood.[5,17] Early surgical intervention and comprehensive rehabilitation are important to prevent complications and social problems, thereby enhancing their quality of life.[5,26]

The patient was booked for an orthodontic assessment, referral letter was written to orthopaedic surgery for further surgical intervention for the syndactyl. However, there were defaults in keeping appointments. Several calls were made to remind them and encourage them to keep to their appointments, but the reasons for missed appointments were relocation outside Lagos and financial constraints.

CONCLUSION

A multidisciplinary team of health care professionals (Geneticists, Paediatricians, Orthopaedic surgeons, Neurosurgeons, Maxillofacial surgeons, Paediatric dentists, Orthodontists, Otolaryngologists, Speech pathologists, Audiologists, Nurses, Neuropsychologists) should be involved in giving early diagnosis and surgical intervention. Correction of defects in the craniofacial, dental, digit and other involved structures is needed to improve the quality of life of individuals with Apert syndrome.

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Conflict of interest

None declared

REFERENCES

1. Gazi SR, Manu PS, Vijayalakshmi S. Apert's Syndrome - A Rare Craniofacial Anomaly. South-East Asian J Case Rep Rev. 2014;3(1):657–67.
2. Chonka Z, Beall D, Jennings B, Wu D, Ly J, Wolff J. Apert's syndrome. Appl Radiol. 2004; 33:42–5.
3. Kilic I, Baykara Y, Semerci N, Ergin H, Lale N, Tufan S. Apert Syndrome. Turkish J Med Sci. 2004; 34:405–8.
4. Kana MA, Baduku TS, Baduku AS. A 37-year-old Nigerian woman with Apert syndrome – medical and psychosocial perspectives : a case report. 2018; 12: 126–130.
5. Wenger TL, Hing A V, Evans KN, et al. "Apert Syndrome." GeneReviews®, edited by Margaret P Adam et. al., University of Washington, Seattle, 30 May 2019.
6. Ornitz DM, Marie PJ. FGF signalling pathways in endochondral and intramembranous bone development and human genetic disease. Genes Dev. 2002; 16:1446–65.
7. Das S, Munshi A. Research advances in Apert syndrome. J Oral Biol Craniofacial Res [Internet]. 2018; 8:194–9. Available from: http://dx.doi.org/10.1016/j.jobcr.2017.05.006
8. Slaney SF, Oldridge M, Jane A, Hall CM, Poole MD, Morriss-kay GM. Differential Effects of FGFR2 Mutations Palate in Apert Syndrome Syndactyly and Cleft. Am J Hum Genet. 1996; 58:923–32.
9. Cohen MJ, Kreiborg S, Lammer E. Birth prevalence study of Apert syndrome. Am J Med Genet. 1992; 42:655–59.
10. Ofodile FA, Adeloye A. Acrocephalosyndactyly with hydrocephalus and dextrocardia in a Nigerian child. J Natl Med Assoc. 1982; 74:800–3.
11. Saberi BV, Shakoorpour A. Apert Syndrome : Report of a Case with Emphasis on Oral Manifestations. J Dent Tehran Univ Med Sci. 2011; 8:90–5.
12. Dixit S, Singh A, Gs M, S Desai R, Jaju P. Apert's Syndrome: Report of a New Case and its Management. Int J Clin Pediatr Dent. 2008; 1:48-53.
13. Bhatia P V, Patel PS, Jani Y V, Soni NC. Apert's syndrome : Report of a rare case. 2013; 17(2):294–7.

14. Khan S, Chatra L, Shenai P, Veena KM. Apert Syndrome : A Case Report. 2012; 5:203–6.
15. Batra P, Duggal R, Prakash H. Dentofacial characteristics in Apert syndrome: A case report. J Indian Soc Pedod Prev Dent. 2002; 20:118–23.
16. Pereira V, Sacher P, Pereira V SP, Ryan M, Hayward R. Dysphagia and nutrition problems in infants with Apert syndrome. Cleft Palate Craniofac J. 2009; 46:285–91.
17. Carinci F, Pezzetti F, Locci P, Becchetti E, Carls F, Avantaggiato A, et al. Apert and Crouzon syndromes: Clinical findings, genes and extracellular matrix. J Craniofac Surg. 2005; 16:361–8.
18. Quintero-Rivera F, Robson C, Reiss R et al. Apert syndrome: what prenatal radiographic findings should prompt its consideration? Prenat Diagn. 2006; 10:966–72.
19. Quintero-Rivera F, Robson C, Reiss R et al. Intracranial anomalies detected by imaging studies in 30 patients with Apert syndrome. Am J Med Genet A. 2006; 140:1337–8.
20. Athanasiadis A, Zafrakas M, Polychronou P et al. Apert syndrome: the current role of prenatal ultrasound and genetic analysis in diagnosis and counselling. Fetal Diagn Ther. 2008; 24:495–8.
21. Ramond F, Whittaker J, Jenkins L, Lench N, Chitty L. Molecular prenatal diagnosis: the impact of modern technologies. Prenat Diagn. 2010; 30:674–81.
22. Pius S, Ibrahim HA, Bello M, Mbaya K, Ambe JP. Apert Syndrome : A Case Report and Review of Literature. Open Journal of Pediatrics. 2016; 06:175-184
23. Jose B, Emmatty TB, Methippara JJ, Kumar K, Thampi NM. Apert Syndrome : An Insight Into Dentofacial Features. Cureus. 2021; 13(9):1–13.
24. Ferraro N. Dental, orthodontic and oral/maxillofacial evaluation and treatment in Apert syndrome. Clin Plast Surg. 1991; 18:291–307.
25. Kreiborg S, Cohen MJ. The oral manifestations of Apert syndrome. J Craniofac Genet Dev Biol. 1992; 12:41-48.
26. Maliepaard M, Mathijssen I, Oosterlaan J, Okkerse J. Intellectual, behavioural, and emotional functioning in children with syndromic craniosynostosis. Pediatrics. 2014; 133:1608–1615.