Dentofacial Characteristics of a 12 year old child suffering from GAPO syndrome –A Rare Case Report

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ABSTRACT
Growth retardation, alopecia, pseudoanodontia and optic atrophy are all characteristics of the GAPO syndrome, a group of several congenital defects involving connective tissue. This case reports a 12 year old female patient with GAPO syndrome highlighting the dentofacial characteristics of this condition
Keywords: Alopecia, Autosomal recessive disorder, GAPO, Growth retardation, Pseudoanodontia

INTRODUCTION

Growth retardation (G), Alopecia (A), Pseudoanodontia (P) and Optic atrophy (O) (GAPO) syndrome is an autosomal recessive disease. [1] Since its original description in 1947, approximately 45 instances have been recognised globally. It is a very rare hereditary condition. [2]

Andersen and Pindborg were the first to describe this disorder in 1947[3], and Tripton and Gorlin gave it the label GAPO syndrome in 1984. [4] Clinically those who suffer from this syndrome have a geriatric appearance. Patients may experience various signs in addition to the traditional ones, such as endocrine, opthalmic, vascular, and CNS manifestations. [2]

GAPO syndrome is thought to be caused by homozygous nonsense or splicing mutations in the anthrax toxin receptor 1 (ANTXR1) gene, formerly known as tumour endothelial marker 8 (TEM8), which result in a truncated isoform of the ANTXR1 protein. However, the candidate gene is still unknown.[5,6] They have a shorter life expectancy and typically pass away in their third or fourth decade of life from widespread interstitial atherosclerosis and fibrosis.[5] The objective of this report is to present the dentofacial characteristics of GAPO syndrome in a 12 year old child.

CASE REPORT

A 12-year-old female child reported to the Department of Pediatric and Preventive Dentistry, with a chief complaint of pain in lower right back tooth region since 1 week. Prior to the onset of the current condition, the child appeared healthy up to age 2, at which point the parents noticed a slower growth

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How to cite this article: Yadav A, Kumari R, Khan Z, Mishra P, Chaudhary R. Dentofacial Characteristics of a 12 year old child suffering from GAPO syndrome –A Rare Case Report. TMU J Dent 2024; 11(1): 22-25.
Submitted: 22 Aug 2023 Revised and accepted: 23 Aug 2023
Doi: https://doi.org/10.58358/tmujd.ped11103c

spurt. Parents also noticed a progressive decline in eye sight and no teeth erupting. During her pregnancy, the mother did not experience any prenatal, antenatal, or postnatal difficulties, and her birth was uneventful. The child was already diagnosed with GAPO syndrome from the 2nd year of her life based on past medical history and reports.

The patient's general physical examination in question has an oddly geriatric appearance. Due to postnatal growth retardation, the child had a small stature with a weight of 28 kg and a height of 120 cm, which was below the usual parameter. Hyperextensible joints and osseous anomalies, such as short-length long bones, were discovered during an examination of the hand and leg joints. Legs and hands had small fingers, and koilonychia could be seen in the nails. Growth retardation and developmental milestone delays were seen in the child. Examination of the face and head-neck region revealed hypertelorism, a prominent scalp, and a high, bossy forehead while eyebrows and eyelids showed total alopecia, the scalp hair showed complete loss. (Figure 1)

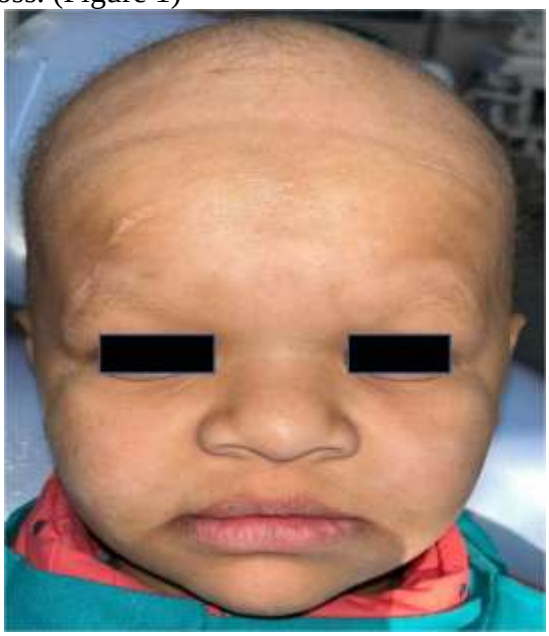

Figure 1- Facial profile of the patient

Low-set ears and an early sign of ageing, including superfluous hyperelastic skin and odd creases, were both present. Wide nostrils, a low nasal bridge, and a lengthy philtrum were present. The bottom lip was thick and everted, but the upper lip was normal. Both of the child's eyes exhibited gradually worsening near- and farsightedness. During performing an intraoral examination, it was discovered that the patient had pseudoanodontia, pseudomacroglossia, and micrognathia. The gingiva had a light, pinkish hue. (Figure 2)

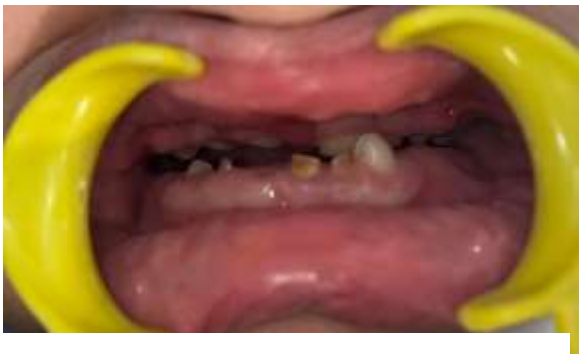

Figure 2- Intra- oral view showing thick alveolar ridges with partially erupted teeth and retruded maxillae

Primary teeth that were visible in the oral cavity were only partially erupted. The patient's widely spaced dentition was the more pronounced feature. The upper and lower alveolar ridges have thickened abnormally. The following teeth were present and partially erupted: 54 64 71 72 73 74 82. (Figures 3 and 4).

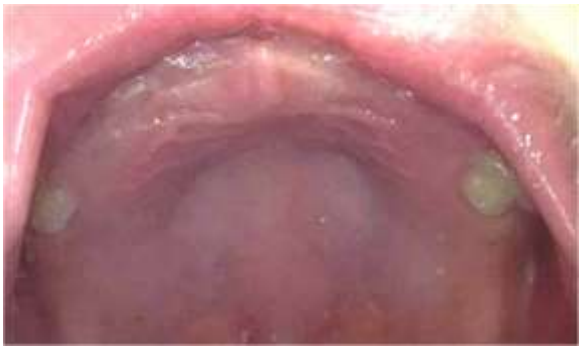

Figure -3- Occlusal view of maxillary arch showing partial anodontia

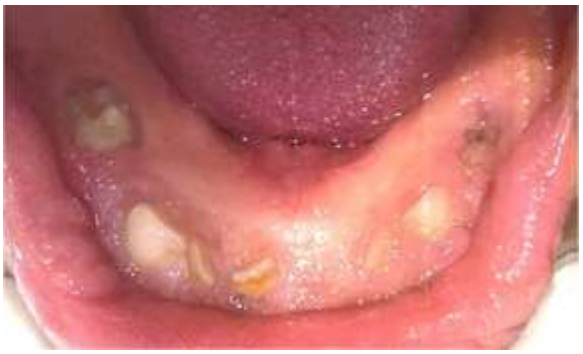

Figure-4- Occlusal view of mandibular arch showing partial anodontia

Nevertheless, stains and calculus were visible despite the absence of caries. Orthopantomogram of the patient revealed the presence of several impacted primary and permanent teeth. (Figure-5)

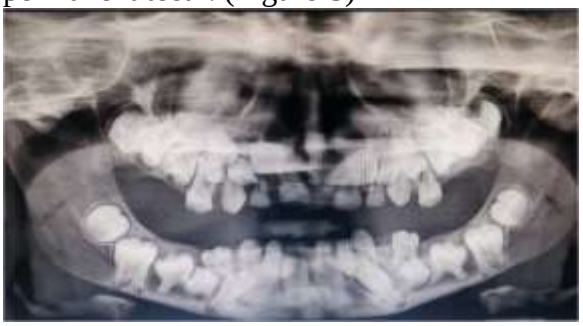

Figure-5 – Orthopantomogram of the patient showing multiple impacted primary and permanent teeth

For this condition, there is no effective cure. The majority of management strategies involve symptomatic therapy of the several health issues together with ophthalmologic, neurologic, and auditory surveillance. The pain was being caused by the eruption of the lower right molar because there were no signs of oral or dental pathology. So the case was managed conservatively by prescribing topical anaesthetic gel for the relief from pain and suggested her to undergo routine follow-up to evaluate the progress of development and eruption of permanent teeth. If neurological abnormalities do not predominate in the early years of infancy, the prognosis of patients with GAPO syndrome can be deemed fair due to a shorter lifespan (until their fourth to sixth decade of life).

DISCUSSION

Although the craniomaxillofacial area and pseudoanodontia have consistently been identified as a defining feature of this syndrome, there have not been many reports of it in the dental literature. The patient in this report displayed the typical anomalies and symptoms of GAPO syndrome. Her eyelids, brows, and hair were all affected by alopecia as shown in figure 1.

The optic nerve atrophy that results from nerve constriction in GAPO disease patients is thought to be the cause of their decreased visual acuity. [7] Clinical signs include early craniosynostosis, frontal bossing, early fusion of calvarial sutures, and epiphyseal plates are thought to be caused by an abundance of homogenous, amorphous hyaline material that is present in all organs, interstitial spaces, and serosal membranes. Premature fusing of the growing bone ends may result from the buildup of excess hyaline material. In light of this, it may be said that in these patients, dwarfism, short height, and growth retardation are causally related. Interesting dental discoveries were found in this patient. Although the teeth were there,

they did not emerge properly, leading to pseudoanodontia and an increase in the volume of the ridge bone leading to pseudoanodontia. [8-10]

The inability of primary and permanent teeth to erupt in the oral cavity, which can result in partial or total anodontia, is linked to an overabundance of extracellular connective tissue matrix, which builds up over the course of life and obstructs all tissues' and organs' normal functions. [10] We were particularly struck by the patient's unusually thick buccal and lingual frenii.

CONCLUSION

Though the GAPO syndrome has received extensive research, there are still some scientifically unexplored aspects of the syndrome. Yet, in order to help such individuals live their lives with the least amount of suffering, rigorous assessment and appropriate diagnosis are required.

REFERENCES

1.Wajntal A, Koifmann CP, Mendonca BB, Epps Quaglia D, Sotto MN, Rati PBU et al. GAPO syndrome (McKusick 23074).A connective tissue disorder: Report on two effected sibs and on the pathologic findings in the older. Am J Med Genet 1990; 37: 213-23.

2. Nanda A, Al-Ateeqi WA, Al-Khawari MA, Alsaleh QA, Anim JT. GAPO syndrome: A report of two siblings and a review of literature. Pediatr Dermatol 2010; 27: 156-61.

3. Andersen TH, Pindborg JJ. El tifaelde at total "pseudoanodonti" inforbindelse med kraniedeformitet, dvaergvaekstogektodermaldisplasi. Odontol Tilster 1947; 55: 484-493.

4. Tripton RE, Gorlin RJ. Growth retardation, alopecia, pseudoanodontia, and optic atrophy - The GAPO syndrome. Am J Med Genet 1984; 19: 209-216.

5. Salas Alanís JC, Scott CA, Fajardo Ramírez OR, Duran C, Moreno Treviño MG, Kelsell DP. New ANTXR1 Gene mutation for GAPO syndrome: A case report. Mol Syndromol 2016; 7: 160-3.

6. Bayram Y, Pehlivan D, Karaca E, Gambin T, Jhangiani SN, Erdin S, et al. Whole exome sequencing identifies three novel mutations in ANTXR1 in families with GAPO syndrome. Am J Med Genet A 2014; 164A: 2328-34.

7. Lei S, Iyengar S, Shan L, Cherwek DH, Murthy S, Wong AM. GAPO syndrome: A case associated with bilateral interstitial keratitis and hypothyroidism. Clin Dysmorphol 2010; 19: 79-81.

8. Manouvrier Hanu S, Largilliere C, Benalioua M, FarriauxJP, Fontaine G. The GAPO syndrome. Am J Med Genet 1987; 26: 683- 8.

9. Suri L, Gagari E, Vastardis H. Delayed tooth eruption: Pathogenesis, diagnosis, and treatment. A literature review. Am J Orthod Dentofacial Orthop 2004; 126: 432- 45.

10. Dahake PT, Kale YJ, Dadpe MV, Kendre SB. A rare Indian case of GAPO syndrome with dental and other findings. Int J Pedod Rehab 2020; 5: 25-8.