World Journal of Advanced Research and Reviews
eISSN: 2581-9615 CODEN (USA): WJARAI
Cross Ref DOI: 10.30574/wjarr
Journal homepage: https://wjarr.com/

(CASE REPORT)

Check for updates

A Case Report of Apert Syndrome in a 13-Year-Old Girl Presenting with Dental Pain

Bayar A Qasim [1], Rende SA Kochary [1], Halder J Abozait [2,*] and Anees M Mudhir [3]

[1] *Department of Medicine, College of Medicine, University of Duhok, Duhok, Kurdistan Region, Iraq.*
[2] *Department of Biomedical Sciences, College of Medicine, University of Zakho, Zakho Independent Administration, Kurdistan Region, Iraq*
[3] *Department of Prevention, Orthodontic & Pedodontics, College of Dentistry, University of Duhok, Duhok, Kurdistan Region, Iraq.*

World Journal of Advanced Research and Reviews, 2025, 28(01), 1631-1635

Publication history: Received on 30 August 2025; revised on 15 October 2025; accepted on 18 October 2025

Article DOI: https://doi.org/10.30574/wjarr.2025.28.1.3519

Abstract

Introduction: Apert syndrome is a rare congenital craniosynostosis syndrome caused by mutations in the FGFR2 gene, with an incidence of 1 in 65,000 to 200,000 live births. It is characterized by premature cranial suture fusion, midfacial hypoplasia, and syndactyly of the hands and feet. In addition to craniofacial and skeletal anomalies, patients often present with dental, airway, and ophthalmological complications requiring long-term multidisciplinary management.

Case Presentation: We describe a 13-year-old female who presented with progressively worsening dental pain over one month, associated with temporal headaches and difficulty chewing. Examination revealed brachycephaly, midfacial hypoplasia, exophthalmos, and significant oral findings including high-arched palate, malocclusion, and dental crowding. Bilateral surgical scars from previous syndactyly correction were present, with residual deformities of the hands and feet. Laboratory evaluation demonstrated normal thyroid and gonadotropin profiles. Pelvic ultrasound showed normal uterus and ovaries with multiple immature follicles. She was referred for orthodontic assessment and multidisciplinary follow-up.

Conclusion: Apert syndrome is a multisystem disorder that requires coordinated, lifelong management. This case emphasizes the importance of early and continuous dental surveillance, endocrinological monitoring, and psychosocial support in optimizing outcomes for adolescents with Apert syndrome.

Keywords: Apert syndrome; Craniosynostosis; Syndactyly; Dental crowding; Endocrinology

1. Introduction

Apert syndrome, or acrocephalosyndactyly type I, is a rare congenital disorder within the group of syndromic craniosynostoses. It has an estimated prevalence of 1 in 65,000 to 200,000 live births and demonstrates no ethnic or geographic predilection (1). The syndrome is caused by mutations in the fibroblast growth factor receptor 2 (*FGFR2*) gene, which lead to abnormal signaling in cranial and limb development (2). Inheritance is autosomal dominant; however, most cases arise sporadically due to de novo mutations, often associated with advanced paternal age (3). Clinically, Apert syndrome is characterized by the triad of premature cranial suture fusion (craniosynostosis), midfacial hypoplasia, and symmetrical syndactyly of the hands and feet (4). Craniosynostosis typically involves the coronal sutures, resulting in brachycephaly, turribrachycephaly, or acrocephaly. Midfacial hypoplasia contributes to shallow orbits, exophthalmos, and a depressed nasal bridge. The shallow orbits predispose to ocular complications, including keratitis and vision impairment, due to proptosis and incomplete eyelid closure. Dental manifestations are also common and may include maxillary hypoplasia, cleft palate, high-arched palate, dental crowding, and delayed eruption (5). These

* Corresponding author: Halder J Abozait

features contribute to feeding difficulties, recurrent infections, and reduced oral health. Early surgical intervention to correct cranial and limb anomalies has improved outcomes; nevertheless, patients often require lifelong multidisciplinary management given the multisystem nature of Apert syndrome (6). In this report, we present the case of a 13-year-old female with Apert syndrome who developed significant dental pain secondary to her craniofacial abnormalities. This case underscores the importance of long-term dental surveillance and illustrates the interplay between craniofacial structure, endocrine function, and psychosocial well-being in adolescents with this syndrome.

2. Case presentation

We describe a case of a 13-year-old female who presented with a one-month history of gradually worsening dental pain associated with temporal headaches. The pain was aggravated by chewing hard or coarse foods but was not associated with fever, weight loss, or generalized fatigue. Her appetite was preserved, although oral discomfort limited intake. She also described nasal obstruction, dry eyes with irritation, incomplete eye closure during sleep, difficulty swallowing, and a reduction in voice pitch. Additionally, she reported mild shortness of breath with moderate exertion but denied cough, chest pain, or palpitations. She has experienced irritability and increased emotional lability over the month before consultation.

Her past medical history revealed complications beginning at birth. She was delivered at term by vaginal delivery after a pregnancy complicated by maternal preeclampsia and polyhydramnios. The neonate required two days of NICU admission due to poor cry at birth. Apert syndrome was diagnosed clinically, and her neonatal course was complicated by feeding difficulties and suspected cerebrospinal fluid rhinorrhea, which resolved with conservative management. She experienced recurrent ear infections and upper respiratory tract infections throughout childhood. At the age of three, she underwent corrective hand surgery for syndactyly of the right hand, followed by surgery on the left hand six months later. Both procedures were uneventful.

Developmental history showed delays: she sat independently at one year, walked at two years, and spoke her first words at 18 months. Despite initial developmental lag, she was socially interactive and integrated into schooling at age seven, though she repeated fourth grade due to learning difficulties. Her academic performance remains average. There is no family history of Apert syndrome or similar conditions. Her family background is notable for hypertension and type 2 diabetes in relatives and poor socioeconomic circumstances.

On examination, the patient had brachycephaly with a flat occiput, prominent forehead, and midfacial retrusion. Exophthalmos was evident. Oral examination revealed dental crowding with malocclusion, underbite, and high-arched palate (Figure 1). Oral hygiene was poor, and she was noted to be a mouth breather. Her hands exhibited bilateral surgical scars from syndactyly release, and she was unable to fully form a fist (Figure 2). Residual syndactyly was observed in the feet, with flattening of the arches, particularly on the left (Figure 3). Chest and abdominal examinations were unremarkable.

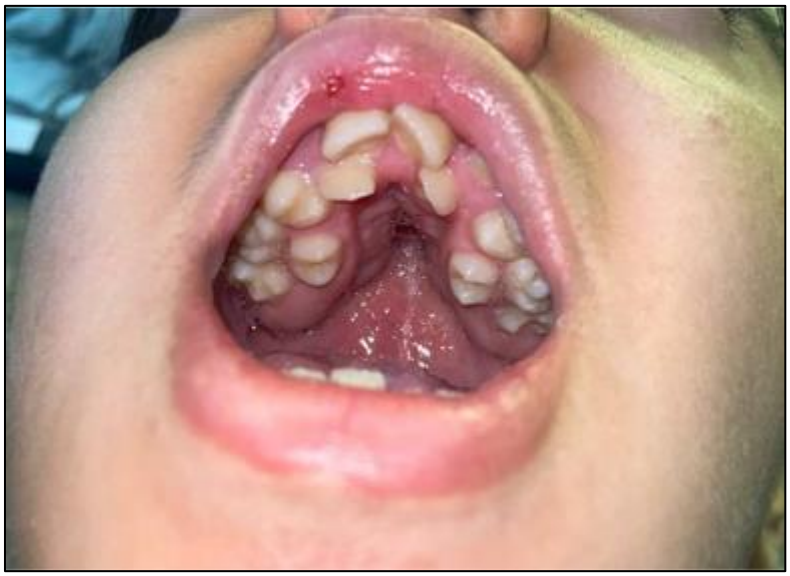

Figure 1 Dental crowding with high-arched palate in a 13-year-old girl with Apert syndrome

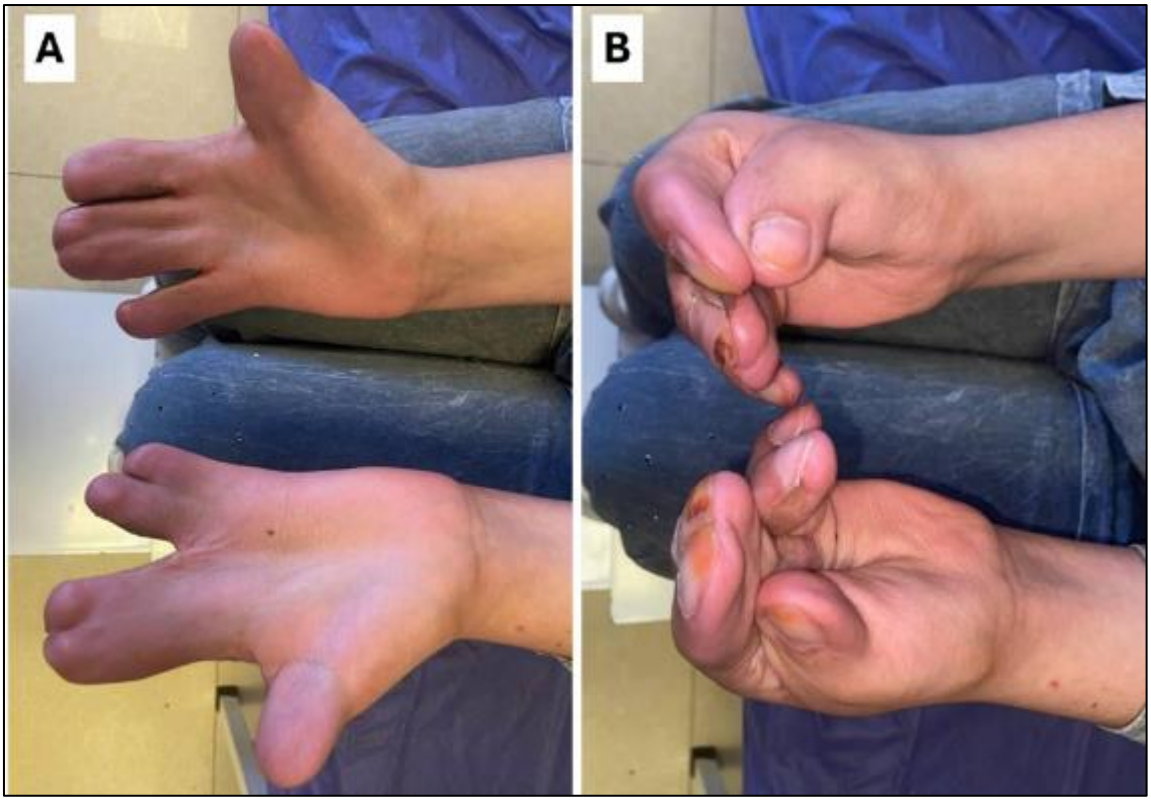

Figure 2 Bilateral upper extremity syndactyly with inability to close hands in a 13-year-old girl with Apert syndrome

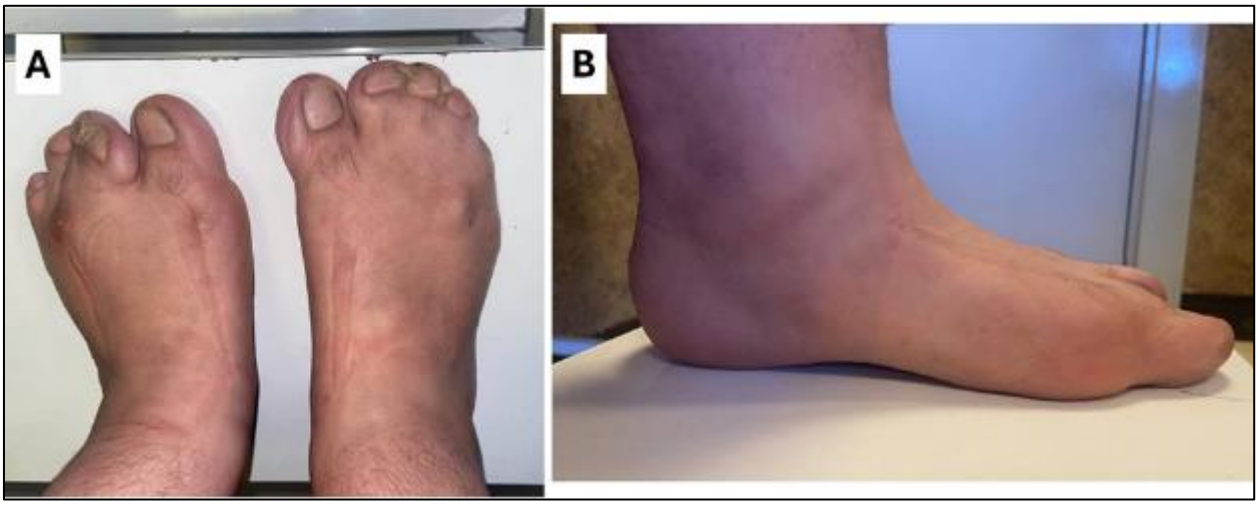

Figure 3 Bilateral lower extremity syndactyly with flat foot in a 13-year-old girl with Apert syndrome

Laboratory investigations performed revealed a TSH of 0.52 μIU/mL (Reference range (RR): 0.36-5.8) and free T4 of 21.2 pmol/L (RR: 12-22). Pituitary–gonadal axis hormones revealed a serum LH of 4.29 mIU/mL (RR: 0.5-9) and FSH 4.06 mIU/mL (RR: 0.5-9) but estradiol was relatively low at 28.5 pg/mL (RR follicular-phase: 87.7-173). Notably, her evening cortisol was elevated at 668.9 nmol/L (RR: 64-327), with an ACTH of 40.6 pgmL (RR: 7.2-63.3), warranting further endocrinological evaluation, but the overnight dexamethasone suppression test was negative. Glycemic control was normal with HbA1c at 5%. Pelvic ultrasonography revealed a normal uterus and ovaries with multiple immature follicles but no cysts or masses. She was referred to an orthodontist for further evaluation and possible interventions.

3. Discussion

Apert syndrome is a classic example of a rare disorder in which craniofacial anomalies extend far beyond cosmetic concerns to affect multiple aspects of health. Our patient demonstrated the characteristic craniofacial features, including brachycephaly, midfacial hypoplasia, and exophthalmos, along with the hallmark syndactyly of the hands and feet. However, her presenting complaint of dental pain reflects one of the less often highlighted but clinically significant aspects of Apert syndrome. Dental abnormalities are common in Apert syndrome and are often underestimated in clinical care (7). The presence of a high-arched or cleft palate, malocclusion, and dental crowding predisposes to caries, periodontal disease, and recurrent infections (8). In our patient, oral pain interfered with nutrition and daily activities, reinforcing the role of dentists and orthodontists as integral members of the multidisciplinary care team.

Endocrine findings in this case add another dimension. The patient's gonadotropin levels were within normal limits, but estradiol was lower than expected for her age, suggesting possible pubertal delay. Reports on the endocrinological aspects of Apert syndrome are limited, but some studies have suggested that craniosynostosis syndromes can be associated with hypothalamic–pituitary dysfunction, particularly when midline craniofacial defects are present (9). This highlights the importance of long-term endocrine monitoring in syndromic craniosynostoses, especially during adolescence, when pubertal development and psychosocial adaptation are critical.

Psychological and social aspects are equally important. Adolescents with Apert syndrome frequently struggle with self-esteem issues due to facial differences and functional limitations, such as residual syndactyly affecting fine motor skills (10). Our patient displayed emotional lability and frustration, which may reflect the psychosocial burden of her condition in addition to the physical symptoms. Early integration of psychological support and social services is crucial to address these issues, reduce the risk of isolation, and promote adaptive coping strategies.

This case also illustrates the importance of early and continued multidisciplinary follow-up (11). In infancy and early childhood, craniofacial surgeons, neurosurgeons, and pediatricians play a dominant role in preventing complications of raised intracranial pressure and airway obstruction. As the child grows, dental specialists, orthodontists, ophthalmologists, and endocrinologists become increasingly important in managing the evolving complications of the syndrome. Our patient's delayed referral for dental intervention underscores a gap in continuity of care, which is common in resource-limited settings.

4. Conclusion

This case highlights the complexity of Apert syndrome, particularly the burden of dental and craniofacial manifestations in adolescence. Our patient presented with dental pain, but her history and clinical features underscore the need for comprehensive, multidisciplinary care. Regular dental follow-up, endocrinological evaluation, and psychosocial support remain central to improving quality of life in individuals with Apert syndrome.

Compliance with ethical standards

Disclosure of conflict of interest

The authors declare no conflicts of interest.

Statement of ethical approval

The ethics committee of the College of Medicine, University of Duhok approved the publication of this case report.

Statement of informed consent

Informed consent was obtained from the parents or legal guardians of the pediatric patient to permit the release of their anonymized clinical information and their participation. To protect patient confidentiality, all personal information was removed. The Nature of this study is retrospective analysis of clinical data, with no experimental interventions exceeding standard medical care.

References

[1] Karsonovich T, Patel BC. Apert Syndrome. StatPearls. Treasure Island (FL) ineligible companies. Disclosure: Bhupendra Patel declares no relevant financial relationships with ineligible companies.: StatPearls Publishing Copyright © 2025, StatPearls Publishing LLC.; 2025.

[2] Kumari K, Saleh I, Taslim S, Ahmad S, Hussain I, Munir Z, et al. Unraveling the Complexity of Apert Syndrome: Genetics, Clinical Insights, and Future Frontiers. Cureus. 2023;15(10):e47281. doi: 10.7759/cureus.47281.

[3] Glaser RL, Broman KW, Schulman RL, Eskenazi B, Wyrobek AJ, Jabs EW. The paternal-age effect in Apert syndrome is due, in part, to the increased frequency of mutations in sperm. American journal of human genetics. 2003;73(4):939-47. Epub 2003/08/06. doi: 10.1086/378419. PubMed PMID: 12900791; PubMed Central PMCID: PMCPMC1180614.

[4] Kana MA, Baduku TS, Bello-Manga H, Baduku AS. A 37-year-old Nigerian woman with Apert syndrome - medical and psychosocial perspectives: a case report. J Med Case Rep. 2018;12(1):126. Epub 2018/05/14. doi: 10.1186/s13256-018-1638-7. PubMed PMID: 29753329; PubMed Central PMCID: PMCPMC5949149.

[5] López-Estudillo AS, Rosales-Bérber MA, Ruiz-Rodríguez S, Pozos-Guillén A, Noyola-Frías M, Garrocho-Rangel A. Dental approach for Apert syndrome in children: a systematic review. Medicina oral, patologia oral y cirugia bucal. 2017;22(6):e660-e8. Epub 2017/10/21. doi: 10.4317/medoral.21628. PubMed PMID: 29053644; PubMed Central PMCID: PMCPMC5813983 potential financial, academic, or personal interest inherent to this manuscript.

[6] Fowler P, Hallang S, Snape L. Apert syndrome: an informative long-term dentofacial outcome. BMJ case reports. 2022;15(3). Epub 2022/03/04. doi: 10.1136/bcr-2021-245224. PubMed PMID: 35236672; PubMed Central PMCID: PMCPMC8895904.

[7] Dalben Gda S, das Neves LT, Gomide MR. Oral findings in patients with Apert syndrome. Journal of applied oral science : revista FOB. 2006;14(6):465-9. Epub 2006/12/01. doi: 10.1590/s1678-77572006000600014. PubMed PMID: 19089249; PubMed Central PMCID: PMCPMC4327301.

[8] Mustafa D, Lucas VS, Junod P, Evans R, Mason C, Roberts GJ. The dental health and caries-related microflora in children with craniosynostosis. The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association. 2001;38(6):629-35. Epub 2001/10/30. doi: 10.1597/1545-1569_2001_038_0629_tdhacr_2.0.co_2. PubMed PMID: 11681997.

[9] Guilmin-Crépon S, Garel C, Baumann C, Brémond-Gignac D, Bailleul-Forestier I, Magnier S, et al. High Proportion of Pituitary Abnormalities and Other Congenital Defects in Children with Congenital Nasal Pyriform Aperture Stenosis. Pediatric Research. 2006;60(4):478-84. doi: 10.1203/01.pdr.0000238380.03683.cb.

[10] Netherton J, Horton J, Stock NM, Shaw R, Noons P, Evans MJ. Psychological Adjustment in Apert Syndrome: Parent and Young Person Perspectives. The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association. 2023;60(4):461-73. Epub 2021/12/31. doi: 10.1177/10556656211069817. PubMed PMID: 34967688.

[11] Varlas VN, Epistatu D, Varlas RG. Emphasis on Early Prenatal Diagnosis and Perinatal Outcomes Analysis of Apert Syndrome. Diagnostics (Basel, Switzerland). 2024;14(14). Epub 2024/07/27. doi: 10.3390/diagnostics14141480. PubMed PMID: 39061616; PubMed Central PMCID: PMCPMC11276282.