Awareness, Knowledge and Presumption About Orofacial Syndromes among Dental Students

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Abstract

Orofacial syndromes are complex conditions where the concurrence of oral and systemic findings often serves as a critical diagnostic indicator. For dental professionals, the transition from theoretical awareness to clinical **"presumption"** the ability to anticipate secondary complications and long-term management needs is vital for early diagnosis and multidisciplinary care. This study aimed to evaluate the levels of awareness, scientific knowledge, and management presumption regarding orofacial syndromes among undergraduate dental students.

Keywords

Orofacial Syndromes; Dental Education; Clinical Presumption; Ectodermal Dysplasia; Pierre Robin Sequence; Diagnostic Awareness; Oral Manifestations; Management Gap; Chi-square Analysis.

Introduction:

The term “syndrome” is derived from the Greek word syndromos, meaning “running together,” and refers to a recognizable complex of symptoms and physical findings that characterize a specific condition[1]. In the field of oral medicine, many systemic syndromes present with distinct orofacial manifestations, ranging from skeletal disharmonies to unique dental morphologies[2]. For the dental professional, identifying these signs is not merely an academic exercise; it is a critical diagnostic responsibility, as the mouth often serves as a “sentinel” window to underlying systemic health[3].

Craniofacial and orofacial syndromes encompass a wide clinical spectrum. Conditions like Treacher Collins Syndrome and Apert Syndrome involve skeletal malformations such as midfacial hypoplasia, which leads to predictable occlusal challenges like Class III malocclusion[4]. Other conditions, notably Ectodermal Dysplasia, are identified by pathognomonic dental markers, including hypodontia and conical-shaped teeth[5]. Because these conditions are often multi-systemic—affecting vital functions such as hearing, breathing, and nutrition—the dental surgeon must be prepared to act as part of an interdisciplinary medical team[6].

The diagnostic intuition of a dentist can be life-saving. For example, the recognition of multiple jaw osteomas may be the primary clinical indicator for Gardner Syndrome, a condition that carries a nearly 100% risk of intestinal malignancy if not addressed[7]. Similarly, identifying early-onset periodontitis in Papillon-Lefèvre Syndrome or mucosal changes in Dyskeratosis Congenita is vital for preventing severe systemic morbidity[8].

Despite their significance, these conditions are often categorized as "rare diseases," which can lead to significant gaps in the presumption the clinical ability to anticipate long-term complications among undergraduate dental students[9]. Educational research indicates that while students may have a high baseline awareness of general terms, their technical knowledge of treatment modalities and rare syndrome complications remains inconsistent[10].

This study evaluates the awareness, scientific knowledge, and clinical presumption of orofacial syndromes among dental students. By identifying specific areas of deficiency, this research seeks to underscore the necessity of integrating comprehensive syndromic education into the dental curriculum to ensure holistic and proactive patient care.

Materials and Methods

A descriptive, cross-sectional observational study was conducted among 100 dental students to evaluate the levels of awareness, scientific knowledge, and management presumption regarding orofacial syndromes among undergraduate dental students. A validated, self-administered questionnaire consisting of 16 multiple-choice questions (MCQs) was used. The survey covered three domains: Awareness,Scientific Knowledge,Clinical Presumption.

Statistical Analysis:

To evaluate the participants' responses, a Chi-Square Goodness-of-Fit Test was performed for each of the 16 items. The test compared the observed frequency of responses against a theoretical uniform distribution. A p-value < 0.05 was considered statistically significant.The significant deviation from a uniform distribution ($p < 0.05$) confirms that the students' choices were influenced by their existing level of knowledge, clinical reasoning, or common misconceptions. This adds statistical weight to the identified "knowledge gaps," as the incorrect answers were also chosen in a non-random, clustered manner

Results

The overall mean score across all domains was 59.6\%. The Chi-Square test revealed that response distributions for all 16 questions were highly significant ($p < 0.001$), indicating that choices were driven by existing knowledge or specific misconceptions rather than guessing.

Awareness (68.3\%): 83\% correctly defined "syndrome" . 62\% correctly linked Apert syndrome's fused fingers to oral hygiene challenges .

Knowledge (56.6\%): Students excelled in identifying Treacher Collins hearing loss (75\%) and Gorlin-Goltz cysts (63\%). However, only 47\% identified the hallmark of Ectodermal Dysplasia .

Presumption (53.7\%): This was the weakest domain. While 72\% knew immediate infant care for clefting , only 41\% chose correct prosthetic management for ED , and 48\% identified the risk of hypoxia in Pierre Robin Sequence .

Q1. Medical context of "syndrome" ($p < 0.001$): Shows high baseline awareness, indicating students have a strong conceptual foundation of the multi-systemic nature of syndromes.

Q2. Papillon-Lefèvre Syndrome features ($p < 0.001$): While half linked skin issues with periodontitis, significant confusion remains regarding other similar dermatological-oral conditions.

Q3. Hearing loss in Treacher Collins Syndrome ($p < 0.001$): Demonstrates a good understanding of conductive hearing loss resulting from craniofacial anomalies.

Q4. Hallmark sign of Ectodermal Dysplasia (ED) ($p < 0.001$): Less than half identified hypodontia, suggesting a need for better education on primary markers for ED.

Q5. Anterior dentition in ED ($p < 0.001$): Students recognize shape anomalies but struggle with precise technical terminology like "conical" or "peg-shaped" (only 46% correct).

Q6. Treatment consideration for ED ($p < 0.001$): 43% incorrectly chose "orthodontics only," failing to recognize the necessity of prosthetic rehabilitation for missing teeth.

Q7. Midfacial hypoplasia in Apert Syndrome ($p < 0.001$): Strong recognition of how this leads to Class III malocclusion.

Q8. SCC risk in syndromes with white patches ($p < 0.001$): Less than half identified the high risk of oral squamous cell carcinoma, highlighting a gap in awareness of pre-malignant potential.

Q9. Gardner Syndrome features ($p < 0.001$): Most students successfully recognized the classic triad of jaw osteomas and intestinal polyps.

Q10. Cysts in Gorlin-Goltz Syndrome ($p < 0.001$): A majority correctly identified Odontogenic Keratocysts (OKCs) as the characteristic lesion.

Q11. Classic triad of Pierre Robin Sequence ($p < 0.001$): Shows a good understanding of the anatomical sequence (micrognathia, glossoptosis, and cleft palate).

Q12. Long-term complication of Pierre Robin Sequence ($p < 0.001$): Only 48% identified the risk of hypoxic injury, while many focused on less serious issues.

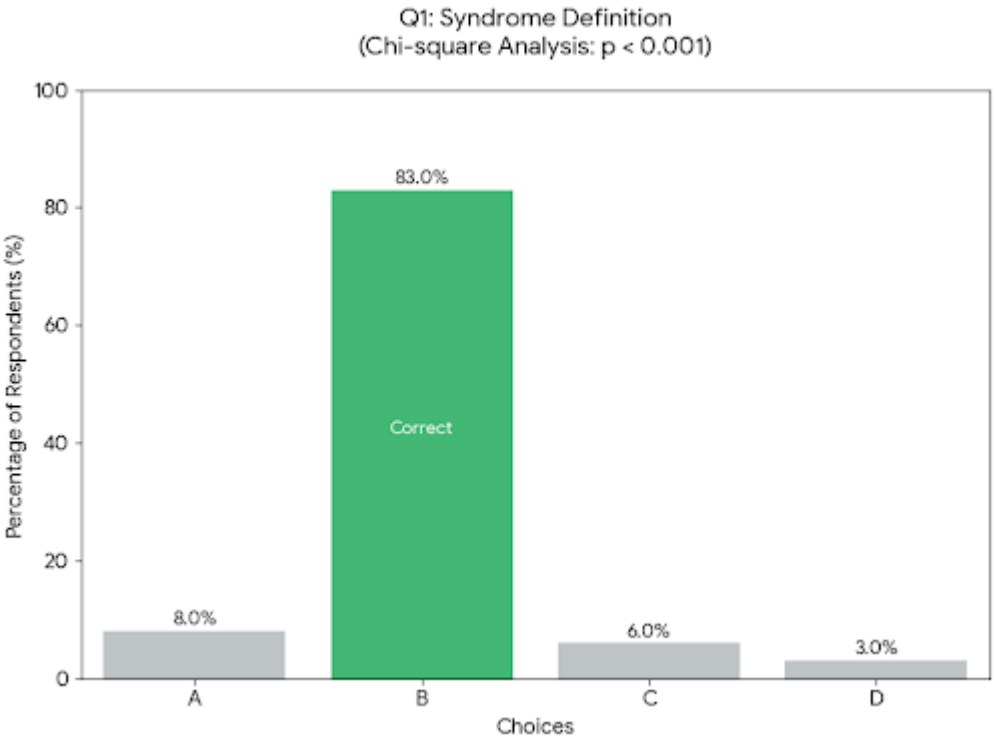
Q13. Primary concern in orofacial clefting at birth ($p < 0.001$): Excellent recognition of immediate feeding and aspiration risks in infants.

Q14. Identification of rare syndromes ($p < 0.001$): Under 50% could identify rare syndromes based on specific eye and finger anomalies (e.g., Oculodentodigital dysplasia)

Q15. Role of dental professional in identifying syndromes ($p < 0.001$): Most understand their referral role, but 28% remain unsure about the full scope of their professional duty.

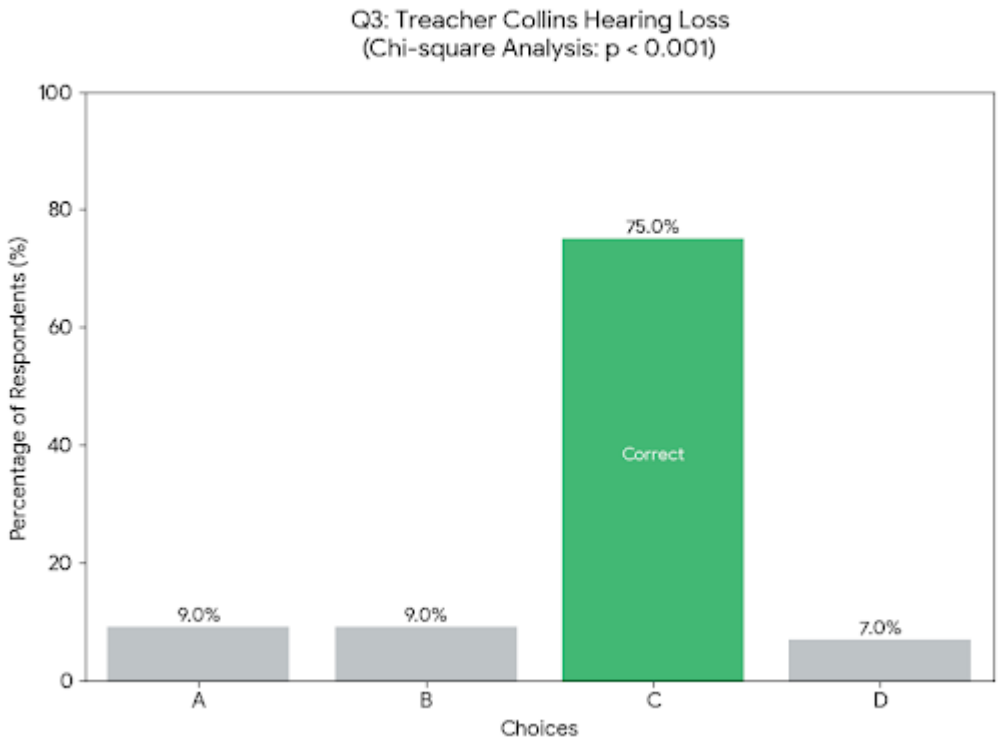
Q16. Importance of Apert Syndrome awareness ($p < 0.001$): Most students correctly understood how physical hand anomalies (syndactyly) impact a patient's ability to maintain their own oral health.

1.What does the term “syndrome” mean in a medical context?



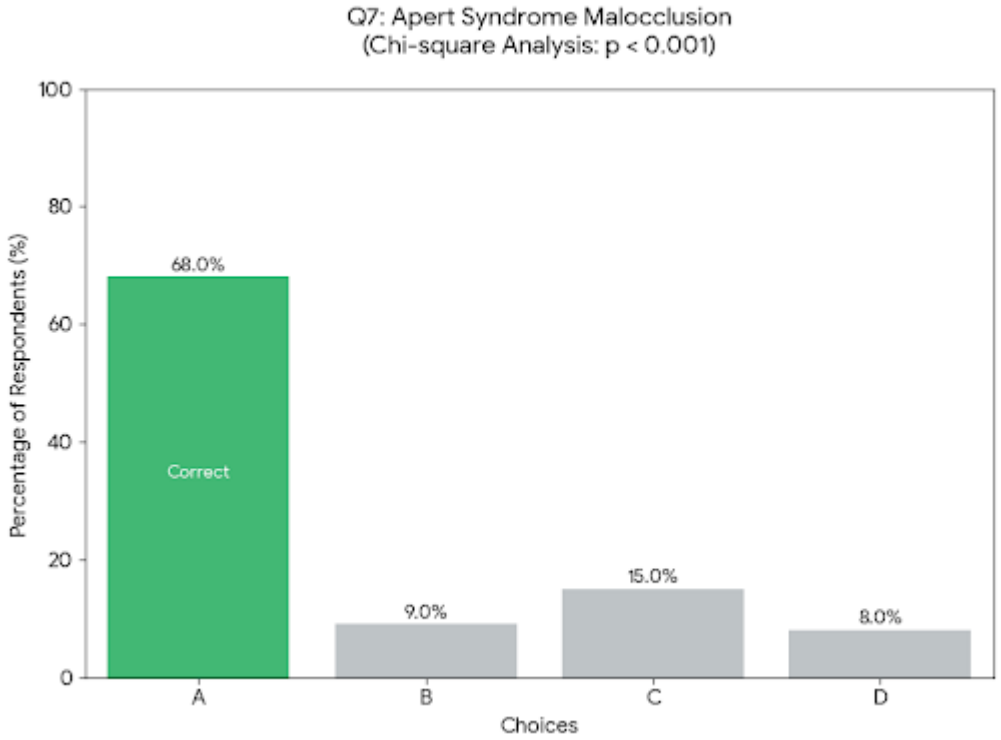
High Baseline Awareness: Most students understand the multi-systemic nature of syndromes, providing a strong conceptual foundation.

3.What type of hearing loss is most commonly associated with Treacher collins syndrome (TCS)?



Good understanding of the conductive hearing loss resulting from craniofacial developmental anomalies.

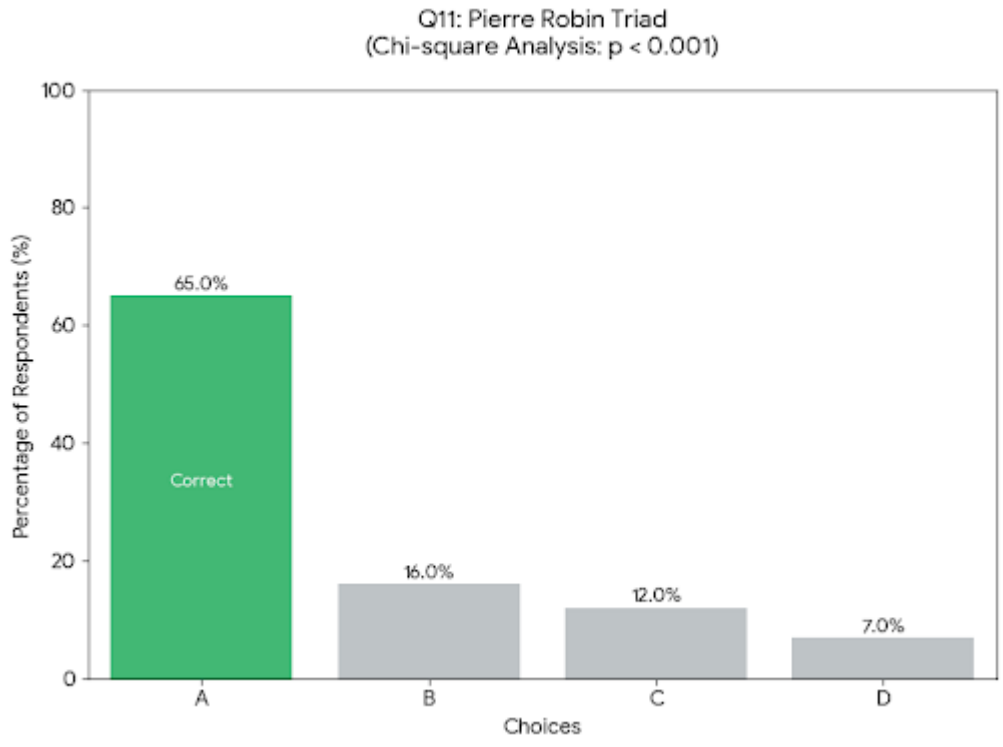
7.Midfacial hypoplasia in Apert Syndrome typically leads to



Strong recognition of the midfacial hypoplasia leading to Class III malocclusion.

11.Which of the following describes the classic clinical trial of Pierre Robin Sequence?

Good understanding of the anatomical sequence (micrognathia,glossoptosis,and cleft palate).



Discussion

The results demonstrate a clear "Recognition-Management Gap." While students are proficient in identifying common syndromes (60-70\% accuracy)[16], they struggle significantly when asked to plan treatment or anticipate systemic risks[17].

The 41\% accuracy in Ectodermal Dysplasia treatment suggests a misconception that orthodontics alone can manage extensive tooth agenesis, whereas prosthetic rehabilitation is often the gold standard[8'25].This mirrors findings by Bahuve et al. (2022) and Shilpa et al. (2014), which emphasize that management of ED must move beyond simple orthodontics to include complex prosthetic and surgical strategies due to extensive tooth agenesis.

The low awareness (45\%) of SCC risk in Dyskeratosis Congenita is a critical finding, as dentists play a life-saving role in early oncological screening[22].Al-Herz et al. (2014) highlight that identifying mucosal changes in this syndrome is vital for preventing severe systemic morbidity, as dentists serve a life-saving role in early oncological screening.

Furthermore, the moderate scores for Gardner Syndrome (58\%) suggest that many students do not yet view oral osteomas as "sentinel" markers for intestinal malignancy[23]. This mirrors findings in literature that rare diseases are often underemphasized in the standard curriculum[17'18]. Kobayashi et al. (2015) and Peker et al. (2009) underscore that the dental professional's diagnostic intuition in recognizing these radiographic findings is critical for life-saving medical intervention.

62% of students correctly linked the fused fingers (syndactyly) in Apert Syndrome to challenges in maintaining oral health. This aligns with research by Babu and Kumar (2022) regarding the level of awareness of Apert syndrome manifestations among dental students.

Pierre Robin Sequence: While students understood the anatomical triad (micrognathia, glossoptosis, cleft palate), only 48% identified the long-term risk of hypoxic injury related to airway obstruction. Butow et al. (2009) stress the importance of anticipating these long-term respiratory complications as part of a multidisciplinary care approach.

The significant Chi-Square values confirm that students hold specific "common errors" for instance, favoring orthodontic treatment in syndromic cases where it is contraindicated alone.

The significant Chi-Square values ($p < 0.001$) for all questionnaire items indicate that student responses were not random, but were driven by specific knowledge or clustered misconceptions. This trend is supported by Motta et al. (2021) and Hala et al. (2020), who noted that rare diseases are often underemphasized in standard dental curricula, leading to inconsistent knowledge regarding treatment modalities. As suggested by Grover et al. (2015), there is a clear need for curricula to shift toward integrated, case-based learning to bridge this gap between theoretical awareness and clinical management

Conclusion

This study reveals that while dental students possess a satisfactory baseline awareness of orofacial syndromes, there are significant deficiencies in clinical management and the presumption of systemic complications. To bridge this gap, dental curricula should move beyond descriptive pathology toward integrated, case-based learning that emphasizes the dentist's role in interdisciplinary medical management.

References:

1.Srivastava, A., et al. (2011). Orofacial Syndromes: A Review.

2. Navarro, M., et al. (2024). Prevalence of Developmental Dental Anomalies and Assessment of Students' Knowledge.

3.Wong, J. W., et al. (2012). Oral and Maxillofacial Considerations in Gardner's Syndrome.

4.Babu, S. S., & Kumar, M. P. (2022). Knowledge and Awareness of Apert Syndrome among Dental Students.

5. Reyes-Reali, J., et al. (2018). Hypohydrotic ectodermal dysplasia: clinical and molecular review.

6. Hanoi Medical University (2025). Knowledge and Awareness on Orofacial Clefts Among Healthcare Students.

7. Peker, I., et al. (2009). One of the Rarest Syndromes in Dentistry: Gardner Syndrome.

8. Shilpa, B. S., et al. (2014). Dental Management of Ectodermal Dysplasia.

9.StatPearls (2023). Pierre Robin Syndrome: Continuing Education Activity.

10. Sujatha, S., et al. (2021). Knowledge and awareness about dentistry among medical students and residents.

11.Neville, B. W., et al. (2015). Oral and Maxillofacial Pathology. Elsevier Health Sciences.

12.Kaur, G., et al. (2016). The Role of the Dentist in Recognizing Multi-systemic Syndromes. Journal of Oral and Maxillofacial Pathology.

13.Mishra, R., et al. (2021). Anticipatory guidance and oral health management in children with special healthcare needs. International Journal of Clinical Pediatric Dentistry.

14.Borglum, A. D., et al. (2003). Molecular genetics of orofacial clefts and associated syndromes. Journal of Dental Research.

15.Vasilakos, K., et al. (2017). Interdisciplinary management of patients with orofacial clefts. Journal of Craniofacial Surgery

16.Grover, N., et al. (2015). Knowledge, attitude and awareness of dental students toward oral manifestations of systemic diseases. Journal of Indian Academy of Oral Medicine and Radiology.

17.Motta, A., et al. (2021). Rare diseases in dental education: A survey of dental students' knowledge and perceptions. European Journal of Dental Education.

18.Hala, J., et al. (2020). Assessment of Knowledge and Awareness regarding Genetic Disorders among Dental Students. International Journal of Dentistry and Oral Science.

19.Deepshikha, C., et al. (2019). Awareness of Syndromes with Oral Manifestations among Undergraduate Dental Students. Journal of Clinical and Diagnostic Research.

20.Khadkhoda, Z., et al. (2023). Evaluating the knowledge of dental students about the oral manifestations of systemic diseases: A cross-sectional study. BMC Oral Health.

21.Desai, V. G., et al. (2014). Papillon-Lefèvre syndrome: A report of two cases and a review of the literature. Journal of Indian Society of Periodontology.

22.Al-Herz, W., et al. (2014). Dyskeratosis congenita: A review of the literature with emphasis on dental manifestations. Journal of Dental Research, Dental Clinics, Dental Prospects.

23.Kobayashi, E. T., et al. (2015). Gardner's syndrome: Clinical and radiographic dental findings. Brazilian Dental Journal.

24.Grippaudo, C., et al. (2016). Apert syndrome: Orthodontic and surgical management of a case. European Journal of Paediatric Dentistry.

25.Bahuve, J., et al. (2022). Management of Ectodermal Dysplasia in children: A review of current prosthetic and surgical strategies. Journal of Prosthodontics.

26.Evans, C. A. (2010). Craniofacial syndromes: Diagnostic and therapeutic challenges in dentistry. American Journal of Orthodontics and Dentofacial Orthopedics.

27.Boutayeb, S., et al. (2019). Oculodentodigital dysplasia: Case report and review of the dental literature. Journal of Oral Biology and Craniofacial Research.

28.Gorlin, R. J., et al. (2001). Syndromes of the Head and Neck. Oxford University Press. (Classic textbook reference).

29.Yadav, S., et al. (2014). Gorlin-Goltz Syndrome: A Rare Case Report and Review of the Literature. Case Reports in Dentistry.

30.Butow, K. W., et al. (2009). Pierre Robin sequence: Analysis of diagnosis and treatment methods. Journal of Cranio-Maxillofacial Surgery.