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Most Frequent Oral and Craniofacial Clinical Characteristics Associated with Syndromes

Características Clínicas Orales y Craneofaciales más Frecuentes Asociadas a Síndromes

Cynthia Guadalupe Torres-Avalos[1]; Alma Graciela Garcia-Calderón[1]; Juan Carlos Cuevas-Gonzalez[1]; Maria Verónica Cuevas-Gonzalez[1]; León Francisco Espinosa-Cristóbal[1]; Rebeca Alejandra Barrio-Soulé[1]; Omar Alejandro Hernández-Cepeda[1] & Luis Felipe Fornelli Martin Del Campo[1]

TORRES-AVALOS, C. G.; GARCIA-CALDERÓN, A. G.; CUEVAS-GONZALEZ, J. C.; CUEVAS-GONZALEZ, M. V.; ESPINOSA-CRISTÓBAL, L. F.; BARRIO-SOULÉ, R. A.; HERNÁNDEZ-CEPEDA, O. A & MARTIN DEL CAMPO, L. F. F. Most frequent oral and craniofacial clinical characteristics associated withsyndromes. *Int. J. Odontostomat., 19(4)*:369-375, 2025.

ABSTRACT: A syndrome is defined as a set of distinctive and clinically recognizable traits, characteristics, or multiple anomalies that occur simultaneously in an individual from birth. Patients with Down, Edwards, Patau, Treacher Collins, Oro-digito-facial, and Cri-du-Chat syndromes commonly present with one of the most frequent congenital nasal anomalies: nasal hypoplasia. This condition is characterized by atrophy of the subcutaneous tissues, skin, alar cartilage, and/or bone components of the nose. Other syndromes, such as Wolf–Hirschhorn, Apert, Crouzon, Cornelia de Lange, and Robinow, often exhibit a depressed or broad nasal bridge, another frequent nasal abnormality. Additionally, a pointed or broad nasal morphology is commonly observed in Rubinstein–Taybi syndrome, as well as in Nance–Horan and Opitz syndromes, which are likewise associated with a prominent nasal appearance. Given these diverse presentations, it is essential for dental practitioners to be familiar with the craniofacial manifestations of syndromic conditions. This knowledge not only facilitates accurate diagnosis but also promotes the formation of multidisciplinary teams capable of providing comprehensive care to affected patients. Due to the complexity and nonspecificity of available information focused solely on the oral region, further research is required to expand current understanding and better equip dentists to deliver optimal clinical outcomes.

KEY WORDS: Craniofacial syndromes, nasal anomaly, physical manifestation.

INTRODUCTION

A syndrome is described as a set of distinctive, clinically recognizable traits, characteristics, or anomalies that occur simultaneously in an individual from birth (Anuthama *et al*., 2013; Gil, 2019; Gedamu *et al*., 2021). According to the World Health Organization (WHO), congenital anomalies occur in approximately 2 % to 3 % of live births. In 2011, it was estimated that between 900 and 5,000 genetically based syndromes were associated with dental, oral, and maxillofacial anomalies (Salerno *et al*., 2021). For this reason, identifying clinical features within the oral cavity and the head and neck region is essential, as some may represent the initial manifestation of an underlying syndrome.

The role of dentists and oral health professionals is therefore critical. Accordingly, the objective of this article is to identify the most frequent oral and craniofacial clinical characteristics associated with syndromic conditions.

SUBJECT DEVELOPMENT

A syndrome is defined as a group of distinctive and clinically recognizable traits, characteristics, or anomalies that occur simultaneously in an individual from birth (Anuthama *et al*., 2013; Gil, 2019). According to the WHO 2 % to 3 % of live births present congenital anomalies, which have been identified as one of the ten leading causes of mortality in children under five years of age (Gedamu *et al*., 2021; Cavaliere *et al*., 2024). In 2021, Salerno *et al*. (2021) reported an incidence of 40 cases per 100,000 inhabitants.

[1]Professor of the Department of Stomatology, Institute of Biomedical Sciences, Universidad Autónoma de Ciudad Juárez, México.

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Although the etiology of congenital anomalies remains unknown in approximately 60 % of cases, several exogenous factors have been implicated, including exposure to drugs, chemicals, alcohol, tobacco, ultraviolet light, ionizing radiation, viral infections, and maternal chronic diseases during pregnancy. Additionally, genetic factors account for nearly one-third of all anatomical anomalies (Vodicka *et al*., 2018; Viotti, 2020; Zylbersztejn *et al*., 2020). Defects in the cell cycle and DNA repair mechanisms increase the likelihood of cellular loss within the 46 chromosomes that constitute a normal human somatic cell, leading to numerical or structural chromosomal abnormalities (Siri *et al*., 2021).

Among numerical anomalies, aneuploidy is particularly noteworthy. This refers to a variation in the number of chromosomal copies that are multiples of 23, with trisomy being the most common form (Vodicka *et al*., 2018; Viotti, 2020; Meler *et al*., 2020; Torres, 2023). Structural anomalies, on the other hand, directly affect chromosomal morphology and arise from defects in the rejoining of chromosomal segments or from the loss of one or more parts of these segments (Miralles *et al*., 2022).

These chromosomal imbalances give rise to a wide spectrum of morphological features and clinical characteristics across various organs and tissues, including the oral cavity and associated craniofacial structures (Viotti, 2020; Salerno *et al*., 2021). For analytical purposes, these manifestations can be classified according to anatomical region, thereby facilitating their systematic study (Fig. 1).

According to the literature, more than 500 syndromes involving craniofacial anomalies have been identified. Craniosynostosis is a prominent feature within the skeletal category and is defined as the abnormal, premature, and non-physiological fusion of one or more cranial sutures. Clinically, it manifests as an abnormally shaped skull, frequently accompanied by restricted brain growth and facial dysmorphism.

Syndromic craniosynostosis is most associated with mutations in fibroblast growth factor receptor (FGFR) genes. Syndromes in which craniosynostosis constitutes a key clinical characteristic include Crouzon, Apert, Pfeiffer, Muenke, Saethre–Chotzen, and Jackson–Weiss syndromes (Bhattacharjee *et al*., 2022; Tibesar & Scott, 2024). Brachycephaly has been reported in individuals with Down syndrome, Angelman syndrome, Apert syndrome, and cleidocranial dysostosis (Corsello & Giuffrè, 2012; Benacerraf *et al*., 2019).

Hypoplasia of the middle and lower thirds of the face is another clinical manifestation reported in multiple syndromes. This condition involves maxillary retrusion and hypoplasia, zygomatic hypoplasia, underdevelopment of the orbital rims, external ear anomalies, a small and depressed nose, and mandibular retrusion, which collectively contribute to malocclusions, speech disorders, and a concave facial profile (Ponce *et al*., 2010; Feregrino *et al*., 2019). These craniofacial deficiencies are frequently reported in conditions such as Pierre Robin sequence, Down, Edwards, Turner, Treacher Collins, Hallermann–Streiff, Sotos, Rubinstein–Taybi, Smith–Lemli–Opitz, Robinow, Nager, DiGeorge, Goldenhar, Cornelia de Lange, Muenke, and various dysplasia and craniosynostosis syndromes (Gorlin & Sedano, 1971; Shiang, 2014; Cammarata-Scalisi *et al*., 2018; Mouthon *et al*., 2019; Marszalek-Kruk *et al*., 2021).

Just as a comprehensive clinical examination of the facial structures is essential, identifying alterations within the oral cavity and surrounding tissues provides additional insight into the development and potential secondary manifestations of syndromic conditions. In syndromes such as Goldenhar, Patau, and Van der Woude, the presence of cleft lip can significantly compromise feeding, breathing, phonation, and dental development (Babai & Irving, 2023; Jayaprakasan *et al*., 2023;).

According to the American Academy of Periodontology (AAP), patients with systemic disorders, genetic conditions, and syndromes often develop a more aggressive and generalized form of periodontal disease. This increased susceptibility results from a combination of factors, including inadequate oral hygiene, alterations in dental morphology, abnormalities of the gingival tissues, changes in saliva composition, macroglossia, immune system deficiencies, and other systemic influences. Syndromes commonly associated with heightened periodontal risk include Down Ehlers–Danlos, Leopard, Marfan, Papillon–Lefèvre, Turner, and Goldenhar syndromes.

Consequently, the implementation of preventive strategies aimed at improving oral hygiene practices is essential to reduce the risk of tooth loss and its subsequent negative impact on quality of life (Ferreira *et al*., 2016; Pinna *et al*., 2019; Díaz-Quevedo *et al*., 2021; Fernández *et al*., 2021; Scott *et al*., 2023).

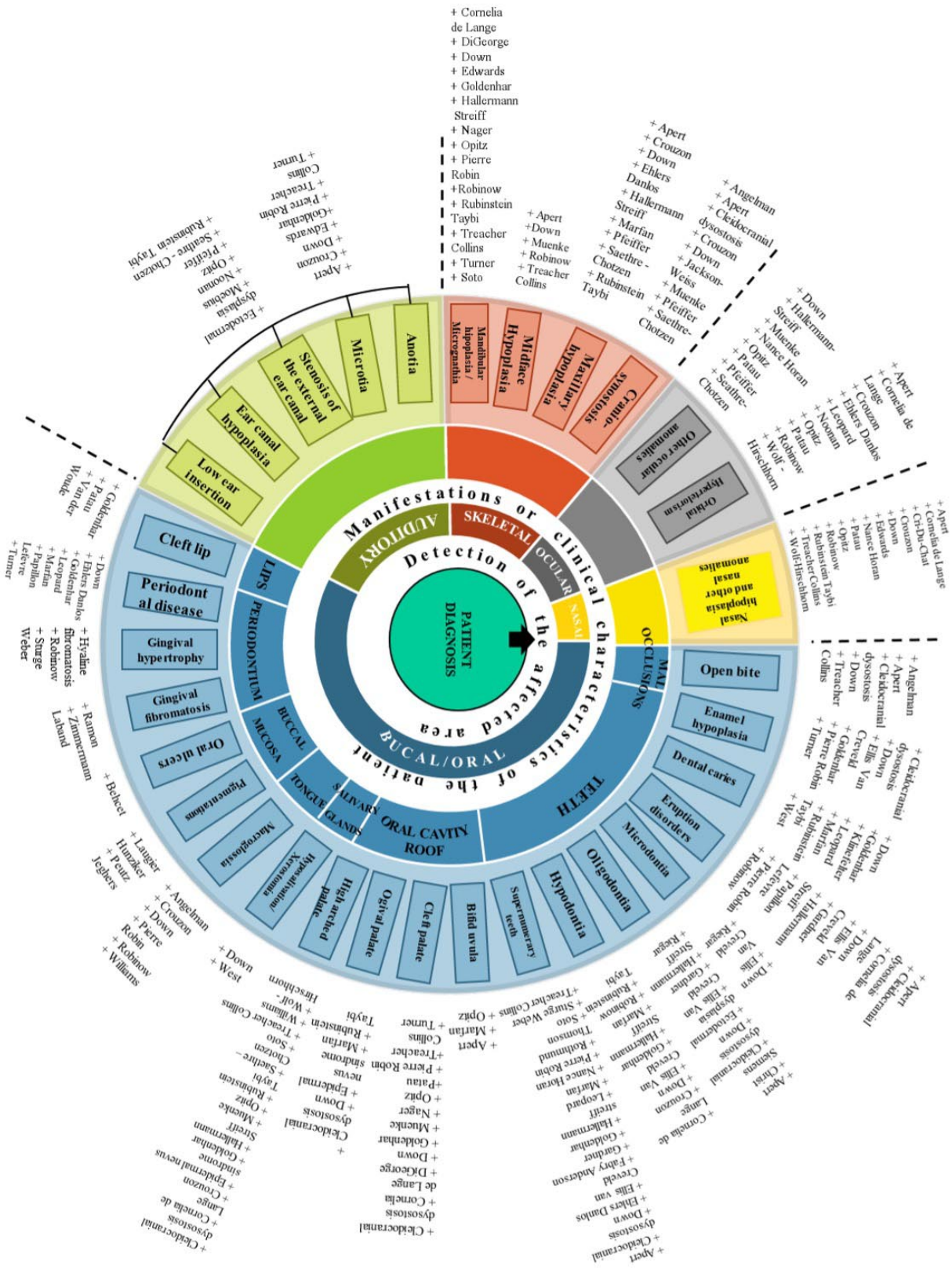

Fig. 1. The most common oral and craniofacial characteristics observed in syndromes divided by area.

Conversely, hereditary gingival fibromatosis and hyaline fibromatosis are characterized by asymptomatic, slow, and progressive gingival enlargement, without changes in periodontal tissue coloration or involvement of the alveolar bone. As a result, affected patients are at increased risk for gingivitis, the formation of pseudo-periodontal pockets, halitosis, impaired mastication, and delayed eruption of permanent teeth. Syndromes commonly associated with this condition include Zimmermann–Laband and Ramon syndromes (Guglielmi *et al*., 2019; Shirian *et al*., 2019; Knezevic *et al*., 2020; Costa *et al*., 2021).

Among the intraoral regions susceptible to alteration in numerous syndromes, the palate is particularly significant. Clinical findings may include a high-arched or deep palate, an ogival configuration, and cleft palate. These alterations can lead to malocclusions, dental malpositions, respiratory difficulties, and feeding challenges due to direct communication with the nasal cavity. Notably, cleft palate is associated with more than 200 syndromes. Syndromes frequently presenting palatal abnormalities include Goldenhar, Van der Woude, Patau, Edwards, Wolf–Hirschhorn, Epidermal Nevus syndrome, Down, Williams, Hallermann–Streiff, Sotos, Crouzon, Rubinstein–Taybi, Opitz, Treacher Collins, Cornelia de Lange, Marfan, Turner, Pierre Robin sequence, Muenke syndrome, and cleidocranial dysostosis (Mouthon *et al*., 2019; Atarbashi-Moghadam *et al*., 2020; Marsza?ek-Kruk *et al*., 2021; Smarius *et al*., 2021; Van Gils *et al*., 2021; Herrera *et al*., 2022; Jayaprakasan *et al*., 2023; Babai & Irving *et al*., 2023; Popescu *et al*., 2023).

Alterations in the development or maturation of the tissues that compose the dental organs have substantial implications for the health of syndromic patients, as they compromise oral function through malocclusions, dental hypersensitivity—often leading to difficulties in feeding and aesthetic impairments (Bilge *et al*., 2018). The most frequently reported anomalies include microdontia, hypodontia, oligodontia, supernumerary teeth, enamel hypoplasia, and disturbances in dental eruption. These dental manifestations are commonly observed in individuals with Down syndrome, Crouzon, Apert, Treacher Collins, Pierre Robin sequence, Ellis–van Creveld, Papillon–Lefèvre, Klinefelter, Rieger, Hallermann–Streiff, Marfan, Gardner, Cornelia de Lange, and Goldenhar syndromes. Additional conditions associated with similar alterations include Rubinstein–Taybi, Sturge–Weber, Fabry–Anderson, Ehlers–Danlos, Nance–Horan, LEOPARD, Christ–Siemens–Touraine, West, Angelman, Rothmund–Thomson, Sotos, Robinow, hypohidrotic ectodermal dysplasia, cleidocranial dysostosis, and craniofacial dysostosis (Ponce-Bravo *et al*., 2010; Anuthama *et al*., 2013; Al-Ani *et al*., 2017; Inoue *et al*., 2017; Gjørup *et al*., 2017; Camaratta-Scalisi *et al*., 2018 Bilge *et al*., 2018; Masood & Benavides, 2018; Feregrino *et al*., 2019; Diaz-Quevedo *et al*., 2021)

It is noteworthy that mucocutaneous lesions and pigmentary alterations are often among the earliest clinical manifestations of systemic diseases and syndromes. Within this category, Behçet's syndrome is characterized by recurrent oral and/or genital ulcers, ocular inflammation, and may also involve vascular, articular, dermatologic, and neurological complications (Saccucci *et al*., 2018; Bettiol *et al*., 2020; López-García *et al*., 2020). Peutz–Jeghers syndrome and Laugier–Hunziker syndrome are distinguished by the presence of lenticular pigmentations, with the former also conferring an increased risk for malignant neoplasms. Additionally, Addison's disease typically presents with hyperpigmentation in sun-exposed areas of the skin (Latchford *et al*., 2019; Pinna *et al*., 2019).

Various syndromic conditions also present distinctive craniofacial features, many of which arise from abnormal development of the branchial arches during embryogenesis. Malformations of the external and middle ear, eyes, and nose such as microtia, stenosis or hypoplasia of the external auditory canal, low-set ears, and canal atresia are commonly reported in syndromes including Goldenhar, Crouzon, Treacher Collins, Pierre Robin, Down, Edwards, Turner, Rubinstein-Taybi, Noonan, Apert, Pfeiffer, Opitz, Saethre-Chotzen, Moebius, and hypohidrotic ectodermal dysplasia (Gorlin & Sedano, 1971; Ponce-Bravo *et al*., 2010; Corsello & Giuffre, 2012; Camaratta-Scalisi *et al*., 2018; Ocak *et al*., 2019; Marszalek-Kruk *et al*., 2021).

These patients frequently present atypical language development during childhood, which may lead to deficits in phonology, processing speed, and comprehension, as well as academic difficulties and behavioral, emotional, and social disturbances (Quantin *et al*., 2018). In the ocular region, hypertelorism is commonly observed in Crouzon, Noonan, Opitz, Apert, Patau, LEOPARD, Ehlers–Danlos, Robinow, Cornelia de Lange, Wolf–

Hirschhorn, and orofaciodigital syndromes (Buchanan *et al*., 2017; Bhattacharjee *et al*., 2022; Corsello & Giuffre, 2012; Ferreira *et al*., 2016; Popescu *et al*., 2023). Additionally, cataracts and lens opacification are frequently reported in Hallermann–Streiff, Down, Opitz, Martsolf, and Nance–Horan syndromes (Gjørup *et al*., 2017; Khokhar *et al*., 2017; Sharma *et al*., 2017)

Patients with Down, Edwards, Patau, Treacher Collins, orodigitofacial, and Cri-du-Chat syndromes frequently exhibit one of the most common congenital nasal anomalies: nasal hypoplasia. This condition is characterized by atrophy of the subcutaneous tissues, skin, alar cartilage, and/or underlying bone (Gorlin & Sedano, 1971; Shiang, 2014; Moczulska *et al*., 2022). Other syndromes, such as Wolf–Hirschhorn, Apert, Crouzon, Cornelia de Lange, and Robinow syndromes, often present with a depressed or widened nasal bridge, which constitutes another prevalent nasal abnormality. A pointed or broad nasal morphology is a characteristic feature in patients with Rubinstein–Taybi syndrome and is also observed in individuals with Nance–Horan and Opitz syndromes, in whom a notably prominent nose is commonly described (Ponce-Bravo *et al*., 2010; Ferreira *et al*., 2016; Gjørup *et al*., 2017; Cammaratta-Scalisi *et al*., 2018; Feregrino *et al*., 2019; Atarbashi-Moghadam *et al*., 2020; Van Gils *et al*., 2021; Popescu *et al*., 2023; Reyes-Saberbein *et al*., 2016).

CONCLUSION

This study highlights the most frequent oral and craniofacial clinical characteristics associated with syndromic conditions, underscoring their diagnostic value and clinical relevance. Recognizing these manifestations is essential for dental professionals, as it supports timely diagnosis and promotes the integration of multidisciplinary approaches for comprehensive patient management. The current limitations and non-specificity of available information focused solely on oral findings reveal a clear need for further research in this field. Expanding the evidence base will not only strengthen dentists' understanding of syndromic manifestations but also improve the quality of care and overall outcomes for affected patients.

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RESUMEN: Un síndrome se describe como un conjunto de rasgos, características o anomalías múltiples, distintivas y clínicamente reconocibles, que ocurren simultáneamente en un individuo desde el nacimiento. Entre los pacientes con síndrome de Down, Edwards, Patau, Treacher Collins, Orodigitofacial y Cri-du-Chat, una de las anomalías congénitas más comunes en la región nasal es la hipoplasia nasal, cuya manifestación física incluye la atrofia de los tejidos subcutáneos, la piel, el cartílago alar y/o el tejido óseo. Asimismo, síndromes como Wolf-Hirschhorn, Apert, Crouzon, Cornelia de Lange y Robinow suelen presentar una estructura nasal deprimido o ensanchado, constituyendo otra alteración frecuente. Por otro lado, una morfología nasal puntiaguda o ancha es característica en pacientes con síndrome de Rubinstein-Taybi, así como en aquellos con los síndromes de Nance-Horan y Opitz, los cuales también se asocian con una nariz prominente. Para el odontólogo, comprender estas manifestaciones es fundamental no solo para favorecer un diagnóstico preciso, sino también para promover la conformación de equipos multidisciplinarios orientados al tratamiento integral de pacientes con síndromes. Dada la complejidad e inespecificidad de la información disponible centrada exclusivamente en la región oral, se abren importantes oportunidades de investigación que permitirán ampliar el conocimiento y mejorar la preparación profesional, con el fin de brindar una atención de mayor calidad y beneficio para estos pacientes.

PALABRAS CLAVE: Síndromes craneofacial, anomalía nasal, signos clínicos.

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Corresponding author:
Juan Carlos Cuevas Gonzalez
Department of Stomatology
Institute of Biomedical Sciences
Universidad Autónoma de Ciudad Juárez
MEXICO

E-mail: juan.cuevas@uacj.mx