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Crouzon Syndrome: A Case Study Of Siblings

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Abstract:
The Crouzon syndrome is typically diagnosed in early childhood due to its characteristic craniofacial anomalies, but in rare cases, diagnosis may occur later, especially when facial features become apparent post-childhood growth or due to secondary complications like dental issues. This clinical case involves two siblings showing significant dental eruption delay and atypical facial anomalies discovered during orthodontic evaluation. Clinical and radiographic examinations confirmed specific facial morphological features, leading to the suspicion of Crouzon syndrome, further confirmed through comprehensive evaluation, including targeted genetic testing. Interdisciplinary collaboration between orthodontists and rheumatologists is pivotal for the early diagnosis of this condition, enabling early medical and surgical interventions to enhance patients' quality of life and functional outcomes.

Keywords: *Crouzon syndrome, orthodontic, craniofacial anomalies.*

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I. Introduction:

The development of craniofacial structures represents an exceptionally complex process, characterized by rapid and precise orchestration of mesodermal cells and craniofacial neural crests, all orchestrated within a complex signaling network. Syndromes affecting the first and second branchial arches manifest as combined hypoplasia and aplasia of their derivatives, encompassing structures such as the face, external ear, middle ear, as well as the maxillary and mandibular arches during early embryonic development. These syndromes constitute the second most common craniofacial malformation after cleft lip and palate. Derivatives of the first arch include the mandible, middle ear ossicles, masticatory muscles, and the mandibular nerve. Based on clinical features, anatomy, and embryological knowledge, it is now evident that all anomalies stemming from disruption of first arch development encompass a wide spectrum of conditions, including Treacher Collins syndrome (mandibulofacial dysostosis), mandibular dysostosis, Pierre Robin syndrome, external and middle ear alterations, congenital deafness-muteness, hypertelorism, cleft lip and palate, and a recently characterized syndrome combining congenital deafness and hypertelorism.

Identifying Crouzon syndrome is typically a diagnostic challenge from early childhood, given its characteristic clinical presentation involving fairly typical craniofacial anomalies. Also known as craniofacial dysostosis, it stands out as an autosomal dominant condition affecting the first branchial arch, which prefigures both the maxilla and mandible. In 1912, French neurologist Octave Crouzon provided the first description, outlining a set of cranial deformations, facial anomalies, and proptosis. This condition results from a mutation in the fibroblast growth factor receptor (FGFR) -2 and -3 gene, located on chromosome 10. Its frequency is estimated at 1 in 60,000 births, with a worldwide prevalence of about 1 in 25,000, with no known racial or gender predilection. It is the most common syndromic craniosynostosis, resulting from premature fusion and ossification of at least two sutures, most commonly the coronal and sagittal sutures, resulting in brachycephaly, midface hypoplasia, and a broader anterior skull base.

However, there are exceptional circumstances where this syndrome may be diagnosed at a later stage, when craniofacial features become apparent only after the child's growth period or due to the onset of secondary complications, notably dental or orthodontic problems, prompting thorough medical evaluation.

The clinical case we present highlights an atypical scenario involving two siblings. These patients exhibited unusual facial morphological characteristics, significant maxillofacial dysmorphosis, and substantial dental eruption delay, identified during orthodontic evaluation. This late discovery raised questions about the precise nature of their disorders and led to a comprehensive medical investigation.

II. Case Presentation:

M.S, a 13-year-old girl, and M.H, a 10-year-old boy, are two siblings in a family of three children. They were recently faced with an intriguing diagnosis: unusual craniofacial anomalies associated with significant delay in dental eruption. This discovery stemmed from meticulous orthodontic evaluation, revealing highly distinctive facial morphological features. These included maxillary hypoplasia, hypertelorism, and maxillofacial dysmorphosis. These findings strengthened the initial suspicions of Crouzon syndrome. The patients were referred to a rheumatology specialist and underwent a comprehensive clinical examination, including detailed assessment of facial morphology, thorough radiographic analysis, and targeted genetic investigations to search for mutations in the FGFR2 gene. The results of these investigations all converged to confirm the diagnosis of Crouzon syndrome.

III. Discussion:

The clinical cases presented demonstrate the role of orthodontics in diagnosing Crouzon syndrome. Maxillary hypoplasia, characterized by underdevelopment of the upper jaw, is one of the most striking features. It often leads to severe dental malocclusion, affecting chewing and aesthetics. Hypertelorism, manifested by unusual spacing between the eyes, is another distinctive element. Lastly, maxillofacial dysmorphosis gives their faces singular proportions, characteristic of Crouzon syndrome. These clinical findings raised suspicions regarding the possible presence of Crouzon syndrome. In this regard, the patients were referred to a rheumatology specialist, an essential step to deepen the understanding of their condition.

Role of Orthodontics in Diagnosis:

Orthodontic evaluation plays a crucial role in the early and accurate detection of late-onset forms of Crouzon syndrome, even in cases where the disease was not identified during early childhood by pediatricians or school medicine. This critical importance of orthodontic evaluation is particularly illustrated by the cases of two patients, M.H and M.S, within a sibling group of three children, where the youngest boy has a normal morphology. These two patients exhibit atypical craniofacial anomalies, characterized by maxillary hypoplasia, albeit discreet hypertelorism, and maxillofacial dysmorphosis. These distinctive features, though visible, were not detected early in their development. Early childhood medical visits, pediatric consultations, and school health examinations did not identify these anomalies. One reason for this late detection lies in the fact that Crouzon syndrome is a rare and complex genetic disease that can present subtle or variable clinical manifestations. In many cases, characteristic signs of the disease only become apparent as the child grows, making early detection particularly challenging. Additionally, Crouzon syndrome can be confused with other craniofacial conditions, further complicating its diagnosis. This is where orthodontic evaluation becomes invaluable. By focusing on craniofacial morphology and dental anomalies, orthodontists are well-equipped to spot subtle clinical signs of Crouzon syndrome. They can not only identify anomalies but also differentiate them from other possible conditions, allowing for prompt and appropriate management. The history of M.H and M.S highlights the importance of orthodontic evaluation in detecting late-onset forms of Crouzon syndrome. It also underscores the need for increased awareness of subtle clinical manifestations of this disease, even in children who were not diagnosed early. Through close collaboration among healthcare professionals, it is possible to detect, diagnose, and treat Crouzon syndrome more effectively, thereby offering patients a better quality of life and more promising prospects. The discovery of Crouzon syndrome is exceptionally late, with few late-onset cases described in the literature, such as the case of a six-year-old girl diagnosed in a pediatric consultation due to a particular facial appearance. During physical examination, suspicion of Crouzon syndrome was raised and confirmed by head computed tomography, revealing asymmetric thickening of the calvarium, diffuse indentation of the inner table of the skull, and moderate hydrocephalus with a large cyst in the posterior fossa.

Role of Additional Examinations:

Radiographic examinations have proven crucial for a thorough understanding of the pathology. Panoramic radiography provided an assessment of bone structures. Genetic investigations were an essential step in diagnosis. The FGFR2 gene is closely linked to Crouzon syndrome, and specific tests were conducted to search for mutations in this gene. The results converged to an undeniable conclusion: M.H and M.S were indeed affected by Crouzon syndrome.

Management of Crouzon Syndrome:

Crouzon syndrome, though exceptionally complex, is not insurmountable. Significant advances in medical and surgical treatments have opened new perspectives for patients with this condition. Craniofacial surgery is a crucial aspect of managing Crouzon syndrome. It aims to correct severe craniofacial anomalies observed in patients, including premature closure of cranial sutures and maxillofacial dysmorphosis. These interventions are often performed in multiple stages, depending on the child's growth, to achieve the best

possible results. Orthodontics plays a key role in managing dental problems associated with Crouzon syndrome. Orthodontic appliances are used to correct dental malocclusions and improve chewing function. Regular orthodontic follow-up is necessary to monitor dental development. Regular medical follow-up throughout life is essential for patients with Crouzon syndrome. It allows for monitoring the evolution of craniofacial anomalies, early detection and treatment of other potential health problems, and providing psychosocial support to patients and their families. Plastic surgery interventions may be considered to improve aesthetic appearance. These may include facial feature correction surgeries, such as rhinoplasty or genioplasty, to enhance facial symmetry and harmony. Management of Crouzon syndrome should also consider addressing potential complications. For example, respiratory problems related to nasal obstruction may require specific interventions to improve breathing. Crouzon syndrome is generally inherited in an autosomal dominant pattern, meaning a affected parent has a 50% chance of passing the genetic mutation to each child. Therefore, genetic counseling is crucial to help families understand the risk of disease transmission and make informed decisions about family planning. This rare and complex syndrome affects craniofacial morphology, dental development, and patients' quality of life. Multidisciplinary management, including surgical, orthodontic, and genetic interventions, is essential to improve patients' prospects. Regular medical follow-up and psychosocial support are also important elements of comprehensive management of this condition. Through collaborative efforts among healthcare professionals and families, it is possible to improve the quality of life of individuals with Crouzon syndrome and offer them a promising future.

Conflict of Interest: The authors declare no conflicts of interest related to this article.

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