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Larsen Syndrome: A Rare Case Report" – A Call for Greater Awareness of Orofacial Manifestations in Rare Genetic Disorders

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To the Editor,

We are writing in response to the article "Larsen Syndrome: A Rare Case Report" by Poudel et al. [1], published in Vol. 7, No. 1 (2024) of the Journal of BP Koirala Institute of Health Sciences. The authors have presented a compelling and clinically significant case of a five-year-old female diagnosed with Larsen syndrome, emphasizing the rarely reported dental manifestations associated with this condition. As a dental professional and advocate for early diagnosis and multidisciplinary collaboration, we commend the authors for highlighting a neglected yet critical aspect of this rare genetic disorder: its oral and craniofacial manifestations.

Larsen syndrome, characterized by musculoskeletal abnormalities, is often diagnosed based on distinctive skeletal and craniofacial features such as joint hypermobility, equinovarus deformity, hypertelorism, and a depressed nasal bridge [2]. However, the condition's orofacial features—particularly dental anomalies—remain underreported, often leading to delays in dental intervention that can compromise a child's function and quality of life. The case documented by Poudel et al. [1] is especially important because it adds to the limited body of literature acknowledging the significance of these oral findings.

In the presented case, the child exhibited oligodontia, abnormal tooth morphology, and malocclusion—all of which are consistent with other isolated reports of Larsen syndrome [3, 4]. These anomalies not only impair mastication and phonation but also affect psychosocial development, especially in early childhood when peer interactions are heavily influenced by facial appearance. Unfortunately, such manifestations are frequently dismissed as secondary concerns in genetic disorders where life-threatening complications may overshadow functional and aesthetic impairments.

This case brings to light two important clinical implications. First, it affirms the necessity of early dental screening in children with known syndromic diagnoses, particularly those with midface hypoplasia or known genetic mutations such as FLNB. Second, it underscores the role of pediatric dentists within the larger multidisciplinary team. As the article indicates, interventions such as the restoration of malformed teeth and discussions around prosthetic rehabilitation significantly improved the child's oral function and comfort. These interventions are not merely cosmetic; they are essential to overall health and development.

Additionally, we applaud the authors for including a brief genetic explanation of the FLNB gene mutation—a missense mutation (3-58084597-C-A variant) causing a threonine-to-asparagine substitution—which was confirmed by exome sequencing. This reinforces the growing role of genomic diagnostics in oral medicine and pediatric dentistry. Understanding the genetic etiology of such disorders allows for precision in diagnosis, counseling, and even treatment planning. As genetic technologies become more accessible, their integration into dental settings should be encouraged.

Despite these strengths, it would have been better if authors could have highlighted the uniqueness of the case. The article could have been enhanced with a longer-term follow-up of the prosthetic management plan. While the family declined interim prosthetic treatment,

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Declarations

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or speech development assessments could have provided a more holistic understanding of the patient's quality of life.

Nonetheless, this report significantly contributes to the medical and dental communities by reminding practitioners to consider systemic syndromes when encountering multiple missing or malformed teeth. As dental professionals, we are often the first to observe such anomalies, and raising awareness of their potential systemic implications is crucial. The article also aligns well with the recommendations by the National Organization for Rare Disorders (NORD), which advocates for early multidisciplinary interventions in rare conditions such as Larsen syndrome [5].

In conclusion, the article by Poudel et al. [1] should be recognized not only for its clinical detail but also for drawing attention to a much-needed area of focus in pediatric oral health. It encourages clinicians to think beyond traditional silos, underscores the importance of early genetic and dental evaluations, and advocates for integrated care models in managing rare syndromes. We hope this case inspires further research and reporting on the orofacial aspects of rare genetic conditions so that children like the one presented can benefit from timely, comprehensive care.

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