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THE EDITOR'S CORNER

Trisomy 21

Down syndrome, also known as trisomy 21, is a common genetic condition caused by an extra copy of chromosome 21. Named after British physician John Langdon Down, who formally described it in 1866, the condition is associated with distinct physical and oral characteristics. Orthodontists typically see a few adolescent patients with Down syndrome each year. While these individuals may benefit from orthodontic care, their unique dental and muscular features present significant treatment challenges.

Humans typically have 46 chromosomes, arranged in 23 pairs, with each parent contributing half. These chromosomes are composed of DNA, the blueprint of life that carries instructions for growth and function. Trisomy 21 results from chromosomal nondisjunction during meiosis, so the fertilized egg carries 47 chromosomes instead of the usual 46. This additional genetic material influences cognitive development, physical traits, and specific health characteristics.

Down syndrome occurs in approximately one in 700 live births in the U.S., a rate comparable to the global incidence of cleft lip and palate. It affects all racial and ethnic groups, though studies suggest a higher prevalence among Hispanics in the U.S. and a slight male predominance. The likelihood increases with advancing maternal age, particularly after 35. The average life expectancy is around 60 years, primarily due to complications from congenital heart disease, immune deficiencies, and early-onset Alzheimer's.

Individuals with Down syndrome typically experience mild to moderate intellectual disability, along with distinct physical characteristics, including a flattened facial profile, small ears, upward-slanting eyes, shorter stature, low muscle tone (hypotonia), joint laxity, and congenital heart defects. Oral manifestations often include a protruding or enlarged tongue, Class III skeletal malocclusion due to midface hypoplasia, delayed tooth eruption, taurodontism, and an increased risk of periodontal disease and sleep apnea.

Parents of children with Down syndrome may pursue orthodontic treatment not only to address functional concerns but also to ensure their child receives the same level of care and opportunities as their siblings. Consequently, orthodontists may feel obligated to proceed, especially if they have previously treated other family members. Treating patients with Down syndrome presents a unique dilemma, however, as the potential benefits of orthodontics must be carefully weighed against the associated risks.

While this may not be a common stance, I have largely stopped providing orthodontic treatment for patients with Down syndrome due to the increased risk of root resorption and near-complete orthodontic relapse caused by tongue pressure. When I do proceed, I exclusively use aligners, as I believe they minimize the risk of resorption while also providing occlusal coverage against the grinding often seen in these patients. Despite a clear orthodontic need, in my experience, the risks generally outweigh the benefits.

Others may take a different approach and successfully treat many patients with Down syndrome in their practice. They will readily attest that these individuals are among the most wonderful and kindhearted patients, making their care especially rewarding for both the orthodontist and their team. For those looking to do the same, my best advice for guiding your team in interacting with young patients with Down syndrome is simple: closely watch how their siblings naturally care for them. They will show you.

Trisomy 21 is more than an extra chromosome—it's a spark that makes individuals with Down syndrome truly extraordinary. Their orthodontic care should honor that uniqueness, requiring a thoughtful, tailored approach to support their skeletal, dental, and myofunctional needs. Orthodontists who provide this care understand it's more than just aligning teeth; it's about embracing their resilience and celebrating the joy they bring into the world. These qualities aren't just part of who they are—it's in their DNA.

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