

# Craniofacial Features and Specific Oral Characteristics of Down Syndrome Children

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## Abstract

**Aim:** This article intends to describe the characteristics of Down syndrome children in order to facilitate their management in the dental office.

**Methods:** A review of literature was made limited to articles published between 2003 and 2013. The article is based on a literature search in PubMed and the authors' clinical experience with the patient group.

**Discussion:** Individuals with Down syndrome are clinically characterized by generalized hypotonia, neurological, cardiac and respiratory problems and changes in their stomatognathic system. Almost all these patients have an occlusal disharmony, including anterior open bite, anterior and posterior crossbite, anterior proclination and underdevelopment of the maxilla and the midface. Dental anomalies such as hypodontia, peg-shaped teeth and enamel hypocalcification are also found. The dentomaxillofacial features have clinical implications in speech, nutrition, posture, ventilation and aesthetics, with enormous consequences in growth, development and social integration.

**Conclusions:** When planning the dental treatment of patients with Down syndrome, dental practitioners should always consider their general health, in order to achieve a holistic and interdisciplinary approach.

*Key Words: Down syndrome, craniofacial abnormalities, pediatric dentistry*

## Introduction

Down Syndrome (DS), also referred as Trisomy 21, is a congenital autosomal anomaly characterized by generalized hypotonia, neurological changes with intellectual impairment, great risk of respiratory problems and infections as well as dental anomalies. Approximately 95% of the DS cases have an extra chromosome 21, resulting in a total of 47 chromosomes. Other chromosomal abnormalities include translocation (3%) and mosaicism or partial trisomy (2%) [1,2].

Because of the medical advances and improvements in the medical field, the mean survival of individuals with DS has increased considerably [2,3]. Their life expectancy has substantially increased over the last decades and they often live until their 6th decade [3-5]. Thus it's important for the health care providers and educational and social services to improve the quality of life of these individuals [2,6,7].

Children with DS have a well-recognized phenotype, including external characteristics, specific physical problems and intellectual impairment with delayed cognitive and motor development [8,9]. The different abnormalities that characterize this syndrome are classified as general and craniofacial features.

## Aim

This article intends to describe the craniofacial characteristics of Down syndrome children in order to facilitate their management in the dental office.

## Methods

A PubMed database research was conducted. The search was limited to articles published between 2003 and 2013 in Portuguese and English.

The article is based on a literature search in PubMed and the authors' clinical experience with the patient group.

## Discussion

### General characteristics

The Down syndrome entails clinical complications (such as cardiac, neurological, endocrinal, respiratory, hematologic, ophthalmologic and gastrointestinal problems) that affect the overall development of the child. These patients often have a short stature, obesity, infertility, epicanthus, oblique palpebral fissures, hypotonic tongue, shortening of the extremities (hands, feet, fingers, ears and nose) and a single palmar crease [1,8,10]. Due to the improvements in the medical field a decrease in infant morbidity and mortality occurred [11-13].

Patients with DS have a high prevalence of specific otorhinolaryngologic pathology, including recurrent sinusitis and chronic nasal obstruction, as a consequence of hypotonia and craniofacial malformations [1,8,14]. These problems, together with their short eustachian tubes, predispose them to chronic media otitis with effusion and conductive hearing loss, which interferes with their language acquisition [7,8,11]. This group of people also has a high incidence of recurrent infections, particularly of the upper airway [1,7,8,11]. Sleep apnea is diagnosed in more than 50% of the patients [2,14] and may adversely affect behavior, growth and neurodevelopment [15].

The atlanto-axial joint, which is responsible for promoting communication between the first and the second vertebrae, is unstable in about 20% of the individuals [1,14]. This defect may cause spinal cord compression during sudden movements of flexion and extension and, therefore, the dentist must be very careful while handling their necks [15].

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Another common abnormality is the dysfunction of the thyroid gland. Individuals with DS tend to present hypothyroidism [14,16] and it is related to an underdevelopment of the bones and the teeth and to a delayed tooth eruption [16].

When compared to the general population these children have up to 20 times higher risk of developing leukemia [3]. The dentist should be alert to the presence of persistent lesions and spontaneous gingival bleeding as it may be a sign of leukemia [17].

Mental health and behavioral problems, including attention deficit disorder, hyperactivity, obsessive-compulsive disorder and depression are common among individuals with DS. Most of them also develop Alzheimer's disease [15,17-19] around the fourth or fifth decade of life [19]. This degenerative disease is related to an over-expression of  $\beta$ -amyloid precursor protein ( $\beta$ APP) which is the expression of one of the triplicated genes in DS [17].

#### **Craniofacial features**

The most common craniofacial features observed in children with DS are: small nose, low nasal bridge, narrow, short, deep and high palate, bifid uvula, underdeveloped jaw, cleft lip, incomplete lip closure, hypotonic lips, fissured tongue, inaccurate and slow tongue movement and changes in temporary and permanent tooth eruption [1,6,7,20].

Brachycephaly can be found [6,7,18] and the base of the skull, the frontal bone and the paranasal sinus are significantly small, leading to a decrease in the size of the sella turcica. There's a flattening of the cranial base as a result of vertical hypoplasia of the structures of the skull [4].

Tongue gives the impression of being abnormally large on account of muscle weakness and of an anterior and low position in the mouth (relative macroglossia) [7,21].

As DS patients are mouth breathers, exhibit open bite and their orofacial muscles are hypotonic, there's an incomplete closure of the lips [4,12,22]. It causes an imbalance in orofacial development which leads to malocclusion [7,12,23] and craniofacial malformations such as the hypoplasia of the midface [6,23-25].

Due to the presence of a protruding tongue and a muscular hypotonicity, these children have oral-motor problems (seen during swallowing, chewing and sucking) [12,23-25]. Hypotonicity is associated with ligament laxity, easily visible throughout the body [3]. It induces hyperflexible joints, which can compromise the periodontal ligaments. Excess of saliva on the labial commissure is also related to the muscle hypotonicity and can lead to irritation, cracking (angular cheilitis), aphthous ulcers and infectious conditions like candidiasis [15,26,27].

#### **Occlusal anomalies**

Underdevelopment of the midface bones is common, causing a shortened palate in the anteroposterior dimension [24-26]. The most frequent occlusal abnormalities stem from variations in vertical and transversal dimensions (anterior open bite, posterior crossbite and reductions in the maxillary arch) [4,6,7]. These anomalies cause problems related to oral functioning (chewing, swallowing and speaking) [28].

Oredugba found 51% of class I dental malocclusion and 47% of class III among 43 individuals with DS and only 5% of class III in the control group (individuals without

DS), concluding that class II has a higher incidence in DS individuals than in the general population [20].

Mushish [29] and Soares et al. [30] have concluded that class III is more frequent in DS individuals. Anatomically, the facial mid-third is underdeveloped but the mandible follows normal development (pseudo-progeny). This midface dysplasia also contributes to the narrow maxilla.

Mandible measurements are not significantly different from normal subjects. However, a transverse expansion may occur due to lingual pressure. This intermaxillary discrepancy prevents the optimal intercuspal position to occur, which is needed to stabilize the mandible and the hyoid bone during mastication and swallowing [30].

Oliveira et al. [7] found a 31% prevalence of posterior crossbite and 33% anterior crossbite in the group of DS children and Soares et al. [30] identified posterior crossbite in 39% and anterior crossbite in 26% of their sample. Anterior crossbite is primarily attributed to the anteroposterior deficiency of the maxillary arch development, resulting in a crossbite of the mandible, projecting the jawbone arch towards the front of the maxilla [30].

An anterior open bite is normally found in the DS children. Oral and facial musculature, in particular tongue and lips, are hypotonic. Tongue thrust and posture might hamper enough eruption to cause anterior open bite and to influence the shape of the dental arch and the position of the teeth. Normally these children have a high palate, hypertrophy of the tonsils, hypotonia and nasal obstruction, which lead to mouth breathing and therefore an anterior open bite is common [7].

Malocclusion comes from vertical and/or horizontal changes and its occurrence in DS patients increases with age. This happens due to craniofacial growth restriction, oral-motor dysfunction and generalized orofacial hypotonicity. On the other hand, insufficient bone development associated with impaired facial muscles can lead to a weak lip closure strength, drooling and occlusal abnormalities [7,23]. All these factors, associated with hypotonic and protruding tongue lead to respiratory and orthodontic disorders and speech, suck, chew and swallow problems [23].

To improve the oral health of people with DS, health programs must incorporate intervention methods to prevent and treat malocclusion as early as possible. Therefore, there's a need to do a complete radiographic examination, an early mixed dentition orthodontic examination and an airway assessment, including consideration of tonsillectomy, palate expansion and tongue crib appliances. An occupational therapy is also important to strengthen orofacial musculature [9,23,31].

#### **Dental anomalies**

Dental anomalies are very common, both in the primary and permanent teeth and occur with an incidence five times greater in DS individuals than in general population [1,6].

Abnormalities in the number (fewer), size (smaller) and morphology and the timing of their development (late dentition) are constant features of this syndrome. In the primary dentition, the most commonly absent teeth are lateral incisors, while in the permanent dentition, third molars, second premolars and lateral incisors, in this sequence, are the most frequently missing teeth [6].

Patients with DS have complete tooth mineralization,

delayed tooth eruption (six to eighteen months) [1,27] changes in the sequence of eruption (mainly of the temporary teeth), high incidence of impacted teeth (incisors and canines) and teeth agenesis [1,6,9]. Microdontia, enamel hypoplasia, hypodontia of deciduous teeth and oligodontia are the most common dental anomalies [1,9]. Structural abnormalities include taurodontia, peg-shaped teeth, fusion and gemination. Canines are the most affected regarding shape and size [1,6].

#### **Parafunctional habits**

Bruxism is quite common in this population, since a very young age, and often persists throughout life [1]. The different ages studied may be one of the causes of the different results. Patients with DS have chronic anxiety, underdeveloped nervous system, malocclusion and TMJ dysfunction due to hypotonicity, hyperflexibility and laxity of the supporting ligaments, leading to an increased frequency of bruxism [27].

Initially it creates an erosion of the pits and fissures of the occlusal surfaces (that become smoother), enabling self-cleaning with tongue and facilitating oral hygiene. On the other hand it can lead to an overloading of the supporting tissues and subsequent teeth fractures [1].

These patients should be monitored through a regular program to allow an early diagnosis of the problems related to bruxism. In cases where bruxism is diagnosed it is necessary to reposition the jaw and to decrease teeth grinding. Unfortunately, patients with a severe bruxism are the ones with more neurological problems and the treatment may not be successful [1].

#### **Oral diseases**

**Caries:** The majority of the published studies has reported that patients with DS have lower rates of caries than those without DS [27,32,33]. The literature attributes the low prevalence of caries in individuals with DS to factors such as: eruptive pattern (teeth erupt later and so they are exposed to caries' etiological factors for less time); high prevalence of bruxism (flatter occlusal surfaces facilitate self-cleaning and oral hygiene, eliminating food debris that could be adhered to the sulcus and serve as a substrate for oral bacteria); dental morphology (microdontic teeth and diastema allow an early detection of caries with a simple clinical examination and without a radiological examination); salivary composition and differences in the composition of the microbiota (saliva buffer capacity of the individuals with DS appears to be higher when compared to general population); visit the dentist early in life (these children have several health problems and their parents seem to be easily warned of the oral risk factors) [1,15,20,27,32-34].

**Periodontal disease:** Periodontal disease is the most significant oral health problem in people with DS. Manual

dexterity difficulties may lead to a poor oral hygiene. Plaque and debris accumulation, gingivitis and periodontal disease are common. Consequently, a large number of young people with DS lose their permanent anterior teeth in their early teens [15,20].

There is also a high incidence of aphthous ulcers, oral candidiasis and acute ulcerative gingivitis [35].

The increased incidence of periodontal disease can be explained by the muscular hypotonicity and its consequences, dento-alveolar joint laxity, lack of understanding of the needs of oral hygiene, impaired dexterity, compromised immune system, low T cells count and leukocyte dysfunction [15,20,35,36].

Difficulty in gargling and swallowing, associated with a poor chewing ability reduces the natural teeth cleaning. Consequently, patients with this syndrome have halitosis, discomfort during chewing and early loss of permanent teeth [1,15].

Gingivitis and periodontal disease start early in life and severity of these diseases increases with age. The prevalence of periodontal disease in adolescents with DS is 30% to 40%. In individuals in their thirties the incidence rises up to nearly 100% [37-39].

Cichon et al. suggested that severe periodontal destruction that occurs in individuals with DS is compatible with aggressive periodontitis [39].

### **Conclusion**

Patients with Down syndrome present peculiar orofacial features that, when not corrected, may interfere with their physical, psychological and social development.

Children with this syndrome have a high risk of developing malocclusion and periodontal problems and these should be the main concerns in their treatment needs.

When planning the dental treatment of patients with Down syndrome, dental practitioners should always consider their general health, in order to achieve a holistic and interdisciplinary approach.

### **Contribution of each author**

Viviana Macho: responsible for the conception and design; Viviana Macho and Ana Coelho were responsible for the data collection and manuscript redaction;

Ana Coelho, Cristina Areias and Paula Macedo were responsible for the critical revision of its contents;

David de Andrade was responsible for the critical revision of its intellectual contents and final approval of the version to be published.

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