Dental and skeletal characteristics of patients with Down Syndrome

Aspectos dentários e esqueléticos de pacientes com a Síndrome de Down

Fernanda Gumes de FARIA[1]
Roberta Andrade LAURIA[2]
Marcos Alan Vieira BITTENCOURT[1]

ABSTRACT

Down syndrome, also known as Trisomy 21, is a very common genetic alteration, which has very apparent clinical, dental and facial characteristics and a wide range of systemic manifestations. These patients have their physical development delayed and exhibit changes in normal bone growth pattern. One method to perform this analysis is by associating skeletal and chronological ages through the evaluation of ossification centers in hand and wrist radiographs. In general, a poor development of the midface occurs, contributing to a high rate of malocclusion. However, there are some individual variations. This article, therefore, aimed to conduct a brief review of some of the features of Down syndrome that play an important role in establishing correct clinical diagnosis. We also sought to make a comparative analysis of the skeletal and dental patterns of two patients with Down syndrome through clinical evaluation and using panoramic and hand and wrist radiographs. It was found that, while younger patients (12 years 3 months) displayed bone and skeletal growth retardation, chronologically older subjects (14 years) had their development accelerated.

***Indexing terms**: Bone development. Down syndrome. Malocclusion.*

RESUMO

A síndrome de Down, também conhecida como Trissomia do 21, é uma alteração genética muito comum, que apresenta características clínicas, dentárias e faciais bastante evidentes, além de diversas manifestações sistêmicas. Os portadores desta anomalia possuem retardo no desenvolvimento físico e alterações no padrão normal de crescimento ósseo. Uma forma de fazer esta análise é associar as idades cronológica e esquelética, por meio da avaliação dos centros de ossificação em radiografia de mão e punho. De modo geral, um pobre desenvolvimento do terço médio da face está presente, o que contribui para o elevado índice de maloclusões. Contudo, existem algumas variações individuais. Assim, neste artigo, teve-se como objetivo fazer uma breve revisão acerca de algumas características da síndrome, importantes para que o cirurgião-dentista estabeleça um correto diagnóstico clínico. Procurou-se, também, fazer uma análise comparativa dos padrões dentários e esqueléticos de dois pacientes portadores da síndrome de Down, através da avaliação clínica e por meio de radiografias panorâmica e de mão e punho. Evidenciou-se retardo nos crescimentos ósseo e esquelético no paciente mais jovem (12 anos 3 meses), enquanto o que apresentava maior idade cronológica (14 anos) registrou uma aceleração em seu desenvolvimento.

Termos de indexação: Desenvolvimento ósseo. Síndrome de Down. Má oclusão.

INTRODUCTION

Amongst the congenital defects which have aroused most clinical and scientific interest is Down syndrome, whose most common manifestations were first described in 1866 by John Langdon Down, for whom the condition is now known[1]. Desai & Flanagan[2] stated that Down syndrome is also known as Trisomy 21 because approximately 95% of affected individuals have an extra chromosome 21, thus having 47 chromosomes in total. They added that other chromosome abnormalities include translocation (3% of individuals) and mosaicism (2%). According to Kaye et al.[3], this syndrome occurs in around one in every 600 to 1,500 live births. It is estimated that Brazil has some 300,000 individuals with Down syndrome[4]. Fiske & Shafik[5] state, for reasons which are not completely understood, the chance of a woman having an affected child increases with age, particularly above the age of 35, whereas the age of the father is less significant.

De Mari[6] stated that, based on data available at the time, the life expectancy of Down syndrome patients leaped from 10 years in the 1920s to 60 years, which is relatively close to that of the Brazilian population in general.

The diagnosis of Down syndrome may be carried out on the basis of clinical signs arising from the vast array of disturbances and anomalies found[1]. It is common to find retardation in pre- and post-natal development, as well as microcephaly, a flattened face, small nose, depressed nasal

[1] Faculdade Bahiana para o Desenvolvimento das Ciências, Salvador, BA, Brasil.
[2] Universidade Federal da Bahia, Faculdade de Odontologia, Departamento de Ortodontia. Rua Augusto Viana, Canela, 40110-909, Salvador, BA, Brasil. Correspondência para / *Correspondence to*: MAV BITTENCOURT. *E-mail*: <alan_orto@yahoo.com.br>.

bridge and a short, wide neck. The epicanthic folds and the slanting fissure with high external angles give an almond-shaped appearance to the eyes. The ears are dysplastic, small, low-set and with a deformed helix. Additionally, in the majority of cases, there is a delay in starting to speak, which only occurs at around four years of age, on average, slowing the pace of learning to read and write[6].

According to Oliveira et al.[7], dental anomalies are also quite frequent, occurring in 44% of sufferers investigated. Most evident are the congenital absences, microdontia, retarded root formation, enamel hypoplasia and fusion[1]. Desai & Flanagan[2] observed that the teeth most affected by these absences are the lower central incisors, followed in order of magnitude by the upper lateral incisors, second upper and lower premolars, canines and, in rare cases, the molars. Following the chronological pattern of physical development, they present with a delay in dental eruption, as well as altered sequences[2], in both dentitions[1].

As far as caries is concerned, it was noted that there is a reduced risk in these patients, in both the deciduous and permanent dentitions[8]. The authors attribute the low risk to a number of factors such as the increase in the pH of the saliva and the level of bicarbonate present, the reduction in the number of Streptococos mutans and even the morphology of the teeth, which present with few pits and fissures and with flatter surfaces due to the bruxism which is usually present. The opposite is the case when it is a question of periodontal disease, which is present in almost all individuals afflicted by the Syndrome, which is due far more to the defective immune system than to poor dental hygiene[9-11]. Due to this impairment, Pilcher[12] reports that studies show the incidence of periodontal disease in between 90% and 96% of adults suffering with the syndrome. She also adds that there is a high prevalence of acute necrotizing ulcerative gingivitis (ANUG) as well as aphthous ulcers and oral candidiasis.

There is consensus in the literature that these individuals have a brachycephalic facial pattern[5,9,13], where the occipital bone presents in flattened form and with a prominent fontanelle[9,14]. As far as these authors are concerned, there is a third or fourth fontanelle present, all of which are spacious and open for an extended period of time. Moreover, they state that the frontal and sphenoidal sinuses are absent and the maxillary sinus is hypoplastic in 90% of patients. One fairly common finding is the poor development of the midface, causing relative mandibular prognathism[5,9,15]. Consequently, patients usually present with Angle class III malocclusions[1]. Oliveira et al.[7] found a high prevalence of anterior crossbite (33%), posterior crossbite (31%) and anterior open bite (21%), in 100 individuals with Down syndrome.

The deficient development of the midface also causes atresia of the palate[16]. On occasions, bifurcated uvulas and cleft lips or palates may occur[9]. Due to the narrowing of the nasopharynx and the hypertrophied tonsils and adenoids, there is an impact on the passage of air through the upper airways, leading to inadequate lip posture[9]. Consequently, they are chronic mouth breathers[11], presenting with labial incompetence when at rest, aggravated by a true, or relative, macroglossia where the tongue juts out[9]. The amount of space to accommodate the tongue is therefore reduced, impacting speech and chewing[12].

The chewing and facial muscles are seen to be hypotonic[16]. Pilcher[12] states that this, associated with mouth breathing and the lingual posture, contributes to the disequilibrium in the muscular forces between lip and tongue, which constitute decisive factors in the development of anterior open bite.

Sannomiya et al.[17] conducted a study to evaluate bone age in individuals with Down syndrome by means of x-rays of the hand and wrist, selecting 81 children in the 6 to 15 age range. They found that the bone age was close to chronological age in the group of children between 6 and 10 years old. In the group of children between the ages of 10 and 13, bone age was seen to be approximately 12 months ahead of chronological age in females, while in males this was delayed. In the 13 to 15 age group, bone age was more advanced than the chronological age in both genders.

Kushnick[18] emphasized that the centers of ossification in mongoloid children are normal at birth, however over time, bone age usually becomes retarded. Pozsonyi et al.[19] stated that there is a delay in skeletal maturation up to 8 years of age, though subsequently patterns are assumed that are consistent with normal children. The authors also noted that skeletal maturation stops at around 15 years of chronological age in mongoloid children while, for normal individuals, it is expected to continue until the age of 18.

The aim of this study is to perform a comparative analysis of dental and skeletal patterns in two patients with Down syndrome, by means of a clinical examination and x-rays, and comparing the characteristics obtained with data in the literature.

CASE REPORT

Case 1

Male patient aged 14 with Down syndrome, presenting with flattened face, small nose, depressed nasal bridge, increased intercanthal distance, prominent, slanting eyes, with a convex palpebral fissure. The facial pattern is of the mesocephalic type with a straight profile (Figure 1).

From a dental perspective, he falls within the permanent dentition phase with all teeth erupted, with the exception of the third upper and lower molars which are still in Nolla development stages 8 to 9 (Figure 2). According to the hand and wrist x-ray, the osteocartilaginous development shows an age of 16 (Figure 2).

In both arches, eight rotated teeth can be observed as well as severe crowding, especially in the anterior region, and linguoversion of the posterior teeth in the lower arch. He suffers from Angle Class II malocclusion, presenting with posterior crossbite between teeth 16 and 46 and exaggerated overbite. He has a V-shaped atresia of the palate and non-coincident midlines (Figure 3).

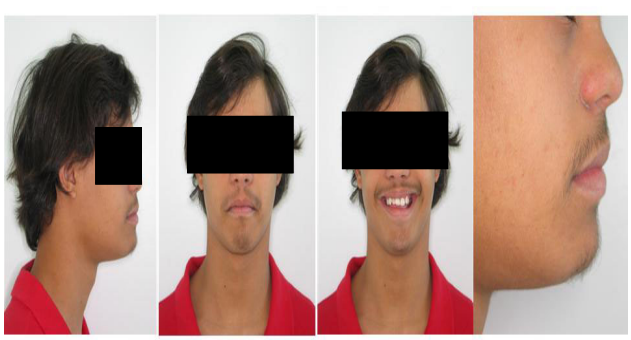

Figure 1. Facial appearance of patient 1.

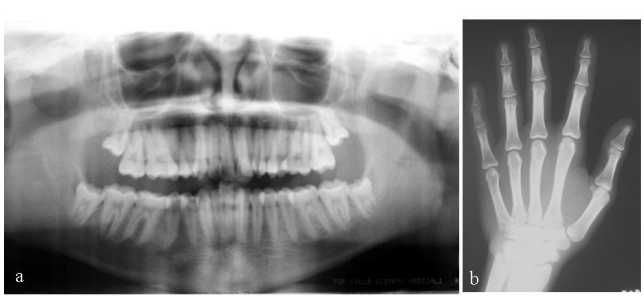

Figure 2. A) Panoramic radiograph of patient 1. Note that all the permanent teeth have erupted except for the third molars; B) Hand and wrist radiograph of patient 1, presenting with calcification of the epiphyses with diaphysis on the middle finger and the presence of a sesamoid.

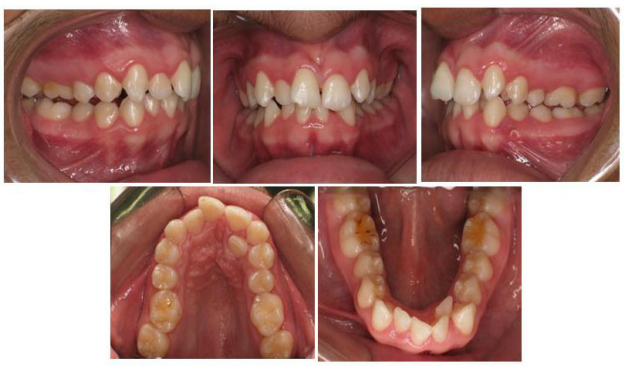

Figure 3. Intraoral photographs of patient 1 in centric occlusion and upper and lower occlusals.

Case 2

Male patient aged 12 years and 3 months, also with Down syndrome, presents with several typical alterations of the anomaly, such as small nose with depressed nasal bridge, increased intercanthal distance and protruding eyes. He has a mesocephalic facial pattern, convex profile and difficulty with labial sealing (Figure 4).

He is in the mixed dentition phase, more precisely in the second transitional period with deciduous canines and molars still present (Figure 5). According to the analysis of the hand and wrist x-ray (Figure 5), his bone age is around 11.

A number of dental anomalies can be observed, such as the absence of tooth 12 and microdontia in tooth 22. The patient suffers from Angle Class I malocclusion, with diastemas in the upper arch and lingual inclination of the posterior teeth, particularly in the lower arch (Figure 6).

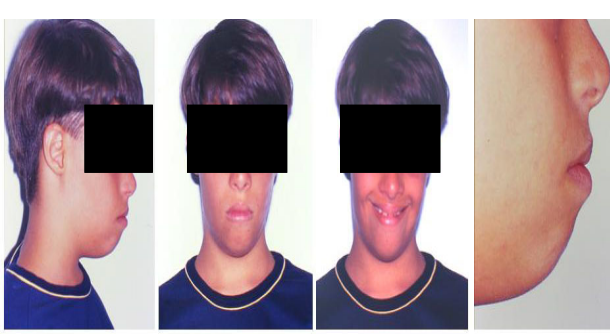

Figure 4. Facial appearance of patient 2.

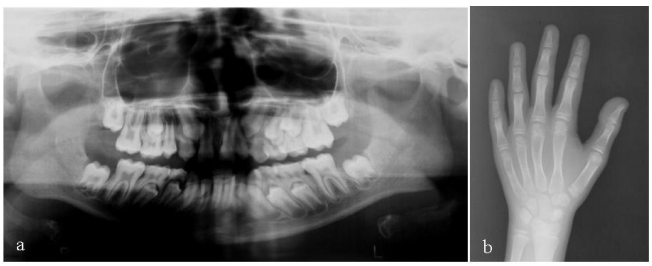

Figure 5. A) Panoramic radiograph of patient 2. Note the presence of deciduous canines and molars and the absence of tooth 12; B) Hand and wrist radiograph of patient 2, with as yet uncalcified epiphysis and diaphysis on the middle finger and without the sesamoid.

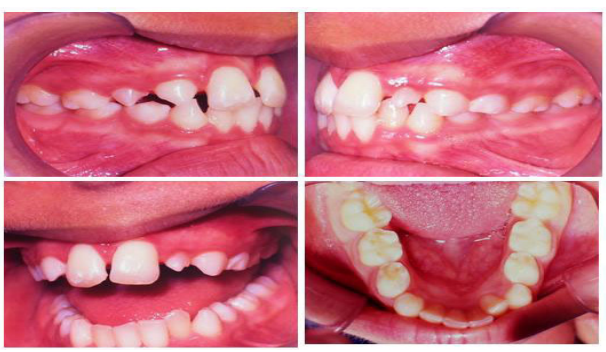

Figure 6. Intraoral photographs of patient 2 in centric occlusion and lower occlusal. sesamoid.

DISCUSSION

Genetic content, as reported in the literature, is one of the factors responsible for the morpho-functional determinism of individuals. Down syndrome, also known as trisomy 21, is one of the most frequently reported genetic alterations. Its occurrence causes the development of individuals with peculiar appearance and similar morphological characteristics, pattern of development and behavior. Therefore, the previously described facial appearance, such as flattened cheeks, small nose, depressed nasal bridge, wide gap between protruding, slanting eyes, are typical of patients with this anomaly[1].

Patient 1 has good labial sealing in repose as well as an erect profile. However, the second patient suffers from a convex profile and the absence of labial sealing in repose. Mouth breathing, the presence of deleterious habits and hypotonicity of the perioral muscles, may be associated with this feature[7,20].

The mesocephalic facial pattern of the two patients differs from reports in the literature, where the brachycephalic aspect of the skulls is added to the reduction in their bone base, which produces individuals with lower total facial height in comparison with normal individuals[21].

Oral manifestations involve certain peculiar characteristics such as agenesis and alteration to the positioning and shape of the teeth[2,5,7,12,22]. According to Vittek et al.[23] and Ondarza et al.[22], the frequency of these anomalies is significantly higher in individuals with this syndrome, when compared to individuals with other types of deficiency as well as being greater than in normal individuals. According to Desai & Flanagan[2], agenesis and defects in development are ten times more common in patients with Down syndrome than in the overall population. In patient 2, one can observe the absence of tooth 12 and microdontia in tooth 22, which is also reported quite frequently, as 35% to 55% of individuals with Down syndrome present with microdontia in the deciduous or permanent dentition2. The presence of these alterations contributes to a positive dental discrepancy, with generalized diastemas in the upper arch. In the lower arch, the posterior teeth (deciduous molars) are seen to be lingualized, which was also noted by Ondarza et al.[22].

As for patient 1, he did not present with these dental anomalies, however he did exhibit a number of teeth with some form of positional alteration. The linguoversion of tooth 16 caused a crossover between this and its antagonist. There is some controversy in the literature as to the most prevalent type of malocclusion. Vittek et al.[23] found, in their sample of patients with syndromes, a total of 53.7% who suffered from Angle Class I malocclusion while Desai & Flanagan[2] considered Class III to be the most prevalent, occurring in 65% of the evaluated population.

Children with Down syndrome present a greater tendency to acquire abnormal sucking habits, mainly finger sucking, contributing to 69% of cases of anterior open bite[2]. This habit and malocclusion were found in patient 2, however they were absent in patient 1. Moreover, these individuals usually present with a narrow V-shaped palate resulting from the hypodevelopment of the midface[2,16], a feature that is present in both patients.

As for dental development, a big difference can be seen between the two patients. Patient 2 is in the mixed dentition phase, more accurately in the second transitional period. According to Proffit[24], a dental age of 12 is characterized by the presence of all permanent teeth already erupted. Therefore this patient presents with a small delay between chronological age (12) and dental age, as his upper and lower canines and premolars are still in the phase of eruption, which is characteristic of an 11 year-old individual. Patient 1, on the other hand, at 14 years of age, has all the permanent teeth erupted and well-

formed apices, except for the third molars, which are stil. l being formed, in Nolla stages 8 to 9[25]. So it may be said that, according to Proffit[24], he has a dental age consistent with an individual of 20 years of age. In relation to skeletal development, however, according to the analysis of the osteocartilaginous development of the hands and wrists, patient 1 exhibits a skeletal age of 16. On the other hand, patient 2 has bone development compatible with an individual aged 11 years and 3 months, an age range in which his dental development is also situated[24], i.e. delayed in relation to his chronological age.

It can be seen therefore that there is a big discrepancy between the two cases. Patient 2, younger, at 12 years of age, has delayed dental and bone ages while patient 1, aged 14, shows accelerated growth. A similar situation transpired in a study conducted by Sannomiya et al.[17], in which, upon evaluating bone age in individuals with Down syndrome by means of hand and wrist x-rays in a selection of 81 children in the 6 to 15 age range, he found that in the 10 to 13 age range in males, bone age was seen to be approximately 12 months behind chronological age. In the 13 to 15 age group, however, bone age was seen to be ahead of chronological age, in both sexes.

CONCLUSION

Although the facial, dental and skeletal characteristics of individuals with Down syndrome are quite peculiar, it can be seen that, between the two cases reported, there are still great differences. Bone age, obtained via x-rays of the hands and wrist, subsequently assumes patterns consistent with normal individuals. This might explain the fact that patient 2 (12 years and 3 months) demonstrated a delay in development, unlike patient 1 (aged 14), who had already achieved development beyond that expected for his age.

Collaborators

FG FARIAS took part in the data collection, the detailed description of the clinical cases and the composition of the article. RA LAURIA took part in the discussion, comparison of cases and the composition of the article. MAV BITTENCOURT directed the research and took part in the composition of the article.

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