Down´s Syndrome – Aspects of interest to dentists

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ABSTRACT

Down's syndrome is a chromosomal problem caused by trisomy 21. Among its various alterations, mental retardation and morphofunctional disturbs, with corresponding physical appearance. The purpose of this paper is to review the literature on the etiology, general and oral characteristics, emphasizing the important aspect to dentists. This will help the professional to deal with his patients' genetic condition.

Key Words: Down's Syndrome, trisomy, dentistry, oral alterations.

INTRODUCTION

Reports on Downs's syndrome (DS) go back to the 19th century. Thompson et al. (1993) report that Down's syndrome was first described in 1866 by John Langdon Down. However, only in 1932 did a Dutch ophthalmologist propose that this syndrome could be caused by a chromosomic anomaly. A century elapsed between this description and the discovery that in DS the majority of children have 47 chromosomes and that the trisomy was due to an extra acrocentric chromosome – the chromosome 21.

The incidence of DS is 1/800 being greater in newborns or fetuses of mothers age 35 years old or above (Thompson, 1993). This syndrome can be caused by three types of chromosomic alterations: free trisomy, mosaicism and translocation. The exceeding chromosomic material has paternal origin in 20% of the cases, being the remaining maternal (Thompson, 1993; Kumasaka, 1997; Mustachi & Peres, 1999; Schwartzman, 1999).

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Cases of free trisomy are caused by non-disjunction of the chromosome 21. Non-disjunction is a phenomenon in which there is no segregation of the homologue chromosomes in the first meiotic division or of the sister chromatides in the second meiotic division or in the mitosis, which alters the number of chromosomes. The probability of this defect attaining other children is very low. It is during the ovogenesis that the majority of these cases occur, which is favored by older ovocytes. However, non-disjunction may occur also in the spermatogenesis or in the first cleavage of a normal zygote. Free trisomy is present in around 95% of syndromic individuals. In women younger than 30 years there is a 0.2% risk of generating a baby with DS. In women above 45 the risk is 4%. In the mosaic trisomy the person has two cell progeny, normal and trisomic. This alteration is due to a post-zygotic non-disjunction and occurs in circa 2% of cases of DS (Thompson, 1993; Mustachi & Peres, 1999).

Usually, cases of translocation are transmitted by young and normal mothers and account for 3% of the affected cases. The analysis of the cariotype of these mothers points to an overlap of a segment that refers to the material of the chromosome 21 in the chromosomes of groups D (13, 14 or 15) or G (21 or 22). In this way, the translocation is balanced and thus the woman is normal. Translocation is the change of segment among non-homologue chromosomes. There are two main types: reciprocate and robertsonian. Reciprocate translocation results in a break in the non-homologue chromosomes with a mutual interchange of segments, without alteration in the number of chromosomes. Robertsonian translocation involves two acrocentric chromosomes that fuse near to the region of the centromer, loosing the short arm. The bearer of a translocation is a normal person who nevertheless presents a risk for the decendency due to the formation of non-balanced gametes (Thompson, 1993; Mustachi & Peres, 1999).

The difference between free trisomy of the 21 and translocation is that the latter is not related to the mother's age although showing a high risk of recurrence when one of the parents is a bearer, mainly the mother. The gamete with translocation, when combined with a normal one, produces three different types in equal proportions. In this case the risk for the decendency is 1:3 (Thompson, 1993; Mustachi & Peres, 1999).

The chromosome may also undergo translocation 21q21q, which is formed by two long arms of the chromosome 21. It is accepted that the 21q21q has origin as an isochromosome, that is, a chromosome in which one of the arms is absent and the other is duplicated. This is a rare anomaly in which the individual can form two types of gametes (i) with the chromosome 21q21q in double dosage of the genetic material of the chromosome 21, which will lead to a case of DS or (ii) absence of the chromosome 21 leading to a monosomy of the chromosome 21 which is lethal. Therefore, all individuals with isochromosome will not have normal children (Thompson, 1993; Mustachi & Peres, 1999).

The aim of this article is to increase the awareness of dentist to the general and oral characteristics of Down's Syndrome. In this regard these professionals would be apt to provide adequate care to individuals with this syndrome.

GENERAL CHARACTERISTICS

Down's syndrome can be diagnosed early due to its dismorphic characteristics. The facial features are typical and the clinical signs are due to a delay on the pre and postnatal development. Mental retardation is the most severe consequence. Muscular hypotony is one of the first anomalies observed in the newborn. Patients show reduced height, oligofreny, brachicephaly and mild microcephaly. The skull wall is thin and there is a delay in the closure of the fontanel. The neck is short and wide, being the skin loose in it posterior region. The nasal bridge is flat with a tendency for internal epicanthus and ears have a low implantation presenting a characteristic fold. In the eyes, there are spots in the iris (Brushfiel spots), hypoplasia of the peripheral zone of the iris, internal epicanthal folds and dull crystalline. The metacarpals and phalangeal bones in the hand are short and widened and there is a transversal palmar crease (simian crease) and chlinodactily in the little finger. In all fingers the dermal crests show ulnar loop and the axial palmar triradium is in a distal position. Feet show a wide space between the first and second toes and all dermal crests are in the big toe. There is hypoplasia of the iliac bones, which are widened, and the acetabular angle is short.

Congenital cardiopathy is present in 1/3 of the newborns. The skin is dry and, later on, becomes hyperkeratotic. Hair is thin, silky and scarce in most cases. Pubic hair is straight. Men are sterile and women show hypogonadism and primary amenorrhea. Some of the women may become pregnant but 50% of descendents are affected due to the trisomy. In comparison to other syndromes, DS shows a greater occurrence of duodenal atresia and tracheoesophagical fistula. Risk for leukemia is high as well as for epilepsy and Alzheimer (Thompson, 1993; Sigal & Levini, 1993; Mustachi & Peres, 1999; Schwartzman, 1999, Regezi & Sciubba, 2000).

ORAL CHARACTERISTICS

There are a variety of oral manifestations in DS. They include short mandible and oral cavity; a high, narrow and curved palate; the tongue is fissured and large with the possible occurrence of benign migratory glossitis (geographical tongue) (FIGURE 1). The nasopharynx may be narrow and with hypertrophic tonsils and adenoids. The protrusion of the tongue and the oral breathing leads to dryness and fissures in the lips.

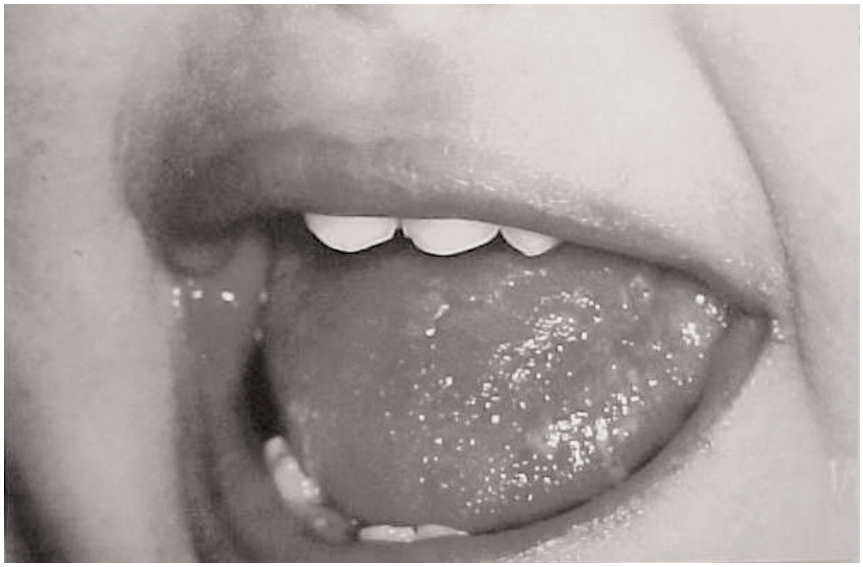

FIGURE 1 – Dorsal surface of the tongue showing small fissures and migratory benign glossitis (lateral side).

There is angular cheilitis in the labial comissures due to the difficulty of closing the lips (FIGURE 2) (Regezi & Sciubba, 2000).

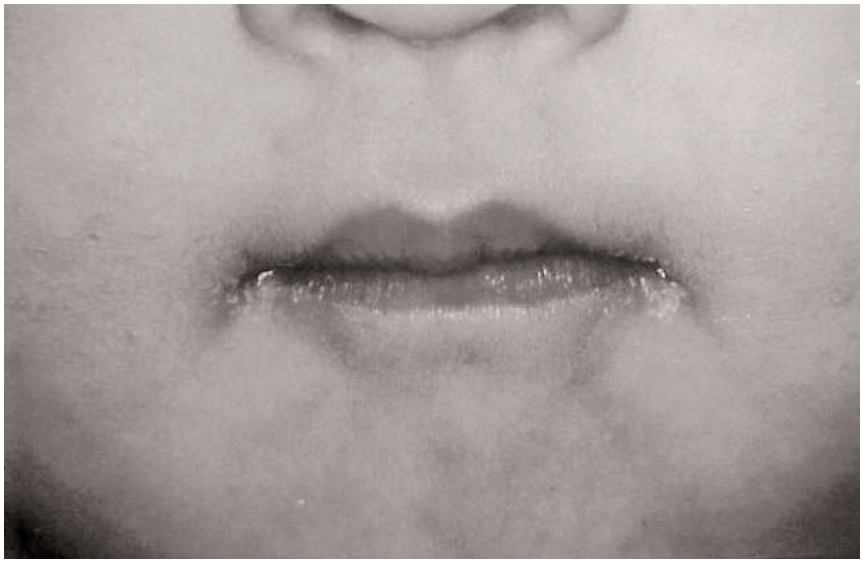

FIGURE 2 – Angular lateral cheilits. Lesions sho wing fissures and erithematous and flaking alterations.

Dentition shows characteristic anomalies and periodontal disease is prevalent. Most frequent anomalies are oligodonty , microdonty, hipodonty, fusion and taurodonty . Hipodonty occur in both dentitions and the microdenty is the most prevalent in the observed alterations. Dental developmental anomalies such as coronal and radicular malfor mation are also common. Morphometric alterations in the deciduous second molars are

frequent among DS patients. (Townsend, 1983; Townsend & Brown, 1983; Brown & Townsend, 1984; Peretz et al., 1996; Peretz et al., 1999).

Bell et al. (1989) e Alpöz & Eronat (1997) conducted X ray examinations and evaluated the prevalence of taurodonty in the inferior molars of children with DS. Results showed that the incidence of taurodonty is 36.4% e 66%, respectively. Rajic; Metrovic (1998) studied 43 patients with DS for taurodontic teeth, its intensity , distribution by sex and involved tooth. The incidence was 55.8%, being 32.6% in females and 23.2% in male. The second molar was the most affected (54.2%) followed by the first molar (40.0%) and the third (6.5%). In relation to the form, the mesotaurodontic was most frequent (72.5%). Hypotaurodonty was observed in 9.7% and the pyramidal tooth in 17.7% among the studied cases.

Mestrovic et al. (1998) reported that in a group of 112 patients with DS hipodonty was observed in 38.6%. The lateral superior incisive was the most affected tooth. Kumasaka et al. (1997), in a radiographic evaluation of the prevalence of oligodonty in 98 cases with DS, with a control of 150 individual with normal caryotype, found that the frequency of this alteration in permanent teeth was respectively 63% and 17%. Most frequent involved teeth were the lateral incisive and the premolars, both superior and inferior. These results are similar to those previously reported by Russel & Kjaer (1995), as this alteration occurred in 81% of the cases.

Occlusal disharmonies, posterior cross bite, aperognaty and marked crowding of teeth are common among these patients (Reuland-Bosma & Van Dijk, 1986, Bell et al.,1989; Alpöz & Eronat, 1997; Mestrovic et al., 1998; Regezi & Sciubba, 2000). The high level of malocclusion is more frequently related to the superior arch, particularly in the area of the incisives and canines. According to Ondarza et al. (1995), this characteristic is due to small and ogival maxillar arch, associated to macroglossy and other exogenous factors (FIGURE 3).

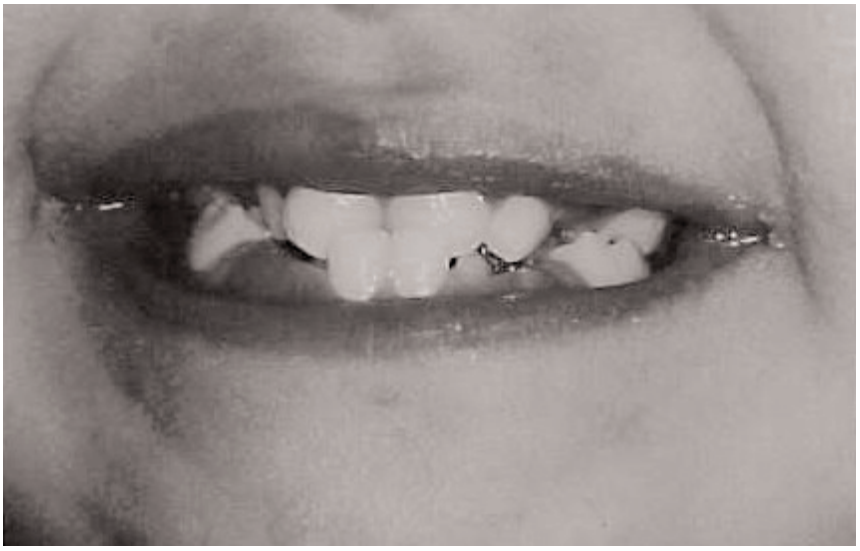

FIGURE 3 – Occlusion of a patient 2 years and 6 months old. Notice the presence of a false Class III.

Delay in dental eruption is a common finding among DS patients. It usually occurs at the 6th month and is present both in the deciduous and permanent dentition (Reuland-Bosma & Van Dijk, 1986; P eretz et al., 1996; Ondarza et al. 1997; Mustachi, 1999; Schwartzman, 1999; Regezi & Sciubba, 2000).

Salivary flow in patients with DS is 50% lo wer than in nor mal children; such reduction is related to the metabolism of the parotid. Additionally, the pH is higher as w ell as the le vels of sodium, calcium and bicarbonate. Therefore, the b uffer condition is high, w hich could lead to a lo w incidence of ca vities. However, the incidence of caries seems not to be higher than in nor mal children. Recently, Gabre et al. (2001) conducted a longitudinal study on incidence and pre valence of caries, teeth mor tality and inter proximal bone loss in 124 adults with mental retardation (including DS cases) for 8.5 y ears. The results revealed that the incidence and pre valence of caries in indi viduals with DS is lower than the incidence and pre valence in the remaining g roups. However, these later show a high level of bone loss.

On the other hand, DS patients show a signif icant tendency to periodontal disease, which increases with age (Reuland-Bosma & Van Dijk, 1986, Ulseth et al., 1991; Morinushi et al. 1997; Cichon et al., 1998; Agholme et. Al., 1999; Gabre et al. 2001). There is earl y periodontal involvement with extensive inflammation of the gum, w hich progresses rapidly if compared to normal individuals. The inferior quadrant (region of the incisive) is more prone than the superior (re gion of the molars). Reuland-Bosma et al. (1988) studied the mor phological aspects of the gum tissue of children with DS, as w ell as the histolo gical alteration of this tissue during the development of the dental plaque. They also investigated whether these str uctural alterations w ere related to the clinical features of the periodontal disease. The results re vealed that the histo - logic structure in syndromic children w as similar to the control g roup, although the de velopment of gingival inflammation appeared earlier (7 days in DS and 14 da ys in the control g roup) and showing a rapid progression. According to these authors, these f indings could be due to a more virulent bacterian plaque and/or the host response. Exogenous factors related to the de velopment of the periodontal disease in children with DS include lack of oral h ygiene, presence of materia alba and calculus, malocclusion, macro glossy and the f act that the mouth remains not fully closed. The ulcerative necrotic acute gingi vitis (GUNA) and bruxism are other related alterations to DS children (Reuland-Bosma & Van Dijk, 1986; Reuland-Bosma et al., 1988; Gabre et al. 2001).

DISCUSSION

The care pro vided to DS cases is quite limited. The uncoordinated action among health professionals results in poor preparation and e ven

absolute lack of knowledge on how to deal with these individuals. Once aware of the general, bucal and behavioral characteristics of these patients, the dentist is able to act with readiness and competence. In most instances there is fear and prejudice in regards to treating DS cases. Only in rare cases do some patients need special care such as procedures under general anesthesia and control of convulsions. Most of the time, these patients are docile, easy to manipulate and quite cooperative despite their own limitations. Therefore, the reaction of some dentists may be due to an inadequate preparation in the Dentistry School to cope with these special patients.

Some authors have raised the question whether the susceptibility to periodontal disease in DS individuals is related to morphological modifications in the gingival epithelium, such as a discrete anoxia due to poor local circulation or some morphological alteration in the microcirculation. According to Morinushi et al. (1997), Cichon et al. (1998), Agholme et. al. (1999) and Gabre et al. (2001), the increase in bone loss due to periodontal disease in DS individuals should not only be attributed to a poor bucal cleansing but also to immunological modifications. Cichon et al. (1998) refer that the patter n of the periodontal disease in DS is similar to that of juvenile periodontitis. In a study by Gabre et al. (2001), among individuals that have lost 6 or more teeth due to periodontal disease, 44% have Down's Syndrome.

An interesting fact reported by Reuland-Bosma & Van Dijk, in 1986, is that DS children living in nursing homes showed a higher index of calculus if compared to those living with their own parents. Ulseth et al. (1991) e Gabre et al. (2001) do not agree with these results. In the studies of these authors, the results indicate a reduction of the periodontal disease in individuals under care in nursing homes. Diet and hygienic habits are factors to be taken into consideration in this differentiation. Additionally, it is important to note that institutions have a multidisciplinar team and an adequate physical structure, providing a better care to these individuals.

In general, it may be said that it is not possible to correlate the occurrence of calculus and bacterian plaque to the severity of periodontal disease.

Although the literature states that DS patients show low incidence of caries, this assertive may be questioned. In the studies, it is necessary to take into consideration the analyses of the total number of teeth in the oral cavity. If one takes into consideration the number of caries in relation to the number of teeth, the difference in the incidence of caries among normal individuals and syndromic patients disappears or becomes hardly significant (Reuland-Bosma & Van Dijk, 1986; Schwartzman, 1999, Gabre et al., 2001). In this conception, in comparative studies one should take into consideration the absolute number of teeth in the DS group, once the eruption of teeth in these patients is delayed, this could lead to false results. According to Gabre et al. (2001), another factor to be considered in the low index of caries in these cases

is the use of an ideal concentration of fluoride in the water supplied to these patients. In the study by these authors, they call special attention to DS patients, once they consider the high risk for poor bucal cleansing.

Associated to these factors one should stress that, in the response to inflammatory and infectious process in the oral cavity of DS patients, some aspects are important in the evolution and repair of these occurrences, namely: alteration in the function of polymorphonuclears and monocytes, mainly in the chemotaxy and opsonization phase; atypical pattern of T cells and modification in the biosynthesis of collagen (Reuland-Bosma & Van Dijk, 1986, Reuland-Bosma et al., 1988).

The morphometric variations observed in DS patients include alteration in the shape and size of teeth, represented by an increase in the intercuspidal distance and modification in the external diameter of teeth (MD and VL). According to the literature there is a delay in the proliferative activity of cell in charge for the development of teeth, posterior to an acceleration of the initial cellular activity (Townsend, 1983; Townsend & Brown, 1983). This leads to the formation of greater deciduous teeth and smaller permanent ones. The main morphological alterations observed in the permanent first molars are located in the distal oclusal portion of the crown characterized by a significant reduction of the size of distal and distal lingual cusp (Brown & Townsend, 1984). Some authors associated the morphological variations in the permanent inferior incisive with a predisposition to develop severe periodontal disease in this region (Townsend & Brown, 1983; Reuland-Bosma & Van Dijk, 1986). The teeth are smaller with a coronal mesial diameter greater that the vestibulo lingual in comparison to teeth in patients whit normal caryotype (Townsend & Brown, 1983). Furthermore, the incisives may show a conic shape with small roots, which could speed up the loss of bone support (Reuland-Bosma & Van Dijk, 1986). The study conducted by Agholme et al. (1999), in which was made a evaluation of the periodontal condition of patients with DS for a period of 7 years, confirmed that the area of greater loss of alveolar bone in that of the inferior incisives.

In these individuals, teeth more frequently involved by dental anomalies are the lateral incisives and molars. More prevalent alteration in molars is taurodonty and anodonty and microdonty in the incisives. Taurodontic teeth deserve some care during endodontic treatment due to the modification of the root canal system, making dificult the biomechanics and the obturation of these canals. With the advance of dental cosmetics, the alteration in the incisives can be easily corrected or ameliorated. The fact to be considered is the adequate choice of restorative material since the cleansing in these patients is not satisfactory.

Since the tongue is fissurated and there is macroglossy, cleansing of the tongue should be emphasized, aiming the reduction of retention of food and bacterian locus. In more severe cases of DS the cleansing by the patient, in most cases, is almost impossible and parents should be aware of that. Education of parents should be made in a clear and objec-

tive way with follow-up to ascertain that the instructions are being followed. The correction of occlusal disharmonies by orthodontic treatment is not always possible. It depends on each case and on the cooperation of the patient.

Elderly people with DS may develop Alzheimer. This association will lead to a motor deficiency with marked interference in the bucal cleansing, leading to an increase in the rate of caries and periodontal disease, halitosis and bucal infections (Sigal & Levine, 1993; Regezi & Sciubba, 2000). Periodontal disease is the most common cause of early loss of teeth. All these factors will lead to a worsening in the general health condition of the individual and, thus, in his/her quality of life. The increased risk for developing leukemia in these patients is probably due to the fact that this disease is causes by a combination of environmental and genetic factors. Some types show specific chromosomic anomalies. The acute lynphocytary leukemia is an example, showing an increased incidence in DS patients (Regezi & Sciubba, 2000).

CONCLUSION

Trisomy 21, which causes DS, is the autosomic aneuploidy more commonly found in newborns. Circa 95% of DS cases are caused by non-disjunction, 3% by translocation and 2% by mosaicism. In the later, the phenotype is mild. Although there is a variety of general and oral characteristics in DS cases, there is a marked presence of characteristics that helps the clinician to set the diagnosis.

The most frequent oral alterations are: macroglossy, fissured tongue, angular cheilitis, retardation in the dental eruption, dental anomalies (hypodonty, microdenty and taurodonty) and a trend to develop cavities and, mainly, periodontal disease.

Undoubtedly, periodontal disease is the most important alteration since it leads to early loss of teeth. The family support is essential to any dental treatment. If early preventive care is provided, these cases may have an adequate oral health and a better quality of life.

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