Malocclusion in Down syndrome - a review

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D Rao,[1] S Hegde,[2] S Naik,[3] P Shetty[4]

ABSTRACT

Down syndrome or Trisomy 21 is a genetic disorder, which results in intellectual impairment, typical craniofacial features and a wide spectrum of phenotypic abnormalities. Characteristic features of midface hypoplasia in Down syndrome include smaller maxilla, presence of 'stair' palate and severe malocclusion. Generally, the most frequent malocclusions stem from variations in vertical and transverse occlusions, identified mainly as Angle's Class III molar relations, anterior open bite, anteroposterior crossbite, and proclination of the anterior teeth. By familiarising themselves with these features, general dental professionals, paediatric dentists and orthodontists can plan a varied combination of treatment modalities to prevent and correct occlusal anomalies.

Key words: Down syndrome, malocclusion, review, Trisomy 21

INTRODUCTION

Referred to by various names as, 'trisomy 21' or 'trisomy G', Down syndrome is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21. In fact, it is the most common and well known of all human malformation syndromes, characterized by central growth deficiency, which is evidenced by delayed physical and mental development.[1]

In 1866, John Langdon Down described some of the characteristics of the syndrome which now bears his name. Down syndrome is an easily recognized, congenital, autosomal anomaly characterized by generalized growth and mental deficiency.[2] According to Molteno *et al.*,[3] in 1997, the overall birth prevalence rate of Down syndrome in Cape Town, South Africa, was 1.49 per 1000. Delport *et al.*,[4] documented an incidence of 1.33 per 1000 livebirths in a Pretoria urban academic hospital, and Venter *et al.*,[5] recorded a figure of 2.09 per 1000 livebirths in a rural South African hospital. Kromberg *et al.*,[6] reported an incidence of 1.67 Down syndrome infants per 1000 livebirths at an academic hospital in Johannesburg, South Africa.

1. **D Rao:** *PhD Student.* The Pacific Dental College and Hospital, Pacific University, Udaipur, Rajasthan State, India.
2. **S Hegde:** Professor and Head, Department of Paediatric Dentistry. The Pacific Dental College and Hospital, Pacific University, Udaipur, Rajasthan State, India.
3. **S Naik:** *PhD Student.* The Pacific Dental College and Hospital, Pacific University, Udaipur, Rajasthan State, India.
4. **P Shetty:** Professor, Department of Orthodontics. The Pacific Dental College and Hospital, Pacific University, Udaipur, Rajasthan State, India.

Corresponding author

D Rao:
Pacific Dental College & Hospital, Pacific University, Udaipur. Rajasthan State, India. E-mail: pedodinesh2003@yahoo.co.in.
Tel: +91 9414158235. Fax: +91 294 2491508

It is believed that approximately 96% of all cases of Down syndrome result from meiotic nondisjunction. According to Mattei,[7] nondisjunction may occur during the first and the second meiotic division in either the female or the male patient. Although the syndrome occurs in offspring of mothers of all ages, the risk increases with increasing maternal age, as evidenced by Morris *et al.*,[8] who determined that the chance of birth with Down syndrome is 1 in 1441 when the mother is 20 years of age, one in 959 when the mother is 30 years old, one in 84 at 40 years of age and one in 44 at 50 years or higher. There appears to be no racial, socio-economic or gender predilection. According to three different studies reported from South Africa, the incidence of Down syndrome infants born to the mothers aged 35 years or more was found to be 52%, 56%, and 55%, respectively.[4-6] According to Verma *et al.*,[9] approximately 93% of Down syndrome cases have an extra chromosome 21, making the chromosome count 47 instead of the normal 46. The remainder presented translocation (4%), mosaicism (2.6%) and additional karyotypic abnormalities along with trisomy 21 (0.3%).

In spite of advances in the health care of the subnormal and a gradual increase in the life span of those affected with Down syndrome, the average life expectancy is still 35 years. The periods of highest mortality risk are in infancy when congenital heart disease, leukemia and respiratory diseases are frequent threats and in late adulthood when Alzheimer's disease and declining immunological function are significant factors. There has been a long standing belief that subjects with Down syndrome reach a 'plateau' in adolescence, beyond which further developmental change is not possible. The findings of longitudinal studies illustrate the complex, varied, and changing relationship between persons, their contexts and the effects of experience across the life-span.[10]

CRANIOFACIAL DISCREPANCIES

Trisomy 21 is marked by craniofacial characteristics that make it well defined and readily identified. The features include a brachycephalic cranium, diminished anterior cranial base with a round face, flat cheekbones, flat bridge of the nose, narrowed oropharynx and oblique orbits, with the external epicanthus higher than the internal epicanthus.[11,12]

Observations of midface retrusion and reduced maxillary length among Down syndrome individuals have been reported previously in the literature.[13-15] Several authors[11,13-17] have described the cranial base as small in all dimensions and the cranial base angle as increased. However, many aspects of the craniofacial morphology remain unclear. Some investigators have described the mandible as small,[16,18] while others have found it to be similar to those of unaffected normal individuals.[11,14] Detailed morphologic features of the jaws including the dimensions and relations of the mandible, the anterior maxilla, and the alveolar structures have not been well described. In fact, one cephalometric report described both the maxilla and mandible to be similar in size to published mesofacial norms for similar ages.[11]

Fink *et al*.[16] also observed a significant degree of deficiency in the midfacial area, cranial base, frontal bone, and paranasal sinuses in cases of trisomy 21. These cranial characteristics lead to vertical hypoplasia of the central structures of the cranium, with lowering in the position of sella turcica and consequential flattening of the cranial base. This has led many investigators to point towards authentic cranial dysplasia, with a high incidence of Angle's Class III malocclusion.[19]

The analysis of craniofacial developmental in trisomy 21 subjects by Fischer-Brandies[14] found that the linear length of the anterior cranial base was shorter than normal, with underdevelopment in the midfacial area. The cranial base flexure angle was seen to be obtuse, indicating a flat cranial base, a morphology known as platybasia. Roche *et al*.[20] postulated that the flattening of the cranial base is due to increased cranial pressure associated with growth. However, the angle of the cranial base was found by Fischer-Brandies[14] to drop with age.

Suri *et al*.[21] found that Down syndrome subjects have a larger cranial base angle, reduced elevation of sella from Frankfort horizontal plane, reduced anterior and posterior cranial base lengths and facial heights, smaller maxilla with reduced anterior basal and apical dimensions, and smaller mandibular ramus and body. Anterior open bite was frequently noted with a forward rotation pattern of both maxillary and mandibular planes.

Recently, Koravem and AlKofide[22] described the cephalometric characteristics of Down syndrome subjects, citing a shorter anterior and posterior cranial base length and a backward rotation of the posterior cranial base. The maxilla was found to be deficient, which predisposed to a Class III pattern. Lower facial height was increased with an increase in the mandibular plane angle and a tendency to skeletal open bite. Bimaxillary dental protrusion was evident with protruded and proclined upper and lower incisors leading to an acute nasolabial angle and protruded upper and lower lips.

General hypotonicity of the musculature of the orofacial region, tongue, upper lip, and ligamentary tissue of the temporomandibular joint is also seen among Down syndrome individuals.[19] According to Pilcher[23] and Guimaraes[24] the tongue may seem too large, which is due not to macroglossia, but as a result of midface hypoplasia and a small oral cavity.

OCCLUSAL MALRELATIONS

Occlusal anomalies have considerable impact on individuals with trisomy 21, causing problems in their daily activities, including discrimination based on physical appearance, and difficulties related to oral functioning such as chewing, swallowing, and speaking. Generally, the most frequent malocclusions stem from variations in vertical and transversal occlusions, identified mainly as anterior open bite, anteroposterior crossbite, and proclination of the anterior teeth.[11,12,25]

Whilst the literature[14,26] suggests that trisomy 21 children exhibit asymmetric micrognathic jaws, tooth malformation and congenital absence of teeth, the data, however, is insufficient to permit an accurate assessment of the extent of these abnormalities. Furthermore, there is no agreement as to whether these abnormalities present in all individuals with Down syndrome.

MESIO-OCCLUSION

An Angle Class III malocclusion has been recorded in a high proportion of Down syndrome subjects. The following percentages have been reported: Cohen and Winer - 37.7%, Gullikson - 50%, Brown and Cunningham - 49%, Swallow - 61% in institutionalized and 26% in non-institutionalized Down syndrome children, Gorlin - 60% and Patel and Boghani - 44.3%.[27-32] This increased occurrence of Class III malocclusion in Down syndrome children can probably be attributed to any of the following: an underdeveloped maxilla, an enlarged and forwardly positioned tongue, or a relatively large and prognathic mandible.[33,34] Bauer *et al*.[35] considered a decrease in flexion of the cranial base as one of the etiologic factors of a skeletal Class III pattern.

ANTERIOR OPEN BITE

Amongst the variety of recorded malocclusions, that having the greatest negative impact on Down syndrome individuals may be an openbite, considering that it is the malocclusion calling for the most complex treatment. The prevalence of openbite in Down syndrome individuals was reported to be 4.8% by Cohen and Winer, 15% by Brown and Cunningham, 20% by Carlstedt *et al*., 21% by Oliveira, 30% by Gullikson and 38% by Vigild.[27-29,38-40] Jensen *et al*.[1] observed anterior open bite in 62% of males and 48% of females, while posterior openbite was seen in 8% of males and 25% of females.

Open bite is a multifactorial phenomenon and possible reasons for the high incidence in Down syndrome can be quite varied.[36] According to Alió Sanz[37] the characteristic macroglossia and forward placement of tongue found in this syndrome tends to produce a dental biprotrusion accompanied by an anterior open bite. Another cause can be the deficient development of the mandibular condyle, resulting in a characteristic micrognathia that favours the subsequent appearance of an openbite.[19] Advanced periodontal diseases, frequently overlooked as contributing factors, may render the maxillary and mandibular incisors more susceptible to the effects of any co-existing skeletal and muscle imbalances, leading to anterior open bites.[39]

ANTERIOR CROSSBITE AND OVERJET

The prevalence of crossbite in Down syndrome was reported to be 15.4% by Cohen and Winer, 46.4% by Gullikson, 56% by Brown and Cunningham, 65% by Vigild and 100% by Kisling and Krebs.[27-29,38,41] Quintanilla *et al*.[42] observed an anterior crossbite in 38.4% of Down syndrome patients with lower incisor protrusion in 84.6%. Bauer *et al*.[35] reported a 67% prevalence of anterior crossbite of more than one tooth in a Down syndrome group. Oliveira *et al*.[40] have recorded cross bite of all anterior teeth in 33% of their Down

syndrome subjects. Jensen *et al.*[1] observed some form of crossbite in approximately 90% of their Down syndrome sample, both sexes being affected similarly. The majority of posterior crossbites were bilateral (females-68.5% and males-42.5%), while anterior crossbite represented the second largest category (males-20% and females-10%). There were approximately equal percentages of unilateral right-sided and left-sided crossbites. These findings could be partially explained by the fact that in Down syndrome subjects, mandibular arch widths tended to exceed those of the maxilla. Crossbites tend to increase with increasing age in both sexes.

Meštrović *et al.*[43] have observed that centric occlusion is absent in subjects with Down's syndrome,. The mandible is permanently pushed to the right, to the left or forward, always reacting with a different occlusion on demand. Because of maxillary deficiency, the subjects tend to mandibular prognathism. As would be expected, the authors found that overbite and positive overjet were much smaller for Down syndrome individuals, being a reflection of the openbite and Angle's Class III tendency. Although no significant changes in overjet with age could be detected, it was apparent that in the younger Down syndrome sample, overjet had a tendency to be larger than in older. This finding could be a result of tongue function and posture, the anteroposterior size of the maxillae and vertical growth insufficiency.[29] Vigild reported mandibular overjet in 41% of Down syndrome individuals.[38]

ANTERIOR CROWDING AND SPACING

Gorlin[31] in 1965 described irregular alignment of teeth and crowding in 65% of individuals presenting Down syndrome. Jensen *et al.*[1] and O'Donnell and Cohen[44] have identified the presence of a decreased arch length and arch circumference as well as asymmetrical dental arches.

According to Ondarza *et al.*[45] and de Moraes *et al.*,[46] the frequency of tooth anomalies like agenesis and altered shape are significantly higher in individuals with Down syndrome, when compared with individuals with other types of deficiency, as well as being greater than in normal individuals. According to Desai and Flanagan,[47] agenesis and defects in development are ten times more common in patients with Down syndrome than in the overall population. The higher frequency of mal-alignments in Down syndrome individuals could well be related, however, to a decrease in size and asymmetry of the dental arches.[45]

These dental anomalies along with mandibular prognathism result in spacing in the dentition.[48] Occurrence of microdontia among Down syndrome subjects was variable from 2.04% to 55%. According to Townsend,[49] permanent tooth dimensions were significantly smaller in the Down syndrome group and also tended to show greater variability. According to Suri *et al.*,[50] the most frequently agenetic teeth among Down syndrome were, in descending frequencies, maxillary and mandibular third molars > maxillary lateral incisors > mandibular second premolars > mandibular incisors > maxillary second premolars > maxillary second molars. Maxillary hypodontia was not associated with significant regional craniofacial differences, while mandibular hypodontia was associated with decreased mandibular length and increased ramus body ratio. According to Desai,[51] interdental spacing among Down syndrome individuals is common because of microdontia and can be corrected either by restorations or orthodontic intervention.

NARROW PALATAL VAULT

Quantitative assessments of the shape and dimensions of the hard palate in Down syndrome subjects are limited. [52-55] Investigations of Down syndrome individuals have found a shorter hard palate,[53,55] decreased width, and height,[55] and an elliptic paraboloid palatal shape.[53, 55] Early hypotonia in Down syndrome and lingual diastasis were recognized as etiological factors of this specific palatal morphology which is associated with soft tissue prominence along the palatal surfaces of the maxillary dental arch,[56-58] giving rise to the description of the palate as shelf-like or a 'stair' palate.[55] The frequency of shelf-like palate is higher among the youngest age group of Down syndrome individuals.[39,51,53] Overall, all palatal dimensions have been found to be somewhat larger in Down syndrome males than in females with permanent molar teeth.[51,56]

MANAGEMENT OF MALOCCLUSION

Unfortunately, the extent of mental deficiency in Down syndrome young individuals has often been exaggerated, and this may have caused dental professionals to shy away from managing the associated malocclusions. Though most orthodontists have Down syndrome patients in their clinical practices, few cases of successful orthodontic treatment have been published.[47, 59-61] With medical advances and an improvement in dental material science, and an enhanced recognition of the oral characteristics of these individuals, better dental as well as orthodontic management is possible. Dental professionals can eliminate or reduce the chances of malocclusion among Down syndrome individuals by undertaking preventive orthodontic procedures like caries control, managing premature loss of primary teeth, management of oral habits, extraction of supernumerary or retained primary teeth etc. It is also advisable to undertake certain interceptive procedures like space regaining, correction of occlusal interferences, interception of skeletal malrelations etc., when malocclusion has already developed or is developing. Some studies have mainly focused on behaviour management needs[62] and orthodontic diagnostic methods, but not on orthodontic therapy outcomes.[63]

Before beginning orthodontic treatment on a patient with Down syndrome, it is important to determine the patient's level of tolerance and cooperation.[62] Case reports published in the literature have described the requirement of general anesthesia to perform certain phases of orthodontic treatment, especially when using fixed multibracket appliances.[62,63] Outumuro *et al.*[61] exclusively used psychological techniques of behaviour modification while making dental impressions for diagnosis and for appliance design as well as the periodic revisions. Many clinicians could improve the orofacial function, facial appearance and prevent secondary conditions like pseudoprognathism, malocclusions, open bite and pseudomacroglossia etc by following the "Orofacial Regulation Therapy for children with Down syndrome" developed by Castillo-Morales in the mid-1970s.[64-66] The specially designed acrylic plate designed by Castillo Morales is used in cases with hypotonic, inactive upper lip and broad, hypotonic tongue, which is often positioned between dental arches, as the appliance enhances the position of lips and tongue due to lack of acrylic material in the anterior part of palate.[67]

The successful delivery of treatment is within reach for many orthodontic patients with Down syndrome, depending

upon effectively addressing some of the requirements like motivation, oral hygiene, behaviour management and manual dexterity.[60] By familiarising themselves with different kinds of malocclusions, dental professionals, orthodontists and pediatric dentists can plan a varied combination of treatment modalities. In this way, they may broaden the therapeutic options for many of the more difficult cases, hitherto too frequently denied professional attention.

CONCLUSION

Generally, there appears to be a simple correlation between mental status and the severity of craniofacial handicap. But in the case of Down syndrome the effect of chromosomal aneuploidy coupled with mental deficiency manifests not only as anomalies of dental occlusion, but also discrepancies of craniofacial complex. Careful attention, prevention and management of occlusal aspects that are common in Down syndrome can promote considerable improvement in the quality of life of these individuals.

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