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Review Article

Down Syndrome: A Review

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ABSTRACT:
Down syndrome, a genetic condition characterized by the presence of an extra 21st chromosome, significantly impacts various aspects of health, including dental care. Individuals with Down syndrome exhibit distinctive oral and dental challenges that necessitate specialized attention from dental professionals. These challenges include delayed dental development, increased susceptibility to periodontal disease, and a higher prevalence of dental anomalies such as hypodontia and malocclusion. Additionally, individuals with Down syndrome often present with anatomical and physiological variations that complicate dental procedures, including altered facial morphology and reduced muscle tone. Effective dental management of patients with Down syndrome requires a comprehensive approach that addresses both preventive and therapeutic aspects. This includes tailored oral hygiene instruction, early and regular dental check-ups, and a multidisciplinary approach to address any co-existing medical conditions. By understanding the unique dental needs of individuals with Down syndrome and implementing individualized care strategies, dental professionals can enhance oral health outcomes and improve the overall quality of life for this population.
Keywords: Down syndrome, Chromosome 21, Dental anomalies

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INTRODUCTION

Down Syndrome is a genetic disorder, which is due to the presence of 47 chromosomes instead of 46, with an extra copy of chromosome 21.It is also known as Down's syndrome or trisomy 21 and was discovered by John Langdon Down in 1866. The extra copy of chromosome 21, which may either be full or partial, depending on the variant, causes the abnormality and associated structural and functional anomalies of the bodily systems.In 1866,John L. Down published an article accurately describing some of the characteristics of this syndrome that today bears his name.[1]It is consistent in the literature that advanced maternal age (AMA) is a primary risk factor in down syndrome births.A major cause of foetal death in humans, about 50% of spontaneous foetal loss during pregnancy (before 15 weeks of gestation), are related to down syndrome.[2]

The nature of the abnormality and the physical characteristics make the individuals with down syndrome resemble one another rather than their own family members. However, only less than 5% is hereditary. Down syndrome has considerable health cost implications, given the individual and socio-economic consequences. Down syndrome is found in all races, nationalities, religions or socio-economic levels.[3] The life expectancy and quality of life for affected people have increased remarkably due to improved medical care, general awareness, as well as increased social interactions. The estimated life expectancy of persons with down syndrome has increased from just 12 years in the 1940s to an average of 55–60 years in the present decade, particularly in developed countries.[4]

GENETICS

Prenatal testing

For pregnancies, the high risk of down syndrome is evaluatedby fetal sample's analyzing after invasive chorionicvillus sampling (CVS) and amniocentesis,and by applying laboratory techniques such asconventional cytogenetic analysis

(karyotype),Fluorescence *in Situ* Hybridization (FISH), QuantitativeFluorescence-Polymerase Chain Reaction(QF-PCR), Multiplex Ligation Probe Assay (MLPA)and array Comparative Genomic Hybridization(CGH), which are common techniques used forprenatal diagnosis of down syndrome and each of them presentingwith advantages and disadvantages.There is also a noninvasive technique for detectionof trisomy 21 by Next Generation Sequencing(NGS) technology, known as Non-InvasivePrenatal Diagnosis (NIPD). The process is basedon analysis of extracted cell-free fetal DNA screeningfrom maternal plasma samples.[5]

Postnatal testing

Down Syndrome is caused by trisomy of chromosome21. Mainly there are three cytogeneticforms of down syndrome:

1. Free Trisomy 21 consists of a supplementarychromosome 21 in all cells.[5]
2. Mosaic Trisomy 21 means that there aretwo cell lineages, one with the normal number of chromosomes and another one with an extranumber of chromosomes 21.[6] The mechanism of occurrence consists of an error or misdivisionafter fertilization during cell division.
3. Robertsonian Translocation Trisomy 21 occursonly in 2-4% of the cases. The long arm of chromosome 21 is attached to another chromosome,generally an acrosome, mainly chromosome14.[7]

Systemic conditions associated with Down Syndrome

Heart defects: Congenital cardiac anomalies are present in about 40% of infants with Down Syndrome. In decreasing order of frequencies are the ventricular septal defects, A/V communis, arterial septal defects, and patent ductus arteriosus. All of these cardiac anomalies can be corrected with surgery during infancy, which often results in a very good prognosis.[8]

Vision problems: Good vision is very important to the development of a child, especially a child with developmental problems such as those associated with down syndrome. More than half of children with down syndrome have ocular abnormalities. In addition to ocular features related to down syndrome such as epicanthal fold Down Syndrome, narrowed or slanted palpebral fissures (the mongoloid slant) and Brushfield spots (38–85%), these vision disorders include strabismus (20–47%), nystagmus (11– 29%), congenital cataract (4–7%), acquired cataract (3–15%), blepharitis (7–41%), refractive errors (43–70%) and glaucoma (0.7%).[9,10]

Hearing loss: Hearing impairment and otologic problems are prevalent in children with down syndrome, and these problems correlate substantially with developmental problems. Midface hypoplasia is common in children with down syndrome and consists of abnormalities of the nasopharynx, abnormal Eustachian tube anatomy, abnormal tooth development and agenesis of the teeth.[11]

Speech: Expressive language of Down Syndrome children is more delayed than receptive language. This is related to mental deficiency, hearing problems, aphasia, excessive salivation, poor oral closure, dry and thickened mucous membrane, a relatively large tongue in a small oral cavity, high vault, dental anomalies, and generalized muscle hypotonia.[12]

Leukemia: Children with Down Syndrome are at greater risk of developing leukemia, usually the acute lymphocytic type. The dentist should be aware of such abnormal findings, carefully review the medical history, and consult with the patient's physician if suspicious lesions or symptoms appear.[13]

Hypotonia (poor muscle tone): Poor muscle tone and low strength contribute to the delays in rolling over, sitting up, crawling, and walking that are common in children with Down Syndrome. Despite these delays, children with Down Syndrome can learn to participate in physical activities like other children. Poor muscle tone, combined with a tendency for the tongue to stick out, can also make it difficult for an infant with Down Syndrome to feed properly, regardless of whether they are breastfed or fed from a bottle.[14]

Physical findings in Down Syndrome

Although the phenotype is variable, there typically are multiple features that enable the experienced clinician to suspect the diagnosis.

Among the most common physical findings are

- Hypotonia
- Epicanthic fold Down Syndrome (extra skin of the inner eyelid, which gives the eyes an almond shape)
- Upslanting palpebral fissures (slanting eyes)
- Brachycephaly (a smaller head that is somewhat flattened in the back)
- Flat nasal bridge
- Brushfield spots
- Small mouth
- Small ears
- Excessive skin at the nape of the neck
- Single transverse palmar crease
- Short fifth finger with clinodactyly and wide spacing, often with a deep plantar groove between the first and second toes.

ORAL MANIFESTATIONS

Fissured Tongue

Fissured tongue (FT) is a benign, usually asymptomatic condition characterized by deep grooves on the dorsal surface and margins of the tongue.[15,16] It is the most common oral soft-tissue anomaly present in down syndrome patients accounting for 73% of cases and gender-wise comparison showed about 80% of fissured tongue in males and 59.4% in females.

Everted lower lip
Everted lower lip was the second most anomaly present in 61% of cases, Study conducted by Al Maweri confirmed the same.[17]

Angular cheilitis
Accounted for 51% of cases. Previous studies done by Al-Maweri *et al*., compared lip and oral lesions between control and Down's syndrome patients showed 38% of angular cheilitis lesions. Increased incidence of angular cheilitis in this study may be caused by *Candida albicans* as a result of drooling and immune defects in these children.[18]

Lip Fissures
Lip fissures, also called cracked or fissured lips, have been reported in three papers. Camacho et al., found them in 20% of 15 down syndrome subjects under the age of 16 and none in 57 other down syndrome subjects aged between 2 and 29 years. Al-Maweri et al., reported a rate of 64% in 50 subjects aged 6–18 years, while Scully et al., reported a rate of 17% in 24 down syndrome subjects aged 0–20 years.[19]

Macroglossia
It was the third-most common anomaly present in our study which accounts for 53% of our cases the results of our study showed slightly lower percentage than the previous study. Previous studies revealed that the tongue size does not vary significantly from the general population. Due to small size of oral cavity tongue looks macroglossic.[15]

Geographic Tongue
Geographic tongue (GT) was reported in two cohort studies and one case-control study. GT is a benign condition of uncertain aetiology affecting the dorsal tongue, its margins and, rarely, other oral sites. The percentages of GT reported were: 11% of 71 down syndrome subjects (age range 0.17–25 years) by Ercis et al., 4% out of 100 down syndrome subjects (age range 3–20 years) by Daneshpazhooh et al., and 2% out of 50 down syndrome subjects by Bilgili et al., (age range 0–11 years).[20]

Hypotonic tongue: Tongue protrusion or thrust during drinking, while sucking a pacifier, eating, and speaking is reported in the presence of a hypotonic tongue. Midline junction of the tongue is weak (lingual diastasis) with excessive concavity of the frontal two third Down Syndrome of the tongue and weak frenulum.[21]

Hypoplasia: Hypoplasia and hypocalcification are common. Tetracycline staining may occur as a result of the frequent necessity for antimicrobial chemotherapy in early life.Infants show generalized or localized congenital dental malformation ranging from intrinsic discolorations that are smooth to overt defects that are easily detected by a dental instrument. Hypoplastic defects are frequently the result of significant illnesses or prolonged fevers.[22]

Malocclusion: The following factors play an important role in malocclusion: mouth breathing (96%), improper chewing (60%), evidence of bruxism (45%), tooth agenesis (12.7%), midline deviation in maxillary arch (80%), an anterior open bite (45%), dysfunction of temporomandibular joint (24%), delayed eruption and exfoliation of both primary and secondary dentition, characteristic tongue thrust, hypotonic ligamentary apparatus of mandibular joint, developmental disturbances of the mandible (platybasia) and maxilla (midfacial complex), and the jaw relationships.[23]

ORAL CANDIDIASIS
Candida is one such endogenous pathogen, the prevalence of which has been reported to be 45-65% in healthy children and 30-45% in healthy adults. Opportunistic infections occur mainly when the host defenses are inadequate, candidiasis is the most common among them. Streptococci form a large portion of the resident oral micro flora. In the general population, half the isolates from the tongue and saliva are Streptococci, but in Down Syndrome individual's oral streptococcal levels are relatively lower.[24]

DENTAL MANAGEMENT OF DOWN SYNDROME CHILDREN
Communication
Language delay and impairment in down syndrome is a major barrier to effective communication, and social interaction and development. Children with down syndrome thus tend to depend on nonverbal skills for longer period Down Syndrome than typical children. These patients are likely to have language deficits, particularly in expressive language, and poor speech intelligibility.[25]

Restorative
Children with down syndrome should limit caries risk factors such as frequent sweets intake and prolonged use of a bottle. Early education of the children and parents/caregivers about prevention should be instituted. All restorative treatments should be performed, as for normal individuals, using behavioral management techniques. Some individuals with down syndrome require sedation or general anesthesia.[26]

Endodontics
Pulp therapy depend Down Syndrome on many factors: IQ, physical status, and periodontal status of the patient. Pulp therapy should be avoided in children with down syndrome with heart defects .Differences in root canal anatomy among the down syndrome population has not been fully studied. Kelson and colleagues[27]

Periodontal Treatment

Both surgical and nonsurgical therapies showed a comparative clinical effect. The immunologic impairment can be overwhelmed and does not interfere with proper clinical healing and maintenance.Consequently, the principal issue in the management of periodontitis in patients with down syndrome is prevention. Prevention includes all the measures specified for prevention, along with the use of medicaments as needed.In circumstances in which vulnerability to periodontitis is encountered, an aggressive treatment should be instituted.[28]

Orthodontic Intervention

Malocclusion and craniofacial characteristics of individuals with down syndrome clearly indicate that these patients benefit from orthodontic correction and intervention. The level of mental insufficiency may have caused dental experts to be timid in dealing with the related malocclusions.

The following has been recommended in the literature to assist in orthodontic treatment:

1. Before commencing, decide on the level of tolerance and cooperation.
2. The use of behavioral management and psychological approaches.
3. The use of fast-setting impression materials with pleasant flavours.
4. Easy bonding of brackets.
5. The use of memory-type wires and self-ligating brackets.
6. Use of the current and advanced planning and techniques in orthognathic surgery.
7. Implant replacement of congenitally missing teeth. Usage of temporary anchorage devices.
8. Early appliance therapy using a Castillo-Morales plate to stimulate and improve orofacial musculature function. Oromotor therapy can be combined with other functional orthodontic treatments, speech therapy, and physiotherapy.[29]

Dental implants

The literature contains few case reports of successful results. Partially or completely edentulous patients with implant-supported prostheses have achieved high success rates and patient satisfaction compared with traditional techniques.[30]

Prosthetic Treatment

In general, patients with down syndrome do not favour removable dentures. In contrast, prosthetic techniques may present difficulties in patients with down syndrome. The use of sectional impression procedures and a McKesson mouth prop to keep the mouth open is effective. A fast-setting impression material should be used, with care taken to avoid the impression material entering the pharynx or remaining in the mouth.[31]

General Anesthesia Management

General anesthesia can be used for the treatment of some patients with down syndrome, but it is crucial to recognize differences between patients and their implications. For example, airway anomalies, endocrine disorders, and congenital heart disorders may require some modifications or consideration for general anesthesia. These patients are also more prone to hypothermia during surgery or to developing spinal cord damage from atlantoaxial subluxation. Preoperative and postoperative morbidities are also more common.[32]

CONCLUSION

Dental care for the patient with Down Syndrome can be achieved in the general practitioner's office in most instances with minor adaptations. Children with down syndrome have several down syndrome-specific morbidities and screening programmes are available to support and educate patients and their families. Although the most frequently occurring morbidities are emphasized, a potential drawback is that a child with down syndrome might have rare down syndrome specific problems, but children with down syndrome can also have the same problems as their healthy peers.

There should be a focus on probable changes in long-term down syndrome morbidity. Furthermore, we need to address the quality of this longer life span. Although this population has some unique dental care need Down Syndrome, few patients require special facilities in order to receive dental treatment. Adequate dental health care for persons with developmental disabilities is a major unmet health need. It is hoped that the pediatric dentists will encourage general practitioners to be willing to provide comprehensive dental care to the children with Down Syndrome.

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