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Research paper

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Down's Syndrome: A Study of the Available Research Rajiv Ahluwalia¹, Sanjeev Tomar², Priyanka Aggarwal³, Natasha Gambhir⁴, Sumita Giri⁵, Priyanka Thukral⁶

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ABSTRACT:-

This overview of the literature on Down syndrome focuses on the syndrome's clinical symptoms, systemic and oral defects, and advice for those who have it. Up until recently, chromosomal subgroups were distinguished by karyotyping, but nowadays, increasingly advanced diagnostics and therapies have an impact on the lives of people with Down syndrome. Current trends of normalisation and deinstitutionalization of these individuals are the consequence of medical advancements, specialised educational initiatives, and rising community acceptance of people with disabilities. These people can receive conventional dental care once a dentist is informed with their medical history and takes the necessary safeguards.

Keywords: Oral Pathology, RadiolEndod Oral, Oral Surgery, Oral Medicine

INTRODUCTION: -

Trisomy 21, trisomy G, and mongolism are other terms used to describe Down syndrome. Esquirol presented the earliest description of a youngster who appeared to have Down syndrome in 1838. Eight years later, Seguin wrote about a patient who appeared to have characteristics of the defect that would subsequently be referred to as Down syndrome. John L. Down accurately described some of the traits of the syndrome that carries his name today in a piece he wrote in 1866. Trisomy 21 was found to be the cause of Down syndrome by LeJeune and Jacobs in 1959. In 1974, Nebuhr hypothesised that the "Down syndrome phenotype" might be brought on by a partial duplication of the band q22 on chromosome 21,



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which makes up roughly one-half of the long arm. The genetic causes of Down syndrome have continued to be discovered by researchers. Down syndrome is a readily identifiable congenital, autosomal (non-sex chromosomal), defect that affects 1 in 600 to 1 in 1000 live births and is characterised by global growth insufficiency and mental deficiency. The extra chromosome 21, present in approximately 95% of Down syndrome patients, results in a chromosomal count of 47 instead of the normal 46. Other chromosomal abnormalities, such as translocations (3%) and mosaicisms (2%) or partial trisomies, account for the remaining 5%. In cases of translocation, the additional chromosome is not free but is instead translocated on another chromosome, typically on the 13- or 15-group chromosomes (D), leaving the count at 46. Some cells in mosaicism contain 46 chromosomes, while other cells have 47. These aberrations are modest, and their IQ is normal-looking. Cellular mosaicism, tissue mosaicism, and chimerism are the three different forms of mosaicism. The prevalence of Down syndrome is significantly influenced by the age of the mother.

SYSTEMIC ANALOGIES:-

Cardiovascular abnormalities

Of newborns with Down syndrome, roughly 40% have congenital heart abnormalities. [6] Ventricular septal defects, A/V communis, arterial septal defects, and patent ductus arteriosus are listed in decreasing frequency order. 6 Surgery can be performed to treat all of these cardiac defects in infants, which frequently yields a very positive prognosis. According to Barnett et al. [7], persons with Down syndrome may have a mitral valve prolapse (MVP) at a rate of 5% to 15% higher than that of the general population, and it can occur without any obvious auscultatory signs. If the MVP need antibiotic prophylaxis, speaking with the patient's doctor is advised. The American Heart Association's recommendations for antibiotic prophylaxis should be followed when appropriate.

Hemotropic anomalies

Uncertain factors may contribute to people with Down syndrome being more susceptible to various infections. Neutrophil leukocytes are undoubtedly dysfunctional and transient. 8In addition to reports of lymphopenia and eosinopenia, cell-mediated immunity is hampered and the patterns of serum immunoglobin are off. Dermal, mucosal, gastrointestinal, and respiratory infections are the most frequent conditions in people with Down syndrome. [8] It is possible to plan an early preventive health programme with recall and maintenance visits as frequently as every three to four months. These examinations could uncover any abnormalities in the oral cavity. Children with Down syndrome are more likely to develop acute lymphocytic leukaemia, which is the most common kind. [9] An estimated 1 in 200 children are impacted, which is a 10- to 15-fold increase over normal kids. [9] This heightened frequency only affects children under 18 years old. Leukemia symptoms include persistent lesions and sudden gingival bleeding. If suspicious lesions or syrup lumps develop, the dentist should be aware of such aberrant findings, carefully evaluate the patient's medical



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history, and contact the patient's doctor. Patients with Down syndrome who live in institutions may be seven times more likely than the general population to carry the hepatitis virus.

Dicks and Dennis found that the likelihood of a community-based Down syndrome patient who has not spent more than three weeks in an institution spreading hepatitis B viruses is no higher than that of any member of the general population. The same measures that are used to treat hepatitis C patients from the general community can also be used to treat hepatitis C patients with Down syndrome.

Skeletal abnormalities in the muscles

Transverse ligaments between the atlas and the odontoid processes of the cervical vertebrae and between the atlas and occipital condyles at the base of the skull exhibit increased laxity in 12–20% of people with Downsyndrome. A series of lateral radiographs of the spine are often used by a radiologist to make the diagnosis. Injury from neck hyperextension, radical flexion, or direct pressure on the neck or upper spine is possible if there is atlanto-occipital and rotational instability or if the gap between the odontoid process of the axis and the anterior arch of the atlas exceeds 4.5 mm. [9]This may harm the spinal cord permanently. Dentists should be mindful of this risk when manipulating the patient's neck if there is a history of atlantoaxial laxity. Although there is a very minimal chance that general anaesthetic could cause spinal cord damage, the anesthesiologist and their team must be aware of the possibility and make a plan to prevent it.

There is relative prognathism and underdevelopment of the midface. Due to septal deviation and mucosal thickness, the normally small nasal airways are occasionally partially blocked. The result is frequently mouth breathing. The tongue is frequently pushed past the lips when the mouth is left open. [6]

Neurological abnormalities

Younger individuals typically have delayed motor function, which can cause reduced coordination. Age does, however, lead to improved coordination. Until the person develops the necessary skills, oral hygiene may need to be the primary caregiver's responsibility. [9]

Alzheimer's disease affects about 30% of Down syndrome patients. Many people experience neuropathy changes beyond the age of 35 that are comparable to Alzheimer's disease. [9] 70% don't, however, show any clinically discernible behavioural alterations. The mental deficiencies displayed by these people differ greatly, from virtually normal to profoundly retarded. Physical abnormalities and mental capacity do not always correlate. Every patient should have a unique evaluation. Given their elevated risk of Alzheimer's disease, removable prosthodontics should be avoided in order to preserve the natural dentition. [9] The patient's primary carer should be consulted and involved to ensure a successful outcome.

Children with Down syndrome express their language more slowly than they can receive it. Aphasia, excessive salivation, poor oral closure, a dry and thickened mucosal membrane, a



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comparatively large tongue in a tiny oral cavity, a high vault, dental anomalies, and widespread muscle hypotonia are all associated with this. Lack of incisors makes articulation challenging and significantly alters the characteristics of sibilant sounds. Younger patients should wear space maintainers or, if necessary, be referred to speech and orthodontic therapy. The speech would sound better if anterior teeth that were missing were replaced.

The ability to engage in unrestricted enjoyment of life's blessings, natural spontaneity, genuine warmth, penetrating clarity in relationships with others, tenderness, patience, and tolerance have all been noted in young Down syndrome patients. [11] Due to these traits and the rarity of severe physical impairment, many Down syndrome individuals can get straightforward care in a general clinic.

Oral anomalies

Compared to the mandible, the midface has not fully developed. The length, height, and depth of the palate are reduced as a result of this inadequate development (midface complex), but the width is not significantly altered. The palate has a "stair palate" appearance due to a significant reduction in length. It also has a high arch, and there may occasionally be folds that resemble palatal clefts. [11] Additionally, high vault palates with a V shape may have a soft palate deficiency and have a lower retention rate for maxillary dentures. Patients should be assessed for surgical and orthodontic treatment. Depending on the patient, prosthetic devices or implants may be taken into consideration.

The orbicularis, zygomatic, masseter, and temporalis muscles can become hypotonic, which can cause various important face traits. With passive elevation of the hypotonic top lip and thinning of the lateral aspects, the mouth's angle is lowered. Additionally hypotonic and becoming more and more everted, especially when the tongue protrudes, is the lower lip. The comparatively broad tongue in a small oral space gives the impression that the mouth is open. Mouth breathing, drooling, a chapped lower lip, and angular cheilitis are the results of this. Infections of the respiratory tract and persistent periodontitis are caused by mouth breathing.

Bifid uvulae, lip and palate clefts, and enlarged tonsils and adenoids are uncommon birth defects. Early in life, due to the xerostomia-related decrease in salivary flow, the mucosal lining of the oral cavity thins.

Unusual tongue pressure leaves a distinctive pattern on the teeth that looks like depressed ovals surrounded by a raised, white scalloped border. The causes of this condition include diastema, tongue pushing, tongue sucking, clenching, or a swollen tongue. It can be bilateral, unilateral, or isolated. It has no symptoms and is safe.

During a tongue is hypotonic, tongue protrusion or push is observed when drinking, eating, speaking, and sucking a pacifier. With an excessive amount of concavity in the front two thirds of the tongue and a weak frenulum, the midline junction of the tongue (lingual diastasis) is weak. [5]



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Dental abnormalities

Between 35 and 55 percent of people with Down syndrome have microdontia in their primary and secondary dentition. Clinical crowns usually have conical shapes and are smaller and shorter than usual. They were "stunted with short, tiny crowns and roots," according to Spitzer (1963). All teeth, with the exception of the upper first molars and lower incisors, had smaller diameters, according to Kissling's (1966) analysis of tooth diameters, although the creation of the roots was always complete. Age was correlated with the root and crown ratios of central and lateral incisors. It was thought that incisor teeth with short roots and an unfavourable crown/root ratio may be more prone to movement and eventual tooth loss. The mandibular bicuspid is smaller but still has a regular form. There are a few cases of shovel-shaped incisors and peg-shaped lateral incisors. [12]

Hypocalcification and hypoplasia are frequent. The incremental lines were shown to be inflated in the Down syndrome group by Johnson et al. in 1965. Due to the frequent requirement for antimicrobial treatment in infancy, tetracycline staining may happen. Infants display generalised or specific congenital dental malformations, which can range from subtle intrinsic discolorations to obvious deformities that can be seen with a dental instrument. Major infections or protracted fevers can lead to hypoplastic abnormalities. Teeth with hypocalcifications need to be monitored for the earliest signs of deterioration. General suggestions can range from sealants to smooth rough flaws to bonding or restorations to full crown coverage, depending on the degree of hypoplasia. As a preventive measure, topical fluoride application should be taken into consideration.

Taurodontism affects people with Down syndrome more frequently than the general population (p 0.001); prevalence ranged from 0.54% to 5.6%. [13] In bifurcated or trifurcated roots, taurodontic teeth exhibit expanded pulp chambers and apical displacement. The tooth in the dentition that is most usually affected is the second molar on the mandible. When the epithelial diaphragm fails to invaginate at the correct horizontal level or when its invagination is postponed, a tanrodontic tooth develops. The invagination of the diaphragm is postponed if the diaphragm cells are changed and cell proliferation rates are slowed, as in Down syndrome. [12] Taurodontism is not treatable in any way. [6]

The majority of crown changes are found on the labial surfaces, incisal edges of front teeth, and changedcuspal inclines on canines, absent or diminished distolingual cusps on maxillary molars, and displaced distal cusp on mandibular molars. [7] Reduced mitotic activity of dental progenitor cells during embryogenesis is the cause of common variants of reduced size, agenesis, and high frequency of crown abnormalities. [7] Depending on the individual's needs, crown imperfections might be corrected with bonding or full crown coverage for aesthetic reasons.

A positive element in the clinical management of people with Down syndrome is the low occurrence of dental caries. In a study by Orner, who compared the dental caries experiences



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of Down syndrome patients with those of their siblings, the researcher discovered that Down syndrome patients had fewer dental caries than their unaffected siblings by less than one third. Shapira et al. [1] discovered that as compared to patients with dental caries, persons with Down syndrome who were caries-free had considerably lower Streptococcus mutans levels. The low prevalence of tooth decay is attributed to a number of causes.

Defects in the periodontium

When compared to individuals with identical plaque levels, those with Down syndrome experience earlier onset, more severe gingivitis, and generalised, rapid periodontal deterioration as adults. [14] Ages 6 to 15 are prime starting points for periodontal disease. Extreme tooth movement, which leaves no other option but extraction, is the most common issue by the mid-30s. Marginal gingivitis, acute and subacute necrotizing gingivitis, advanced periodontitis, gingival recession and pocket formation, horizontal and vertical bone loss with suppuration, bifurcation and trifurcation involvement in the molar area, marked mobility of posterior and anterior teeth, and frequent tooth loss, particularly in the mandibular anterior area, are all common conditions seen in people with Down syndrome.

Anomalous occlusions

Patients with Down syndrome had a higher frequency of malalignments in both the deciduous and permanent dentition than those without Down syndrome, according to a research by Ondarza et al. The central, lateral, and canine incisors are the teeth that are affected the most commonly.

In Down syndrome patients, the midface is less developed than the mandible. [8] Instead of a restriction in the transverse dimension, the anteroposterior deficit of the maxillary arch is predominantly blamed for the anterior cross bites. A lack of vertical development of the maxilla, which causes overclosure of the mandible and projects the mandibular arch forward in relation to the maxilla, further supports this. The apparent prognathism should be primarily attributable to the maxillary insufficiency rather than to expansion of the jaw because the mandible is not noticeably impacted.

CONCLUSIONS:-

The number of Down syndrome patients in office settings has significantly increased in the United States as a result of deinstitutionalization and the growth of community housing. This article discusses the systemic (cardiovascular, hematopoietic, musculoskeletal, nervous) and oral (palate, oral opening, tongue, dental, periodontal, and occlusion) anomalies that can occur in people with Down syndrome and their clinical implications. Each person is only allowed to have a small number of aberrations, which vary in severity and with age. This article highlights the anomalies that can call for medical advice while highlighting the fact that these patients can typically be treated in an office setting.



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