Case Report

Down Syndrome: A Case Report with Concise Review

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ABSTRACT:

Down Syndrome is a common disorder with phenotypes including leukemia, congenital heart defects and Alzheimers disease. It is a chromosomal abnormality which leads to mild to moderate intellectual ailment and this impairment causes dysfunctioning of multiple organs including the orofacial region. The extra genetic material can result in disturbed mental and physical growth of the child. Mental retardation, delayed language and motor development, delay in fetal brain growth, prenatal and postnatal growth deficiency, gastrointestinal malformations, congenital hematologic disorders, and cardiac abnormalities are observed in this disorder. Dental and craniofacial features include brachycephaly, flat occiput, mandibular prognathism and reduction in parotid salivary flow rate. Necrotizing ulcerative gingivitis, periodontal disease and delay in eruption of both deciduous and permanent teeth is observed in this disorder. This article aims to discuss the clinical features and management of a six year old male child showing classical signs of this disorder.

KEY WORDS: Down's syndrome, disorder, growth, deficiency

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INTRODUCTION:

Down syndrome is a common chromosomal anomaly in which distinctive phenotypic features and psychomotor developmental delays are observed. There is presence of all or portion of third copy of chromosome 21, and that is why it is also known as trisomy 21 or trisomy G. A Britisher named John Langdon Down first observed and identified this disease in the year 1886, and hence it is known as Down Syndrome.[1] The incidence of Down Syndrome is calculated to be 1 per 800 to 1000 births. In India, the frequency is 2.2 per 1000 live births. It is an abnormality of chromosome 21,which is the smallest human chromosome. It ranges over 48 million base pairs which are the building blocks of DNA and it represents 1.5 to 2 percent of total DNA in cells.[2] Various medical complications like neurological, cardiac, cognitive and orthopaedic disorders can be observed in this disorder. There is increased risk of Alzheimers disease, congenital heart disease, leukemia, dementia, seizure disorders, vision and hearing difficulty, gastrointestinal complications, sleep apnea, disturbance in endocrine system, musculoskeletal deformity and excess mobility of atlas and axis are observed in this disorder.[3] Severe degree of intellectual disability, physical growth disturbance, neotenization of brain and mental retardation are common in this syndrome. The orofacial features include high arched or oval palate, maxillary canine impaction and underdeveloped maxilla.[4] Development of this syndrome is associated with decrease in the neuronal number and abnormal neuronal differentiation. In a patient suffering from this syndrome,neurons deteriorate and undergo apoptosis. Busciglio J et al observed that deterioration of neurons can be prevented by treating with free radical scavengers.[5]

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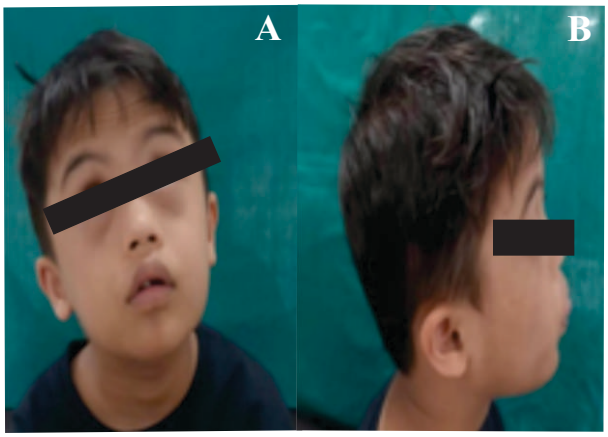

Figure 1: Frontal & side view showing features of Down's syndrome.

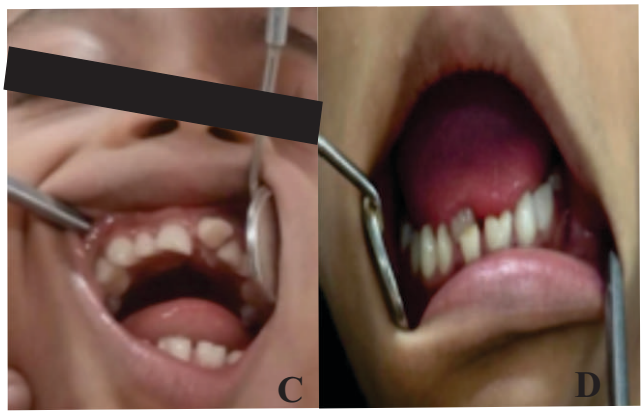

Figure 2: Intraoral examination of maxillary &mandibular arches showed malocclusion with delayed tooth eruption, retained central incisors, lingually placed 61,71 & 81 and peg shaped maxillary lateral incisors.

CASE REPORT:

A six year old male patient reported to the Department of Oral Medicine and Radiology for routine dental check-up. Despite having poor speech ability, the patient was able to understand the given instructions. Frankl behaviour rating scale indicated the child's behaviour as definitely positive. Medical history revealed that the patient was diagnosed with Down syndrome by his physician 5 years ago and surgical management of the patient has been done. On general examination, patient was found to be conscious and cooperative but he was short statured and mentally challenged. Extraoral examination showed hypertelorism, squint in the eyes, and upward inclination to the eyes, depressed nasal bridge, slanting of palpebral fissures, medial epicanthal folds and brachydactyly (Figure 1). On intraoral examination, malocclusion was observed with delayed tooth eruption. There was retained deciduous teeth that is retained central incisors in maxilla and mandible. The maxillary central incisors on the left side of the patient were palatally placed (61) and the mandibular central incisors were also found to be lingually placed (81 and 71).Maxillary lateral incisors were found to be peg shaped. High arched and constricted palate was observed in the patient. Tongue appeared to be fissured which protrudes on mastication (Figure 2). Anterior open bite was observed. Dryness of mouth due to mouth breathing was present. Due to severe malocclusion, erythematous gingival with bleeding on probing, stains and calculus was present. This showed poor oral hygiene with halitosis. However, no hearing loss was observed in the patient. A provisional diagnosis of Down Syndrome was given and radiograph was advised.

RADIOGRAPHIC FEATURE:

Orthopantomograph showed short mandibular ramus with a steep mandibular angle and mild prominence of the antegonial notch. Crowding of teeth, underdeveloped condylar and coronoid process were observed. Inverted cone shape was seen with respect to 51 and ill-defined periapical radiolucency was seen with respect to 81 and 71. Coronal radiolucency was seen involving enamel, dentin and pulp. There was generalized decrease in the height of the alveolar bone extending below roots of most teeth which indicated vertical bone loss. (Figure 3)

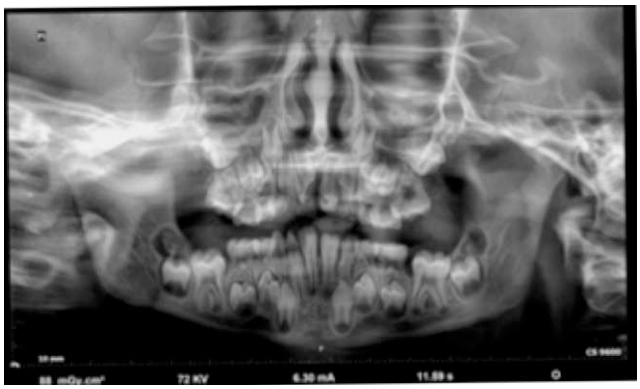

Figure 3: OPG showed crowding of teeth, underdeveloped condylar and coronoid process.

Correlating the clinical and radiological features, a diagnosis of Down syndrome was made. Oral prophylaxis was done and orthognathic surgery was advised. Psychological counselling was given to the patient and parents.

DISCUSSION:

Down Syndrome is a chromosomal disorder and can cause intellectual deformity, congenital heart disease, Alzheimer's disease, leukemia and Hirschsprung disease. Low muscle tone, folding of skin on the inner corner of the eyes, upward-slanted eyes,

abnormal shaped ears, deeply felt wrinkle over the focal point of the palm, and enlarged tongue with a little nose are some of the features.[6] There are 3 genotypes of Down Syndrome and they are trisomy 21,translocation and mosaicism, out of which trisomy 21 is most common.[1] Premature birth,prenatal and postnatal deficiency of growth, delayed osseous maturation, flaring of iliac wings, brachymesophalangy of the fifth finger, poor verbal short term memory, disturbed auditory sequencing, disturbed color retention, disturbed visual-motor task, atrial septal defect, gastrointestinal disturbances such as tracheo-esophageal fistula, pyloric stenosis and annular pancreas are commonly observed in this disorder. Hyperkeratosis, seborrheic dermatitis, leukemia and lymphoma can occur. Retinoblastomas, pancreatic and bone tumors are also observed in this disorder.[2] In this syndrome, age of mother conceiving pregnancy is an important risk factor because as age increases risk of chromosomal abnormality increases.[7] Dental and craniofacial features include deep and high palate, bifid uvula, underdeveloped nose, low nasal bridge, cleft lip, cleft palate, underdeveloped jaw, abnormal lip closure, hypotonic lips, abnormal movement of tongue, fissured tongue, anterior open bite and muscle hypotonia.[6] There is reduction in flow rate of saliva. There is absence of frontal and sphenoidal sinus but fontanelles are large. Bony midface hypoplasia, mandibular prognathism and ocular hypotelorism is common. In most cases, maxillary sinuses are hypoplastic. Brachycephaly is common in such patients. Periodontal disease and necrotizing ulcerative gingivitis are common in such patients. There is delay in eruption of both deciduous and permanent teeth. The eruption process is disturbed because in some cases, deciduous first molars exist before incisors. Third molars, second premolars and lateral incisors are mostly absent in permanent dentition. Deciduous lateral incisors can also be absent. Sometimes, the maxillary lateral incisors are peg shaped. Enamel hypoplasia, enamel hypocalcification, widely spaced teeth and posterior crossbite can occur in such patients.[2]Dental caries can be observed in such patients due to immunosuppression where pathogens colonize and demineralise the tooth surface causing cavitation. Macroglossia, malocclusion and bruxism can also be present.[1]In such patients, there can be difficulty in providing strong anesthetic effect to patient because these patients are comparatively more sensitive to pain than other individuals, so there can be delayed but magnified pain response. Microdontia, taurodontism, reduced occlusal vertical dimension and atypical shape of dental crowns can be observed.[8] Ultrasound and maternal blood screening are some of the noninvasive diagnostic methods which can indicate this syndrome. Amniocentesis and chorionic villus sampling are diagnostic methods which can confirm the presence of an extra chromosome 21. Karyotyping, which is a blood test, can also be helpful diagnostic tool in identifying this syndrome.[9] Regular medical check ups and monitoring should be done in such patients. Early intervention services are useful in such patients because they help in identifying delay in normal physical and mental growth of the patient. Speech therapy, occupational therapy, behaviour therapy and social skills training can be helpful in such patients. A psychologist can monitor such patients so that they can deal with day to day challenges. A balanced and nutritious diet is important for such patients.[10]

Ventricular septal defect(VSD) is common in such patients.It is a cardiac disorder in which a hole is present in the septum which seperates the two ventricles of the heart. In such patients, the oxygen rich blood travels from left ventricle, through the opening in the interventricular septum and then mix with blood having less oxygen in the right ventricle. Reduced oxygenated blood provides less oxygen to body tissues resulting in hypoxia, disturbed cellular metabolism and decreased rate of respiration. Treatment of such patients using general anesthesia should be avoided as it can cause serious side effects such as sudden increase in pulmonary vascular resistance, hypoxia, hypercapnia, hypothermia, acidosis and hyperinflation of lungs. Non pharmacological behaviour management methods such as voice control and tell-show-do (TSD) are useful.[11] The orofacial features caused by genetic disorder and those acquired in later life must be identified. For specific treatment of patients suffering from Down Syndrome, it is important to identify features which are epigenetic. Large cranial base angle, reduced size of cranial base, decrease in length of maxilla and increase in length of mandible leads to skeletal class III malocclusion. According to Moss's functional matrix theory, development of the alveolar process occurs because of inductive growth potential of the teeth.[12] Chavan et al conducted a study to assess and observe the effect of Pilates training on children suffering from Down Syndrome. In this study, children suffering from this disorder, from 8 to 18 years of age, were randomly divided into two groups. Pilates training was provided to group A and group B was control group. The exercises were conducted for 12 sessions, five days in a week. The session duration was 30 minutes including warm up and cool down period. It was found that Pilates is a successful training program to increase the cardiopulmonary functions in children suffering from this syndrome.[13] Nuchal translucency (NT) is helpful

in identifying about 83% of trisomy 21 pregnancies in the first trimester. NT is a psysiological process marker in the fetus that reflects the fetal lymphatic and vascular growth in head and neck region.[14] Our case report further emphasizes the importance of dental consultation and further requirement of invasive surgical procedures to be important in the management of the cases of Down Syndrome.

CONCLUSION:

Early diagnosis of Down Syndrome and identification of its orofacial features is important for improving physiological, psychological, medical and dental quality of life. In such patients, it is important to execute preventive methods to avoid oral health issues and address long term consequences. A tailored prevention strategy is required for such patients. This includes parental education and participation, regular orofacial assessment starting at 12-18 months, providing balanced diet, oral prophylaxis and hygiene motivation. For oral hygiene, there can be modification of toothbrushes to help such patients. Electric toothbrushes can be helpful in such cases. Interim therapeutic restoration, using materials such as glass ionomers that release fluoride, play an important role in both preventive and therapeutic techniques. Some patients may show anxiety with uncooperative behaviour. Behavioural reinforcement techniques such as tell-show-do, desensitization, sedation, verbal and non verbal communication, distractions and euphemisms may be helpful. During first six months, surgical management of cardiac defects is advised. Antibiotics, leukemia therapies and anticonvulsive drugs also play an important role in the management of patients suffering from this disorder. Careful monitoring of such patients by specialists such as cardiologist, pediatrician, neurosurgeon, orthopaedic surgeon and psychiatrist should be done.

DECLARATION OF PATIENT CONSENT:

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s)/guardian has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients/guardian understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of Interest

There are no conflicts of interest.

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