CASE REPORT

Down Syndrome Patient With Double Row Dentition - A Rare Case Report

Balamanikandasrinivasan Chandrasekarran*, Ramesh Kumaresan*, Mohan N**

Abstract

Down syndrome is chromosomal disorder named after British physician John Langdon Down. This syndrome commonly occurs due to improper cell division which possesses extra genetic material in chromosome 21 and hence called as trisomy 21. Though classical features present in down syndrome have been widely documented, the physical features and associated medical problems can vary from mild to severe among the affected population.

With increased prevalence of Down syndrome, general dentists being the first line in providing oral health care, have more chances to meet and treat such patients. Arrival of the final diagnosis should not be based on clinical components alone but should also consider the collective information obtained from the history and investigations.

Hereby we present a case report of a 19 year old male patient with double dentition having features of Down syndrome. The patient had significant features with strong family history. Along with screening options, management of such patients with respect to dentition was also discussed briefly.


Key words: Down syndrome, Double dentition, Trisomy 21

Introduction

Down syndrome is the most common genetic disorder caused by abnormalities that occur in chromosome 21. The incidence of this syndrome ranges from 1:600 to 1:1000 in live births across the globe. From review of literature it can be inferred that, though the classical features of this syndrome were well documented, still variations do occur between one affected individual to the other. Therefore the clinician should be competent enough to recognize the clinical characteristics pertaining to this syndrome. Hereby a case report of Down syndrome has been presented. To our knowledge, no such case of Down syndrome has been reported in the literature with double row of dentition.

Case Report

A nineteen year old male patient (Figure 1) accompanied by his father came to Department of Oral Medicine and Radiology with chief
complaints of retained upper and lower primary teeth. He was seen with moderate built, short stature and normal gait. The patient was seen with poor memory and his behavior and speech implied that the intelligent quotient was below to his age.

Family history

The patient was born as the third child to a non-consanguineous couple through forceps delivery as reported by his father. The paternal and maternal ages corresponded to 42 and 38 years respectively at the time of his birth. The patient had an elder brother and sister, where the later expired within one year of birth due to unknown reason.

Patient’s elder brother, mother, and patient’s maternal uncle also had similar presentation. His uncle expired eight years back due to respiratory disease, mother and elder brother expired five and three years back respectively due to cardiovascular problems. The patient had multiple systemic problems and was treated for ventricular septal defect during infancy. He suffered from brain fever and jaundice at the age of three and six years correspondingly. Epilepsy was evident till the age of twelve years. Besides that he also suffered from dyspnoea and improper bowel movements.

Extra oral features

Extra orally the patient was seen with brachycephalic head, marked frontal prominence, wide intercanthal space, depressed nasal bridge and retrognathic maxilla, prognathic mandible with everted lower lips. Besides that he had short neck with unequal spacing between his fingers. (Figure 2).

Intra oral features

Intra oral examination revealed the presence of both primary and permanent teeth. Total number of teeth present in patient’s mouth was thirty four. Twelve teeth were found to be retained which included primary incisors, canines, molars in maxilla and mandible (Figure 3 & 4).
first molar and central incisor took place around 8 years of his chronological age.

The patient presented with enamel hypoplasia and stains in most of the teeth. Oral hygiene was compromised with the presence of plaque and calculus. Pocket formation was evident in relation to his mandibular incisors and maxillary molars. The patient was seen with macroglossia and high arch palate. He was treated for ankyloglossia at the age of ten years. Examination revealed that the patient had the habit of mouth breathing and snoring while sleeping.

It was disclosed through his father that he started to speak at the age of three. Currently his speech was slurred and the answers were not relevant to the questions asked.

**Investigations**

Orthopantomogram depicted the presence of retained deciduous teeth, erupted and unerupted permanent teeth whereas in lateral cephalogram nasal bridge depression and retrognathic maxilla with everted lower lips were evident (Figure 5 & 6).

**Diagnosis**

Considering the history and clinical features in account a provisional diagnosis of Down syndrome was made.

**Discussion**

**History and Etiology**

Down syndrome (Trisomy 21, Trisomy G) is one of the most common chromosomal anomalies seen worldwide irrespective of gender, race and economic predilection. Though first described by Esquirol in 1838 and recognized as an anomaly by Seguin, it was John L. Down who precisely documented and published a paper describing some of its features as syndrome and hence named after him in 1886. Lejune and Jacobs et al. through their independent research found out Trisomy 21 as the cause of Down syndrome.

The etiology of this congenital autosomal anomaly has been related to non disjunction of chromosome 21 during oogenesis. Three types of Down syndrome have been documented though no clinical difference was found among them:

i) Trisomy 21 was the commonest among the three (about 95%) where extra chromosome (three instead of two) was seen in 21st chromosome making the total number of chromosomes to 47 (instead of 46)

ii) Translocation occurs roughly in about 2-3% where extra chromosome was translocated to another chromosome (usually to 14) thus referred to 14/21 translocation. These individuals possess normal 46 chromosomes.

iii) Mosaicism- A nondisjuntion type of Down syndrome found to occur in later stages of cell division in 1-2% where some cells have 46 chromosomes and others have 47.

**Influence of paternal and maternal age**

Down syndrome is well recorded to have higher incidence with increase in maternal age. According to Hayes et al, risk for women aged between 35 -39 years old rises 6.5 times when compared to 20-25 yrs. Children born to women aged 40-45 yrs have 20.5 times increased chances for getting this syndrome.

Besides that, study conducted in University of Pennsylvania signifies the influence of parental age...
on Down syndrome. According to them the effects of paternal age are found to be contributory when there is increase in maternal age more than 35 years and older. The paternal contribution to Down syndrome was found to increase by 50% in this situation. The influence of paternal and maternal age as established in the literatures correlates with the patient described above.

Diagnostic features

Presence of the extra chromosome in this syndrome affects almost every important systems of the human body. Several oral and skeletal anomalies have been documented in Down syndrome patients.

Oro facial anomalies includes brachycephalic head, flat face, large anterior fontanelles, and open sutures due to delayed closure. Small slanting eye with epicanthal folds, strabismus, under development of mid face, prognathic mandible, cleft lip and palate, hypotonic upper and lower lips were also documented in many cases.

Authors differ in their opinion regarding the presence of macroglossia as true or relative in Down syndrome patients. According to some, macroglossia may be due to reduced size of oral cavity and others opinion that it is due to inadequate lymphatic drainage. Apart from that tongue thrust leading to mouth breathing, chronic periodontitis and respiratory tract infections were as well recorded in various literatures.

Dental anomalies includes microdontia in primary and permanent dentition (35-55%). Enamel hypoplasia and taurodontism (0.54 to 5.6%) were high among the affected persons. Often patients with Down syndrome presents with partial anodontia (50%) and tooth agenesis. Agenesis of teeth was found to be ten folds greater in males than females, in mandible than maxilla, in left side than right side. Patients with down syndrome were documented with low incidence of dental caries due to delayed eruption of teeth, alteration in chemical composition of saliva, increased space found between the dentition.

Eruption pattern and sequence were found to be delayed in both primary and permanent dentition. First eruption of primary teeth in the affected individuals occurs by 12 months but can delay until 24 months. In permanent teeth, it is normal to wait until 8-9 years of age for the first eruption to occur. The delayed eruption of teeth reported in the literature correlates with our patient.

Retention of anterior maxillary and mandibular primary teeth is a common feature which is also seen in our patient also. The succeeding permanent teeth erupt before shedding of deciduous counterpart. Due to this factor, the dentist needs to extract the retained teeth in these patients. This disturbed eruption pattern also contributes to malalignment and malocclusion in developing dentition. Variations in jaw relation were mostly associated with mandibular over jet, anterior open bite and posterior cross bite in Down syndrome patients.

Almost 40% of affected individuals have congenital cardiovascular anomalies such as ventricular septal defect, auricular septal defect, patent ductus arteriosus and mitral valve prolapsed. Our patient had ventricular septal defect by birth and was treated appropriately thereafter. Both brother and mother of our patient who had similar features died due to cardiovascular anomalies which confirm the prevalence of these types of anomalies in these type of patients.

Patient with Down syndrome were documented with impaired immunity due to defective, short lived neutrophils, lymphopenia, eosinopenia and impaired cell mediated immunity. Besides that, increased risk of leukemia was also recognized in down syndrome patients. Common skeletal abnormality includes increase in laxity of transverse ligaments between atlas (C1 vertebrae) and odontoid processes of other cervical vertebrae and also between atlas and occipital condyles. Characteristic features such as short broad hands, short incurved fifth finger has also been reported in many literatures.

Many nervous system anomalies such as delayed motor function, phonation and low intelligent quotient (IQ) were noticed among affected population which was found in our patient also. About 8% of affected population has gastrointestinal problems. Several other anomalies...
such as endocrine (mostly hypothyroidism),\textsuperscript{33} chronic respiratory diseases are linked with Down syndrome.

It is known fact that in syndrome cases, all the physical features need not be manifested within an individual.\textsuperscript{34} But most of the characteristic findings reported in the literatures such as mental retardation, short stature, brachycephalic head, deficient mid face growth, macroglossia, enamel hypoplasia, malocclusion, delayed eruption, retained deciduous teeth were well recognized in our patient which facilitated our provisionally diagnosis.

**Prenatal screening**\textsuperscript{15,35}

Prenatal screening gives significant details with respect to the risk but does not confirm the diagnosis. The following prenatal tests can provide some clue in identifying the abnormalities in growing fetus.

Karyotyping of the fetus - can be used as single modality or in combination with prenatal screening. It is proven to be 99\% accurate.

Ultrasonographic examination done in second trimester can diagnose the presence of nuchal translucency, cardiac abnormalities, short femur.

Maternal serum marker tests- alpha fetoprotein, unconjugated estriol are found to be lower and beta-human chorionic gonadotropin are often higher in mothers carrying down syndrome children when compared their normal counterparts.

Screenings of these substances are collectively referred as “Triple Screen” usually done in second trimester.

Amniocentesis (done in 16-18 weeks of gestation) for karyotype evaluation and chorionic villus sampling can be done for determination of karyotype (done during 9-11 weeks of gestation).

**Postnatal screening**\textsuperscript{15}

When suspected postnatally, a thorough examination should be commenced from general appearance to systemic examination.

Diagnosis should be based on the presence of constellation of characteristic signs of this syndrome among which hypotonia is most often noted. Serum chromosomal karyotyping of the infant should be done for confirmation of the diagnosis.

Complete blood tests and thyroid function tests are also recommended.

**Dental Management care in Down syndrome patients**\textsuperscript{14}

Though the overall goal in management of these patients is to provide comprehensive treatment as far as possible, it has to be individualized and mostly involves the need of multidisciplinary approach. The influence of medical history, systemic problems and increased susceptibility to infections has to be taken in account before initiation of the dental care.

Most of the dental procedures such as scaling, restorations can be accomplished in general dental set up itself with some minor modifications. Extractions can be done under local anesthesia. But in difficult patients, alternative approaches such as conscious sedation and general anesthesia are found to be useful. Physical fitness has to be obtained from physician and other relevant specialties when necessary.

Diet can be altered wherever needed since most of the patients have high rate of missing teeth. Since low IQ is prevalent in these patients, the role of family member or care giver becomes crucial as proper communication forms the base for success in their management. Besides that, the care giver should be also educated about the importance of oral hygiene for effective management of periodontal diseases in these patients.

In our patient, after oral prophylaxis consent was obtained from general physician and extractions were done under local anesthesia. Patient was accompanied by his father. Two to three teeth were extracted in each appointment. Post operative instructions were given and the importance of having good oral hygiene was emphasized to patient’s father. After two weeks follow up as clinical healing was satisfactory, patient was further referred to speech pathologist for appropriate training. Considering the family history, we also recommended genetic counseling for this patient. But unfortunately, since patients
father was reluctant and required only symptomatic treatment, denied our proposal.

Conclusion

A case report of Down syndrome with highly significant family history has been presented to facilitate a better understanding for general dentists. Dental management in such patients also been discussed briefly. Most of the patients can be treated in dental office itself as mostly the routine management doesn’t require any specialized skills or equipments.

References


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