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Trisomy 21: Genetic Analysis Aided Diagnosis

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Authors' contributions

This work was carried out in collaboration between all authors. Author AD designed and wrote the manuscript. Authors AKN and SR analyzed and reviewed the manuscript. Author NS did literature research and patient management. All authors read and approved the final manuscript.

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Case Study

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ABSTRACT

Being the most common yet rare genetic syndrome in the field of dentistry Trisomy 21 also known as the Down's syndrome named after John Langdon Down has its own importance in dentistry. It has characteristic physical, genetic and mental features which could remain undiagnosed particularly in low socio-economic group areas in countries of southern asia. Dental professionals could be the first ones to diagnose this problem as it shows unique intra-oral and extra-oral features. So we report a classic case of Down's syndrome which remained undiagnosed throughout his life till he visited a dental professional.

Keywords: Chromosome 21; Down syndrome; Trisomy 21; chromosomal analysis; simian crease.

1. INTRODUCTION

Down syndrome or Trisomy 21 is a disorder of genetic origin associated with the disturbances in chromosome 21. Patients suffering with Down syndrome will have three copies of chromosome 21 instead to two [1]. Every one child in 800-1,100 births is at risk of developing this disorder in 21st century and the incidence has shown to be increased with females of

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older age group giving birth. Presence of an additional genetic material results in a disruption in the normal course of development, projecting exclusive features of this condition [2].

The medical problems associated with Down syndrome will have a huge impact on patient's social and educational lifestyle due to the poor developmental state. These problems could be acute but mostly occur in long terms, projecting wide variability and involving multiple organ systems [3]. People with this condition have higher risk for heart anomalies, digestive problems such as gastro-esophageal reflux or celiac disease, hearing disturbances, and blood cancer tissue (leukemia). Also, some patients with will have hypo-functioning thyroid gland (hypothyroidism) [2].

Dental anomalies are also quite frequent, showing prevalence of 44% in sufferers investigated. The commonest are the congenital anomalies, generalized microdontia, retarded root formation, enamel hypoplasia and fusion [4]. Most of the patients will only visit a doctor for their odontogenic complaint and dental professionals could be the first ones to assess, diagnose and manage these patients.

2. CASE HISTORY

A 27 year old male reported to the department of Oral medicine and Radiology with a chief complaint of mobility of teeth in upper left posterior region since 3 months. Medical history revealed that he was admitted to the hospital a year ago for the treatment of jaundice and also history of weakness, breathlessness and hyper-flexibility of joints were evident. It was patients first visit to the dentist and he was unaware of his condition. Patient elicited history of loss of few teeth due to periodontal reasons in a span of 3 years and was not able to practice proper oral hygiene.

2.1 Examination

Extra oral features revealed flat nasal bridge, slanted epicanthal folds giving almond like shape; Mongolian folds, strabismus of left eye, brachycephaly and a hint of "brushfield spots" (Fig. 1), yellow discoloration of sub palpebral conjunctiva was also appreciated. The overall appearance of a patient was of short stature with a flat head and shorter neck. Single "simian crease" transverse was noted on his left and right palms. The IQ of the patient was found to be less than normal.

Fig. 1. Facial profile of the patient

Intra Oral examination revealed generalized microdontia, compromised periodontal status, poor oral hygiene, macroglossia with fissured tongue and high arched palate (Figs. 2 and 3). Disto-proximal caries were detected with respect to 25 and partially edentulous maxillary and mandibular arches were also observed.

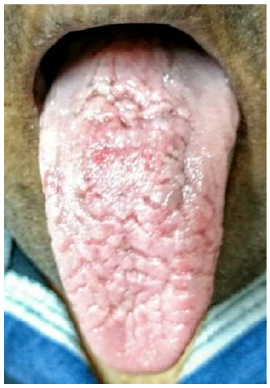

Fig. 2. Macroglossia with fissured tongue

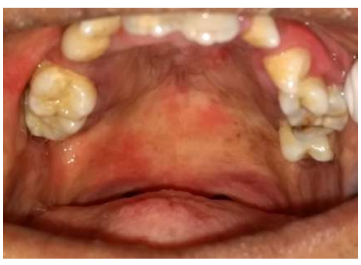

Fig. 3. High arched palate and compromised periodontal status

Skull projections and Intra oral radiographs revealed open mid sagittal sutures and heavy alveolar bone loss with respect to 27 giving it a floating tooth appearance (Figs. 4 and 5). Mesio-proximal caries with respect to 25 with ill-defined periapical radiolucency was seen. OPG examination was not possible as the patient got uncooperative with time. On the basis of general clinical and radiographic examination a provisional diagnosis of Down syndrome was considered and further investigations were advised to the patient in order to confirm the diagnosis.

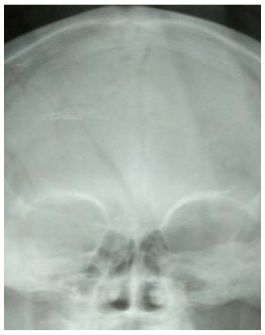

Fig. 4. Open mid-sagital sutures

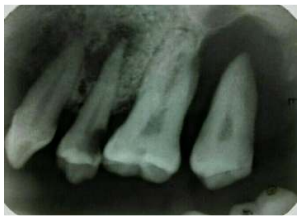

Fig. 5. IOPAR showing floating tooth appearance

2.2 Investigations

Complete blood picture revealed Hemoglobin levels of 7 gm %. Thyroid examinations revealed elevated TSH levels. Glycosylated hemoglobin gave fair results of 5.2%. Chromosomal analysis (GTG-Banding)) revealed male karyotype with trisomy 21 pattern in all the metaphases analyzed which confirmed Down syndrome (Fig. 6). As these findings were confirmed patient was sent for general cardiac examination to a cardiologist.

3. DISCUSSION

An extra chromosome is solely responsible for all the ill effects on multiple systems of human body in Down syndrome which leads to various abnormalities in skeletal and oral systems. Craniofacial anomalies ranges from brachycephalic head, flat face and open sutures because of delay in closure. Strabismus, under-developed mid face, prognathic mandible, cleft lip and palate, hypotonic upper and lower lips are also common in many patients [5]. The average IQ of a Down syndrome patient is equivalent to that of a 8 or 9 year old which is about 50, but variations are evident [6].

Irrespective of race and ethnicity some exclusive combination of facial features are seen in patients with Down's syndrome. Most cases appear short statured with a short neck and hypoplastic mid-face, with epicanthal fold placed higher over inner canthus giving a characteristic slanted-eyes appearance. Ophthalmic features are the "Brushfield spots" (due to connective tissue deposition) and less frequently eye infections. Hypoplasia of middle face could be related to the presence of underdeveloped para-nasal sinuses, giving rise to a sloping forehead and a flat face. Malalignment of teeth (Class III malocclusion), high arched "stair palate" and mandibular prognathism are commonly added in the facial appearances of these patients [7].

Down syndrome patients have an increased sub gingival bacterial growth pertaining to loss of periodontal attachment which in turn leads to compromised periodontal health [8]. Mental retardation is exclusively seen in all patients with Down syndrome. Mild effect of this condition will allow them to perform normal functions in a workshop like environment. Dementia is seen in 30% of patients with Down syndrome, and premature aging is common. Dental therapy is aimed at treating dental caries and correcting periodontal disease. Orthodontic intervention and surgical procedures can be performed in well functioning individuals only, if required. Guidelines established by the American Heart Association for antibiotic prophylaxis should be followed [9].

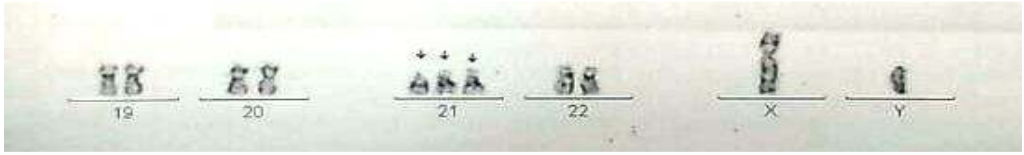

Fig. 6. Genetical analysis showing trisomy 21

Cardiac risk is estimated at 71% in these patients. Increased susceptibility to infection, autoimmune disorders, malignancies and acute lymphoid and non-lymphoid leukemia (1.5%), congenital cardiac anomalies with or without other lesions (45%) is seen [10]. Most affected embryos abort spontaneously early in the first trimester and those that survive into the second trimester experience high risk of infant death. Advanced maternal age is a strong factor which has shown prevalence increasing from 0.6 to 4.1 per 1,000 between age 15-45 [11,8]. Our patient was not aware of his condition. Huge risk of life and lack of awareness could be fatal in such patients and they must be treated like normal patients. Proper intra oral and extra oral examination along with diagnostic tests confirmed the diagnosis of Down syndrome in our case.

4. CONCLUSION

Down syndrome is not just a disease it's a complex of disorders. Usually, this condition is highlighted congenitally but lack of awareness among low socio-economic groups and less advanced medical facilities can make this condition go unnoticed. Dental professionals very often deal with patients of Down syndrome because of its exclusive intra-oral features and treatment should include a multi-disciplinary approach. In most cases dental specialists are the first ones to diagnose this condition as seen in this particular case. Hence, thorough knowledge of Down syndrome is of utmost importance in the field of dentistry.

CONSENT

All authors declare that 'written informed consent was obtained from the patient (or other approved parties) for publication of this paper and accompanying images.

ETHICAL APPROVAL

All authors hereby declare that all experiments have been examined and approved by the appropriate ethics committee and have therefore been performed in accordance with the ethical standards laid down in the 1964 Declaration of Helsinki.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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