



Case Report

JMR 2017; 3(3): 107-109

May- June

ISSN: 2395-7565

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www.medicinarticle.com

Received: 01-06-2017

Accepted: 26-06-2017

Clinical Presentation of Down's syndrome: A case report

Anoop Kurian Mathew¹, Antony Shijoy Amaladas², Anis Ahmed¹, Shaul Hameed³

¹ Reader, Department of Oral Medicine and Radiology, Indira Gandhi Institute of Dental Sciences, Ernakulam, Kerala, India

² Senior Lecturer, Department of Orthodontic and Dentofacial Orthopedics, Sree Mookambika Dental College, Kulasekharam, Tamil Nadu, India

³ Senior Lecturer, Department of Oral Medicine and Radiology, Yenepoya Dental College, Yenepoya University, Mangalore, Karnataka, India

Abstract

Down's Syndrome or Trisomy 21 is the most common chromosomal abnormality occurring in human leading to mental retardation and stunted growth. It is considered as a genetic disorder with a defect in all or a part of a third copy of chromosome 21. This article highlights varied clinical presentation of 18 year old patient with Down's Syndrome.

Keywords: Chromosome, Trisomy 21, Syndrome.

INTRODUCTION

Down's syndrome or Trisomy 21, previously known as "Mongolism" ^[1], is the most common chromosomal abnormality among humans. The condition was named after a British physician, John Langdon Down, who coined and described the syndrome in 1866. The most prominent characteristics of the condition includes physical growth delays, subnormal mentality and a severe degree of intellectual disability which is one of the most common feature.

CASE REPORT

Here, we present an 18 year old female [fig 1] who reported to our department with a complaint of painful decayed teeth in her mouth. Patient gave a history of pain for the past 1 month and underwent medication for the same. On general examination, patient is poorly oriented, short stature and is mentally challenged. On extra oral examination, classical signs appreciated which include the following:

Face:

- Brachycephalic skull [fig 1]
- Flat facies with ocular hypertelorism
- Slanting palpebral fissures
- Saddle nose deformity
- Midface deformity with underdeveloped nasal bone [fig 2]
- Flattening of nasal bridge [fig 2]
- Retruded maxilla which makes a protruded mandible

Extremities:

- Broad fingers and palms [fig 3]
- Broad toes
- Wide spacing between first and second toes [fig 4]

On Intraoral examination, multiple teeth were decayed including root stumps [fig's 5,6] and few missing. With respect to 36, there was a sinus opening with pus discharge on the buccal aspect. Tongue appeared to be fissured & is larger than normal (macroglossia) [fig 7] which protrudes on mastication. Overall poor oral hygiene leading to periodontitis noticed.

*Corresponding author:

Dr. Anoop Kurian Mathew

Reader, Department of Oral Medicine and Radiology, Indira Gandhi Institute of Dental Sciences, Indira Nagar, Nellikuzhy P.O, Kothamangalam, Ernakulam District, Kerala, India

Email:

drakm2017[at]gmail.com

Radiographic finding include panoramic radiograph status which revealed multiple decayed teeth with evident periapical abscesses and few missing. Also there is generalized reduction in the alveolar bone height extending below the middle third of roots of most teeth suggestive of vertical bone loss [fig 8].

Lateral skull view revealed a hypoplastic maxilla (underdeveloped middle third face) which makes a prognathic mandible [fig 9].



Figure 1: Downs Syndrome patient with Brachycephalic skull

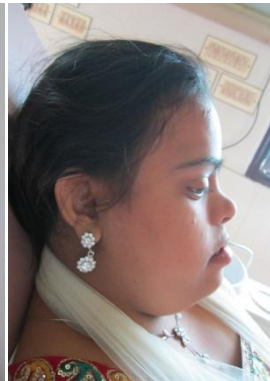


Figure 2: Mid face deformity



Figure 3: Broad fingers and palms



Figure 4: Spacing between first and second toe



Figure 5: Maxilla showing grossly decayed teeth



Figure 6: Mandible showing grossly decayed teeth



Figure 7: Macroglossia



Figure 8: Panoramic radiograph of the patient



Figure 9: Lateral skull radiograph showing hypoplastic maxilla

DISCUSSION

Down's syndrome is the most common chromosomal abnormality associated with a number of other phenotypes including learning disability, heart problems, leukemia in childhood as well as Alzheimer's disease [2]. Its incidence is around 1:660 live births [3]. Development of Down's syndrome brain is associated with reduction in the neuronal number and abnormal neuronal differentiation. It has been previously reported that Down's syndrome neuron degenerate subsequently and undergo apoptosis. However, Busciglio J *et al*, reported that degeneration of these neurons can be prevented by treating with free radical scavengers [4].

Hackshaw AK *et al* in their study, proposed a new screening method in which measurements obtained during 1st and 2nd trimester are

integrated to provide the risk status of having pregnancy with DS. Moderate to severe intellectual disability occur as a constant feature, with IQ's ranging from 20 to 85 [5]. Seizure disorders are present in 5-10% of patients. Infantile spasms are the most common seizures observed in infancy [5]. In the present case, seizure history has been reported by the patient before 10 years of age.

Previous study by Bertelli *et al* reported that the occurrence of Down's syndrome independent of maternal age presents an evidence for other risk factors [3]. Kennard *et al* in his review stated that there are a number of ultrasound markers in Down's syndrome which includes nuchal fold thickness, cardiac abnormalities, duodenal atresia, femur length & pyelectasis [6].

According to Wishart *et al*, from a very early age it would appear that these patients avoid opportunities for learning new skills ^[7]. Incidence of congenital heart diseases in Down's syndrome is about 60% ^[3]. However, present case has no history of any congenital heart diseases.

CONCLUSION

Down's syndrome or Trisomy 21 is considered as the most common chromosomal abnormality occurring in new born infants. Several theories have been put forward to increase our understanding regarding the insight of the disease. This case report clearly dictates most varied clinical aspects of the conditions which helps in the proper diagnosis of the condition.

Conflicts of Interest:

The authors do not state any conflict of interest.

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