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CLNICAL PRESENTATION OF DOWN'S SYNDROME : A TRISOMY CASE REPORT

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Abstract:

Downs syndrome is defined as a genetic disorder caused by the presence of all or a part of a third copy of chromosome 21. Downs syndrome is the most common chromosomal abnormality in humans. This article reports a case of downs syndrome in a 21 year old female patient who came to our Department of Periodontology and Oral Implantology describing the classical signs.

Key Words: Chromosome 21 trisomy, mental retardation, stunted growth

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INTRODUCTION

Downs syndrome is defined as a genetic disorder caused by the presence of all or a part of a third copy of chromosome 21. Downs syndrome is the most common chromosomal abnormality in humans. Downs syndrome was named after John Langdon Down, the British physician who described the syndrome in 1866. This syndrome is also known as Trisomy 21, named by Dr.Jecome Lejeune as a common chromosomal abnormality.

This syndrome is typically associated with physical growth delays, a particular set of facial characteristics and a severe degree of intellectual disability, which is opredominant characteristic found in all most all of them associated with this syndrome. This syndrome can be identified in a newborn by direct observation or in fetus by prenatal scanning.

CASE REPORT

Here, we present a 21 year old female patient who reported to the Department of Periodontology and Oral Implantology, Sri Guru Ram Das Institute of Dental Sciences & Research, Sri Amritsar, with a compliant of painful decayed teeth with bleeding gums in her mouth. Patient gave a history of pain for the past 1 month and underwent medication for the same. On general examination, patient was short statured and mentally challenged. On extra oral examination, patient had saddle nose deformity, midface hypoplasia that is retruded maxilla and protruded mandible. On extra oral examination, classical signs appreciated which include the following:

Face:

- Brachycephallicshull [fig 1]
- Flat facies with ocluar hypertelorism
- Slanting palpebral fissures
- Saddle nose deformity
- Midface deformity with underdeveloped nasal bone

- Flattening of nasal bridge
- Retruded maxilla which makes a protruded mandible

Extremities:

- Broad toes
- Wide spacing between first and second toes [fig 2]
- Broad fingers and palms [fig 3]

On Intraoral examination, multiple decayed teeth including root stumps [fig 4]. Tongue appeared to be fissured & larger than normal (macroglossia) which protrudes on mastication. Overall poor oral hygiene leading to periodontitis noticed.

Radiographic finding include panoramic radiograph status which revealed multiple decayed teeth. 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 5]. This case was presented for its rarity and classical clinical presentation.

DIFFERENTIAL DIAGNOSIS

Before confirming a diagnosis of Down syndrome, the following conditions are ruled out:

Trisomy 18: This condition, which also is called Edward syndrome, is caused by an extra copy of chromosome 18. Babies with Trisomy 18 usually have severe mental retardation and heart problems. They also may have problems of the stomach, digestive tract, reproductive organs, and urological tract. Trisomy 18 often is fatal in infancy or early childhood.

Multiple X chromosomes: Males normally have one X chromosome and one Y chromosome. (Females have two X chromosomes, but this condition does not occur in females.) In some cases, male infants are born with one or more additional X chromosomes in each cell, increasing the number of chromosomes beyond the normal total of 46. Multiple X chromosomes can cause decreased IQ, a distinct facial appearance and speech, skeletal, and coordination problems in males.

Zellweger syndrome and other peroxisomal problems: Peroxisomes are the part of a cell's structure that helps the body get rid of toxins. Some children with peroxisomal disorders do not have enough of these elements and some do not have any at all. In the case of Zellweger syndrome, peroxisomes are either deficient or missing in the liver, kidneys, and brain. Children with this condition have vision problems, an enlarged liver, and too much iron and copper in their blood. There is no cure for Zellweger syndrome and the condition usually is fatal within 6 months of birth.

Congenital hypothyroidism: Poor feeding, poor growth, hypotonia, constipation, dry skin, fatigue. Associated with the Normal karyotype.

Klinefelter's syndrome : a male is born with an extra X chromosome (XXY). This is Klinefelter syndrome. It's also called Klinefelter's or XXY.

Turner syndrome, a condition that affects only females, results when one of the X chromosomes (sex chromosomes) is missing or partially missing. Turner syndrome can cause a variety of medical and developmental problems, including short height, failure of the ovaries to develop and heart defects.

Down syndrome typically is recognized at birth, Turner syndrome often is not recognized until adolescence,and many men with Klinefelter syndrome are too difficult to get diagnosed.[1]

DISCUSSION

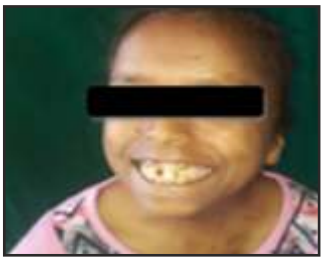

Figure 1

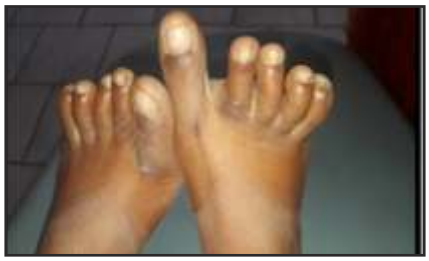

Figure 2

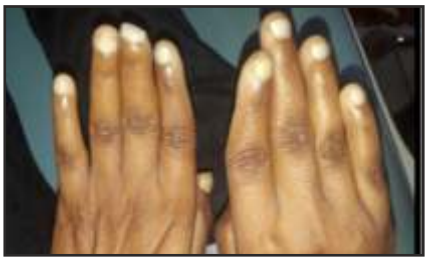

Figure 3

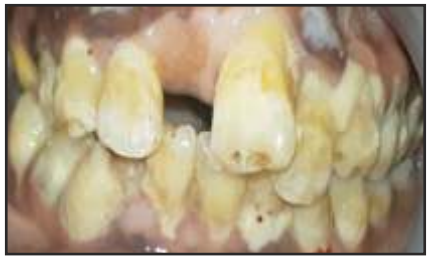

Figure 4

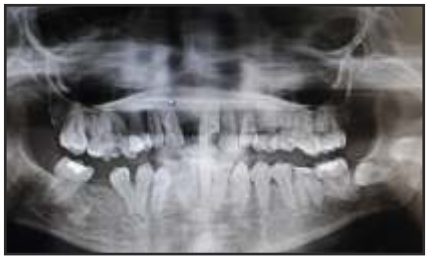

Figure 5

The existence of two chromosomal abnormalities in the same individual is relatively a rare phenomenon. Double aneuploidy leading to trisomy and / or monosomy of two different chromosomes arises because of two meiotic non-disjunctional events. Both these aneuploidies could have the same or different parental origin.[2] The coincidence rate of both Down and Klinefelter syndromes in the same individual is estimated to lie in the range 0.27 to 0.7 × 10-5.[3]However, neonatal survey data has revealed that the incidence of XXY and trisomy 21 double trisomy at birth is higher than expected from the incidence of either alone.[4]On the other hand, lower values of XXY pattern recorded in older boys and men with Down syndrome suggest that there might be an increased selection against these individuals after birth.[5]

The signs and symptoms of Downs syndrome are charaterised by neotenization of brain and bodies. Management statergies such as Early childhood intervension, screening from common problems, medical treatment when indicated, a conductive family environment and vocational training can improve the overall development of children with Downs syndrome. Almost 99.8% of patients with Downs syndrome experience mental retardation and stunted growth.[6] In 69% of high arched or oval palate[7] is seen. This elevated figure can be attributed to an underdeveloped maxilla. In our case,there were features like macroglossia,a common characteristic with fissuring in the anterior 2/3rd of dorsum of tongue. Moderate to severe intellectual disability occur as a constant feature, with IQ's ranging from 20 to 85.[8]According to Wishart et al, from a very early age it would appear that these patients avoid oppurtunities for learning new skills.[9]

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.

The choice for the oral rehabilitation technique is fundamental to treatment success. The dentist must have an interdisciplinary approach to be scientifically based, technically competent and be socially integrated in order to provide the best care possible, and promote the inclusion of individuals with special needs to health services.

Syndromic patients should be treated with all resources provided by modern Dentistry, ranging from a simple tooth extraction to the most audacious rehabilitating procedures, reestablishing the oral function and aesthetics of individuals, regardless of their physical or neurological condition. Although more experience is needed before dental implants can be considered a suitable option during oral rehabilitation in people with Down syndrome, this case report shows a promising outcome.

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