

# **DOWN SYNDROME - A case report**

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## **ABSTRACT**

Down syndrome named after John Langdon Down, the first physician to identify the syndrome, is a chromosomal disorder caused by an error in cell division resulting in the presence of an additional third chromosome 21 or trisomy 21. Down syndrome is the commonest autosomal chromosomal anomaly with an incidence of 1 in 600 to 1000 live births in all races and economic groups. In this condition extra genetic material causes delays in the way a child develops, both mentally and physically.

The physical features and medical problems associated with Down syndrome can vary widely from child to child

This article presents a case report of a 21-year old male patient with the classical features of Down syndrome .The skeletal and soft tissue features, aberrations in dental, periodontal and caries characteristics are discussed.

Key words: Down syndrome, autosomal, Trisomy 21

Introduction:

Down syndrome is an easily recognized congenital, autosomal anomaly characterized by generalized physical and mental deficiencies. It affects between 1 in 600 and 1 in 1000 live births. Down syndrome is named after John Langdon Down, the British doctor who first described the condition in 1887 [1,2].

Down syndrome is a chromosomal disorder caused by an error in cell division that results in the presence of an additional third chromosome 21 or “ trisomy 21.” The incidence of Down syndrome rises with increasing maternal age. The features of Down syndrome can range from mild to severe. Usually, mental development and physical development are slower in people with Down syndrome [1,3].

Most people with Down syndrome have IQs that fall in the mild to moderate range of mental retardation. They may have delayed language development and slow motor development [3].

Some common physical signs of Down syndrome are flat face with an upward slant of the eye, short neck, and abnormally shaped ears, small hands and feet ,deep crease in the palm of the hand, poor muscle tone, loose ligaments and white spots on the iris of the eye [3,4].

The other health conditions that are often seen in people who have Down syndrome include skeletal problems, dementia, thyroid dysfunctions, celiac disease, hearing problems, congenital heart disease, intestinal problems such as blocked small bowel or oesophagus and eye problems such as cataracts[4].

Anomalies related to the dentition:

About 35% to 55% of individuals with Down syndrome present with microdontia in both the primary and secondary dentition. Clinical crowns are frequently conical, shorter, and smaller than normal, and the roots are shorter as well. Tooth agenesis or defective development is more likely in patients with Down syndrome. The teeth most affected by agenesis are mandibular central incisors, followed by maxillary lateral incisors, second premolars, and mandibular second premolars. Canines and first molars are rarely affected. There is a delayed eruption in both the deciduous and permanent dentition. The central incisors still erupt first and the second molars are usually last but in between, the sequence of eruption varies greatly [5,6].

A relatively high frequency of mal-alignment is seen in both the deciduous and permanent dentition in individuals with Down syndrome. Some individuals have open-mouth which causes in-coordination of the lips, and cheeks in swallowing and speech. Individuals with habitual mouth breathing tendencies are more susceptible to periodontal disease. The prevalence of dental caries in patients with Down syndrome is low [5, 6].

Soft tissue features:

Patients with Down syndrome have a high arched V-shaped palate, which is caused by deficient development of the midface, affecting the length, height, and depth of the palate. Perioral muscles are affected by characteristic muscle hypotonia. This leads to a descending angle of the mouth, elevation of

the upper lip, and an everted lower lip with tongue protrusion. The hypotonic tongue shows characteristic imprints of teeth along the lateral border. A scalloped (crenated) and plicated (scrotal) tongue is also common. A small oral cavity with a relatively large tongue causes mouth breathing, which is a common cause of chronic periodontitis and xerostomia[7,8].

Skeletal features:

Most cases of Down syndrome present with mandibular over jet, anterior open bite, posterior cross bite, Class III occlusion and protrusion of the maxillary and mandibular incisors. The freeway space is about three times the normal value of 2 to 3 mm and the mid-face is more deficient than the mandible [5].

Case report:

A 19-year-old male patient, fourth birth order, born to non-consanguineous couple aged 45 and 49 years was referred to the Department of Orthodontics with the chief complaint of spacing between teeth in the upper as well as the lower jaws. The patient also complained of missing teeth and desired replacement of the same.

The patient was moderately built and had mild degree of mental retardation. He had most of the common dysmorphic features of Down syndrome like

short palpebral fissures, epicanthic folds of the eyelid causing almond shaped eyes; mongoloid slant, hypertelorism, depressed nasal bridge, and bilateral clinodactyly. The patient also presented with a short neck, and abnormally shaped ears, small hands and feet, deep crease in the palm of the hand, single palmer fold known as Simian crease, poor muscle tone, and white spots on the iris of the eye known as Brushfield spots. The patient also presented with excessive space between large toe and second toe.

Family history was non-significant. His medical history was unremarkable and he was not on any medication.

The facial profile showed a flat face with an upward slant of the eye and short palpebral fissure. The profile view revealed depressed nasal bridge and infra-orbital margins.

Fig 1a & 1b: Facial view showing a flat face with an upward slant of the eye and a descending angle of the mouth.



Fig. 1A



Fig. 1B

Intra oral examination revealed generalized spacing of teeth in relation to the maxillary and mandibular arches and the clinical crowns were conical and smaller than normal (microdontia). The patient had a high arched palate, relatively small mouth and macroglossia with a crenated tongue. He also presented a descending angle of the mouth, and everted lower lip with tongue protrusion.

Figures 2a, 2b & 2c: Intra-oral view showing generalized spacing in relation to the maxillary and mandibular arches, conical clinical crowns and macroglossia.



Fig. 2A



Fig. 2B



Fig 2C

The patient had a Class III occlusion with protrusion of the maxillary and mandibular incisors. The patient had a relatively low caries incidence and fairly good oral hygiene status.

The orthopantomogram (Figure 3) revealed generalised spacing of teeth in the upper as well as the lower arches, multiple missing teeth and decayed teeth as well.



Figure 3: Orthopantomogram showing generalized spacing and multiple missing teeth.



Figure 4: Eyes showing white spots on the iris known as Brushfield spots.

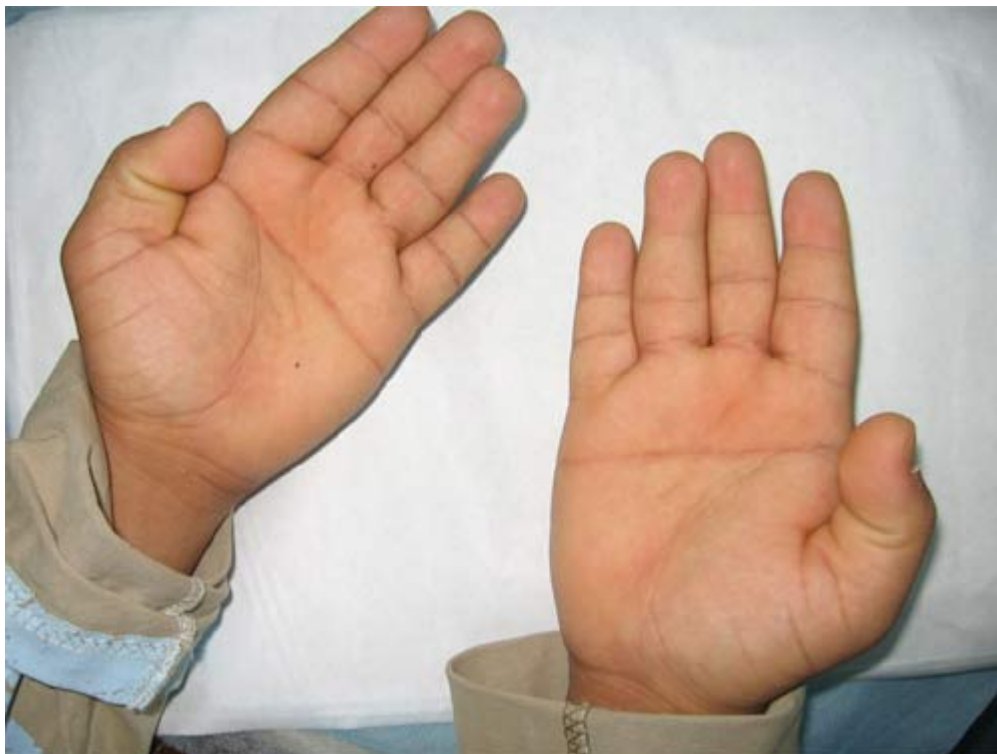


Figure 5: Palms showing deep crease; single palmer fold known as Simian crease.



Figure 6: Feet showing excessive space between large toe and second toe.



Figure 7: Study Model showing generalized spacing in the upper and lower arches.

Considering the physical and mental development, clinical, oral and radiographic findings the patient was diagnosed as a case of Down syndrome and full mouth rehabilitation was scheduled restoring the structural and functional integrity.

Discussion:

Down syndrome is an autosomal chromosomal anomaly resulting from trisomy of all or a critical part of chromosome 21. It affects approximately 1 in 600 to 1000 live births. Despite the development of prenatal diagnosis, the incidence of Down syndrome births is predicted to remain static or even to increase over the next decade, partly due to increased maternal age. Many of the medical and physiological characteristics of this condition have direct consequences for the oral health of subjects affected and indirect consequences for the quality of life of the affected persons and their guardians.

Dental care for the patient with Down syndrome can be achieved in the general practitioner's office in most instances with minor adaptations.

Although this population has some unique dental care needs, few patients

require special facilities in order to receive dental treatment. Adequate dental health care for persons with developmental disabilities is a major health need.

Treatment objectives for any population with developmental disabilities should be the same as that of normal patients. Treatment plans may need to be adapted as necessary due to each individual's condition, but the overall goal should be to provide as comprehensive treatment as possible. Areas of dental care such as cosmetic dentistry, orthodontics, prosthodontics, and reconstructive oral surgery should be considered in these patients.

#### References:

1. Regezi S. Oral pathology. Clinical pathologic correlations, 1st ed. Philadelphia: Saunders, 1989: 450-51.
2. Muller RFO, Young IDO, Emery's elements of medical genetics. 11<sup>th</sup> ed. Churchill Livingstone; 2001.pp-52.
3. Vogel F, Motulsky AGO. Human genetics, problems and approaches. 2<sup>nd</sup> ed. Springer-Verlag; 1996 pp-59.
4. Smith WB. Recognizable pattern of human malformations, 4th edition. Jones LK, ed. Philadelphia: Saunders; 1988: 10-12.
5. Sassouni V, Forrest E. Dentofacial pathology related to malocclusions. Orthodontics in Dental Practice. St. Louis: Mosby, 1971: 169-97.
6. Wilson MD. Special considerations for patients with Down's syndrome. ODA Journal 1994 (winter); 184 (3): 24-25.

7. Russell BG, Kjær I. Tooth agenesis in Down syndrome. *Am J Med Genetics* 1995; 13; 55(4): 466-71.
8. Sterling ES. Oral and dental considerations in Down syndrome. *Down Syndrome Advances in Medical Care*. In Lott I, McCoy E, editors. New York: Wiley-Liss; 1992: 135-45.

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