

## Down's Syndrome - Case Series

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## Abstract

Down's syndrome is a chromosomal disorder caused by an error in cell division which results an additional third chromosome 21 or trisomy 21. The incidence of Down syndrome is 1 in 600 to 1000 live births. This condition leads to developmental delay, both mentally and physically. It is named by a British physician John Lang Down. There is no treatment but can be managed by early childhood intervention and proper care can improve quality of life.

## Introduction

Down's syndrome (DS) 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 Longdon Down, the British doctor who first described the condition in 1887 [1,2].

DS is genetic disorder caused by the presence of all or a part of a third copy of chromosome 21 [3]. Down's syndrome can range from mild to severe. Usually, mental development and physical development are slower in people with DS [1,3].

DS can be identified in a newborn by direct observation or in fetus by prenatal scanning [4]. Some common physical signs of DS are flat face with an upward slant eyes, short neck, abnormal shaped ears, small hands, single crease in the palm of the hand, poor muscle tone, loose ligaments and white spots on the iris of the eye [3,4].

## Anomalies related to the dentition

About 35% to 55% individuals with DS 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 DS. 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 [4,5].

Perioral muscles are affected by characteristic muscle hypotonia. This leads to 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 [6,7].

## Skeletal features

Most cases of DS 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 mid-face is more deficient than the mandible [4].

## Case Report 1

A male baby of 7 months was admitted to Mahatma Gandhi Memorial Hospital, Warangal, Telangana, admitted with complaints of fever, cold and cough. There was no history of consanguinity marriage of their parents. Based on karyotype examination the child observed with down characteristics such as low set of ears, epicanthal folds, simian crease, depressed bridge of the nose, protruding tongue, generalized hypotonia (Figure 1).



Figure 1: Shows downs baby.



Figure 2: Shows downs baby.

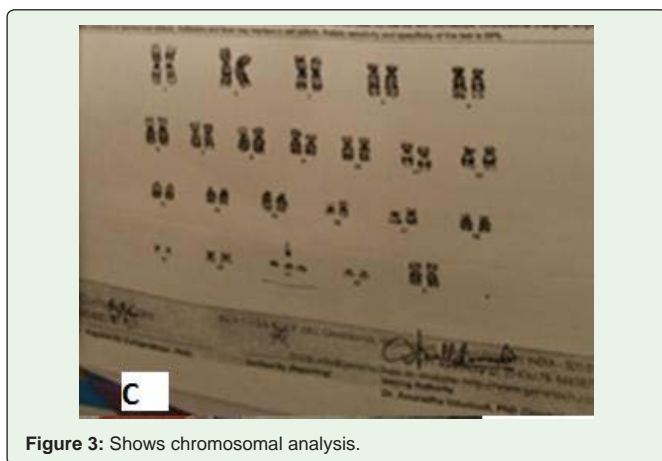


Figure 3: Shows chromosomal analysis.

### Case Report 2

A female baby of 10 months was admitted to Mahatma Gandhi Memorial hospital, Warangal with complaints of fever, cold, shortness of breath, known case of down syndrome, the karyotypic report shows narrow, slanting, closely set palpebral fissures, thick protruding tongue, high palatal arch and flat nasal bridge. No history of consanguinity marriage of their parents (Figures 2 and 3).



Figure 4: Shows gap between the teeth.



Figure 5: Shows downs with developmental delay.

### Case Report 3

A female baby of 10 years was admitted to Mahatma Gandhi Memorial Hospital, Warangal. With complaints of fever, cold, cough since one week. Child phenotypically down's syndrome, history of behavioral abnormalities, developmental delay positive at on treatment for abnormal behavior. Child shows gaps between the teeth, this is one of the common symptoms of the downs (Figures 4 and 5).

### Discussion

The signs and symptoms of DS are characterized by neotenzation of brain and bodies. Management strategies as early childhood intervention, screening from common problems, medical treatment when indicated, a conducive family environment and occasional training can improve the overall development of children with DS. Almost 99.8% [8] of the patients with DS experience mental retardation and stunted growth. In 69% [9] of high arched or oral palate is seen with 1-3% [10] incidence of maxillary canine impactation, unilaterally [9]. Canine impactation more prevalent among the female patients.

### Conclusion

There is no single, standard treatment for DS; People with DS can receive proper care while living at home and in the community.

People with DS are at a greater risk for a number of health problems (heart defects, mental retardation, digestive problems) those who do not have DS. So, in order to avoid these early intervention is needed. Early intervention refers to a range of specialized programs like speech therapy, occupational therapy, physical therapy.

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