

Downs Syndrome: A Case Report

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ABSTRACT

Down syndrome (DS) or Down's syndrome, also known as trisomy 21, is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21. It is a common chromosomal abnormality.

Keywords: Down syndrome, chromosomal abnormality, trisomy 21.

INTRODUCTION

Down syndrome is named after John Langdon Down, the British physician who described the syndrome in 1866. World Down Syndrome day is celebrated on 21st March.¹ Some of the characteristics of syndrome are stunted growth, flattened nose, mental retardation, small teeth, atypical fingerprints, short neck, hypotonia, shortened hands, brachycephaly, congenital heart disease smaller genitalia, single transverse palmar crease, eyelid crease, macroglossia, shortened extremities, epicanthic fold, oval palate, strabismus, low-set and rounded ear, Brushfield spots(iris).

Case Report

A 12 year old female patient visited the dental OPD of Jaipur Dental College with chief complain of pain of tooth in the lower left back region since 1 month. Pain was sharp shooting and intermittent in nature which aggravates while chewing and subsides after the removal of stimuli. Medical, dental and family history was non significant. On extraoral examination shape of the head was brachycephalic with flat nasal bridge and intraorally teeth were small and macroglossia was present. According to psychiatrists prescription which was brought by patients mother to the OPD showed that the patient had stunted growth. Hence based on clinical features diagnosis of Downs Syndrome was given and chronic irreversible pulpitis in relation to 7D. Patient was sent to pedo for the needful.



Fig. 1: showing brachycephalic face, flat nasal bridge, slanted eyes

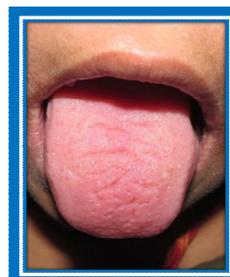


Fig. 2: showing macroglossia and fissured tongue

DISCUSSION

Down syndrome (DS) or Down's syndrome, also known as trisomy 21, is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21.² It is typically associated with physical growth delays, characteristic facial features and mild to moderate intellectual disability³ which was seen in our patient as well. The average IQ of a young adult with Down syndrome is 50, equivalent to the mental age of an 8 or 9 year old child, but this varies widely.⁴ But in our case IQ was not mentioned by the concerned psychiatrist. Down syndrome can be identified

during pregnancy by prenatal screening followed by diagnostic testing, or after birth by direct observation and genetic testing. Since the introduction of screening, pregnancies with the diagnosis are often terminated.^{5,6} Regular screening for health problems common in Down syndrome is recommended throughout the person's life. Proper care along with education have shown to improve the quality of life. Life expectancy is around 50 to 60 years in the developed world with proper health care.⁷

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