Case Report

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A Rare Case of Rubinstein-Taybi Syndrome with Amelogenesis Imperfecta

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Abstract

Rubinstein-Taybi syndrome (RTS) is a rare congenital disorder characterized by typical facial features, broad thumbs and toes, with mental retardation. Additionally, tumors, keloids and various congenital anomalies including congenital heart defects have been reported in RTS patients. Here, we report a case of 11year-old female with a Rubinstein-Taybi syndrome associated with amelogenesis imperfecta having normal mental status with no cardiac abnormality.

Keywords: Rubinstein-Taybi Syndrome**;** Amelogenesis Imperfecta; Congenital Anomalies; Broad Thumbs and Toes.

Introduction

The Rubinstein-Taybi syndrome (broad thumb-hallux syndrome) is a rare congenital disease with prevalence 1:125,000 of live-born children. It is characterized by multiplex malformations, which includes growth and psychomotor retardation. Till date, over 1000 cases have been described in the literature.[1]The pediatrician, Dental surgeon and the orthopedic community should be aware of the syndrome and its associated complications. In this paper we report a case of Rubinstein-Taybi syndrome associated with amelogenesis imperfecta having normal mental status and with no cardiac anomaly.

Case report

A 11 year-old girl reported with her father to the Department of Oral Medicine and Radiology, Bapuji Dental College and Hospital, with the chief complaint of missing upper front teeth since two years. There was no relevant past medical and dental history. Her family history revealed that she was the only daughter of her parents with no history of consanguineous marriage between her parents and born after full term normal delivery with no known developmental defects. All her milestones of growth were reported to be within normal limits.

Patient was conscious, cooperative with well orientation of time place and person having loving, friendly and happy personality with short attention span.

On *General physical examination*, patient was moderately nourished and had short stature (116.5 cm in height) (Fig 1). There was decreased ratio of upper to lower segment of the body. There was no pallor, icterus, cyanosis, clubbing, edema and all her vital signs are within normal limits. Her both upper and lower extremities were plump and shorten from trunk to phalanges (acromelic shortening). Her fingers of hands and toes were normal except the great toe (Fig 3) were short, broad, stubby and plumped (Fig 2). On the palmer aspect of her left and right hand, there was prominent simian crease. All

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