

Broad Thumb-Hallux Syndrome: A Diagnosis Made on Clinical Findings

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An 18-year-old female visited our dental clinic for routine dental checkup. Her medical history was suggestive of low birth weight, delayed milestones. Echocardiography, abdominal ultrasonography and fundus examination of the eyes were normal. Past dental history revealed delayed eruption of primary and permanent tooth. Extra oral examination revealed microcephaly, hypertelorism, broad nasal bridge with the nasal septum extending below the alae nasi [Table/Fig-1a]. She was 122 cm in height [Table/Fig-1b]. She was also noted to have bilateral clinodactyly of fifth fingers of hand with broad thumbs and also had brachydactyly of both feet with broad great toes [Table/Fig-2a&b]. Intraoral examination revealed poor oral hygiene with a high arched palate. Talons cusps were present on the right and left maxillary lateral incisor [Table/Fig-3]. Psychometry revealed low IQ (65). Hand wrist radiograph revealed clinodactyly of fifth finger of right and left hand [Table/Fig-4].

Due to patient's poor economic status and the regional unavailability of genetic test for *CREBBP* and *EP300* genes mutation, the authors were unable to procure genetic analysis for this patient. Thus, based on the available clinical findings and investigation the patient was diagnosed as having broad thumb-hallux syndrome. In this case comprehensive dental treatment could not be planned under



[Table/Fig-3]: Talons cusps in the right and left maxillary lateral incisor.

[Table/Fig-4]: Clinodactyly of fifth finger of right and left hand shown by black arrow in the hand wrist radiograph.

sedation or general anesthesia due to the reluctance of the parents for the procedure. The parents were advised to have regular recall visits.

Rubinstein-Taybi Syndrome (RSTS) or broad thumb-hallux syndrome is a sporadic congenital mental retardation syndrome. Individuals with RSTS have noticeable clinical features which include postnatal growth deficiency with typical dysmorphic facial features. Broad thumbs with broad great toes are present in almost all RSTS cases. In India prevalence of RSTS is 1 in 100,000 to 125,000 [1]. Worldwide prevalence of RSTS is approximately one in every 125,000 newborns [2,3].

RSTS is a rare autosomal dominant disorder characterized by short stature, dysmorphic facial features with prominent forehead, arched eyebrows, slanted palpebral fissures, grimacing or atypical smile, microcephaly, broad thumbs and first toes. Congenital cardiovascular abnormalities of RSTS include ventricular septal defect, patent ductus arteriosus, coarctation of aorta. Additional abnormalities are lacrimal duct obstructions, glaucoma with obstructive sleep apnea. Clinodactyly of fifth fingers and beaked nose with the nasal septum extending below the alae are present in over half the reported cases. There are no standard international guidelines and diagnostic criterion available for diagnosing RSTS [2].

Broad thumb-hallux syndrome is characterized by a variety of orofacial features including limited mouth opening, micrognathia with pouting lower lip, crowding, high arched and narrow palate. Individuals with RSTS have striking oral features including talon cusps, hypodontia with enamel hypoplasia [3].

Kumar S et al., concluded from his retrospective study that RSTS is an archetypical syndrome with multiple congenital malformations. The clinical management for most RSTS cases mainly depends on a multidisciplinary approach. A prior diagnosis of RSTS with medical genetics team counseling helps in better treatment planning and management [1].

Mutations in the genes encoding the Cyclic-AMP-Regulated Enhancer Binding Protein (*CREBBP*) in chromosomal region 16p13.3 and the E1A-binding protein p300 (*EP300*) in chromosomal region



[Table/Fig-1]: a) Broad nasal bridge with the nasal septum extending below the alae nasi; b) Short stature of the patient.



[Table/Fig-2]: a) Clinodactyly of fifth fingers of both hand with broad right and left thumb; b) Brachydactyly of both feet.

22 have been associated with approximately half of RSTS patients. Although, genetic tests act as key tool in diagnosing RSTS, majority of reported RSTS cases are diagnosed based on clinical findings [2]. Majority of RSTS patient require sedation or general anaesthesia for dental treatment. In most RSTS cases dental treatment cannot be performed in routine dental set up due to mental disability to cope with routine dental procedures [3].

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