

LETTER TO THE EDITOR



Broad thumb-hallux syndrome: A rare congenital disorder

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Broad thumb-hallux syndrome (OMIM 180849) is also known as Rubinstein Taybi syndrome. It is a congenital disorder of the rare entity with clinical features showing postnatal growth deficiency, broad thumbs and great toes, mental retardation (intellectual disability) and craniofacial dysmorphism. The etiology for its occurrence may be sporadic or autosomal dominant inheritance. Literature search exhibits very few cases of broad thumb-hallux syndrome cases. Therefore, herewith we aim to present such a rare disorder.

A 12-year-old male patient visited our department for poor oral hygiene. Patient was born to parents with consanguineous marriage. On detailed physical examination, patient exhibited broad and medially deviated right and left thumbs [Figure 1], broad great toes with clinodactyly of left fifth toe [Figure 2]. Patient also showed beaked nose with the columella protruding well below the alae nasi, bushy eyebrows, hypertelorism, folded epicanthal folds [Figure 3]. Patient was mentally retarded but did not show any other systemic problems. On intra-oral examination, high arched

palate with cleft, agenesis of some permanent teeth and crowding were evident.



Figure 2: Patient showing broad great toes



Figure 1: Broad and medially deviated thumbs



Figure 3: Patient with other features: Hypertelorism, bushy eyebrows, folded epicanthal folds and beaked nose with the columella protruding well below the alae nasi

Rubinstein-taybi syndrome (RTS) was first described in 1957, but well-delineated by Dr. Jack Rubinstein and Dr. Hooshang Taybi, in 1963.^[1] The prevalence in the general population is approximately 1 case in 3,00,000 persons and is as high as 1 case per 10,000 live births.^[2] It occurs with equal frequency in males and females and with no racial predilection.^[1,2] All RTS patients exhibit degrees of language, mental, motor and social retardation with the most usual intelligence quotient being in 30-79 range.^[3] The locus of RTS is located on band 16p13.3, which includes a gene encoding binding protein for cyclic adenosine monophosphate-response element binding protein (CREBBP or CBP gene) that is responsible for the phenotype of RTS.^[1,3]

Clinically, this syndrome is characterized by decreased growth, mental retardation, broad mediated deviated thumbs with big toes. Patients also exhibits some craniofacial abnormalities like downward slanted palpebral fissures, microcephaly, posterior rotated ears, hypertelorism, long eyelashes, downward slanted palpebral fissures, pouting upper lip and beaked nose with the columella protruding below alae nose.^[1,2,4] Other clinical features include cryptorchidism, (incomplete or delayed descent of the testes in males) congenital heart problems, gastrointestinal tract abnormalities, recurrent respiratory tract infections.^[2,4]

In the neonate, this syndrome can be confused with the De-lange syndrome, trisomy 13, Apert syndrome and Pfeiffer syndrome. Therefore, it is best to consider individuals without all the classic features of RTS as having an “incomplete form” rather than an incorrect diagnosis.^[5]

Regarding treatment, there is no definitive therapy for RTS.^[1,2] The wide spectrum of clinical manifestations

requires disease management tailored to the problems of each patient.^[3] Physical therapy, speech and feeding therapy and, special education are important supportive measures in infancy.^[3] Individualized surgical treatment based on findings in the patient, for example, a patient with a congenital heart defect may need cardiothoracic intervention. Hand and/or foot surgery frequently improves grasp, oppositional function, and comfort.^[5] The survival rate is good, with frequent reports of adult patients with RTS. Respiratory infections and complications from congenital heart disease are primary causes of morbidity and mortality in infancy.

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