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Rubinstein Taybi Syndrome: A Case Report

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I. Background

Rubinstein-Taybi syndrome (RTS) or Broad Thumb Hallux syndrome was originally described byMichail et al. in 1957. In 1963, American physician Jack Herbert Rubinstein andIranian-American Physician Hooshang Taybi had reported a thorough study of sevenchildrenof similar symptoms which later was named as Rubinstein-Taybi syndrome. It consists of agroup of congenital anomalies consisting of short, broad thumbs and great toes, psychomotorretardation, higharched palates, and histories of recurrent respiratory infections andparticular facial abnormalities.It is one of the classical recurrent pattern multiple congenital anomaly syndromes.

It is an extremely rare (1:300,000 up to 1:1,000,000), AD inheritance which occurs when a mutation of CREB binding protein (CBP) causes genetic defect related to chormosome16. The mutations in the CBP gene are responsible for RTS as well as the t (8; 16) associated acute myeloid leukemia.

	Developmental delay and Intellectual disability is universally present
	Short Stature
Eyes	Downward slant of palpebral fissures , long eyelashes,
Nose	Beaked appearance, Long columella
Ear	Thickened helix, low set ear
Hands and feet	Thumbs and great toes broad, especially distal phalanx, hallux valgus
CVS	ASD, VSD, PDA
Dental	Crowding and malpositioned teeth, caries
Behavioral	Distractibility, aggressive outbursts, and difficulty in sleeping
Neurological	Seizure

Table: Common clinical features of RTS

II. Case

A 16 years old male who did not attend school at all, presented with increasedanger,decreased sleep,physically abusive, wandering behavior for a total duration of 4years,increased for 3months, with history of delayed developmental milestones. Patient is the thirdchild of his parents out of a non-consanguineous marriage,born by spontaneous vaginal deliveryat home, cried immediately after birth, history suggestive ofmacrocephaly at the time of birth,with delay in all developmental milestones,sittingwithout support at 1year, walking at 3yearsof age, control over bowel at 12years of age.

Currently, the patient stays at home. He does simple household tasks, cuts grass for cattle.Butsomeone has to ask him to do the activities. He is able to do self-care activities by himself.He doesnot know how to read and writeand do simple calculations.

On examination, the patient has a low hairline, low set ears,large thumbs and great toes, webs overfeet.

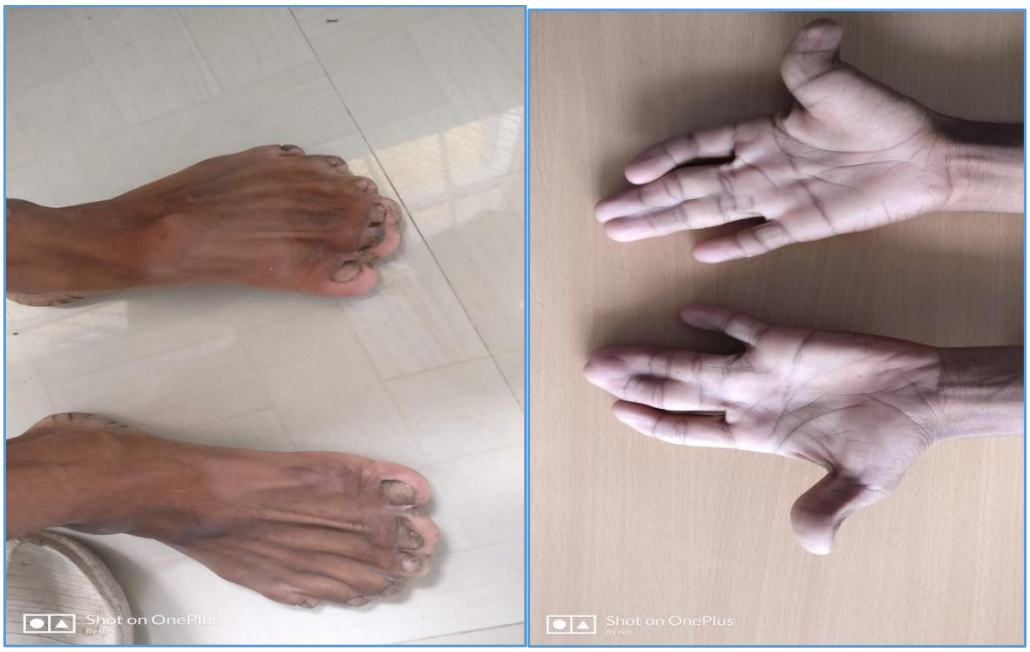

Fig: Broad thumbs and great toes, webbed toes

On mental status examination, the patient is alert and conscious.Rapport not established,eyecontact not maintained. Silly smiling was present throughout the interview. His speech is relevant,incoherent at times and his affect is irritable at times. The Patient had difficulty in comprehending the questionsthroughout the interview.

On Seguin form Board test (SFBT) - Patient does not have the concept of shapes, difficulty incomprehending instructions, stopped doing in between.

Vineland Social Maturity Scale (VSMS) - Social age found to be 7years.The calculated SQ is47, indicating a moderate level of retardation in socio-adaptive functioning.

III. Discussion

In this case report the patient presented with the above-mentioned symptoms with characteristicmorphological findings like a low set ear, low hairline, webbed toes, large thumb and great toeon VSMS indicative of a moderate level of socio-adaptive functioning.

Nayak et al. reported a case of 31years old unmarried male presented with worseningbehavioral problems for 2months,with predominant symptoms ofsuspiciousness,delusion ofreference and persecution,2nd person auditory hallucination, his intelligence quotient is 60,hisphysical anomalies are spiky hair,bushy eyebrows,prominent supraorbital ridges,low setears,long eyelashes,prominent nose,open mouth,carries in teeth talon cusps,broad thumbsand toes,cervical hyperkyphosis and scoliosis. Patient was assessed with MINI Englishversion6.0.0 and was diagnosed lifetime psychotic disorder.[3]

A case report by Cardona T. and Kline A. found with painful toenail, broad hallux,anti-mongoloid slant of eyes, heavy eyebrows, low set ears and beaked nose, plantar cleft tothe plantar first inter-space, hyper-extensible joint with mental retardation. [1]

Case Report by¨unevveroglu and Akg¨o found that 7-year-old girl accompanied by parentswith a complaint of caries and bleeding of the gingiva.She is the second child of a remoteconsanguineous couple short stature with broad thumbs, mental retardation, down the slant of thepalpebral fissures, strabismus and simple ear. The nose has a beaked appearance, broadfleshy bridge, deviated septum, and short low columella and she had cardiacabnormalities.Intra-oral findings were high-arched and cleft palate, a small mouth andmalocclusion. [2]

In a review and meta-analysis of 732 cases by Cantani and Gagliesi found the most evident symptoms arethe thumbs and first toes with broad terminalphalanges (99%). The trunk presentsmany anomalies too, as spina bifida,scoliosis, kyphosis, lordosis, hypotonia, laxligaments and above all stiff gait (83.8%).In adults were noted head circumferenceunder 50th percentile (95%) or microcephaly. Mental, motor, language and social retardationare one of the most common symptomsin RTS; it is present in 98.5% of the patientsand IQ is often under 50(87%). [5]

Based on clinical examination and psychological assessment which is supported by casereports as mentioned reported case is diagnosed as Rubenstein Taybi syndrome.

IV. Conclusion

Most of the patients with mental retardation get under-diagnosed because ofinability on the part of the clinician to detect some subtle morphological features,non-availability of genetic testing, non-affordability on the part of attendants.Therefore, whileevaluating such cases we should focus on morphological features of some commonsyndromes which present as intellectual disability with a behavioral abnormality.

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