Rubinstein Taybi Syndrome: A Case Report

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I. Background

Rubinstein-Taybi syndrome (RTS) or Broad Thumb Hallux syndrome was originally described by Michail et al. in 1957. In 1963, American physician Jack Herbert Rubinstein and Iranian-American Physician Hooshang Taybi had reported a thorough study of seven children of similar symptoms which later was named as Rubinstein-Taybi syndrome. It consists of a group of congenital anomalies consisting of short, broad thumbs and great toes, psychomotor retardation, high arched palates, and histories of recurrent respiratory infections and particular 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 chromosome 16. The mutations in the CBP gene are responsible for RTS as well as the t (8; 16) associated acute myeloid leukemia.

<table>
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<th>Developmental delay and Intellectual disability is universally present</th>
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<td>Short Stature</td>
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<td>Eyes</td>
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<td>Nose</td>
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<td>Dental</td>
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<td>Behavioral</td>
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Table: Common clinical features of RTS

II. Case

A 16 years old male who did not attend school at all, presented with increased danger, decreased sleep, physically abusive, wandering behavior for a total duration of 4 years, increased for 3 months, with history of delayed developmental milestones. Patient is the third child of his parents out of a non-consanguineous marriage, born by spontaneous vaginal delivery at home, cried immediately after birth, history suggestive of macrocephaly at the time of birth, with delay in all developmental milestones, sitting without support at 1 year, walking at 3 years of age, control over bowel at 12 years of age.

Currently, the patient stays at home. He does simple household tasks, cuts grass for cattle. But someone has to ask him to do the activities. He is able to do self-care activities by himself. He does not know how to read and write and do simple calculations.

On examination, the patient has a low hairline, low set ears, large thumbs and great toes, webs over feet.
On mental status examination, the patient is alert and conscious. Rapport not established, eye contact 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 question throughout the interview.

On Seguin Form Board Test (SFBT) - Patient does not have the concept of shapes, difficulty in comprehending instructions, stopped doing in between.

Vineland Social Maturity Scale (VSMS) - Social age found to be 7 years. The calculated SQ is 47, 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 characteristic morphological findings like a low set ear, low hairline, webbed toes, large thumb and great toe on VSMS indicative of a moderate level of socio-adaptive functioning.

Nayak et al. reported a case of a 31-year-old unmarried male presented with worsening behavioral problems for 2 months, with predominant symptoms of suspiciousness, delusion of reference, and persecution, 2nd person auditory hallucination, his intelligence quotient is 60, his physical anomalies are spiky hair, bushy eyebrows, prominent supraorbital ridges, low set ears, long eyelashes, prominent nose, open mouth, carries in teeth talon cusps, broad thumbs and toes, cervical hyperkyphosis and scoliosis. Patient was assessed with MINI English version 6.0.0 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 toe to the plantar first inter-space, hyper-extensible joint with mental retardation. [1]

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

In a review and meta-analysis of 732 cases by Cantani and Gagliesi found the most evident symptoms are the thumbs and first toes with broad terminal phalanges (99%). The trunk presents many anomalies too, as spina bifida, scoliosis, kyphosis, lordosis, hypotonia, lax ligaments and above all stiff gait (83.8%). In adults were noted head circumference under 50th percentile (95%) or microcephaly. Mental, motor, language and social retardation are one of the most common symptoms in RTS; it is present in 98.5% of the patients and 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 of inability 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, while evaluating such cases we should focus on morphological features of some common syndromes which present as intellectual disability with a behavioral abnormality.

References

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