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Case Report:
Rubinstein-Taybi Syndrome

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Abstract:

Rubinstein-Taybi Syndrome is a rare genetic disorder with characteristic featuresincluding downward slanting palpebral fissures, broad thumbs and halluces,and mental retardation. Systemic features may involve cardiac, auditory,ophthalmic, endocrine, nervous, renal and respiratory systems. This syndromeis sporadic in nature and has been linked to microdeletion at 16p 13.3 encoding CREB-binding protein gene (CREBBP). We report a 6-month-old male, who has congenital talipus equino varus, with downward slanting palpebral fissures towardthe ears, hypertelorism, short stature, beaked nose, micrognathia, large toes and broad thumbs.

Keywords: Rubinstein-Taybi Syndrome (RSTS), Broad thumb and halluces, CTEV.

Introduction:

Rubinstein- Taybi Syndrome (RSTS) was initially reported by Michael et al. in1957 as the broad thumb-hallux syndrome and then was described by Rubinstein and Taybi in 1963[1] in children with broad thumbs and toes, facial abnormalities and short stature. Since then, there have been over 250 cases documented in the literature. It has been estimated that 1 per 300-500 institutionalized persons with mental retardation over age 5 have this syndrome. Male and female individuals are affected at equal rates[2].Typical facial features include downward slanting palpebral fissures toward the ears, hypertelorism, long eyelashes, high arched eyebrows, prominent nose, and malpositioned ears with dysplastic helices. In addition, characteristic skeletal findings are broad short terminal phalanges of the thumbs and halluces, and postnatal growth retardation with head circumference below the fiftieth percentile. Dermatologic features include capillary malformation in approximately 50 percent of patients and higher incidence of keloid formation[3] and pilomatricomas[4].There may be systemic involvement of multiple organ systems. Of children with RSTS, 24-38 percent have cardiac abnormalities including atrial and ventricularseptal defects, patent ductus arteriosus, coarctation of the aorta, pulmonic stenosis,and bicuspid aortic valve abnormalities[5]. Feeding difficulties and gastroesophageal reflux disease requires aggressive treatment in young children to prevent nutritional and growth deficits. Cryptorchidism affects 78-100 percent of the male infants[6]. There is an increased incidence of benign and malignant tumors as well as leukemia and lymphoma[3,7]. Other organ systems that may be involved include auditory, ophthalmic, endocrine, neurologic, respiratory[6] and renal systems.

Rubinsten-Taybi Syndrome is associated with a mutation in the CREB-binding protein gene (CREBBP) located on chromosome 16p 13.3. CREBBP which is essential to normal development.

It has been identified as a nuclear protein that participates as a coactivator in cyclic AMP regulated gene expression. The precise relationship between microdeletion in the CREBBP and the phenotype of Rubinstein-Taybi is yet to be elucidated[8,9].

In this report, we introduce a case with features of rubinsten -taybi syndrome with CTEV.

Case Report:

A 6-month-old male child presented with CTEV patient came for further management. On examination patient had a distinctive facial appearance with hypertelorism, a broad nasal bridge, a beaked nose, micrognathia, microcephaly, smile grimacing, a narrow and high arched palate and long eye lashes. On examination, short stature, broad thumbs, large toes, and CTEV.

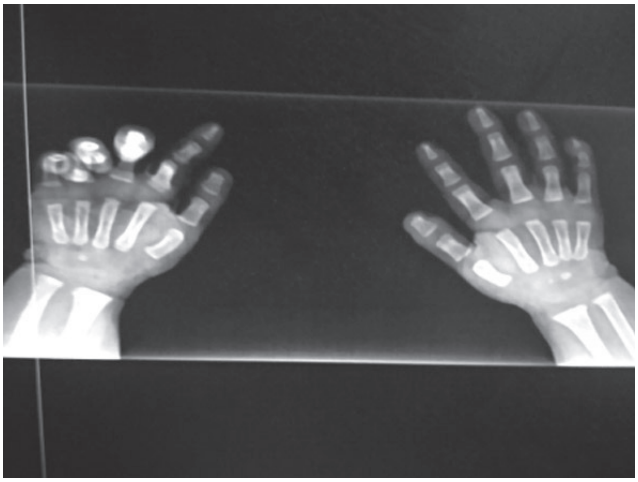
Motor and language developmental delay was noted.

Echo and USG abdomen were done which were normal. Blood investigation done which were within normal limits.

Discussion:

Rubinstein-Taybi Syndrome is a rare congenital anomaly. It appears that there is very little documented evidence regarding this syndrome in pediatric literature[9]. The cause of RSTS is still unclear, but it is associated with a microdeletion at 16p 13.3 region in the CREB binding protein gene (CREBBP) in some patient, suggesting that deletion is the most probable cause of the syndrome[10]. CREBBP is a transcription coactivator and functions as a potent histone acetyltransferase, both of which are essential to normal development[8]. In animal models, the mice with truncated Crb proteindemonstrate clinical features of RSTS observed in humans including growth retardation, retarded osseous maturation, hypoplastic maxilla with a narrow palate, and cardiac and skeletal abnormalities11. More recently, the breakpoint of two distinct reciprocal translocations occurring in patients with the diagnosis of RSTS has been located in the same band 16p 13.3. However, this anomaly cannot be identified in all patients. Clinically, the difference between patients with or without deletion is minimal except for microcephaly. Band 16p 13.[3] seems to be an important locus for mental retardation in patients with correct diagnosis of RSTS. [6,12,13,14]

Our patient, a known case of CTEV, was identified with characteristic features of the RSTS as explained above. In conclusion, RSTS is a rare genetic condition that affects body shape, extremities, and many organs/systems of the body, particularly cardiac, respiratory, nervous, and uro-genital system. This case report can help pediatricians to become more familiar with this syndrome.



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