

Case Report

Nephrotic syndrome in a case of Rubinstein Taybi syndrome: a rare case report

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ABSTRACT

Rubinstein Taybi syndrome (RSTS) also known as Broad Thumb Hallux Syndrome characterized by distinct facial features with beaked or straight nose, short stature, moderate to severe intellectual disability, broad thumb and big toe. We report an interesting case of female child with history and examination suggestive of RSTS with Nephrotic syndrome. Investigation revealed presence of albumin in urine with hypercholesterolemia and hypoalbuminemia. Purpose of this case report is to highlight the distinctive presentation of this syndrome and to update the current state of knowledge.

Keywords: CREBBP, Nephrotic syndrome, RSTS

INTRODUCTION

Rubinstein-Taybi Syndrome was initially described by Michail et al in 19570.¹ In 1963 Rubenstein and Taybi reported seven cases of this syndrome which included a group of congenital anomalies consisting of short, broad thumbs and great toes, poor postnatal height weight growth, psychomotor retardation with distinct facial features.² It is caused either by mutation in Cyclic AMP regulated enhancer binding protein (CREBBP) or EP300 gene at 22q13 or microdeletion at 16p13.3.³ Currently no precise diagnostic criteria are available. The genetic basis was first identified in 1991.⁴ Subsequently affected subjects were analyzed using Fluorescent in Situ Hybridization (FISH). The incidence of the syndrome has been estimated to be 1 in every 300,000 newborns.⁵ There is an equal male and female incidence.⁶

CASE

A 4 year 6 month female 2nd order child product of a non consanguineous marriage that came to us for swelling of abdomen for last 2 yrs followed by face and legs. Head

control was achieved at 7 months of age, crawling at 1 yr, standing with support at 1.5 yr, bisyllable and walking at 2 yr. On detailed clinical examination, she was afebrile with mild pallor with clubbing in all digits with broad terminal phalanges of thumb (Figure 2) and great toe (Figure 3) along with anasarca (Figure 1).



Figure 1: Anasarca.

Vitas are stable. Facial dysmorphism like broad nasal bridge, epicanthic fold, wide open mouth with double chin, antimongoloid slant of palpebral fissure with hypertelorism is seen (Figure 4). Height of the patient is 82cm which falls under 3rd percentile. Abdomen is distended with slit umbilicus with visible veins with fluid thrill (Figure 1). No obvious abnormality detected in cardiovascular or respiratory or ophthalmological examination.



Figure 2: Broad thumb.



Figure 4: Broad great toe.



Figure 4: Facial dysmorphism.

On urine examination albumin was 4+ with plenty of pus cells with culture suggestive of Klebsiella. Complete blood counts revealed neutrophilic leucocytosis with microcytic hypochromic anemia with raised serum cholesterol (418 mg/dl) with decreased serum albumin (1.6gm/dl) with normal creatinine values. Ultrasonogram of abdomen and pelvis revealed ascites. Total T4 count values are on the lower side owing to hypoalbuminemia in a Nephrotic setting. The child was managed in the line of Nephrotic syndrome 1st attack and treated with oral steroids following which the child responded.

DISCUSSION

RSTS is a rare multiple congenital anomaly syndrome.⁷ No precise diagnostic criteria have been defined. It is characterized by typical facial features, microcephaly, broad thumb and 1st toes, intellectual disability and post-natal growth retardation but none are considered as pathognomonic. Many cases are clinically diagnosed although most frequently involved gene is cyclic AMP regulated enhancer binding protein (CREBBP). Renal anomaly are seen in 52% of the cases and in males cryptorchidism is seen in 78-100% of cases.⁸ Cardiac abnormalities are found in 24-38% cases.⁹ Malposition and crowded teeth are present in 62% of pts and increased risk of caries (15-36%).⁵ Cancer of neural and developmental origin(neuroblastoma, medulloblastoma, oligodendroglioma, meningioma, rhabdomyosarcoma, seminoma, choristoma).¹⁰ Nonspecific changes in EEG are seen in 57-66% cases and seizure seen in 25% cases.¹⁰ Iris / retinal/ optic nerve coloboma are more common ocular manifestations followed by glaucoma.

CONCLUSION

In near future multicentric studies are necessary to identify the clinical and genotypic phenotype and to identify new candidate genes. The ultimate goal of our paper is to update our current knowledge regarding this syndrome and to define new guidelines. However, the association between RSTS and Nephrotic syndrome is not researched till yet. Furthermore, case studies will bring a light into this field.

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