# A Rare Case of Rubinstein-Taybi Syndrome With Scoliosis

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## INTRODUCTION:

Rubinstein-Taybi Syndrome (RTS) is a rare and complex neurodevelopmental disorder characterised by facial dysmorphism, growth retardation, and mental deficiency. We report an operated case of RTS patient with scoliosis which achieved a good outcome of balanced spine. This is the third case in the literature after Tatara (2011) and Bounakis (2015).

## CASE REPORT:

A 15-year-old boy with RTS came to us for evaluation of a progressive curvature. He was observed to have short stature with broad thumbs, mental retardation, strabismus, beaked nose, ear anomalies, global developmental delay. He presented to us with kyphoscoliosis, unable to walk without aid but did not manifest any neurological symptoms. In this report we present an operated case on RTS patient with scoliosis: deformity correction with PSIF. Post-operative he was monitored closely in intensive care unit, and later discharged home with satisfactory outcome.

## DISCUSSION:

Rubinstein-Taybi Syndrome (RTS) is a rare autosomal dominant disorder with microdeletion at 16p13.3 or mutations in the CREB-binding protein or EP300 gene (at 22q13). The incidence has been estimated to be 1 in every 300,000 newborn. This patient had striking facial features includes beaked nose, broad nasal bridge, grimacing smile, strabismus. He also had cardiac abnormalities, physical and mental developmental delay. The orthopaedic features include broad thumbs, great toes, with kyphoscoliosis (cobb's angle T1-T12 : $80^0$; T12-L4: $45^0$ ). The objective of surgical intervention is to achieve balanced spine for better sitting positioning on wheelchair and prevent worsening of curvature. There have been reports on surgical intervention to treat congenital dislocation of the patella or thumb deformity, but only two previous literature of RTS patient with scoliosis underwent surgical instrumentation for deformity correction.

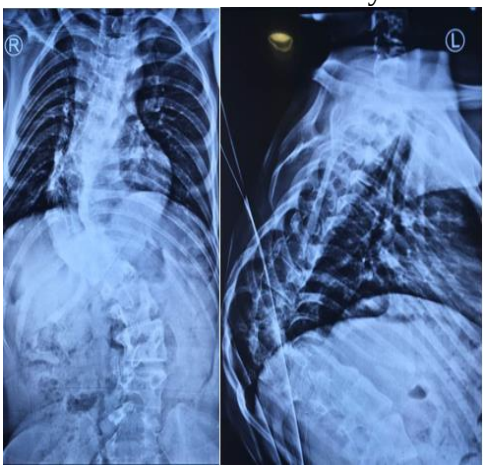

Figure 1: kyphoscoliosis

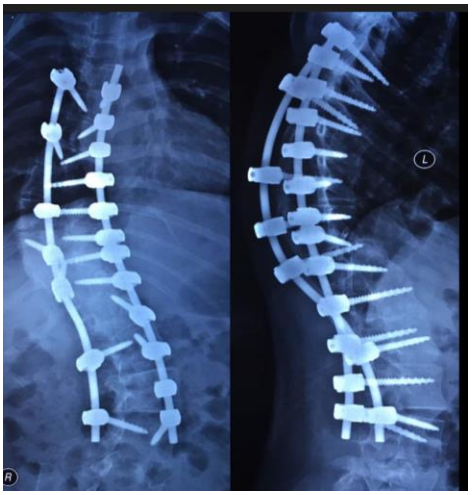

Figure 2: deformity correction and PSIF

## CONCLUSION:

We report this as a third case in literature for surgical intervention of a RTS patient with scoliosis. The great outcome improves quality of life by able to mobilize by wheelchair, and ease of patient care by the family members.

## REFERENCES:

1. Surgical treatment of scoliosis in Rubinstein-Taybi syndrome type 2. N.Bounakis.
2. Rubinstein-Taybi syndrome with scoliosis. Y.Tatara.