

Picture Story

## A case of Rubinstein Taybi syndrome

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Rubinstein Taybi is a short stature syndrome with post-natal growth retardation<sup>1</sup> and unique dysmorphic features.

### Case report

An eight and a half year old boy presented for investigation of short stature. He had delayed gross motor milestones, an unsteady gait, a mental age of 6-7 years and a poor attention span. Social interaction was excellent.

He had a comical grimacing facies (Figure 1).



**Figure 1** Comical grimacing facies

The palate was high arched & narrow with crowded & irregular teeth (Figure 2).

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**Figure 2** High arched narrow palate

There was a pectus carinatum deformity (Figure 3).



**Figure 3** Pectus carinatum deformity

His joints were hypermobile. He had broad thumbs and big toes bilaterally. He also has broad second and third fingers and toes (Figures 4 & 5).



**Figure 4** *Broad thumb and first two fingers*



**Figure 5** *Broad big toe & first two toes*

He was hirsute and had an angulated penis. Systems examination was normal.

He had a bone age of 3-4 years with poorly formed distal phalanges of both hands and feet, bilateral cervical ribs, widened lambdoid and saggital sutures with a hypoplastic maxilla (Figure 6).



**Figure 6** *Hypoplastic maxilla & suture separation of the skull*

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#### **References**

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