A case of Rubinstein Taybi syndrome

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Rubinstein Taybi is a short stature syndrome with post-natal growth retardation1 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).

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

There was a pectus carinatum deformity (Figure 3).

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).

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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 sagittal sutures with a hypoplastic maxilla (Figure 6).

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References