Author’s response to reviews

**Title:** Rubinstein-Taybi Syndrome with Scoliosis

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**Author’s response to reviews:** see over
Thank you for your kind letter of August 08, 2011 regarding our manuscript # MS: 5725430845482390. We are most grateful to you and the reviewers for the helpful comments on the original version of our manuscript. We have taken all these comments into account and submitted the revised manuscript. We improved the manuscript by adding detailed information of the patients.

We have addressed all the comments by reviewers #1 and #2, as indicated on the attached pages, and we hope that the explanations and revision of our work are satisfactory.

We hope that the revised version of our paper is now suitable for publication in Scoliosis and we look forward to hearing from you at your earliest convenience.

Sincerely,

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Response to Reviewer #1

We are grateful to reviewer #1 for the critical comments and useful suggestions that have helped us to improve our paper considerably.

Comments by reviewer #1.

Comment #1.

Does this particular patient have airway anomalies, congenital heart disease, or other major problems?

Response.

No, he doesn’t. We added this information to a subheading of History and Examination of the revised manuscript.

Comment #2.

For each surgery, patient’s age, body height, operation time and blood loss should be added.

Response.

We added this information to a subheading of History and Examination of the revised manuscript.

Comment #3.

Triradiate cartilage seems to be closing on the traction radiograph. When was the patient’s peak height velocity? Was mental retardation the reason for performing the second procedure?

Response.

The preoperative films revealed that the triradiate cartilage was opened and the Risser grade was 4. His secondary sex characteristic had not emerged yet at the time of surgery. The patient’s growth spurt had not reached its peak height velocity, but still he had a large secondary lumbar curve. We had discussed extensively with the parents about the importance and necessity of a second operation. The parents feared that even if the crankshaft phenomenon caused correction loss of the lumbar curve, the patient’s mental retardation might possibly prevent a manifestation of clear symptoms. Therefore, in order to minimize the possibility of a crankshaft, we performed a secondary procedure of anterior lumbar discectomy and fusion. As a result, x-ray films at one year after second surgery demonstrated a stable bony arthrodesis with no loss of initial correction. We stated these in the Discussion section of the revised manuscript.

Comment #4.
The patient could not walk by himself. Are there other findings that suggest the neurological abnormalities? Are there possibilities of myogenic disorders?

Response.

He was unable to walk without aid, but did not manifest any neurological symptoms such as hyperreflexia or abnormal abdominal reflexes; however, there have been no reports that myogenic disorders are involved with Rubinstein-Taybi syndrome.

Many Rubinstein-Taybi syndrome patients have a genetic mutation which has been mapped to chromosome 16p13.3, a genomic region encoding cyclic AMP (cAMP) response element binding protein (CREB) binding protein (CBP). Hallam et al. (references #5) reports that Rubinstein-Taybi syndrome, Huntington’s disease, Alzheimer’s disease and amyotrophic lateral sclerosis all share impaired CBP/CREB function. He summarizes that CBP/CREB dysfunction may be an important factor in multiple neuronal diseases.

Comment #5.

Cobb angles in the figures seem greater especially at the lumbar curve on the traction radiograph. Are the measurements correct? What does “12” on the postoperative radiograph mean?

Response.

Thank you very much for the valuable points you have raised. We remeasured Cobb angles on every x-ray film. We corrected errors and rewrote a subheading of History and Examination of the revised manuscript. We explained numbers in figure 3 in the section of Figure legends.
Response to Reviewer #2

We are grateful to reviewer #2 for the critical comments and useful suggestions that have helped us to improve our paper considerably.

Comments by reviewer #2.

Comment #1.

The authors should mention about his body height, body weight and the degree of skeletal maturity at the time of surgery. Though the authors performed anterior fusion for the lumbar curve to prevent a crankshaft phenomenon, the information regarding the skeletally immaturity of the patient at the time of surgery is not well documented in the manuscript.

Response.

The preoperative films revealed that the triradiate cartilage was opened and the Risser grade was 4. His secondary sex characteristic had not emerged yet at the time of surgery. We judged that the patient’s growth spurt had not reached its peak height velocity.