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Rubinstein-Taybi syndrome with scoliosis treated with single-stage posterior spinal fusion: illustrative case

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BACKGROUND Rubinstein-Taybi syndrome (RTS) is a rare disorder with a range of congenital anomalies. Although 40% to 60% of patients with RTS have scoliotic deformities, few reports discuss the outcomes of correctional surgery and postoperative care. To raise awareness of the clinical features of RTS and surgical considerations, the authors report on the surgical treatment of a pediatric patient with RTS accompanied by scoliosis.

OBSERVATIONS A 14-year-old girl with RTS presented with low back pain associated with progressive scoliosis. Because of jaw hypoplasia, videolaryngoscopy-mediated intubation was chosen. A single-stage T4–L3 posterior corrective fusion with instrumentation was successfully performed. Physical and imaging findings were analyzed up to 2 years after correction. The main thoracic Cobb angle was corrected from 73° to 12° and maintained for 2 years after surgery. The patient's low back pain resolved.

LESSONS Careful consideration of RTS-associated complications and preoperative planning, including the use of videolaryngoscopy-mediated intubation, anesthesia selection, and postoperative care, proved crucial. Scoliosis may appear in many variations in rare diseases such as RTS. Publication of case reports such as this one is needed to provide detailed information about strategies and considerations for correcting scoliotic deformities in patients with RTS.

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KEYWORDS Rubinstein-Taybi syndrome; scoliosis; surgical treatment; posterior; spinal fusion

Rubinstein-Taybi syndrome (RTS) is a rare syndrome, with an estimated incidence of 1 in 100,000 to 125,000 live births.[1] The first genetic cause, which was suggested in 1991, indicated a chromosomal reciprocal translocation in chromosomal region 16p13.3.[2] Later research identified mutations in the cyclic AMP–regulated enhancer binding protein (*CREBBP*) in the same 16p.13.3 region,[3] which has been reported in more than half of the patients with RTS.[4] More recent work has also suggested epigenetic modifications and abnormalities in CREBBP, particularly relating to the p53 pathway.[5] In addition, mutations in the *CREBBP* homolog, such as E1A binding protein p300 (*EP300*) on chromosome 22, also have been suggested as causes of RTS.[5,6] Overall, 55% to 70% of RTS cases can be diagnosed as involving mutations in either of these two genes.[7] Before genetic testing is performed or for the approximately 30% of cases not involving *CREBBP* and *EP300* mutations, the diagnosis of RTS is based on clinical findings.

RTS is generally characterized by specific physical characteristics, including broad thumbs, broad halluces, dysmorphic facial features, and short stature.[8] RTS is also associated with multiple cardiac and neurological complications, which may severely compromise surgical interventions (Table 1).[8] Moreover, RTS is associated with a range of orthopedic disorders (Table 2), including dislocation of the radial head or patella, hypotonia, lax ligaments, tight heel cords, elbow abnormalities, Perthes disease, congenital hip dislocation, slipped capital femoral epiphyses, increased risk of fractures, spinal deformities, spinal cord lesions, and scoliosis.[8] Patients with RTS usually present to an orthopedic specialist either before or after they receive a diagnosis of RTS.[9]

Reports on orthopedic surgical cases for patients with RTS are scarce. Our literature review on spine-related issues that require surgical corrections in patients with RTS (Table 2) produced only 8 identified case reports[7,10–16] and 3 reports that described spinal disorders in patients with RTS who did not undergo surgical

ABBREVIATIONS CA = Cobb angle; CT = computed tomography; ICU = intensive care unit; MRI = magnetic resonance imaging; RH = rib hump; RTS = Rubinstein-Taybi syndrome.

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TABLE 1. Overview of general RTS clinical features and RTS-associated complications

Type	Characteristic Feature	Abnormalities	Present
Clinical features	Typical facial appearance	Arched brows	○
		Down-slanting palpebral fissures	
		Nasal septum extending below alae nasi microcephaly	
		Highly arched palate	
		Grimacing smile	
		Talon cups	
		Frontal protrusion	
		Micrognathia	
		Maxillary hypoplasia	○
		Impaired dentition	○
		Eagle-like nasal apex	○
	Orthopedic abnormalities	Broad thumbs	○
		Broad halluces	○
		Polysyndactyly	
		Chiari malformation	
		Spine curvatures	○
		Cervical vertebral abnormalities	
		Perthes disease	
		Lax joints	
		Dislocated patellae	
	Growth abnormalities	Short stature	○
		Obesity	
	Intellectual disabilities	Mental retardation	○
Associated complications	Heart	Ventricular septal defect	
		Atrial septal defect	
		Patent ductus arteriosus	
	Eye	Strabismus	
		Refractive error	
		Ptosis	
		Nasolacrimal duct obstruction	
		Cataracts	
		Coloboma	
		Corneal abnormalities	
	Skin	Keloids	X
		Pilomatrixoma	
	Genitourinary	Undescended testes	
		Hypospadias	
		Duplex kidney	
		Renal agenesis	
	Cancer	Meningioma	
		Rhabdomyosarcoma	
		Pheochromocytoma	
		Neuroblastoma	
		Medulloblastoma	
		Oligodendroglioma	
		Leiomyosarcoma	
		Seminoma	
		Odontoma	
		Choristoma	
		Leukemia	

The table presents a general list of the most common clinical features and complications associated with patients who have RTS. The Present column indicates features and complications that were observed or diagnosed in our patient during presurgical examination (○) or after surgical intervention (X).

intervention.[17–19] We found only 3 reports of the correction of RTS-associated scoliosis deformity, 2 of which provided clear surgical strategies and outcomes regarding the correction procedure. This lack of information indicates a need for more awareness about surgical experiences and outcomes to develop better insights into spinal correction surgery for patients with RTS, particularly considering the range of additional complications associated with the disorder. We present a case report to raise awareness of RTS and share strategic considerations when planning the correction of surgical scoliosis associated with RTS.

Illustrative Case

Our case involves a 14-year-old girl who presented with imbalanced posture and low back pain, which was attributed to scoliosis. She had short stature (135 cm), low body weight (29.5 kg), maxillary hypoplasia, impaired dentition, eagle-like nasal apex, and broad thumbs and halluces (Fig. 1, Table 1). School underachievement, which was observed from 10 years of age, indicated mild intellectual disability. Scoliosis was diagnosed as dextroscoliosis with an 18° lordotic rib hump (RH). The Cobb angle (CA) from T5 to L2 was 73°, and pelvic inclination was 21° (Fig. 2A). From a radiograph of the sagittal plane with the patient in a standing position, thoracic kyphosis measured 19°, with a sagittal vertical axis of −2 cm. The apical RH was 5.5 cm (Fig. 2B). Using traction radiography, the main curve CA decreased to 50°. Correction of the T4 tilt by left flexion was poor (Fig. 2C–E). Bone maturity was assessed as grade 4 using the Risser classification[20] and as grade 7 according to digital skeletal age (hand radiograph; Fig. 1B).[21]

During surgical planning, a range of complications was observed, which enabled our pediatrics department to diagnose RTS. Genetic testing revealed no abnormalities. Although *CREBBP* or *EP300* domain abnormalities were not examined, as is typical, a diagnosis of RTS was made solely on the basis of clinical findings.[8] Hence, we were unable to confirm an RTS type I or type II classification in our patient.[22] No family or perinatal history of abnormalities was determined. Hormone levels were all within the normal range: growth hormone, 0.72 ng/mL; prolactin, 7.0 ng/mL; adrenocorticotropic hormone, 23.2 pg/mL; luteinizing hormone, 5.2 IU/mL; follicle-stimulating hormone, 5.6 IU/mL; thyroid-stimulating hormone, 2.040 mU/mL; free triiodothyronine, 3.57 pg/mL; free thyroxine, 1.52 ng/dL; cortisol, 3.9 µg/dL; estradiol, 65 pg/mL; and testosterone, 0.23 ng/mL. Cardiac disorders were excluded with the use of electrocardiography and echocardiography. The values for respiratory vital capacity (34.7%) and forced expiratory volume in 1 second (100%) suggested restrictive lung disease. Kidney, urethra, and brain function examinations produced no extraordinary findings. No myelopathies (e.g., tethered cord) were observed using magnetic resonance imaging (MRI), and computed tomography (CT) scans showed no abnormalities other than scoliosis of the spine (Fig. 3). Three months after the diagnosis was made, the major curve had advanced 8°, and surgery was deemed necessary to prevent further progression of scoliosis.

Because of concerns about perioperative risks, including risks related to respiration, circulation, and infectious diseases, as well as concerns about the patient's limited physical strength and anticipated delays in postoperative rehabilitation (in contrast to the general flexible spine indicated on traction radiographs, Fig. 2), single-stage posterior deformity correction surgery was elected. Because of the patient's jaw hypoplasia, endotracheal intubation was deemed risky, and intubation was performed using videolaryngoscopy (Airway Scope, Pentax). Anesthesia was given careful consideration; however, because cardiac disorders had been excluded, general intravenous propofol anesthesia was induced.

With the patient in the prone position, the T4–L3 segment range was exposed using a posterior approach. Anchors were created by inserting pedicle screws and hooks. Facetectomy was performed for posterior release from T9–10 to T11–12, which was followed by instrumented vertebral translation and direct vertebral rotation. Spinal fusion was promoted using combined lamina decortication grafts and hydroxyapatite granules. Monitoring of transcranial electrical stimulation motor evoked potentials revealed no abnormalities during surgery. Surgical deformity correction lasted 4 hours 3 minutes and involved 724 mL of intraoperative blood loss, which was managed during surgery using 800 mL of autologous blood obtained before surgery to avoid the need for transfusion.

Immediately after surgery, the endotracheal tube was removed, and the patient was admitted to the intensive care unit (ICU). No breathing or hemodynamic abnormalities were observed, and she was transferred to the general ward after 1 day in the ICU. The postoperative evaluation revealed improvement in the CA from 73° before surgery to 12° after surgery and improvement in the RH from 5.5 cm to 1 cm (Fig. 2F–I). No wound site abnormalities were observed after 1 week of bed rest. The patient began gait training and was discharged 18 days after surgery. At the 1-month follow-up appointment, keloid scar formation, which is a common observation in patients with RTS,[23] was observed at the site of the surgical incision (Table 1). Because of suspected infection, wound debridement was performed, but no pathogenic bacteria were identified. The symptoms improved, and the patient was discharged 2 weeks later. Two years after correction, fusion of the instrumented segments was observed without instrumentation failure and the established deformity correction was maintained (Supplementary Fig. 1). The patient reported that her low back pain had receded. No signs of infectious diseases were observed, and her general prognosis was good.

Discussion

Observations

RTS was first reported as a malformation of the digits in 1963[24] and has since been considered a disease mainly related to genetic and epigenetic mutations in the *CREBBP* and *EP300* genes, although diagnosis is often based on clinical findings, as occurred with our patient. More specifically, RTS is classified as type I when a *CREBBP* mutation is present or as type II when an *EP300* mutation is present.[22] Type II RTS generally presents with milder phenotypic features than type I RTS.[11,22] RTS in our patient could not be identified as type I or type II because mutations in *CREBBP* or *EP300* were not examined.

Although cases of RTS are uncommon, efforts have aimed to establish medical guidelines for management of patients with RTS,[8,9] which involves complex repeated evaluations by specialists such as orthopedic surgeons, cardiologists, neurologists, ophthalmologists, and dermatologists. No standard therapeutic agent or intervention has been developed for RTS, and optimal treatment for resolving complications that arise from RTS has yet to be established. RTS is often complicated by a range of spinal deformities (e.g., scoliosis, kyphosis, lordosis), craniovertebral junction abnormalities, Chiari malformation, syrinx, low-lying conus medullaris, and tethered cord complications (Table 2). Stevens et al. reported a prevalence of scoliosis of 40% to 60% among patients with RTS, 10% of whom require bracing or surgical intervention.[18] Our review identified a total of 24 patients with RTS (including our patient) with spinal involvement, 10 (42%) of whom were diagnosed with scoliosis and 4 (17%) of whom required scoliotic curve correction (Table 2). If we include the case series by Stevens et al.,[18] we note that 40 of 69 (58%) patients with RTS also had vertebral curve

TABLE 2. Tabular overview of case reports or case series on RTS-associated spinal diseases

Type of Reporting	Report	CVJ Abnormality	Scoliosis	Other Spinal Deformities	CMI	Syrinx	Tethered Cord	Sex	Age (yrs)	Primary Orthopedic Indication	Surgical Intervention	Adjustment to Surgical Intervention for RTS	Reported Complications	Final FU
Reporting on scoliosis surgical correction	Tatara et al., 2011[10]		O					M	14	Rt thoracic scoliosis	Double-stage instrumented double-rod correction & arthrodesis	Maintained on respirator after surgeries	None	1 yr
	Bounakis et al., 2015[11]		O	O				F	15	Double thoracic scoliosis & associated hypokyphosis	Single-stage, single concave rod instrumented correction, arthrodesis, & spinal jacket	Single concave rod instrumented (for skin healing & muscle pain concerns); rescheduled surgery & psychological sessions because of extreme anxiety & poor cooperation; postsurgical nasogastric feeding	None mentioned	2 yrs
	Current study		**O**					**F**	**14**	**Rt thoracic scoliosis**	**Single-stage instrumented vertebral translation & direct vertebral rotation, followed by arthrodesis**	**Video laryngoscope intubation; no use of decannulation**	**Keloid scar formation (resolved)**	**2 yrs**
Reporting on other spinal anomalies in RTS cases	Robson et al., 1980[12]	O						F	14	Tetraplegia caused by C5–6 spondylolisthesis & C6–7 spinal stenosis	Head traction followed by spinal fusion w/ Cloward dowel, which is followed by wearing collar	None mentioned	None mentioned	14 wks
	Yamamoto et al., 2005[13]	O						F	13	Myelopathy caused by compression & stenosis, C1 hypoplasia, C1 occipitalization	Decompression surgery	None mentioned	None mentioned	Unclear

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TABLE 2. Tabular overview of case reports or case series on RTS-associated spinal diseases

Type of Reporting	Report	CVJ Abnormality	Scoliosis	Other Spinal Deformities	CMI	Syrinx	Tethered Cord	Sex	Age (yrs)	Primary Orthopedic Indication	Surgical Intervention	Adjustment to Surgical Intervention for RTS	Reported Complications	Final FU
Reporting on other spinal anomalies in RTS cases (*continued*)	Yamamoto et al., 2005[13] (*continued*)	O						F	19	Atrophy of spinal cord by atlantoaxial subluxation w/ dens hypoplasia	Unspecified	None mentioned	None mentioned	None mentioned
		O						F	3	Cervical cord compression & stenosis w/ C1 occipitalization & dens hypoplasia	Unspecified	None mentioned	None mentioned	None mentioned
		O						M	23	Odontoideum w/ fibrous fusion of atlas, & dens os odontoideum	Unspecified	None mentioned	None mentioned	None mentioned
		O						M	20	Odontoideum & fusion at C2–3	Unspecified	None mentioned	None mentioned	None mentioned
	Tanaka et al., 2006[14]						O	F	8	Symptomatic low-lying conus	One-level laminectomy & durotomy	None mentioned	None mentioned	4 mos
							O	F	2	Symptomatic low-lying conus	One-level laminectomy & durotomy	None mentioned	None mentioned	Unclear
			O				O	M	14	Symptomatic low-lying conus, scoliosis	One-level laminectomy & durotomy	None mentioned	None mentioned	Unclear
							O	M	14	Symptomatic low-lying conus	One-level laminectomy & durotomy	None mentioned	None mentioned	2 days
				O			O	F	7	Symptomatic low-lying conus, hyperlordosis	One-level laminectomy & durotomy	None mentioned	None mentioned	1 mo
							O	M	3	Symptomatic low-lying conus, released tethered cord	One-level laminectomy & durotomy	None mentioned	None mentioned	6 mos
			O				O	F	9	Symptomatic low-lying conus, scoliosis	One-level laminectomy & durotomy	None mentioned	None mentioned	6 mos
							O	M	14	Symptomatic low-lying conus	One-level laminectomy & durotomy	None mentioned	None mentioned	3 mos

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Type of Reporting	Report	CVJ Abnormality	Scoliosis	Other Spinal Deformities	CMI	Syrinx	Tethered Cord	Sex	Age (yrs)	Primary Orthopedic Indication	Surgical Intervention	Adjustment to Surgical Intervention for RTS	Reported Complications	Final FU
Reporting on other spinal anomalies in RTS cases (*continued*)	Kim et al., 2010[17]				O			M	4	Chiari malformation type I	None	NA	NA	NA
	Wójcik et al., 2010[15]				O			F	2	Chiari malformation type I	Suboccipital decompressive craniectomy & decompressive C1 laminectomy	None mentioned	None mentioned	Unclear
	Parsley et al., 2011[16]		O		O	O		F	13	CM, w/ cord syrinx & progressive scoliosis	Chiari decompression, spinal fusion*	None mentioned	None mentioned	Unclear
			O		O	O		F	13	CM w/ cord syrinx & scoliosis	Chiari decompression	None mentioned	None mentioned	Unclear
	Stevens et al., 2011[18] †			O			O	-	18–67	NA	NA	NA	NA	NA
	Giussani et al., 2012[19]	O	O		O	O	O	F	11	Lt thoracolumbar scoliosis, CMI, spinal syrinx from C2, low-lying conus at L2	None	NA	NA	NA
		O	O		O	O	O	M	13	CM, small syrinx T5–7, low-lying conus at L3, double thoracic scoliosis, fusion at C2–3	None	NA	NA	NA
	Hadzsiev et al., 2019[7]		O			O	O	M	3	Myelocele, tethered cord, syringohydromyelia, thoracic scoliosis	Partial lipoma removal	None mentioned	Elevated sphincter tone, increased bladder pressure, vesicoureteral reflux	2 yrs

CM = Chiari malformation; CMI = Chiari malformation type I; CVJ = craniovertebral junction; FU = follow-up; NA = not applicable; O = the presence of the specified spinal complication.

The overview presents results from a literature review on RTS in orthopedics, including the type of spinal deformities or abnormalities diagnosed, surgical intervention applied, and RTS-specific alterations used. The review highlighted limited reports or studies published on RTS and the respective spinal surgical intervention. Most cases involved some vertebral malformation or deformity (craniovertebral junction, vertebral deformities, and Chiari type I columns), highlighting their association with RTS. Boldface text highlights the features of the current study.

* Spinal fusion application was mentioned; however, no report regarding surgical strategies or outcomes was provided.

† Work by Stevens et al. involved a case series of 45 families of adult patients with RTS. No specific information was given for individual patients.

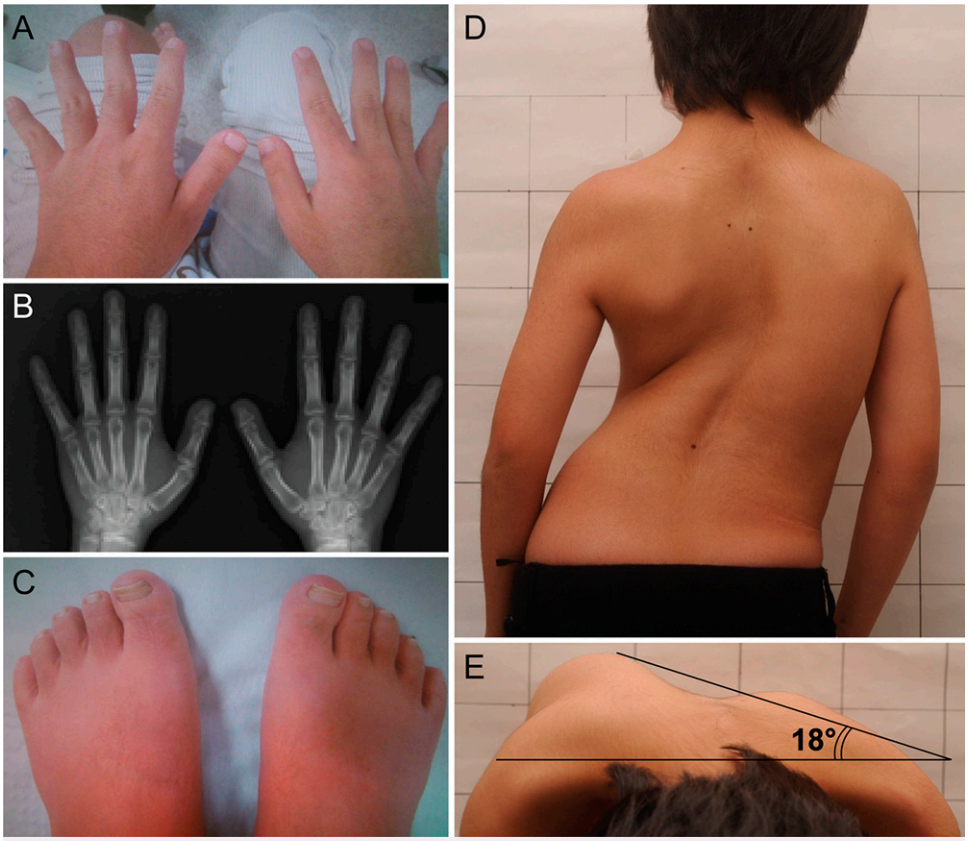

FIG. 1. Clinical presentation of RTS in a 14-year-old girl with broad thumbs (**A** and **B**) and broad halluces (**C**). The patient had short stature, with an asymmetrical waistline and high shoulder blades (**D**) and trunk axis inclined to the right (**E**). With the patient in a forward-bending position, concave protrusion of the ribs was noted in the right lordotic position with an inclined angle of 18°. No skin lesions, arthrochalasis, or evident tumors were observed.

deformities. When taken together, these observations highlight the likelihood that patients with RTS will present to an orthopedic specialist.

Perioperative management of a patient with RTS is difficult, and multiple reports have urged caution.[11,25] Conditions that require particular consideration include intubation impediments because of facies abnormalities, presence of cardiac disease and associated risk of heart failure or arrhythmia during surgery, intellectual disability, and keloid scar formation. Facies abnormalities such as micrognathia, high arched palate, and other maxillofacial malformations are of particular concern because they may hinder the use of surgical anesthesia, such as via tracheal intubation. In our patient, we avoided the risk of intubation complications by using a videolaryngoscope, but no airway narrowing or obstructing deformities were observed during the intubation procedure.

RTS is associated with high rates of congenital cardiac diseases (prevalence of 24%–38%[26]), which raises concerns regarding the risk of heart failure and arrhythmia during or after surgery. When selecting anesthesia, meticulous care is recommended to reduce these risks.[27,28] For example, Stirt and Karahan et al. suggested that anesthetic agents such as atropine, succinylcholine, and neostigmine be avoided because they increase the risk of arrhythmia.[27,28] Preoperative examination excluded heart disease in our patient, and no heart failure or arrhythmia occurred during surgery. Intellectual disability in our patient was mild (an estimated delay of 2–3 years) and allowed for normal communication, thereby limiting the restrictions on peri- and postoperative practices. A particular concern was raised about tube management (whether to perform decannulation), which might prove risky in patients with RTS who have low intellectual capacity, so the procedure was not performed in our patient.

Only 2 case reports on RTS-associated scoliosis surgery were identified in our current literature review (Table 2).[10,11] A case report by Tatara et al.[10] described a 14-year-old boy with RTS who presented with an 84° right thoracic curve and a 63° lumbar curve. The authors decided on a two-stage approach because of the rigidity of the thoracic curve. The first correction, an anterior-posterior surgical approach, involved anterior discectomy from T8–9 to T10–11 followed by posterior osteotomy of the same region. Next, the pedicle screw-anchored instrumentation was applied from T4 through L4, after which arthrodesis was initiated. Surgical correction reduced the thoracic curve to 31° and the lumbar curve to 34°. Nine months after the first correction, the second correction focused on the lumbar region. Using an anterior retroperitoneal approach with rib resection, discectomy of the L1–2 to L3–4 discs was performed, followed by spinal fusion. After both surgeries, the patient was admitted to the ICU and maintained on a respirator for several days. This approach was chosen because of limited communicative abilities as a result of the patient's limited mental

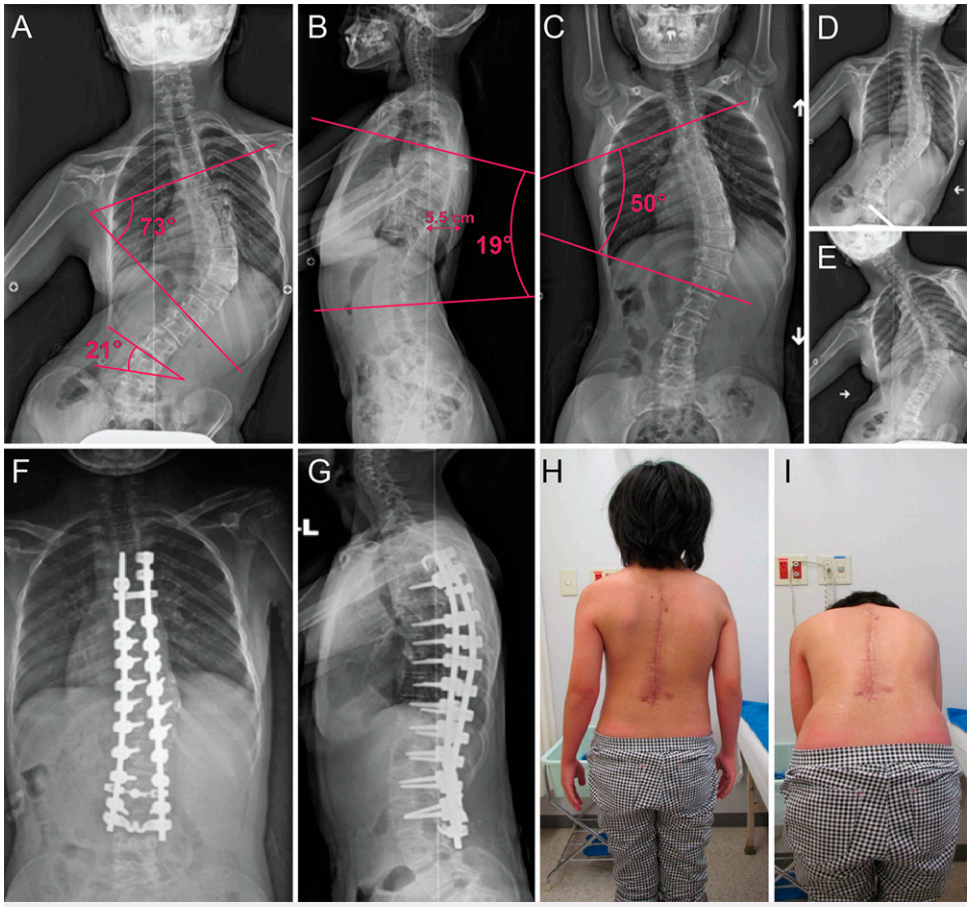

FIG. 2. Pre- and postoperative examination of the patient's spinal deformity. **A:** Anterior coronal plane image revealed a right thoracic curve with a CA of 73° from T5 to L2 and an apical vertebra at T10–T11. The pelvic inclination was 21°. **B:** Sagittal plane image showed thoracic kyphosis of 19°, with a sagittal vertical axis of −2 cm and an apical RH size of 5.5 cm. **C:** Anterior coronal image under traction revealed a reduction in main thoracic CA from 73° to 50°, suggesting a relatively flexible spine. Anterior coronal image in right-leaning (**D**) and left-leaning (**E**) positions. Postoperative spinal deformity correction examination by coronal (**F**) and sagittal (**G**) radiographic images obtained after correction of the spinal deformity, which was accomplished using instrumented vertebral translation, direct vertebral rotation maneuver, and bone grafting of autologous bone graft obtained by decortication of the lamina combined with hydroxyapatite granules to mediate spinal segment fusion. The main thoracic curve with a CA of 73° before correction improved to a CA of 12°. The coronal plane balance improved. Visual examination in standing (**H**) and forward-bending (**I**) positions. RH prominence improved from 5.5 cm to 1 cm.

abilities, which raised concern about his inability to report negative symptoms during surgical recovery.

The second case report by Bounakis et al.[11] involved a 14-year-old girl with RTS who had double thoracic scoliosis curves of 39° and 68° associated with hypokyphosis, which resulted in severely restrictive lung disease. The initial surgery was rescheduled because of her anxiety and poor cooperation on the day of surgery. Before the deferred surgery, the patient was supported with psychological assistance, and anesthesia was induced without active participation (through secured intravenous access) of the now 15-year-old girl. She received a single posterior surgical correction involving single-rod instrumentation on the concave side from T2 to L4 and subsequent autologous arthrodesis. Because the patient was severely underweight, a single rod was used to avoid the anticipated prominent appearance of the instrumentation under the skin. After surgery, the patient remained in the ICU, where she received nasogastric feedings for a few days. She was then required to wear a spinal jacket for 6 months. The thoracic curves, which were corrected to 18° and 30° postoperatively, were maintained at the final 2-year follow-up examination. Mild improvement was seen in pulmonary test results, and no complications were reported.

Although combining our findings with those of previous reports on scoliosis correction in patients with RTS generally suggests mild postoperative complications, the other authors noted the need to adjust standard procedures before, during, and after surgery to limit the risk of complications associated with an RTS phenotype (Table 2). Recurring challenges involve the extent of cooperation and understanding from the patient, which can cause planning delays,[11] as well as preventive measures such as nasogastric feeding tubes[11] or not

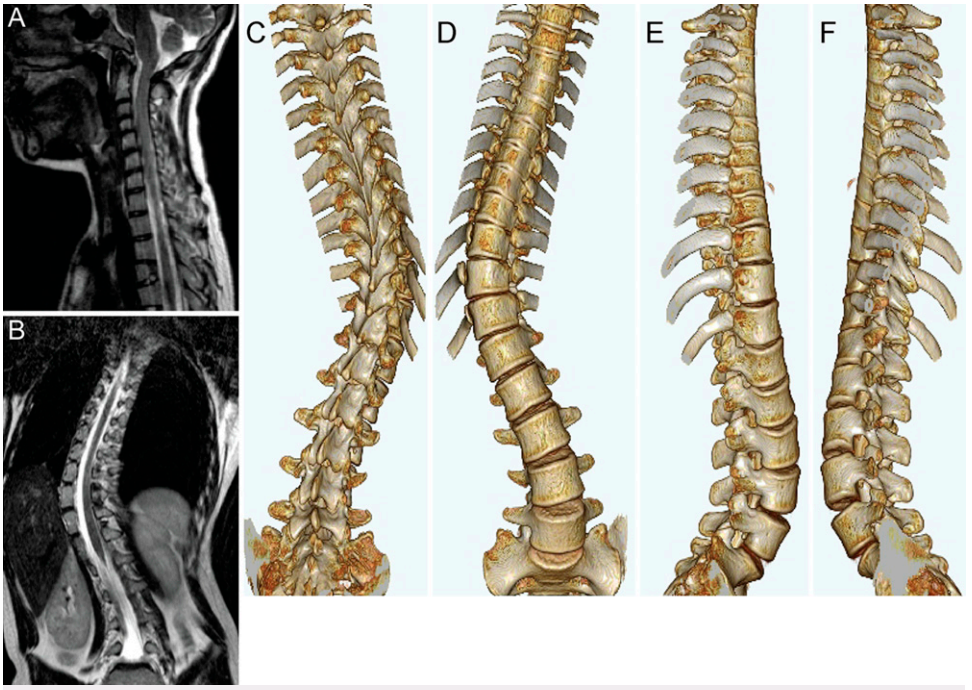

FIG. 3. MRI and CT scan analysis. Cranial (**A**) and abdominal (**B**) MRI scans negated the presence of any myelopathies such as tumor, Chiari malformation, syrinx, low-lying conus medullaris, tethered spinal cord, or the like. CT scan–derived three-dimensional representation of the spine from the posterior (**C**), anterior (**D**), and lateral left (**E**) and right (**F**) perspectives revealed no apparent vertebral column abnormalities other than scoliosis.

using decannulation. Physical concerns, such as the risk of keloid or hypertrophic scar formation, general concerns of facies abnormalities hindering intubation, and careful deliberation about anesthetic agents have been reported. In general, we believe that a careful presurgical plan should be designed to predict potential surgical obstructions and complications and prepare surgical and postsurgical intervention accordingly.

Corrections of deformity were successful in all three reported surgical scoliosis cases associated with RTS and led to clinically significant curve corrections. Currently, follow-up reports have been limited to only 2 years; long-term postoperative follow-up reports are needed to provide a better understanding of the longevity of the corrections and potential long-term complications. We promote the publication of further case reports on RTS-associated orthopedic disorders to provide a better understanding of the strategies and considerations for deformity correction in patients with RTS and to raise awareness of this complex syndrome.

Lessons

In this study, we treated a patient with RTS who had scoliosis. Because RTS is often associated with scoliosis, we deemed it beneficial to raise awareness of this disorder and report our surgical strategy for curve correction. Despite the mild RTS phenotype in our patient, specific adjustments were made to limit risks associated with facies abnormalities and intellectual disability and limit postsurgical complications. Surgical deformities and curve abnormalities were corrected successfully after our careful presurgical planning.

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Disclosures

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

Author Contributions

Conception and design: Sakai, Imai, Nagai. Acquisition of data: Sakai, Imai, Schol, Nagai. Analysis and interpretation of data: Sakai, Imai, Schol, Nagai, Katoh. Drafting the article: Sakai, Imai, Schol, Nagai, Hiyama, Katoh. Critically revising the article: Sakai, Imai, Schol, Katoh. Reviewed submitted version of manuscript: Sakai, Imai, Schol, Sato, Watanabe. Approved the final version of the manuscript on behalf of all authors: Sakai. Statistical analysis: Sakai, Imai, Schol. Administrative/technical/material support: Sakai, Imai. Study supervision: Sakai, Imai, Watanabe.

Supplemental Information

Online-Only Content

Supplemental material is available with the online version of the article.

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