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A case of bilateral elbow dislocation in a patient with Rubinstein-Taybi syndrome

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Rubinstein-Taybi syndrome (RTS) was first described in 1963 by Rubinstein and Taybi in seven patients with psychomotor retardation, unusual facies, and a broad thumb.[9] It is a rare genetic disorder ie, characterized by growth and psychomotor delays, abnormal gross anatomy, and mild-to-severe cognitive disability.[9] It is considered to be caused due to a heterozygous pathogenic variant in the CREB-binding protein gene (CREBBP) on chromosome 16p13.3.[3] Clinical features include a broad hallux, hallux valgus, joint laxity, and an awkward gait. Dislocation of the patella and spinal abnormalities have also been reported to occur relatively frequently.

However, to the best of our knowledge, there have been no reports of posterior elbow instability in patients with RTS. Here, we report a case of bilateral recurrent elbow dislocations in a patient with RTS.

Case report

The patient was a 17-year-old female who was genetically diagnosed with RTS at 5 months of age (Fig. 1). She was found to carry a variant in the CREBBP gene (NM_004380.3: c.5837dup, p.Pro1947ThrfsTer19). She was examined at our hospital after presenting with right elbow pain caused by an unknown injury mechanism. She had a medical history of bilateral recurrent posterior elbow dislocation (4 times on the right side and 3 times on the left side). The first dislocation of the elbow joints was caused by a fall when the patient was 12 years old. Her right elbow joint was swollen, and she had tenderness at the slightest touch. An X-ray of the bilateral elbows revealed a posterior dislocation of the right elbow joint (Fig. 2). However, the subsequent joint reduction was simple. A shallow ulnar trochlear notch on both sides was observed in the radiograph of the right elbow joint after reduction (Fig. 3). In addition, computed tomography revealed a bony component or heterotopic ossification near the lateral insertion of the lateral ulnar collateral ligament (LUCL) (Fig. 4). magnetic resonance imaging and other physical examinations could not be performed because the patient could not remain still. Furthermore, accurate determination of joint range of motion after reduction could not be performed because the patient's own understanding was not obtained.

Institutional review board approval was not required for this case report.

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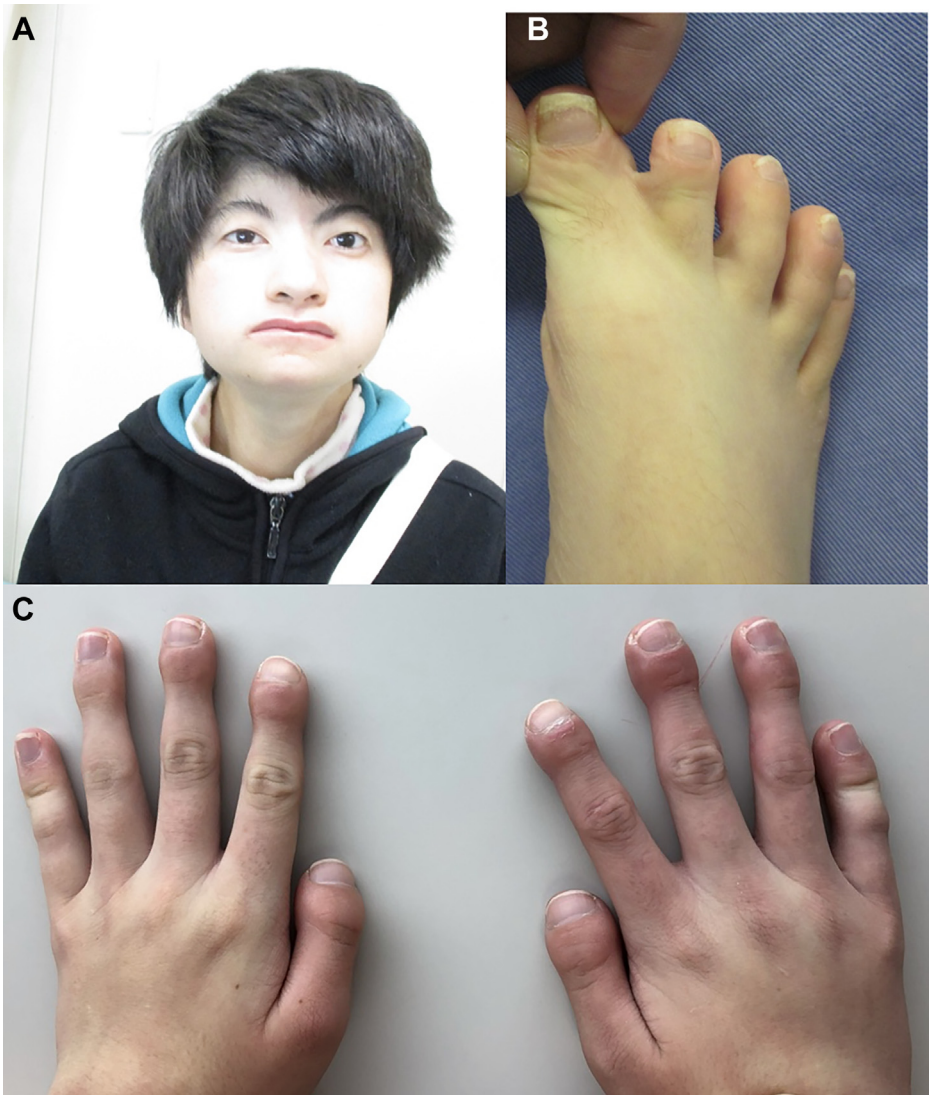

Figure 1 (**A**) Front view of the patient's face showing characteristic features such as a broad nose and thick eyebrows. (**B**) Cutaneous syndactyly between the first and second toes is hypoplastic, indicating a wide hallux. (**C**) Broad thumbs and broadened terminal phalanges on both hands.

The operative method should be selected based on the results of arthrography inspection and posterolateral rotatory instability test carried out before the operation. First, the posterolateral rotatory instability test was performed under general anesthesia and revealed marked instability of the right elbow. Second, arthrography was performed to assess for rupture of the anterior capsule and medial collateral ligaments. However, contrast media leakage from the anterior capsule and medial collateral ligaments was not observed, and since it was only leakage from the lateral collateral ligament, the anterior articular capsule and medial collateral ligament were not repaired; only the LUCL was repaired. The operation was performed using the Kocher approach (Fig. 5*A*), and (Fig. 5*B*). The pit that appeared to be bone component or heterotopic ossification was also confirmed in the lateral humeral condyle. A suture anchor was inserted into the origin of the LUCL (Fig. 5*C*), followed by suturing of the avulsion of the LUCL and the capsule (Fig. 5*D*). A long arm cast was used for 2 weeks postoperatively; subsequently, the patient was allowed full range of motion without restriction. The patient did not undergo formal rehabilitation due to cognitive disability.

No re-dislocation of the right elbow has occurred for five years after the surgery (Fig. 6). During a follow-up, the patient also complained of pain in the left elbow without any apparent cause, and dislocation was discovered. However, since she was scheduled to move from the facility where she was residing, her parents opted for conservative treatment. Reduction was performed without anesthesia in the office. During the reduction maneuver, we first applied gentle longitudinal along the longitudinal axis in the semi-flexed position, without forcing the elbow into extension. Meanwhile, an assistant held the distal humerus near the elbow joint. Once this maneuver had been completed, it felt as if the joint had been reduced; however, it was easily re-dislocated immediately afterward. Therefore, with the cooperation of the anesthesiologist, the patient was sedated and the joint was reduced using an X-ray image intensifier. We found that reduction was easy when we had traction on the forearm, but the elbow became dislocated fully extended. Therefore, cast immobilization was performed for 4 weeks, with the elbow joint flexed at a 90-degree angle. Surgical treatment would be required, however, if the left elbow were to dislocate again. The patient has been followed up regularly since

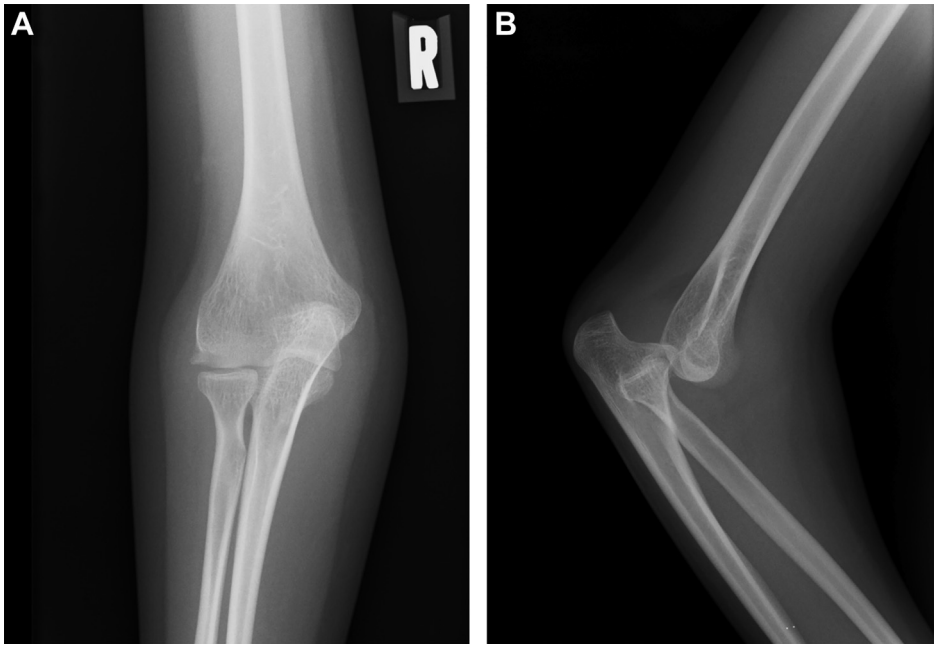

Figure 2 (**A** and **B**) Radiograph showing posterior dislocation of the right elbow joint as seen from (**A**) Anterior view, and (**B**) Lateral view.

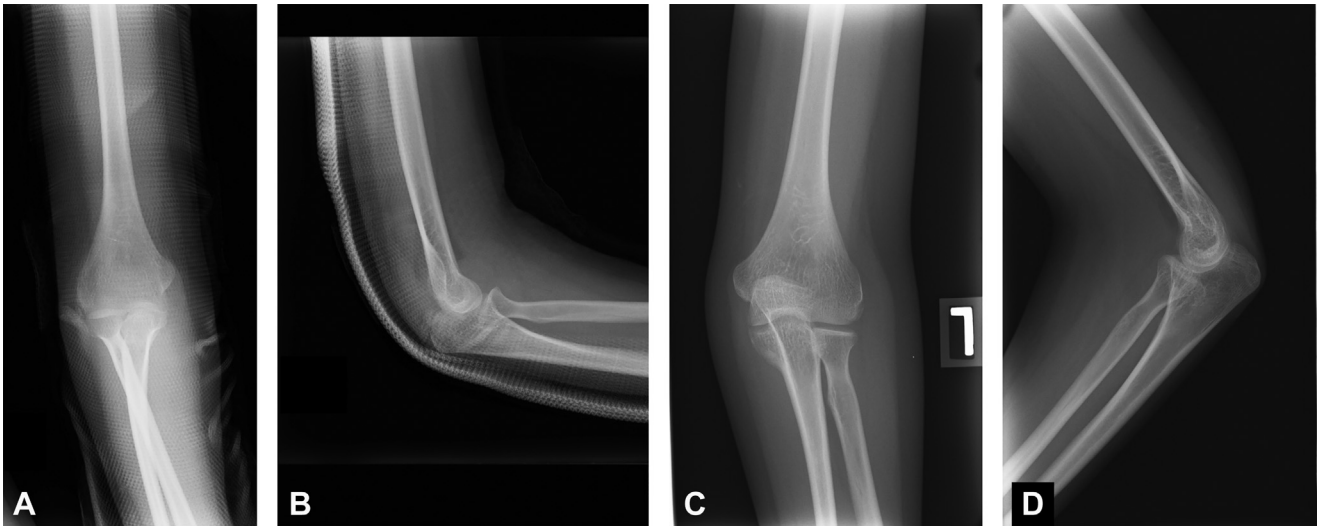

Figure 3 (**A**) Radiograph showing the right elbow after reduction and external fixation with pronation. The carrying angle was 162° immediately after reduction. (**B**) Radiograph showing the lateral view after reduction of the right side elbow dislocation. Radiograph demonstrates shallow ulnar trochlear notches of the ulna. (**C**) The carrying angle was 160° on the left side. (**D**) Radiograph showing the lateral view of the left side elbow demonstrating shallow ulnar trochlear notches of the ulna.

then, but there has been no apparent elbow contracture, nor any complaints from her family about the limited range of motion of the elbow.

Discussion

This patient was found to carry a variant in the CREBBP gene (NM_004380.3: c.5837dup, p.Pro1947ThrfsTer19). This variant has not been reported in any database for the normal population, but it has been reported in three RTS patients including one new patient.[1,8,10] Although this frameshift variant was located at the last exon, escaping nonsense-mediated decay, some downstream variants have been identified as the RTS-causing variants, predicting the loss of an important domain of the CPEBBP protein in the mutant protein. This mutant protein was also found to be 'likely pathogenic' when the variant was evaluated using the American College of Medical Genetics and Genomics and the Association for Molecular Pathology recommendations, with PVS1 Moderate, PM2, PM6, and PP4 applied.

The most common joint dislocations in patients with RTS are patellar dislocations, which have been identified in 3.4% of the patients, with few reports of the treatment of this condition with surgery. While broad thumbs are known features of the upper limbs in patients of RTS, there are no reports of posterior dislocation of the elbow in RTS to the best of our knowledge. In the present patient, anatomical abnormalities of the trochlear notch of the ulna were recognized using X-ray and computed tomography. It has been acknowledged that joint laxity is often present in RTS. Moreover, there are characteristic gait abnormalities in RTS (awkward gait). It was considered that this congenital condition

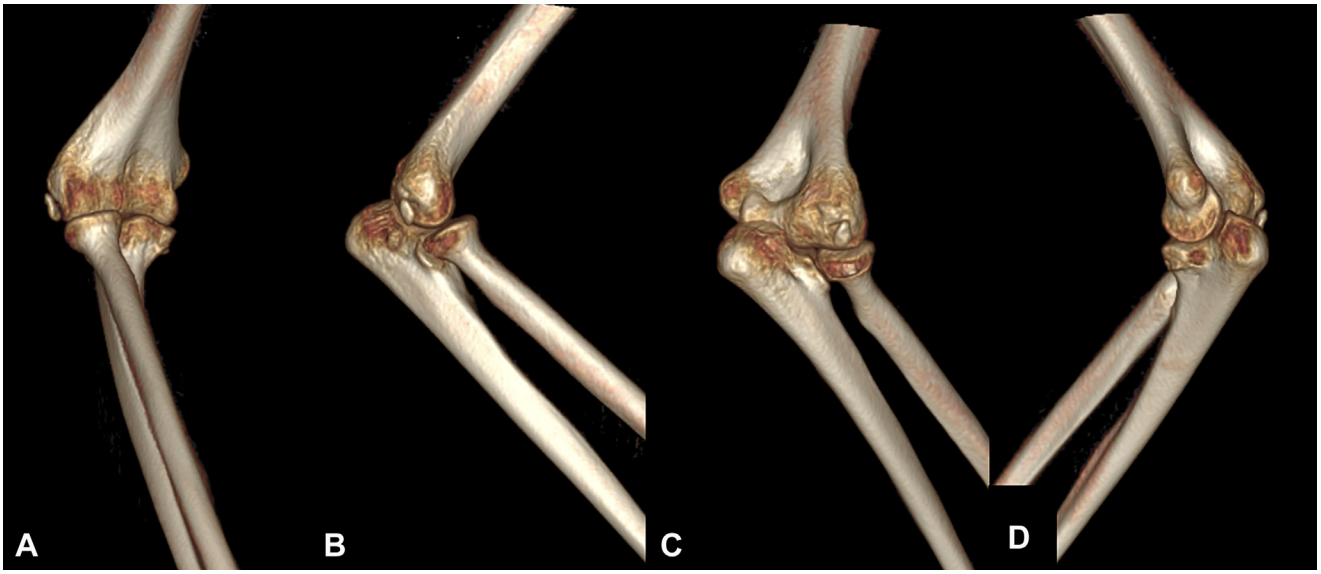

Figure 4 (**A** and **B**) CT image of bone component or heterotopic ossification near the lateral insertion of the lateral ulnar collateral ligament. (**C** and **D**) Shallow ulnar trochlear notches of the left ulna are demonstrated. *CT*, computed tomography.

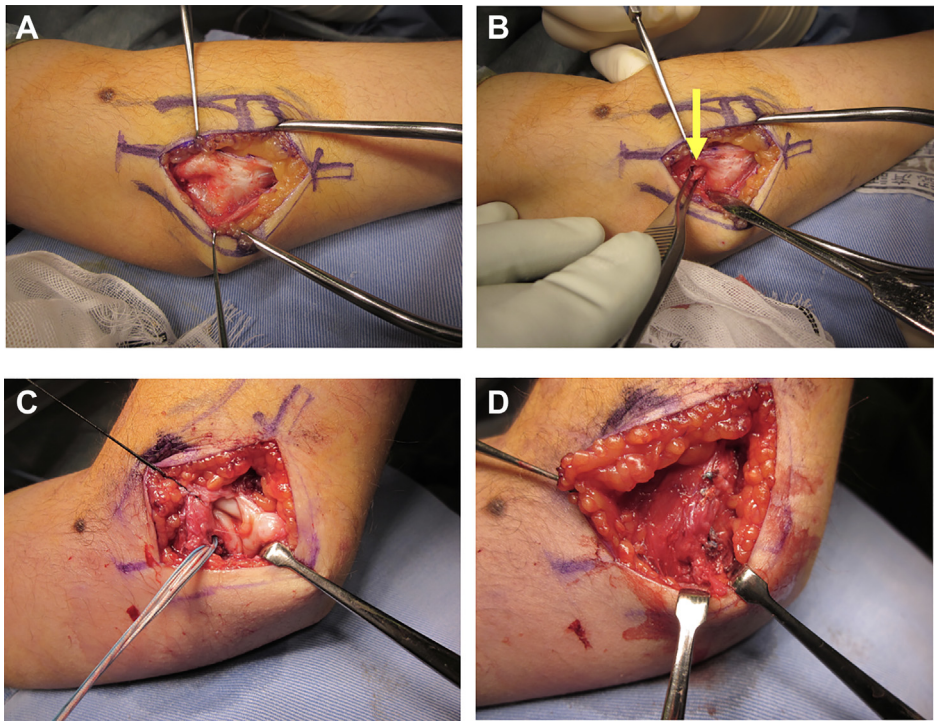

Figure 5 (**A**) The operation was performed using the Kocher approach. (**B**) The spicule to which the ligament component adhered (arrow). (**C**) A suture anchor was inserted into the origin of the lateral ulnar collateral ligament. (**D**) The avulsion of the lateral ulnar collateral ligament and the capsule were sutured.

was the cause of the posterior dislocation of the elbow joint. Initially, the ulnar trochlear notch of the ulna was thought to be responsible for the dislocation. Accordingly, the technique of iliac crest bone grafting in the anterior part of the elbow joint was considered during the operation. There are some reports on the treatment of elbow joint dislocation fracture by iliac bone grafting in the anterior elbow joint.[2] However, in the present case, the patient had never experienced elbow dislocation before the age of 12 years. In addition, as shown in Fig. 3, the carrying angle of this patient was within the normal range. Since cubitus varus contributes to elbow joint instability, O'Driscoll et al have stated that corrective osteotomy into more anatomic valgus is important in this condition.[5] However, in this case, corrective osteotomy for improving the carrying angle was judged to be unnecessary. Therefore, despite the presence of a shallow ulnar trochlear notch of the coronoid process of the ulna, it was considered that both the elbow joints were stable before the injury; thus, an iliac bone graft was not used.

The repair of the torn ligaments and articular capsule was considered, but there was no leakage of the contrast media except on the laterally during intraoperative arthrography. Morrey and An (1983) reported that the anterior capsule plays an important role in providing stability to the elbow joint; however, since there was no leakage of the contrast agent except on the laterally, no suturing of

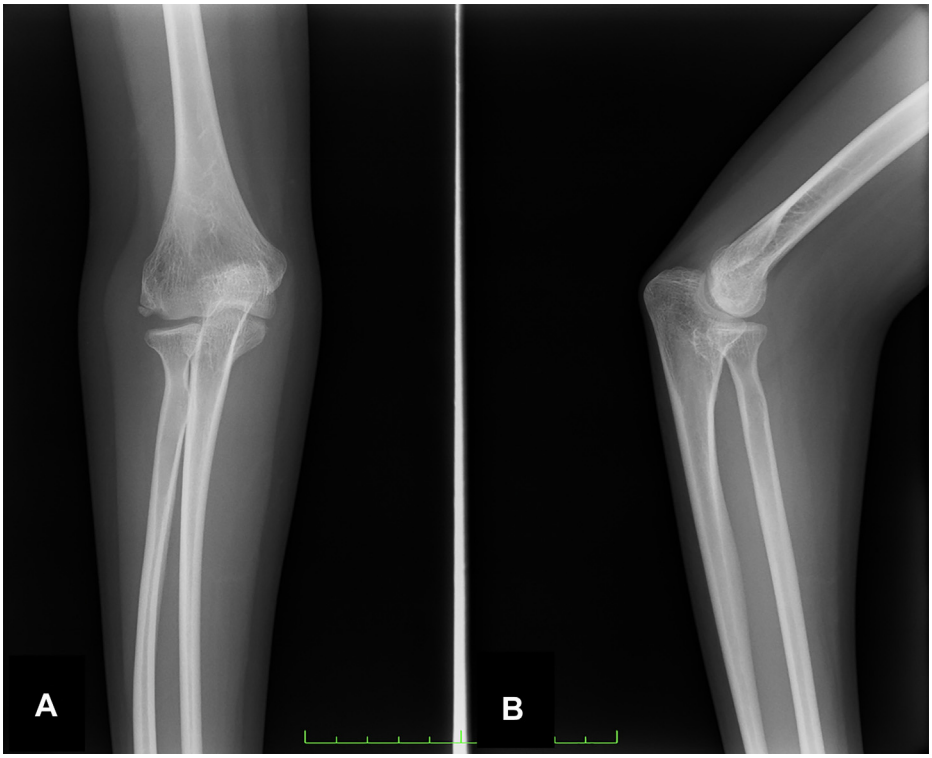

Figure 6 (**A** and **B**) Radiograph showing no posterior dislocation of the right elbow, five years postsurgery, as seen from (**A**) Anterior view, and (**B**) Lateral view.

the anterior capsular or medial collateral ligament was performed in the present case to restore elbow joint stability with a successful result after 5 years.[4]

Fortunately, the patient had no major problem with cardiac function, and the surgery could be performed under general anesthesia. However, since the condition of each patient is unique, a detailed examination is necessary before surgery. The majority of the surgeries that RTS patients undergo are orthopedic, orthodontic, ophthalmic, or heart surgery. The most notable features of anesthetic management of the syndrome are the airway management challenges, the risk of aspiration pneumonia, and cardiovascular pathologies.[6] RTS patients are expected to experience difficulty in airway management due to abnormal anatomical structures, and pretreatments are necessary. However, the response to the anesthetics must first be identified; this is typically an arrhythmia. In addition, efforts to prevent respiratory complications should be made.[7]

Conclusions

We described bilateral recurrent elbow dislocations with shallow trochlear notches in a patient with RTS who underwent LUCL repair on the right elbow. There has been no re-dislocation on the right elbow, five years postsurgery, and we hope that our experience will help clinicians in the future when treating patients with similar conditions.

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