Acute rehabilitation in a pediatric patient with Rubinstein-Taybi Syndrome and spinal cord infarct resulting in paraplegia: A Case Report

College of Medicine
Physical Medicine and Rehabilitation

Christopher James, MD, Nathan Abdelsayed, MD, Sara Shahid Salles, DO

University of Kentucky, Department of Physical Medicine and Rehabilitation, Lexington, KY

Introduction

- Rubinstein-Taybi syndrome (RTS) is a rare genetic disorder impacting an estimated 1/125,000 live births.
- RTS is caused by a mutation in the CREBBP gene and microdeletion of chromosome 16.
- RTS is characterized by intellectual disabilities and physical abnormities including broad thumbs and toes, highly arched eyebrows, down slanting palpebral fissures, high-arched palate, and micrognathia.
- Rubinstein reported that children with RTS could have congenital or acquired scoliosis, kyphosis, or lordosis. The patient in this case report was diagnosed with scoliosis at the age of one and when she was still able to walk without aid. Therefore, we believe that scoliosis in RTS patients may be caused by associated neuromuscular abnormalities.

Case Description

- A 14-year-old female with RTS presented to outpatient orthopedic clinic with a history of urinary retention, spasticity and bilateral lower extremity weakness.
- Patient had undergone a T4-L3 posterior spinal fusion due to progressive scoliosis one month prior to the outpatient visit.
- Pre-operatively, patient was able to ambulate without an assisted device and continent of bladder.
- Neuroimaging revealed spinal cord infarct involving the thoracic spinal cord and was admitted to our inpatient rehabilitation hospital.
- Detailed neurological classification (International Standards for Neurological Classification of Spinal Cord Injury - ISNCSCI) was unattainable due to RTS-related cognitive impairment. However, traces of movements were noted on lower extremities bilaterally at the time of admission.
- Over the course of her inpatient rehabilitation stay, she achieved some functional improvements such as reflex voiding with low post void residuals and inconsistent voluntarily activation of adductor muscles.
- At her 6-month post-operative follow-up, she demonstrated some improvement in hip flexion and knee extension but lacked proper motor control.

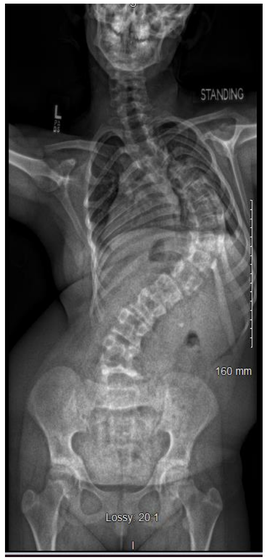

Figure 1
Preoperative AP- Spine XR displaying severe scoliosis

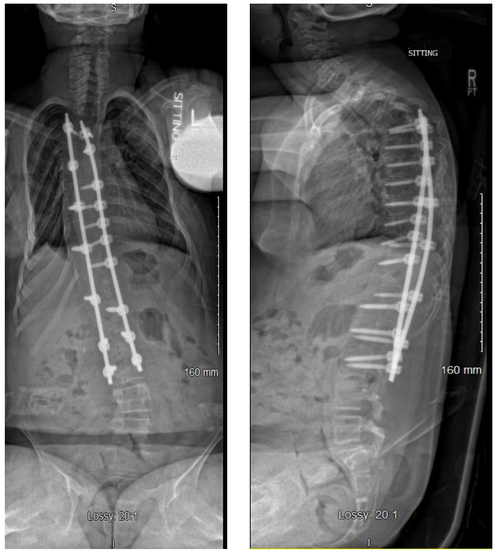

Figure 2 and 3
Postoperative AP/ Lateral Spine XR S/P T4-L3 posterior Spinal fusion and segmental instrumentation

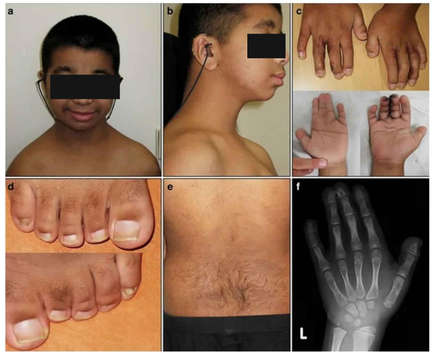

Figure 4
Representation of Typical Phenotypic Presentation. Images unrelated to the patient. (6)

Discussion

- Our patient had symptoms of urinary retention, paraplegia and spasticity secondary to spinal cord infarct.
- Patient's age and intellectual disability complicated her rehabilitation outcomes, leading to an increased burden of care for her family.
- Impaired motor planning further decreased the potential for her to regain prior functional abilities even in the setting of improved muscle strength.
- Significant family teaching was required due to patient's cognitive deficits that was further complicated by splitting time living in two separate households, as well as family disagreements in regard to approach of her care.

Conclusion

- The risk of subacute spinal cord infarct following posterior spinal fusion is extremely low in patients with scoliosis.
- Intellectual disabilities associated with RTS may cause complications during spinal cord injury rehabilitation.
- It is important to acknowledge impairments the patient might have had prior to acute injury.
- Decreased motor planning and intellectual disability may hinder the ability to learn new techniques to optimize independence after a spinal cord infarct and increasing the burden of care to the family.

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