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## CLINICAL VIGNETTE

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# Anesthesia for a Patient with Rubinstein-Taybi Syndrome

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### Introduction

Rubinstein-Taybi Syndrome, also known as 16p13.3 deletion syndrome, is a rare, autosomal dominant, submicroscopic deletion on chromosome 16 that affects multiple organ systems. This condition affects males and females equally with a birth prevalence of 1:100,000 to 1:125,000 and involves mutations in two functionally related genes: CREBBP and EP300.<sup>1</sup> The syndrome is characterized by growth restriction, down slanting of palpebral fissures, microcephaly, dysmorphic features, broad thumbs and toes, and intellectual disability.<sup>2</sup> The characteristic facial features include, but are not limited to, beaked nose with nasal septum protruding below the nares, highly arched eyebrows, micrognathia, and highly arched palate and incisors which may have talon cusps. These individuals can have eye problems such as glaucoma, strabismus, and cataracts and renal malformations. Some have severe scoliosis that require surgical correction.<sup>3</sup> Hirsutism is common and congenital heart disease is present in one-third of these patients.

These patients should have developmental and ophthalmologic evaluations and an echocardiogram performed to assess the presence of congenital heart disease. They are also at increased risk of developing keloids<sup>4</sup> and malignancies.<sup>5</sup> There is only symptomatic treatment at the present time for Rubinstein-Taybi syndrome.

### Case Report

A 13-year-old male with Rubinstein-Taybi syndrome presented to the anesthesia evaluation clinic for preoperative anesthesia evaluation for an electroencephalogram (EEG) and eye exam under anesthesia. He had a history of seizures, for which he had adverse behavioral reaction to levetiracetam. The patient's other medical conditions included developmental delay, strabismus, pharmacologically closed PDA and obesity. He had an echocardiogram after his pharmacologically closed PDA 2 years prior which reported, "No ASD or VSD visualized on parasternal views. No chamber enlargement. Although exam limited, unlikely that there is a VSD or ASD" and that the patient was deemed by his cardiologist as "low risk from a cardiac standpoint for participation in special Olympics."

On the day of the procedure, no midazolam was given to reduce possible adverse interference with the EEG. The patient's mother was allowed to accompany the patient to the operating room to help comfort him. The mother held his hands while we started inhalational induction. After the patient was asleep, the

mother was asked to wait in the waiting room as we proceeded with placement of a peripheral intravenous catheter. Due to some airway obstruction during inhalational induction, the patient needed placement of an oral airway, while maintaining cervical spine neutrality, to relieve the obstruction. The ophthalmologist proceeded with their bilateral eye exam while we placed the peripheral intravenous catheter and started the patient on a total intravenous anesthesia (TIVA) with propofol.

After the technician was satisfied with the EEG results, the propofol drip was turned off and the child was brought to the recovery room. Once the patient arrived in the recovery room, his mother was called immediately to come to his bedside. We communicated with the mother that everything went well and that he will recover in the post-anesthesia care unit (PACU) until he is ready for the next level of care. The patient did well and woke up comfortably in the PACU and was discharged about one hour later.

### Discussion

Rubinstein-Taybi Syndrome is an extremely rare autosomal dominant genetic disease that was first described in 1963<sup>6,7</sup> and no standard diagnostic criteria are currently available. Most cases are diagnosed clinically, however, genetic tests are available and can be useful in making the diagnosis. Despite normal prenatal development, the main features in Rubinstein-Taybi Syndrome include growth restriction, intellectual disability, dysmorphic facial features, microcephaly, and broad thumbs and big toes. These patients are complex and majority of them survive to adulthood and present for medical care and procedures on an ongoing basis.

In addition to thumb and toe abnormalities, patients with Rubinstein-Taybi Syndrome can have skeletal anomalies including scoliosis and cervical vertebral abnormalities, such as C1-C2 instability or fusion of the cervical spine.<sup>8</sup> These are of great concern, along with arched palate and micrognathia, for anesthesiologists, as scoliosis can lead to compromised lung capacities and function and the cervical spine and craniofacial abnormalities can restrict the extension of the neck leading to difficulty with bag mask ventilation and intubation. Our patient had some airway obstruction during inhalational induction of anesthesia that was adequately relieved by the placement of an oral airway without us needing to manipulate the neck. Additionally, it would have been extremely difficult to obtain

flexion and extension x-ray films of the cervical spine in our patient as we sometimes do in adult patients who we suspect may have cervical spine instability. Cerebrovascular abnormalities including cerebral artery dissection have been reported.<sup>9</sup> Furthermore, mood instability, anxiety, depression, and aggressive behavior have also been reported as these patients age.<sup>10</sup> Despite the significant strides that we have made in the past couple of decades, we continue to discover and learn new findings on this intriguing condition.

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