

# Investigation of the Rare Association between Bilateral Vocal Cord Abductor Paralysis and Rubinstein-Taybi Syndrome

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## ABSTRACT

**Background:** Rubinstein-Taybi syndrome is a rare disorder characterized by broad thumbs and great toes, short stature, dysmorphic facial features, eye abnormalities, cryptorchidism, and moderate to severe intellectual disability. Renal, cardiac, and dental anomalies as well as obesity may be associated with the syndrome. The patients may also have behavioral problems. They have a weak laryngeal wall that can easily collapse resulting in swallowing and breathing problems.

**Case report:** We report a case of an infant with Rubinstein-Taybi syndrome. The present case had typical facial features, broad thumbs and great toes, right corneal opacity, and bilateral vocal cord abductor paralysis. To the best of our knowledge, no study has been conducted on bilateral vocal cord abductor paralysis in Rubinstein Taybi syndrome. In addition, the infant recovered with symptomatic treatment without tracheostomy.

**Conclusion:** Rubinstein-Taybi syndrome is associated with various congenital anomalies, however further studies are required to investigate the rare association between Rubinstein-Taybi syndrome and bilateral vocal cord abductor paralysis in future cases.

**Keywords:** Corneal opacity, Rubinstein-Taybi syndrome, Vocal cord abductor paralysis

## Introduction

Rubinstein-Taybi Syndrome (RTS) is a rare genetic syndrome characterized by dysmorphic facial features and other anomalies (1). Airway problems caused by paralysis of the vocal cord are unusual. The airway of infants is anatomically and physiologically different from that of adults. Therefore, diagnosing and managing pathological conditions in the airway of an infant is more challenging. Bilateral vocal cord abductor paralysis is rare in infants which prevents vocal cords from being abducted and resulting in stridor. The etiological factors are usually congenital, such as genetic, traumatic, and neurological factors (2). Idiopathic is the most common cause of congenital vocal cord paralysis (3). The present study aimed to investigate an infant with Rubinstein-Taybi syndrome having

bilateral vocal cord abductor paralysis and associated complications.

## Case report

A male infant with a weak cry, breathing difficulty, cyanosis, stridor, and difficulty with breastfeeding was brought to the neonatal unit of Kasturba Medical College and Hospital, India. He was born to a 26-year-old primigravida woman by emergency cesarean section at 39 weeks of gestation. The indication for cesarean section was an abnormal nonstress test. No history of consanguineous marriage or birth trauma was observed. His birth weight was 2.7 kg, height was 48 cm and head circumference was 34 cm. His heart rate was 140 bpm and his respiratory rate was 46 bpm. Bilateral femoral pulses were well

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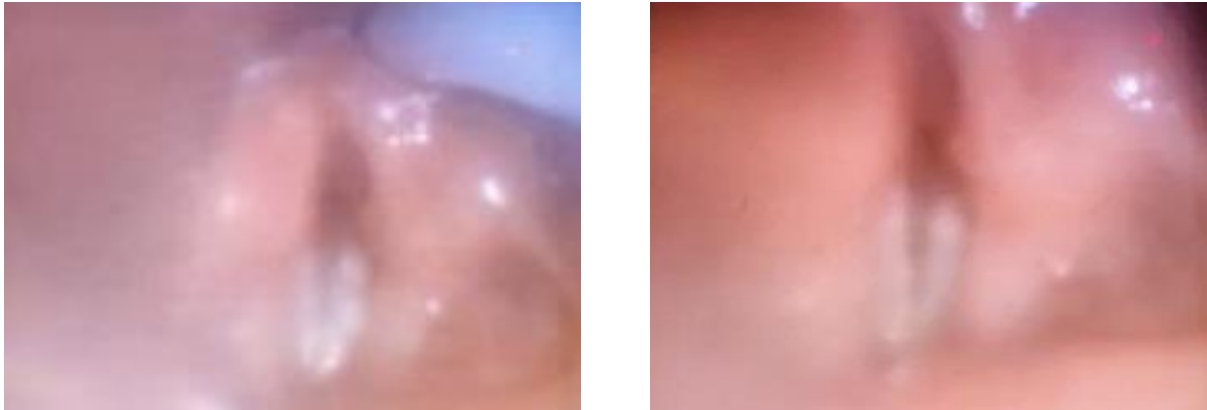
**Figure 1.** Pointed nose, depressed nasal bridge, and right sunken eye

felt. General examination revealed a sunken right eye with corneal opacity, pointed nose, depressed nasal bridge, overhanging columella, high-arched palate, and downslanting palpebral fissures (Figure 1). He also had micrognathia, microstomia, low-set ears, and thick eyebrows. Bilateral upper and lower limbs indicated stubby fingers, flat thumbs, and broad great toes (Figure 2). Ophthalmologic examination revealed right corneal opacity and primary congenital glaucoma. The intraocular pressure was high. The abdomen was soft, not distended, and no organomegaly existed. The infant was diagnosed with Rubinstein-Taybi Syndrome due to clinical features. Baseline investigations indicated polycythemia (Haemocrit 63%) and low blood sugar. A partial exchange transfusion was

performed. Subsequently, the infant was euglycemic. Initial low serum calcium of 6.2 mg/dL was normalized following IV 10% Calcium Gluconate. Chest x-ray showed inhomogenous opacities on the right side. The infant was evaluated for persistent stridor by an ENT specialist. Flexible laryngoscopy confirmed the diagnosis of bilateral abductor paralysis (Figure 3). Flexible laryngoscopy assessed the mobility of the vocal folds on both sides, the adequacy of the glottic chink, and eliminated other laryngeal anomalies such as laryngomalacia, laryngeal cleft, and anterior or posterior glottic stenosis. This case failed to include a fiberoptic endoscopic evaluation of swallowing. The infant was a high-risk case for general anesthesia due to stridor and respiratory abnormalities, therefore rigid endoscopic evaluation was not performed under intravenous anesthesia to remove second laryngeal pathology such as subglottic stenosis and tracheobronchomalacia. Cricoarytenoid joints were normal. The infant was initially managed with invasive mechanical ventilation. Subsequently, he was managed by non-invasive ventilation. As breastfeeding was difficult, expressed breast milk was fed through the feeding tube. Conservative management was performed for about a month without tracheostomy. The infant gained weight during the hospital stay and bilateral vocal cord abductor paralysis gradually resolved. The condition improved and the infant was discharged from the hospital with a feeding tube. Multiplex Ligation-dependent Probe Amplification (MLPA) test failed to detect any common microdeletions. The geneticist also recommended whole exome sequencing, but the test was not performed due to financial constraints.



**Figure 2.** Bilateral stubby fingers, broad thumbs, and great first toes



**Figure 3.** Endoscopy indicates bilateral vocal cord abductor paralysis

## Discussion

Rubinstein-Taybi Syndrome is diagnosed in infants at birth based on abnormalities of the face, hands, and feet. An infant inherits RTS from parents with an autosomal dominant pattern or may be due to mutation. Mutations in the CREBBP2 or the EP3003,4 genes cause this syndrome (4). Prevalence of RTS is one case per 125000 live births (1). Microcephaly, large anterior fontanel, grimace, small right eye, pointed nose, and right eye glaucoma were observed in the present case. Glaucoma was previously reported in RTS (5). The affected infants may have deviated septum, broad nasal bridge, low-set ears, thick high-arched eyebrows, ptosis, epicanthal folds, and nasolacrimal duct obstruction (6). Optic atrophy may occur due to undetected congenital glaucoma. Evaluating intraocular pressure is critical in this situation to prevent additional vision loss.

The respiratory abnormalities include extra small lobulation of the lungs and weak laryngeal walls. The weak and easily collapsible walls of the larynx result in swallowing and breathing difficulties. The present case had stridor and feeding problems mainly due to bilateral vocal cord abductor paralysis. The immobility of the vocal cord is known as bilateral vocal cord paralysis, while decreased mobility of the vocal cord is known as vocal cord paresis. Vocal cord paralysis is often unilateral. The present case had rarer bilateral vocal cord paralysis. The etiology of abductor paralysis may not always be identified. Bilateral vocal cord paralysis is sometimes associated with neurological conditions, especially mediastinal masses or trauma at birth (7). No history of birth trauma or neurological involvement was observed in the present case. Bilateral vocal cord paralysis is therefore idiopathic and may be a rare association with RTS. The condition requires endoscopic

examination. Stridor and respiratory symptoms may predominate if the cords are paralyzed in a more middle position, as in the present case. Other surgical possibilities for vocal cord abductor paralysis include tracheostomy, vocal cord lateralization, posterior cordotomy, arytenoidectomy, posterior cricoid split and grafting, and reinnervation by phrenic nerve transfer to the posterior cricoarytenoid muscle (8). Managing infants affected with RTS includes nutrition and growth monitoring, annual hearing and vision screening, and evaluation of cardiac, dental, and renal abnormalities. Behavioral therapy and special training may be later required. We suspected laryngomalacia, tracheal stenosis, or subglottic haemangiomas as a differential diagnosis when the infant developed stridor. The flexible laryngoscopy confirmed the diagnosis of vocal cord abductor paralysis. Most reports of the cases with RTS published in dentistry and neurology journals were children. Reports involving infants are rare. The rare association of RTS with Dandy-Walker malformation has also been reported (9). In this scenario, vocal cord paralysis is the immobilization of a vocal cord due to disease (10). The present infant improved with conservative management within a period of time. Study limitation of the present study is careful genetic testing has not been performed due to financial constraints. However, most cases of RTS are diagnosed based on clinical features. To the best of our knowledge, this rare association of bilateral vocal cord paralysis in RTS is the first case ever reported in the world.

## Conclusion

The association between bilateral vocal cord abductor paralysis and Rubinstein-Taybi Syndrome is rare and should be investigated in future studies. Affected infants need long-term hospitalization for

effective management of stridor and feeding problems. The syndrome has not been widely studied and many questions remain unanswered due to its rarity. The present case report endeavors to add more literature on this syndrome and the possibility of a rare association between bilateral abductor paralysis and RTS.

### Acknowledgments

None.

### Conflicts of interest

The authors declare that no conflict of interest exists regarding the publication of the present study.

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