ABSTRACT

Background: Spinal muscular atrophy is a rare autosomal recessive neurodegenerative disorder. We report a case of 62-year-old female of spinal muscular atrophy presenting with bilateral vocal fold paralysis, her diagnosis and management.

Objective: Case report of a case of spinal muscular atrophy presenting with bilateral vocal fold paralysis.

Conclusion: Spinal muscular atrophy though rarely associated with laryngeal symptoms should be kept in mind as a possible etiology of bilateral vocal fold paralysis. The surgical decision and the postoperative sequelae will be affected due to the presence of this disease and thus a high index of suspicion is required.

Keywords: Vocal fold, Palsy, Spinal, Muscular atrophy.


Source of support: Nil

Conflict of interest: None

INTRODUCTION

Spinal muscular atrophy (SMA) is a rare, autosomal recessive disorder that involves progressive degeneration of motor neurons in the spinal cord, resulting in weakness of the extremities. It is rarely associated with laryngeal symptoms. We report the diagnosis and management of a patient of SMA who presented with bilateral abductor vocal fold paralysis leading to respiratory distress.

CASE REPORT

A 62-year-old female presented to us with the history of respiratory distress since 6 months which had worsened since the past 3 days. She was a diabetic and a hypertensive on treatment and well controlled. She had a history of ischemic heart disease and had undergone a coronary angiography a year back. She also had noticed a mild weakness of her hand muscles for which an electromyography (EMG) was done recently in another hospital and was reported normal. A laryngoscopic examination revealed a bilateral vocal fold immobility. As a routine investigation for bilateral vocal fold immobility, computed tomogram with contrast with 2 mm cuts of base skull to mediastinum was done, which was otherwise normal.

In our institute, we routinely perform unilateral surgery for such patients of bilateral vocal fold paralysis, so as to preserve the voice to the maximum. The patient is also counselled that in a small percentage, a second stage surgery on the other side may be required.

Thus, in view of the above findings, the patient was posted for a endoscopic laser Kashima’s unilateral vocal fold cordotomy. During preoperative anesthesia evaluation, patient was noted to have a mild intercostal muscle weakness. However, the previous EMG report was normal. She also had CO₂ retention which could be attributed to the bilateral vocal fold palsy.

Patient was taken under general anesthesia. Klinssauser laryngoscope was introduced and suspended which revealed a good view of the posterior segment of the vocal folds (Fig. 1). Bilateral vocal folds were palpated and cricoarytenoid joint fixation was ruled out. Using the CO₂ laser (Acublade, superpulse mode, 2 mm length, 2 mm depth, 10W) a transverse cut was made in the posterior part of the right vocal fold to separate the right vocal fold from the vocal process. The cut was made along the whole thickness and the depth of the vocal fold upto the inner perichondrium of the thyroid cartilage (Fig. 2).

Though the procedure was uneventful, postoperatively, the patient’s CO₂ retention increased. She was shifted to an intensive care unit and was kept on BIPAP support for 3 days. Subsequently, the patient gradually improved with relief of respiratory distress. In view of increased CO₂ retention and finding of intercostal muscle weakness, a neurology opinion was sought and a repeat EMG was done in our hospital.

Neurological examination revealed a muscle weakness in proximal lower limbs and distal hand muscle weakness, tendon areflexia, no thickening of peripheral nerve, intact sensation and normal intelligence. Other cranial nerves were normal.

REFERENCE

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Repeat EMG (Figs 3A and B) done at our hospital showed changes consistent with chronic neurogenic denervation suggestive of anterior horn cell involvement, which was consistent with the diagnosis of SMA. In view of the finding of bilateral vocal fold paralysis, it was diagnosed as SMA type VII.

Retrospectively, on direct questioning, patient did give a history of difficulty in squatting which was largely overlooked by her.

Thus, a diagnosis of spinal muscular atrophy in a patient with bilateral vocal fold paralysis was established.

On follow-up, patient did not have any respiratory distress. She did not complaint of any dysphagia or aspiration. Quality of voice was also satisfactory.

**DISCUSSION**

Spinomuscular atrophy is a autosomal recessive neuromuscular disorder characterized by degeneration of anterior...
horn cells in the spinal cord, in 20% motor nuclei of the brainstem may be involved. In over 95% of cases of spinal muscular atrophy, a genetic autosomal recessive disorder causes a functional loss of the survival motor neuron 1 (SMN1) gene on chromosome V. In 2% of the cases the transmission appears to be either autosomal dominant, X-linked or sporadic. It is characterized by progressive muscular atrophy and weakness resulting in limb weakness with usually the lower extremities being affected first, followed by muscles of upper extremities, spine and neck and, in more severe cases, pulmonary and mastication muscles. There is no sensory component to the disorder. It manifests over a wide range of severity affecting infants through adults and is classified into four types depending on the age of onset of symptom and the milestone achieved.

The association of vocal fold paralysis with distal muscular atrophy is very rare. A review of literature suggests that this association was first described by Young and Harper in 1980. They described this condition in a Welsh family wherein the members presented with progressive distal muscle wasting and weakness together with hoarse voice. The condition was assumed to be a form of distal spinal muscular atrophy associated with vocal fold paralysis. In a subsequent classification of the proximal and distal spinal muscular atrophies the disorder was designated dSMA type VII. In 1989, Boltshauser et al also described distal spinal muscular atrophy with vocal fold paralysis and sensorineural deafness involving three generations in a family. Similar findings were described by Pridmore et al. Roulet and Deonna reported a case of SMA type 1 presenting with vocal fold paralysis. Thaler et al presented a case of SMA with cricoarytenoid joint fixation.

Our case of SMA presented with respiratory distress. Laryngoscopy revealed a bilateral vocal fold palsy which was treated by Kashima’s cordotomy. The main aim of the surgery for bilateral vocal fold palsy is to restore a lumen sufficient to guarantee adequate breathing through the airway, without the patient having to maintain the tracheotomy tube, while preserving acceptable phonatory function and without impairing the swallowing. Kashima and Dennis proposed the CO$_2$ laser posterior cordotomy in 1989. They recommended limiting partial cordotomy to the dorsal glottis in order to maximally preserve the functionally important vibrating anterior part of vocal folds.

An important drawback of the surgery is worsening of voice quality to some extent postoperatively. Hans et al reported that when speech quality was evaluated objectively, an improvement in acoustical and aerodynamic parameters was noted over six postoperative months. Other possible complications of the surgery include granuloma formation, perichondritis of cartilage and ipsilateral false fold hypertrophy (if not removed) causing inspiratory dyspnea.

CONCLUSION

Spinomuscular atrophy though rarely associated with laryngeal symptoms should be kept in mind as a possible etiology of bilateral vocal fold paralysis. The surgical decision and the postoperative sequelae will be affected due to the presence of this disease. Hence, a high index of suspicion ensures an early diagnosis and treatment.

REFERENCES