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Noninvasive ventilation and laser-assisted unilateral posterior cordotomy as novel multidisciplinary approaches for Charcot–Marie–Tooth disease 4B vocal cord paralysis: a case report

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Abstract

Background Charcot–Marie–Tooth disease (CMT) is one of the most common inherited neuropathies. The disease is generally characterized by sensory loss most prominent in distal extremities, muscle weakness, and muscle wasting. There is still no effective therapy for Charcot–Marie–Tooth disease.

Case presentation The patient is a 6-year-old Iranian girl, of Fars ethnicity, who was admitted with a chief complaint of hoarseness and an impression of Charcot–Marie–Tooth disease type 4B. She was initially treated with noninvasive ventilation and, after a year, electively underwent cordotomy as a novel therapeutic approach.

Conclusions Charcot–Marie–Tooth disease type 4B is a less common but important cause of stridor. Noninvasive ventilation treatment and unilateral posterior cordotomy can be utilized for hereditary neuropathies.

Keywords Charcot–Marie–Tooth disease, Vocal cord paralysis, Noninvasive ventilation, Cordotomy, Case report

Background

Charcot–Marie–Tooth disease (CMT) is one of the most common inherited neuropathies, affecting a huge number of people around the world [1]. The disease course is variable due to genotypic and phenotypic heterogeneity of patients [2]. Common manifestations of CMT include sensory loss (especially distal extremities), muscle weakness, and muscle wasting. Meanwhile, vocal cord paralysis presentation is rare [1, 3]. In addition, the presence of focally folded myelin sheaths in nerve biopsy distinguishes Charcot–Marie–Tooth disease type 4B from other subtypes [3].

Currently, there is no effective medication available for CMT; thus, supportive therapy and surgical treatment of skeletal deformities and soft-tissue abnormalities are the best possible choice [2]. In this case report, we present the case of a 6-year-old girl who was admitted with confirmed Charcot–Marie–Tooth disease type 4B with an unusual presentation of vocal cord paralysis that was initially treated with noninvasive ventilation (NIV) and

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subsequently underwent elective cordotomy as a novel therapeutic approach.

Case presentation

Here, we present the case of a 6-year-old Iranian girl, of Fars ethnicity, who was admitted to the emergency ward of the Mofid Children's Hospital, a tertiary medical education center in Tehran, Iran, with a chief complaint of hoarseness. Her past medical, drug, family, and social history was negative. Her vital signs showed a temperature of 36.8 °C, heart rate of 62 beats per minute, respiratory rate of 31 breaths per minute (tachypnea), blood pressure of 92 mmHg, and O_2 saturation of 91%. On physical examinations (P/E), she had suprasternal retraction, and her lung showed auscultation, prolonged inspiration, and high-pitch creaking (stridor) during inhalation were detected. Unfortunately, the patient did not cooperate in neurological examinations. Therefore, she was initially admitted with the impression of croup, and she received glucocorticoid, epinephrine nebulizer, and conservative therapy. However, due to inadequate response to the treatment, a pediatric pulmonology consultation was requested, during which vocal cord pathology was suspected. A fiberoptic bronchoscopy revealed bilateral vocal cord paralysis with the normal subglottic area, trachea, and bronchus. Furthermore, the patient was reevaluated, and in her second comprehensive P/E, mild limb weakness and neuropathy were noted. Noticeably, after a 1-week admission to the hospital, the patient parents mentioned that genetic and nerve conduction studies had been previously performed for her and confirmed Charcot–Marie–Tooth type 4B. It should be mentioned that, initially, her parents denied any past medical history for her daughter.

Regarding vocal cord paralysis, the patient was a candidate for tracheostomy. Since the patient's parents did not consent to the operation owing to social determination of the tracheostomy tube appearance in society, noninvasive ventilation (NIV) with spontaneous/timed (ST) mode was performed as a novel therapeutic approach for her. NIV-ST mode by inducing positive pressure could reduce the patient's muscle force and alleviate the patient's condition. Also, it could prevent apnea in children.

NIV was well tolerated for 1 year till the patient went to school. There, NIV use was problematic, and concerns about NIV use were raised for her parents. Therefore, 2 months after surgical consultation, laser-assisted unilateral posterior cordotomy was performed (Fig. 1). As a result, the patient had acceptable respiratory function and no serious complaints regarding aspiration happened. It should be mentioned that, due to consultation with ENT, the suitable choice for her was tracheostomy, as cordotomy might present severe complications such as severe inflammation of larynx. However, the patient's parents did not assent to tracheostomy; thus, unilateral cordotomy was performed. Furthermore, on the 1-year follow-up, she had stable respiratory status and had no swallowing problems.

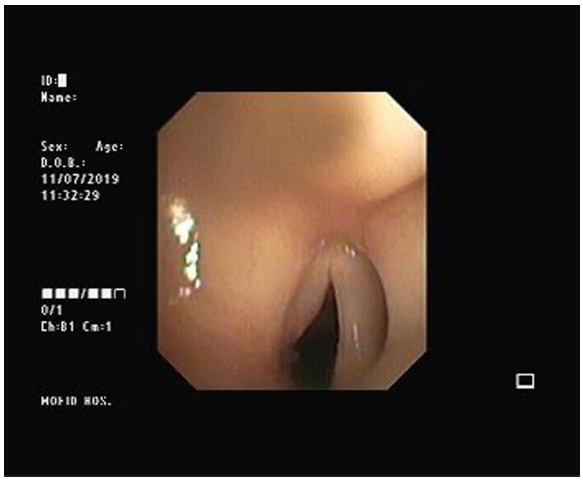

Fig. 1 The 2-month follow-up of unilateral laser cordotomy

Discussion

CMT is one of the most common causes of hereditary peripheral neuropathy [1]. Demyelination and axonal neuropathy are the common pathological presentation of this disease. Approximately 70% of patients with CMT 1 have a demyelination mechanism, which is autosomal dominant [4]. The autosomal recessive pattern accounts for 5% of patients [5]. Eighty genes have been detected for different types of this disease [4].

CMT 4B has an autosomal recessive pattern that manifests as both neuropathy and demyelination. Also, a gene mutation in the myotubularin-related protein (MTMR) gene plays a significant role in the vesicular trafficking of Schwann cells and causes CMT 4B [5].

Several cases of vocal cord paralysis have been reported to occur in the first two decades of life in patients diagnosed with CMT2A, CMT2C, CMT4A, and CMT4B [6]. Although most of the reported patients were within the first 2 years of life, in the current case report our patient presented her vocal cord paralysis at the age of 6. Furthermore, recurrent laryngeal nerve involvement may cause vocal cord paralysis, which may be complicated by aspiration and life-threatening pneumonia [6–8].

Currently, no curative treatment is available for CMT disease. Supportive therapy is performed by multidisciplinary teams to improve quality of life and increase life expectancy. Interestingly, Tyson *et al.* successfully used

NIV to treat a 16-year-old boy with bilateral vocal cord paralysis [9]. The decision to treat a patient with bilateral vocal cord hypomobility depends on the severity of the symptoms, the patient's functionality, and the patient's values. According to Brake and Anderson's article, continuous positive airway pressure (CPAP) could be considered for patients with bilateral vocal cord paralysis who are not candidates for surgery [10]. In our patient, NIV was initiated, since the parents refused tracheostomy, and the patient's symptoms were relieved satisfactorily.

Carmel-Neiderman *et al.* in their case series of 12 patients with bilateral vocal cord hypomobility mention that none had CMT disease and proposed that posterior cordotomy is a safe and effective alternative to tracheotomy when cord mobility is unlikely to return. Also, aspiration pneumonia was not reported in any of their patients. A breathy voice and, in some cases, dysphagia to thin fluids may be expected. In three patients, scarring led to further interventions such as revision of the posterior cordotomy (PC), lateralization, and arytenoidectomy [11]. Among 48 patients with bilateral fold immobility in a referral center reviewed by Brake and Anderson, unilateral cordotomy was performed for 21 patients (48%). In one patient diagnosed with CMT, Reinke's edema recurred, leading to contralateral cordotomy/arytenoidectomy and later vocal cord lateralization via an external approach. Overall, respiratory function did not improve in only two (10%) of the cordotomy patients [10]. In our patient, after a 1-year follow-up, the parents were concerned about the psychosocial difficulties their child would encounter if she was required to use NIV permanently at her school; therefore, an alternative therapeutic plan was needed. Despite the risk of aspiration, a decision was made to perform cordotomy surgery on the patient. The patient's symptoms resolved after surgery. There was no complaint related to aspiration.

To the best of our knowledge, this is the first report on treating a patient with CMT4B via a cordotomy procedure. The vocal cord laterofixation procedure was evaluated by Zambon *et al.* to treat a 3-year-old girl with vocal cord paralysis who was diagnosed with CMT type 4B. However, the procedure unfortunately failed to relieve the symptoms, and a week later, a tracheotomy was performed to relieve the patient's respiratory distress. Active adduction of arytenoids during the whole respiratory cycle was documented by video laryngoscopy as the cause of obstruction recurrence [6].

Moreover, the cordotomy procedure has also been successfully used to treat a 7-month-old patient with congenital bilateral vocal cord paralysis due to CMT type 1 disease. No complaint regarding aspiration was reported until the age of 4 when stridor and respiratory insufficiency symptoms developed again. Following arytenoidectomy, symptoms were relieved until the age of 7 when the patient was followed up [12].

Conclusion

Physicians should consider CMT4B as a rare disease; however, it is an important cause of stridor in pediatrics. NIV treatment could relieve patient conditions with hereditary neuropathies and vocal cord paralysis. In addition, unilateral posterior cordotomy can be considered to safely improve respiratory function while avoiding tracheotomy; however, further research is mandatory to investigate NIV treatment and unilateral posterior cordotomy effects.

Abbreviations

CMT	Charcot–Marie–Tooth disease
NIV	Noninvasive ventilation
MTMR	Myotubularin-related protein
CPAP	Continuous positive airway pressure
PC	Posterior cordotomy

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Author contributions

NF took the history from the patient's parents, examined and managed the patient, and was a major contributor to the writing of manuscript. JKP, MHGZ, AK, and ARB wrote the manuscript. NF and ARB revised the final manuscript. All authors read and approved the final manuscript.

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Declarations

Ethics approval and consent to participate

Informed consent was obtained from the participant included in the study..

Consent for publication

Written informed consent was obtained from the patient's legal guardian for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Competing interests

The authors declare that they have no competing interests.

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References

1. Reilly MM, Murphy SM, Laurá M. Charcot-Marie-tooth disease. J Peripher Nerv Syst. 2011;16(1):1–14.
2. Pareyson D, Marchesi C. Diagnosis, natural history, and management of Charcot–Marie–Tooth disease. Lancet Neurol. 2009;8(7):654–67.
3. Grandis M, Shy ME. Current therapy for Charcot-Marie-Tooth disease. Curr Treat Options Neurol. 2005;7(1):23–31.
4. Jani-Acsadi A, Ounpuu S, Pierz K, Acsadi G. Pediatric Charcot-Marie-Tooth disease. Pediatr Clin. 2015;62(3):767–86.

5. Previtali SC, Quattrini A, Bolino A. Charcot–Marie–Tooth type 4B demyelinating neuropathy: deciphering the role of MTMR phosphatases. Expert Rev Mol Med. 2007;9(25):1–16.
6. Zambon AA, Sora MGN, Cantarella G, Cerri F, Quattrini A, Comi G, *et al*. Vocal cord paralysis in Charcot–Marie–Tooth type 4b1 disease associated with a novel mutation in the myotubularin-related protein 2 gene: a case report and review of the literature. Neuromuscul Disord. 2017;27(5):487–91.
7. Aboussouan LS, Lewis RA, Shy ME. Disorders of pulmonary function, sleep, and the upper airway in Charcot-Marie-Tooth disease. Lung. 2007;185:1–7.
8. Marimbaldo P. Respiratory disease and diaphragm paralysis in Charcot-Marie-Tooth disease. Anales de Pediatria (Barcelona, Spain: 2003). 2010. https://doi.org/10.1016/j.anpedi.2009.11.017.
9. Tyson J, Ellis D, Fairbrother U, King R, Muntoni F, Jacobs J, *et al*. Hereditary demyelinating neuropathy of infancy. A genetically complex syndrome. Brain J Neurol. 1997;120(1):47–63.
10. Brake MK, Anderson J. Bilateral vocal fold immobility: a 13-year review of etiologies, management and the utility of the Empey index. J Otolaryngol Head Neck Surg. 2015;44:1–7.
11. Carmel-Neiderman NN, Chason M, Wengier A, Wasserzug O, Cavel O, Horowitz G, *et al*. Endoscopic posterior cordotomy for treatment of dyspnea due to vocal fold immobility. Multidiscip Respir Med. 2020. https://doi.org/10.4081/mrm.2020.35.
12. Lewis AF, Carron JD, Vedanarayanan V. Congenital bilateral vocal fold paralysis and Charcot-Marie-Tooth disease. Ann Otol Rhinol Laryngol. 2010;119(1):47–9.

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