Laryngeal Electromyographic Findings in Charcot-Marie-Tooth Disease Type II

Todd G. Dray, MD; Lawrence R. Robinson, MD; Allen D. Hillel, MD

Charcot-Marie-Tooth disease is a hereditary motor and sensory neuropathy that exhibits progressive muscular atrophy in the limbs, beginning with the lower extremities. It is now understood to be a heterogeneous group of disorders that can be differentiated both clinically and genetically. In Charcot-Marie-Tooth disease type II C, axonal neuropathy, diaphragm weakness, and vocal cord paralysis are described within kindreds. We used laryngeal electromyography to study a patient with this disorder. This technique has potential in the diagnosis of Charcot-Marie-Tooth disease type II.

Classically, Charcot-Marie-Tooth disease (CMT), also known as hereditary motor and sensory neuropathy or peroneal muscular atrophy, has been considered a hereditary disorder that begins with weakness and atrophy in peroneal muscles. The condition may advance to involve other distal muscles of the upper and lower limbs. Recently, detailed kindred evaluations and genetic linkage studies have demonstrated that CMT is a heterogeneous group of disorders. Type I CMT is a demyelinating neuropathy; type II is a less severe neuronal form and appears to be less common. In general, patients with CMT type II develop less hand involvement, do not have palpably enlarged nerves, and have a later age at onset. It appears to be a genetically distinct entity from CMT type I. Further genetic heterogeneity within CMT type II has been suggested by genetic linkage studies. Several kindreds with features of axonal neuropathy, diaphragm weakness, and/or vocal cord paralysis are described (CMT type II C). In this report, we discuss a patient with CMT type II disease and laryngeal findings consistent with bilateral recurrent laryngeal neuropathy based on history, videostroboscopic examination, and laryngeal electromyography (EMG).

A 39-year-old woman was first examined at our institution in May 1997. She had undergone multiple orthopedic procedures for talipes cavus and hammer toe deformities. In 1981 she had undergone an EMG study of her lower extremities at an outside hospital and was given the diagnosis of probable CMT. In 1988, the patient underwent a nerve conduction study at an outside institution that showed normal nerve conduction velocities of her lower extremities; a diagnosis of probable axonal-type CMT was made. She complained of slowly progressive weakness in both lower extremities since childhood and recent weakness in the upper extremities. Sensory symptoms included intermittent burning in the right fourth and fifth digits and chronic numbness in the left great toe. In addition, the patient reported a 1-year history of vocal fatigue and hoarseness, particularly noticeable while singing at church. Family history showed that the patient's 69-year-old mother had been diagnosed as having axonal neuropathy in 1993.

The patient's workup at our institution started in the multidisciplinary muscular dystrophy clinic. An EMG of the lower extremities was performed and motor-sensory needle conduction velocities were determined. Results of motor and sensory conduction studies were within normal limits for latency, amplitude, and conduction velocities with the exception of absent left...
sural sensory nerve action potentials. Prolonged latency and decreased amplitudes of the right peroneal compound muscle action potential were judged consistent with changes secondary to her foot surgery. The stimuli required to achieve sensory nerve action potentials and compound muscle action potentials were noted to be greater than normal. Needle EMG examination elicited large-amplitude motor unit potentials in both abductor hallucis muscles and the left extensor hallucis longus, suggestive of reinnervation from a chronic neuropathic process. There was no evidence of acute denervation. Taken together with the clinical appearance, these findings suggested mild hereditary motor and sensory neuropathy type II.

The patient was referred to the Voice Disorders Clinic in July 1997 to evaluate her hoarseness. At that time, her voice had a slightly breathy quality. Loudness, nasal resonance, and intonation were noted to be adequate. A full head and neck examination disclosed normal cranial nerve function bilaterally, with the exception of the vagus nerve. On videostroboscopy, full range of true vocal fold abduction and adduction were seen. Glottic closure was incomplete secondary to a small (<1 mm) posterior gap during phonation. The mucosal waves had decreased amplitudes bilaterally. Despite identification of normal vocal fold mobility, subtle signs of laryngeal weakness were noted on videostroboscopy. Consistent with our standard workup in this setting (laryngeal weakness of unknown cause), laryngeal EMG was performed (Table). Findings included (1) no fibrillation potentials and reduced recruitment in the right lateral cricoarytenoid muscle; (2) normal findings in the right cricothyroid muscle; (3) 2+ fibrillation potentials, occasional polyphasic potentials, and normal recruitment in the left thyroarytenoid muscle (Figure); and (4) no fibrillation potentials, large (>1500 mV) motor unit action potentials, and reduced recruitment in the right thyroarytenoid muscle. The laryngeal EMG was consistent with a peripheral neuropathy.

In 1886, Charcot and Marie described a progressive muscular atrophy with a familial tendency that started in the feet and legs. The hands were often involved in later stages of the disease. Fasciculations and atrophic muscles were noted, and, although sensation was usually grossly intact, it was sometimes affected. The disease was also characterized by diminished or absent deep tendon reflexes and talipes cavus. Bell's review of the literature concluded that inheritance could be dominant, recessive, or sex-linked, and some cases could be sporadic. It is now understood that CMT is a hereditary disorder and can be classified with a broad spectrum of neurodegenerative diseases, including the olivopontocerebellar, cerebellopontine, and spinocerebellar disorders.

Dyck and Lambert observed that, on the basis of electrophysiological characteristics, peroneal muscular atrophy kindreds could be separated into groups with markedly slowed nerve conduction velocity (NCV) and normal (or mildly abnormal) NCV. The kindreds with low NCV were classified as CMT type I, having features resembling those classically described by Charcot and Marie as well as by Tooth. The kindreds with normal or borderline-low NCV were subdivided into CMT type II and other degenerative diseases. These observations have subsequently been validated and extended by others. Genetic heterogeneity within CMT type II was suggested as kindreds with features of axonal neuropathy, vocal cord paralysis or paresis, and variable diaphragmatic weakness were described. These kindreds, designated as CMT type II C, underwent visual inspection of vocal cords, without laryngeal EMG description of the laryngeal muscles and nerves. A report by Holinger et al in 1979 described the association of bilateral abductor vocal cord paralysis and CMT in a mother and son. The EMG and NCV stud-

**Table**

<table>
<thead>
<tr>
<th>Muscle</th>
<th>Spontaneous Activity</th>
<th>Voluntary MUAPs</th>
<th>Recruitment†</th>
</tr>
</thead>
<tbody>
<tr>
<td>Right LCA</td>
<td>None</td>
<td>Normal</td>
<td>Reduced</td>
</tr>
<tr>
<td>Right CT</td>
<td>None</td>
<td>Normal</td>
<td>Full</td>
</tr>
<tr>
<td>Left TA</td>
<td>Fibrillations</td>
<td>Increased polyphasics</td>
<td>Normal</td>
</tr>
<tr>
<td>Right TA</td>
<td>None</td>
<td>Large amplitude</td>
<td>Reduced</td>
</tr>
</tbody>
</table>

*MUAP indicates motor unit action potential; LCA, lateral cricoarytenoid; CT, cricothyroid; and TA, thyroarytenoid.*

†Reduced recruitment indicates reduced number of MUAPs firing rapidly.

Electromyogram of the left thyroarytenoid muscle shows polyphasic potential (arrow).
ies verified the diagnoses of CMT, but no laryngeal EMG analysis was performed. Although the cause of the vocal cord paralysis went unknown, the authors correctly postulated that vocal and respiratory involvement might represent an important genetic marker.

Electromyography has proved useful for the evaluation of neurologic diseases involving the laryngeal muscles. This report describes laryngeal EMG identification of a lower motor neuron lesion of the vagus nerve and its laryngeal branches diffusely affecting the intrinsic laryngeal muscles in a patient with CMT. The patient had reported chronic hoarseness, and a laryngology evaluation failed to identify a cause before the EMG study. The finding of large and rapid-firing muscle unit action potentials in this patient confirms a chronic peripheral neuropathic disorder affecting her laryngeal function. In general, peripheral nerve or lower motor neuron denervation demonstrates an EMG response with positive sharp waves and/or fibrillation potentials when the axon loss is recent. Reinnervation patterns of EMG response show polyphasic potentials, or large motor unit action potentials, which are indicative of reinnervation by distal axonal sprouting. In contrast, a central pattern of EMG response appears with normal motor unit action potentials that are decreased in number and fire at a slower rate. In this patient, with known CMT type II, laryngeal evaluation including laryngeal EMG allowed a more precise diagnosis of CMT type II C to be made. Currently no therapeutic intervention has been offered, and the patient will be followed up during the next year. A potentially reversible procedure medializing the vocal fold might be offered in the future to improve her voice. Permanent medialization procedures (arytenoid adduction) will not be considered because of the potential progression of abductor paralysis.

CONCLUSIONS

Although laryngeal EMG is in its clinical infancy, the technique has great potential in enabling the laryngologist to assist in the diagnosis and treatment of neurologic diseases involving the head and neck. In this case, the laryngeal EMG pattern in a patient with CMT disease is presented. Further hoarseness and vocal fatigue in this patient will most likely represent a progressive peripheral lesion and may be amenable to a reversible medialization procedure.

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REFERENCES