**CLINICAL NOTES**

**Orthotic Technique for Dystonia Musculorum Deformans**

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- Tone reducing, inhibitive casting, and orthoses have been effectively used in patients with cerebral palsy and head injury to improve gait patterns and decrease tone. We present a patient with dystonia musculorum deformans who had severe inversion and supination of his left foot with weight bearing. He did poorly with metal double-upright ankle-foot orthoses with lateral T-strap. A tone-reducing ankle-foot orthosis (TRAFO) was successful in decreasing problems with abrasions and allowing him to walk without assistive devices.

**KEY WORDS:** Dystonia; Dystonia musculorum deformans; Orthosis

Dystonia musculorum deformans is a genetic syndrome which has a recessive form seen most commonly in Jews of eastern European descent, and a dominant form which is sporadic. It is thought to be due to a basic neurochemical abnormality of the brain, possibly related to the norepinephrine, serotonin, and dopamine systems of the telencephalon, diencephalon, and lower brainstem. According to Marsden and colleagues, it is defined by (1) the presence of dystonic postures and movements with a normal perinatal history; (2) no history of any other precipitating causes; (3) no signs of intellectual, cerebellar, pyramidal, or sensory deficits; and (4) a negative laboratory examination, including a work-up for Wilson disease. It can present at any age, and is more devastating with younger onset. Often the onset occurs in the left leg and is noted as walking difficulty. It can then spread to the other extremities, trunk, and neck. With severe dystonia, contractures and malformations can occur, which can further impede ambulation and activities of daily living. Attempts to correct gait abnormalities with bracing have generally been unsuccessful.

Tone-inhibiting casting is well described for children with cerebral palsy. This idea was modified into a tone-reducing ankle-foot orthosis (TRAFO) by Zachazewski’s group for use in an adult, closed-head-injury patient. We present a patient with dystonia musculorum deformans whose ambulation improved considerably when changed from a double-upright metal ankle-foot orthosis (AFO) to a modified TRAFO.

**CASE REPORT**

The patient was a 28-year-old right-handed man of Greek, Irish, and English descent. His father had a movement disorder of unknown type which was treated with thalamotomy. His paternal grandmother had a neurologic deficit which was called a “stroke.” The patient’s left leg was turning in. His dystonic movements progressed, eventually involving his trunk, pelvis, mouth, and tongue. The movements were not present when he was asleep. He was treated with a variety of medications including Ariane, Sinemet, L-Dopa, Tegretol, and Valium, none of which was particularly effective. He underwent a right thalamotomy at age 17 and a left thalamotomy at age 19, with some improvement of sitting balance and dystonia after the first thalamotomy, but only minimal change after the second one. When he attempted weight bearing, his left foot inverted and supinated maximally, and could only be everted by hand. An EMG demonstrated few motor units in the left anterior tibialis and left gastrocnemius, and none in the left peronei. Left tibial nerve conduction velocity was 49 meters per second. X-rays were normal.

At age 17 he was fitted with a posterior leg splint for use at night, and a double-upright short leg brace with an outside T-strap for ambulation. Despite the brace, he continued to have some inversion, and a steel plate and strap were added to the brace. Tibial nerve block was attempted but did not improve his gait. At age 28, the patient began to experience painful swelling on the lateral aspect of his left foot. X-rays of the foot and ankle were normal. The patient was evaluated for ankle fusion, but the orthopedic surgeon felt that it was not indicated. A podiatrist placed a fiberglass cast on the left foot to try controlling the inversion and plantar flexion to decrease the lateral foot swelling. The cast helped his gait, but broke after three weeks because of the severe inversion. He presented to our Orthotics/Prosthetics Clinic for further management of his problem.

On physical examination he had dystonic posturing of his entire trunk, with a tendency to flex to the right and to hyperextend as he ambulated. With the brace, his gait and stance were wide based and he had inconsistent step lengths. He had no left-sided heel strike or toe-off. Stepping directly on the sole of his left foot, within the brace, caused foot inversion and plantar flexion, as well as internal rotation at the hip. During the swing phase of gait, there was decreased knee flexion and hiking of his left pelvis. Examination of his left foot revealed a raised, fluctuant, erythematous, tender area on the lateral aspect measuring 3 x 4 cm (fig 1). A fiberglass-impregnated, plaster short leg cast was made with the foot in a neutral position. The maximum range achieved was 80° of dorsiflexion and 5° of inversion. No padding was placed over the bony prominences in order to provide a tighter fit and prevent further callus formation. Casting was performed with a piece of rubber tubing on the anterior leg from the tibial tubercle to the toes to aid in cast removal. While wearing the cast, the patient’s gait improved significantly. He had a narrower base, more regular cadence, and less hip rotation. During swing phase there was more knee flexion and a more normal pattern of pelvic movement.

The patient was then fitted with a bivalved, plastic AFO with solid...
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Fig 1—Patient's left foot, in which lateral swelling and inversion deformity are visible.

ankle and 5° of dorsiflexion (fig 2). The brace had a polypropylene posterior section and a high-density polyethylene anterior section. An ankle check strap was incorporated under the extended bivalved anterior section. The strap aided in keeping the ankle against the posterior section of the AFO. The negative model of the foot was built up over the malleoli and over the area of the abrasion at the styloid process of the 5th metatarsal. A one-quarter-inch lateral heel and sole wedge were added to the shoe. The orthosis was held together by two Velcro straps. Six degrees of varus was required to accommodate the inversion deformity. The finished orthosis was a modified version of the original TRAFO. Ambulation in the brace was similar to the pattern seen with the cast and significantly improved over the double upright AFO.

He wore the brace continuously throughout the day. When examined three weeks later, he continued to walk with a less wide-based gait and with increased knee flexion and more stability. Range of motion in his foot improved to 100° of dorsiflexion and 5° of eversion. Three months later, the fluctuant, swollen area had considerably decreased, and extra padding was required over the 5th metatarsal (fig 3). Six months after the original casting, the patient's gait pattern had improved to the point where a new casting was required, and a new brace was made. He initially had extra folds of skin on the lateral aspect of his foot due to the long-standing foot inversion, but this decreased. The patient then wore the brace all day without discomfort, and ambulated without an assistive device.

DISCUSSION

Inhibitive casting is a well-known technique for treating children with cerebral palsy whose increased tone forces their feet into equino-varus position. The casting is designed to be therapeutic and temporarily serves as an adjunct to therapy. Sussman and Kusick suggested that casting physiologically acts to extend the toes, thus inhibiting the plantar grasp response, which decreases tone. Further, the cast firmly fixates the unstable foot and ankle to give a stable base of support which also serves to reduce tone. Zachazewski reported a case of inhibitive casting in an adult head-injury patient whose difficulty ambulating due to a positive support reaction was helped by inhibitive casting. To maintain the inhibition, a molded orthosis was required which had the same tone-inhibiting characteristics as casting: hyperextension of the toes, pressure under the metatarsal heads, a stable ankle position, and deep tendon pressure along the tendo calcaneus.

We used the TRAFO in this patient as a permanent brace rather than an adjunct to a therapy program designed to decrease tone and normalize gait pattern. We felt that the lack of peroneal muscle activity on EMG indicated that retraining efforts would not be beneficial. Furthermore, sensory feedback therapy via EMG has been shown to be helpful with torticollis, but there are no reports of improving gait in dystonia patients with therapy.

Dystonia musculorum deformans affects gait in 80% of patients to the point where 14% are wheelchair dependent. Twenty
suggests that this method should be applicable to patients with a number of neuromuscular disorders. Preliminary data from patients and patients who had had complete sections of the ulnar or median nerve. After determining the recruitment threshold and the twitch tension (spike-triggered averaging) of a single unit, its nerve was stimulated at the wrist and the elbow using surface electrodes. By using computerized subtraction of responses just above and below threshold for a given unit, the same motor unit could often be identified in response to stimulation at both sites and its conduction velocity determined. The twitch tension and recruitment threshold of the motor units were closely correlated with the conduction velocity of the motor axons in normal subjects. Preliminary data from patients suggests that this method should be applicable to patients with a number of neuromuscular disorders.

CONCLUSION

Dystonia musculorum deformans is a genetic condition which often affects gait. Our patient had extremely severe inversion and supination of his left foot with any attempt at weight bearing. This problem did not respond to numerous pharmaceutical or therapeutic interventions. He did poorly with a conventional double-upright AFO with an outside T-Strap. A modification of a tone-reducing ankle-foot orthosis was successful, decreasing his problems with abrasions and allowing him to walk without assistive devices.

References

ABSTRACTS of selected literature


- Tungsten microelectrodes of the type used for microneurography have been used to record motor units selectively from the first dorsal interosseous and abductor pollicis brevis muscles of normal subjects and patients who had had complete sections of the ulnar or median nerve. After determining the recruitment threshold and the twitch tension (spike-triggered averaging) of a single unit, its nerve was stimulated at the wrist and the elbow using surface electrodes. By adjusting the position of the surface electrode and the stimulus intensity and by using computerized subtraction of responses just above and below threshold for a given unit, the same motor unit could often be identified in response to stimulation at both sites and its conduction velocity determined. The twitch tension and recruitment threshold of the motor units were closely correlated with the conduction velocity of the motor axons in normal subjects. Preliminary data from patients suggests that this method should be applicable to patients with a number of neuromuscular disorders.


- The involuntary angle-specific isokinetic plantar flexor torques of seven male subjects aged 18-21 yr were measured using a Cybex II dynamometer (Lumex) modified by the addition of a strain gauge load cell to improve the dynamic response of the instrument. Supramaximal electrical stimuli were used to evoke a maximal tetanic response from the triceps surae and ensure constant muscle activation at each angular velocity studied. Angle-specific torques were measured over a range (0.5-5.0 rad/s) of preset velocities, torque decreasing in a nonlinear manner with increasing angular velocity. The torque-velocity data was adequately described by an exponential equation of the form: \[ V = P e^{-\frac{a}{b}} - P e^{-\frac{b}{b}} \] where \( V \) = velocity (rad/s), \( P \) = torque (N-m), \( P_0 \) = isometric torque (N-m), and \( a \) and \( b \) are constants. The mean intrasubject coefficient of variation of torque over the range of velocities studies was 7.9 ± 1.88% (SD).