Orthotic management of the lower limb in children with hereditary motor sensory neuropathy (HMSN)

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Abstract
The lower limbs of 55 paediatric patients, with the diagnosis of hereditary motor sensory neuropathy (HMSN) referred to the Orthotics and Biomechanics Department of Hacettepe University, were assessed for appropriate orthotic intervention. Since in the natural history of HMSN symptoms and complaints are variable there is a wide range of interventions possible. The biomechanics of deforming forces and the consequential incidence of deformities in these 55 children, its orthotic implications and the efficacy of orthotic applications are discussed in detail.

Introduction
In the Orthotics and Biomechanics Department of the School of Physical Therapy and Rehabilitation of Hacettepe University many children with neuromuscular disorders are referred and treated from various medical institutions all over Turkey.

Among other neuromuscular diseases hereditary motor sensory neuropathy (HMSN) in the juvenile form makes up a considerable number of the patients. Starting from 1992, 55 paediatric patients with the diagnosis patients with the diagnosis of HMSN were seen.

HMSN, also known as Charcot Marie-Tooth, is an inherited progressive degenerative disorder of the nervous system resulting in distal muscle weakness and atrophy as well as impaired sensation (Walton, 1977; Alexander and Johnson, 1987).

Wasting does not usually appear in the hands until a number of years after its onset in the feet. Occasionally, however, both upper and lower limbs are affected simultaneously; exceptionally the hands suffer first (Walton, 1977).

HMSN has two forms: Type I is autosomal dominant and a family history of thin legs and high arches is common. It may be seen in the adult or juvenile form (Agre and Matthews, 1996). The predominant histological findings in Type I are segmental demyelination and Schwann cell proliferation to form concentric arrays or ‘onion bulbs’ around demyelinated or partially remyelinated axons. Patients from these families may have palpably enlarged nerves (Chance and Pleasure, 1993).

Type II is x-linked recessive or autosomal recessive and also has two forms. Type II is approximately one third as common Type I. In Type II nerve condition velocities are normal or near normal, but motor and sensory potentials are reduced in amplitude. The predominant histological feature is Wallerian degeneration (Chance and Pleasure, 1993). The mild form is seen in 2 to 10 years old children whereas the severe form may be seen in children as small as 4 months old. Severe involvement is usually present within a decade from appearance of the first manifestations and ambulation may become compromised (Agre and Matthews, 1996). Fortunately the early onset form is relatively uncommon.

In HMSN muscle wasting frequently occurs bilaterally and sequentially first in the foot intrinsics, second the peroneals and thirdly in the anterior tibial muscle. The posterior tibial and the gastrosoleus are the last to atrophy (Sabir and Lyttle, 1984). Ultimately the feet may become flail with neither dorsiflexion nor
plantarflexion being possible. There may be a typical stork leg appearance. Contractures are usually slight in proportion to the degree of wasting (Walton, 1997; Dubowitz, 1995).

The tendon reflexes are variable. They are usually diminished or lost in the wasted muscles in proportion to the degree of wasting, but loss of tendon reflexes may precede atrophy. Loss of sensibility and slowing in nerve conduction velocity also varies. Sensibility may be unaffected but there is generally loss of vibration, light touch, pain, temperature and proprioception in the glove-stocking distribution (Walton, 1977; Alexander and Johnson 1987; Williams et al., 1994).

Life expectancy is normal except in some severe juvenile cases (Agre, 1996; Dubowitz, 1995). Abnormal responses to cold stress, Williams et al., 1994), impotence (Bird et al., 1994) and respiratory dysfunction (Carter et al., 1992) may be associated problems.

The initial complaints in HMSN may be general foot weakness and unsteady gait. Parents usually bring in their children because they cannot run and climb stairs like their peers and have frequent falls (Sabir and Lyttle, 1984). Actually curling or clawing of the toes may be the first manifestation but clawing and heightening of the longitudinal arch is rarely a matter of concern for the parents.

The cavus deformity is secondary to muscular imbalances acting on the mid and anterior tarsus and the midtarsal joint (Alexander and Johnson, 1989; Bushbacher, 1996). With atrophy of the intrinsic flexors and the unopposed action of the long extensors the metatarsophalangeal joints go into hyperextension. While the long toe flexors are active at this stage they are unable to flex the metatarsophalangeal joints. This intrinsic minus position of the toes causes obvious clawing which typically upsets the windlass mechanism of the foot structures and accentuates the cavus deformity (Mann, 1992).

Hypersensitivity and corn formation on the dorsal surface of the toes are common, although at this stage this is not a rigid deformity but a dynamic disfiguration and accommodation within the shoe is necessary for comfortable fit.

As the disease progresses, another typical manifestation is an uncontrolled slapping of the feet following heel strike (Bushbacher, 1996). Normally after heel strike the concentric contraction of the anterior tibial muscles controls the speed and magnitude of plantar flexion. As weakening of the tibialis anterior continues, concentric contraction is also affected and any form of heel strike becomes impossible.

The consequential drop foot is either compensated for by steppage gait, in other words with more than normal ranges of hip and knee flexion or causes dragging of the feet during swing (Dubowitz, 1995; Smidt, 1990). Sometimes the compensation for drop foot may be a marionette gait with pelvic elevation and pelvic shift to the swing side (Sabir and Lyttle, 1984).

Together with an ataxic gait due to lack of proprioception, drag of the feet will predispose the patient to frequent falls (Alexander and Johnson, 1987).

The active inadequacy causing drop foot may in time be replaced by a hindfoot equinus due to the unopposed action of the ankle plantar flexors.

In the upper limb, clawing of fingers is the typical deformity due to loss of the intrinsic hand muscles especially the lumbricals. The intrinsic minus position of the hands accompanying narrowing in the web space causes a loss in dexterity. The condition may be accentuated by insufficient proprioception (Bushbacher, 1996). The lack in dexterity is a major concern in school age children because it causes problems in handling pencils and crayons.

In progressive neurological conditions patients may also have a significant risk of acquiring a serious spinal deformity. The curves are difficult to control with bracing and progression does not cease with maturation (Hensinger and MacEwen, 1976).

In the severe form of HMSN, deformity will progress relentlessly.

Method and material

In the Orthotics Department of Hacettepe University in a period of eight years from 1992 to 2000, 55 paediatric patients with HMSN were treated 25 (or 45%) of the patients were girls and 30 (or 55%) were boys. The age at which symptoms first appeared was 5.30±4.02 years. The age at which they began to seek treatment for these symptoms was 9.82±4.89. The discrepancy in these dates is striking. The age of appearance of first symptoms is lower than that cited in the literature. This may be due to the fact
that upon having their child diagnosed as having HMSN the parents might have exaggerated their recollections of their child's motor impairment. The age at which they were able to contact appropriate medical assistance is in accordance with the literature.

At the time patients came to the department the symptoms were clawing toes in 3 (1.65%), pes cavus in 8 (4.4%), difficulty during elevation activities in 32 (17.6%), frequent falls in 15 (8.25%) deviations in the gait pattern such as loss of heel strike, slapping foot flat or steppage gait in 27 (14.85%), equinus in 14 (7.7%), atrophy of the hand muscles in 9 (4.95%) and two patients had become wheelchair bound and had severe scoliosis.

Orthotic management is an important feature in the rehabilitation of these children. The objectives of orthotic management can be defined as:
1. providing a safe, normal and pain-free gait;
2. retarding development of deformities and preserving a plantigrade foot;
3. accommodating rigid deformities and distributing weight-pressure evenly;
4. providing a gait free of excessive energy consumption;
5. maximising functional usage of the hand;
6. when scoliosis or kyphoscoliosis is a problem, bracing is done as a method of buying time; aiming at curve control until surgery becomes necessary but while there is still adequate vital capacity.

Although all 55 patients were referred with the same diagnosis of HMSN their symptoms and complaints were variable. Consequently the orthotic management of patients with HMSN is also variable.

In the mildest form, where clawing of the toes, discomfort and callus formation due to shear forces acting on the dorsal aspect of the PIP joint is the main problem the principle of treating the intrinsic minus hand is utilised.

In the hand of the patients with intrinsic muscle paralysis but functioning extrinsic extensors clawing may be prevented and active IP joint extension facilitated by passive MCP flexion. This is achieved by means of a MCP extension stop assembly, added to the basic opponens orthosis or a knuckle bender (Figs. 1 and 2) (Fishman et al., 1985). Similarly when the problem is hyperextension in the MTP joint a bar lying on the proximal phalanges of the feet will have the same effect of stopping clawing of the toes. Obviously such a bar has to be installed into the inner surface of the vamp of the shoe. Finding the exact location takes some time, but once it is established patients express instant relief (Fig. 3). In order for such a bar to be effective, besides a high toe box, the vamp of the shoe must be made of sturdy material. The authors use polyform about 1cm depth, the length and width cut to cover the first proximal digits of the toes.

For patients with an inclination toward pes cavus a lateral heel and sole wedge is employed. If the cavus has become rigid, the deformity is accommodated. An insert with medial longitudinal and transverse arch support will eliminate the gap between the sole and floor and equalise weight-bearing. If the cavus is accentuated a moulded inlay is utilised (Hsu et al., 1992).

In patients with drop foot a dorsiflexion strap made of leather and elastic strap can be very effective. Children prefer it because the elastic strap is sewn directly onto tennis shoes and the
other end goes through a tongue at the ankle and is buckled to a calf strap made of leather (Fig. 4). Due to this inexpensive and easy application elevation activities are facilitated and the distance walked without tiring is prolonged.

However, the leather calf strap is uncomfortable for some patients and the suspension may be inadequate. For such patients, the calf strap is replaced by a supramalleolar mould resembling the airstirrup and the anterior elastic strap is buckled onto a leather bridge between the upper ends of the airstirrup (Fig. 5).

Together with these assistive orthoses patients are given a rigid AFO supporting the feet in subtalar neutral and the ankle at 90° to be worn at night. Nevertheless, due to the unopposed action of the ankle plantar flexors, deformity may progress relentlessly and a polyethylene AFO is required for ambulation. The design of the AFO is determined by the extent of the deformity. The authors’ preference is a dynamic AFO with or without a plantar flexion stop. But when there is weakness in the plantar flexors as well as the dorsiflexors, ankle instability or gross deformity a fixed AFO is inevitable. Also in cases where the patient has undergone surgery such as a triple arthrodesis a fixed AFO is used.

It is rare that an orthosis other than an AFO is required for ambulation, but in some patients the progression is so abrupt that the child walking with AFOs all of a sudden requires HKAFOs and then becomes wheelchair bound within months. This was so in 2 of the 55 patients. In a survey on outpatient rehabilitation for chronic neuromuscular diseases 94% of the equipment cost was for ankle-foot orthoses and appropriate footwear, wheelchairs were only 2% of the equipment cost for HMSN (Koch et al., 1986).

Discussion

In the authors’ experience, one of the major mistakes made in the orthotic management of patients with neuromuscular disease is a tendency to overestimate the need for orthotic intervention. Other studies have also drawn attention to this danger (Thompson and Robb, 1995; Carmick, 1995).
For instance, both gait and function may be compromised when wearing fixed AFOs it should always be remembered that an equinus gait is able to compensate for weak knee extensors while fixed AFOs shift the ground reaction force posteriorly causing a reduction in knee extensor moment (Rose, 1986).

To counteract the risk of over-treating (bracing) the patient it is important to keep precise records, repeat timed and functional tests at regular intervals. Also manual muscle testing assists in prognosticating and determining when and which sort of bracing may be indicated (Allsop, 1989). The primary aim of rehabilitation in HMSN is maintenance of function since currently there is no known way to alter the progression of the disease.

It should always be remembered that orthotic management is an integral part of the overall rehabilitation programme which should also include stretching, strengthening and range of motion exercises and inspection of hypoaesthetic areas.

**Conclusion**

In conclusion, the orthotic management of HMSN in children requires a team approach consisting of the paediatric neurologist, the orthotist and the therapist. The functional level and exact requirements of the patients should be assessed because there is a wide range of interventions possible starting from a simple bar on the dorsal aspect of the proximal phalanges to bilateral HKAFOs.

It is important to counteract the risk of unnecessarily giving too much orthotic support.

**REFERENCES**


