

# Ballert

# ORTHOPEDIC

## ORTHOTICS AND PROSTHETICS

### ORTHOTIC TREATMENT OF THE SPINAL MUSCULAR ATROPHIES

By Gene Bernardoni, CO

#### Introduction

The Spinal Muscular Atrophies (SMA) comprise a group of hereditary disorders in which there is degeneration of the anterior horn cells of the spinal cord and the motor nuclei of some cranial nerves. Generally inherited as an autosomal recessive gene, the disease poses unique challenges for orthotic treatment.

In recent years, such treatment has been fine-tuned according to the severity of the condition and the age of onset. In this article, I describe an orthotic treatment for each of the four classifications of SMA, based on my 18 years of experience as an orthotist treating SMA patients. My recommendations are based on consultations with other orthotists, including the chief orthotist at Shriner's Children's Hospital and the director of orthotics at Louisiana State University. The resulting ambulation protocols are referred to in this article as Ballert ambulation protocols.

In general, the later the onset of symptoms, the better the prognosis, and the more orthotic alternatives may be effective in treatment.

#### Type 1 SMA: Acute Infantile Spinal Muscular Atrophy

Type 1 SMA is also known as Acute Werdnig-Hoffmann Disease or Severe SMA. The onset of the disease is usually "in utero" or during the first

few months after birth. The mother may notice that the normal kicking of the fetus becomes weak or disappears entirely, or that the child after birth is extremely limp and weak (*Fig. 1*). The symptoms are usually noticed within three to six months of birth.

Typically the child is hypotonic and weak, has poor head and neck control, and is unable to sit unsupported (*Fig. 2*). Bulbar involvement causes difficulty in sucking and swallowing, making feeding a difficult task. Additionally, severe limb and axial weakness causes the characteristic "frog leg posture." (*Fig. 3*.) The internal rotation of the arms of a supine child results in a "jug handle posture." Although the child has poor head control, the facial muscles are relatively unaffected, a fact that differentiates this disease from Infantile Myotonic Dystrophy or Infantile FSH Dystrophy. Diaphragmatic breathing in a supine child results in severe costal recession and a "bell-shaped" chest. (*Fig. 4*.) About half of these patients demonstrate fasciculations, the deep tendon reflexes are usually absent, and sensation is generally normal.

The pathogenesis is characterized by a sharp reduction in the number of axons present in the central nervous system. The inciting event has not been fully characterized, but is currently under intense investiga-

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Fig. 1.—Month-old boy showing poor head and trunk control.

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tion. In general, axons are unable to regenerate, resulting in a permanent loss of these vitally important neural cellular elements. When the axon dies, the remaining axons are asked to supply more muscle fibers through a process known as sprouting. This attempt at healing is incomplete. Even though the disease may be non-progressive, weakness generally increases as a result of growth, because the weakened muscles are asked to move larger and heavier limbs. (Fig. 5.)

Supportive care includes early tracheostomy, assisted ventilation, and G-tube placement to support respiration and nutrition.

Because they never develop truncal control that allows independent sitting, these children tend to collapse onto themselves when brought into a sitting position. This is characterized by a collapsing spinal deformity, either scoliosis, kyphosis, or a combination of these. This adversely affects the pulmonary and gastrointestinal functions.

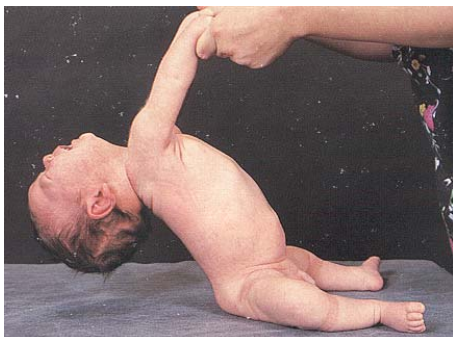


Fig. 2.—Month-old boy unable to sit unsupported and showing neck flexor weakness.

A supported upright sitting posture aids the respiratory and GI systems, so this must be provided by external means. An appropriate seating system is crucial. This may include a custom seating system within a wheelchair. Spinal orthotics may also be effective in providing this sitting support. Most of these patients do not survive long enough to develop clinically significant spinal deformities.

Orthotic treatment of these patients centers around providing sitting balance with such orthoses as a *lumbar corset* or a *soft thoracolumbosacral orthosis*. A hard TLSO is not well tolerated due to breathing constraints. However, a TLSO may be specially constructed with elastic panels or cutouts to optimize the ventilatory function.

#### Type 2 SMA: Chronic Infantile Spinal Muscular Atrophy

Type 2 SMA is also known as Chronic Werdnig-Hoffmann Disease or Intermediate SMA. This form of Infantile SMA is also autosomal recessive, has a later onset of symptoms and is usually noticed at six to twelve months of age. Unlike the acute form, the chronic form allows the child to sit unsupported but prevents him from being able to stand unsupported. Fasciculations often occur and the deep tendon reflexes are diminished. Truncal weakness, hypotonia, joint laxity, and respiratory weakness are less severe than in the acute form, but are marked nonetheless. The SMA child may have an advanced intellect that contrasts with Duchennes Muscular Dystrophy and Myotonic Dystrophy in which the patient may be mildly retarded.

Because these children may survive to late childhood or even early adulthood, the possibility is greater for developing deformities such as scoliosis and limb contractures. The orthotic management of the SMA Type 2 child centers around helping the



Fig. 3.—Typical “frog leg posture.”

child achieve normal developmental milestones. The early achievements of sitting and standing unassisted are beneficial from both a physiological and psychological standpoint. Physiologically, standing prevents ankle and hip flexion contractures and promotes respiratory and urological health, along with bone density and development.

The *caster cart* (Fig. 6) allows mobility in a sitting position; the *prone stander* (Fig. 7) allows graduated weight bearing; and the *A-Frame* (Fig. 8) allows the child to ambulate in parallel bars, so long as there is sufficient upper arm strength. The promotion of standing and ambulation at the appropriate time is of great importance in the treatment of the SMA Type 2 patient. Because of truncal and upper arm weakness, only the exceptional patient will be able to ambulate with

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Fig. 4.—Severe costal recession and bell-shaped chest.

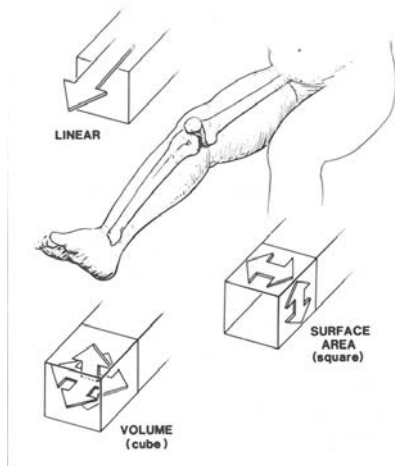


Fig. 5.—The scale effect.

either HKAFO's or KAFO's (Fig. 9).

The *reciprocating gait orthosis* (RGO) (Fig. 10) may be successful in only the strongest of these patients due to both muscle strength and increased motor ability. The patient must use a walker or forearm crutches (Fig. 11) to ambulate with the RGO, and because this requires upper arm strength (especially triceps muscles) very few of these patients will have success. Numerous attempts have been made to adapt the RGO for this use because it is so successful with the spina bifida patient for whom it was developed. Well-meaning caregivers sometimes attempt to give par-



Fig. 6.—Caster cart.

ent and child the expectation of upright ambulation with an RGO, but the results are usually disappointing for all concerned. It is important to set positive but achievable goals for the psychological well-being of both parent and child. The parents of SMA patients are asked to shoulder a great burden and yet be positive and encouraging to their child. Small, positive achievements on a day-to-day basis are needed to encourage both.

There are a number of significant differences between SMA Type 2 and spina bifida. The spina bifida patient has strong upper limbs and trunk, depending on the level of the spinal cord lesion, and therefore can initiate the trunk shift needed for ambulation with an RGO. In contrast, the SMA Type 2 patient, as he grows bigger, becomes weaker in these areas. Nor can he increase upper limb and trunk strength with exercise.

In addition to the psychological impact noted above, there is an economic aspect to consider. Both of these orthotic interventions are quite expensive. It is therefore advantageous to make the right choice the first time to avoid the financial burden of two expensive orthoses.

At Ballert we have developed a protocol for these patients based on a hybrid orthosis fabricated from parts of two different orthoses: the frame of a *parapodium* (Fig. 12A) and the duck feet of an *Orlau walker*. (Fig. 13.) We use a parapodium frame at the first signs of the patient wanting to stand or at a developmental age where standing is appropriate (12 to 24 months). The child is placed in the frame frequently throughout the course of each day and allowed to play with his toys at chair or low table. When the child begins to rock the frame from side to side and thus show interest in mobility we attach the duck feet from an *Orlau walker* to provide the child with mobility (24 to 30 months of age). (Fig. 14.)



Fig. 7.—Prone stander.

This hybrid orthosis may be fine-tuned to the patient's strengths. There should be close lateral contact in order to transmit any side-to-side motion to the duck feet and be converted to forward movement. A sloppy fit causes the patient to waste motion and tire more quickly. It is also imperative to adjust the anterior-posterior tilt by using wedges to facilitate forward lean. A neutral upright position allows both forward and backward movement, whereas a forward lean facilitates forward movement.

Recently at least one other firm has introduced a walker similar to the hybrid orthosis used in the Ballert protocol. This prefab walker does not lend itself to fine-tuning in order to get maximum benefit. Nor does it unlock

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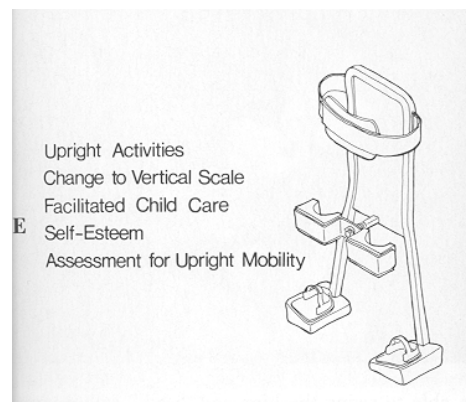


Fig. 8.—A-Frame.

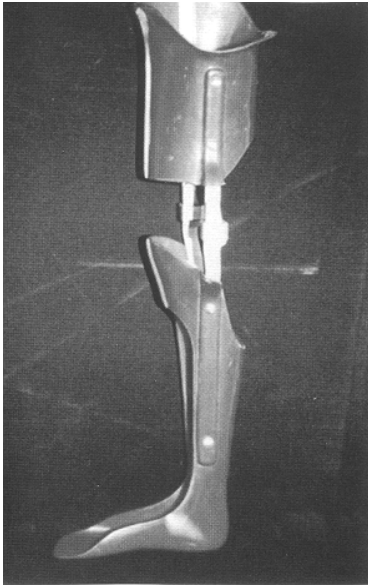


Fig. 9.—Tubular style KAFO.

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at the hips or knees, thereby preventing the child from sitting in the orthosis (Fig. 12B).

In my experience, the Ballert ambulation protocol has several advantages. First, it allows the child mobility with a minimum of side-to-side head and/or upper trunk motion. Second, no upper arm strength is needed for mobility. Third, the child has his hands free to play with his toys. And fourth, we have set achievable goals for psycho-

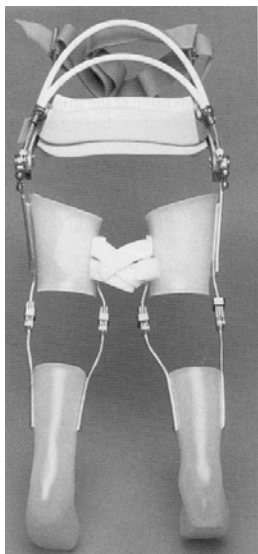


Fig. 10.—ISU Reciprocating Gait Orthosis — posterior view.

logical success. This approach does not eliminate the possible use of an RGO as the child outgrows this orthosis if he has the necessary motor ability and physical strength.

Another major area of orthotic intervention centers on the patient's truncal weakness when seated (Fig. 15). Because he is in a seated position for most of his waking hours, and because of his truncal weakness, the SMA Type 2 child has a high incidence of scoliosis. Growth spurts plus sitting and gravity promote the progression of scoliosis. It is consequently important to initiate TLSO orthotic management early if there is to be any chance of success. As with Duchennes patients, wheelchair sitting and inactivity promote obesity, which in turn creates a problem for TLSO management of scoliosis. We face other challenges when fitting this patient with a TLSO, including severe hypotonia, lumbar and thoracic kyphosis and compromised respiration. The flexion contractures of the knees due to tight hamstrings pull the pelvis into a posterior tilted position that initiates an adverse shift in the normal weight distribution of sitting from the posterior thighs and ischial tuberosities to the coccyx. This is known as sacral sitting, and is associated with pain as well as pressure ulceration. As a result, an obese patient tends to slip out of the TLSO unless the lumbar kyphosis is restored in the fabrication. The pelvic



Fig. 12. — Parapodium. Frame (A) upright position and (B) sitting position.



Fig. 11.—Ambulation with an RGO assisted by forearm crutches.

tilt and lumbar lordosis must be built into the orthosis to encourage as near to normal sagittal plane alignment as possible, even if the patient's legs are pulled under the seat of the wheelchair slightly.

The respiratory difficulty posed by a tight-fitting TLSO for scoliosis must be addressed. Because these children have very low functioning abdominal muscles, the diaphragm is compromised biomechanically. The abdominal viscera have no place to go when the patient inhales using his diaphragm because the TLSO acts as a

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Fig. 13.—Orlau walker combined with parapodium frame.

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mechanical barrier. The child is therefore unable to use his diaphragm, and must use the less effective accessory muscles such as the sternocleidomastoids, the scalenes, and the trapezius muscles to breathe. This becomes evident within 5 to 10 minutes after application of the TLSO by watching the child use his neck and shoulder musculature to breathe.

These respiratory problems may be addressed by using cutouts and/or elastic panels in the TLSO. The best cutout appears to be a circular opening. The superior edge of this opening should be 12 to 24 mm in-



Fig. 15.—These two brothers had SMA of similar severity.

ferior to the xyphoid and the inferior edge should be 24 to 36 mm superior to the symphysis pubis. This cutout allows the diaphragm to displace the viscera during inhalation, and thereby makes breathing easier when wearing the TLSO. The scoliosis is still controlled by its lateral walls acting through the rib cage. Sitting balance is addressed by the high profile design. It is usually helpful to add an elastic abdominal binder (Fig. 16) or a wrap of 6-inch elastic bandage to provide some abdominal control if the abdomen protrudes excessively through the opening. (Fig. 17.) This elastic wrap may be over or under the TLSO opening. It is best, however, not to attach it to the TLSO so it can be changed periodically. As in the children with traumatic spinal cord injury, we believe it is beneficial to initiate TLSO orthotics early to have any chance of stopping or slowing the progression of scoliosis. At this juncture surgery is likely the best option, and TLSO usage is intended as a holding strategy until an optimal age for surgical intervention. Surgery can also be considered earlier if the scoliotic curve is not controllable by orthotic means. The standard surgical treatment is a posterior spinal fusion T2 to the pelvis with spinal implants designed for children weighing between 30 and 60 lbs. Luque-Galveston constructs are typically utilized. Proximal fixation may include screws or hooks. Post-operative TLSOs are required to protect the spinal implant-bone interface during the 6-9 month healing process. It is crucial that the family and the child understand that tracheostomy and ventilatory support may be required, if a surgical course is chosen.

If the patient has missed the opportune time for surgery because of diminished respiratory capacity, TLSO treatment is the only option, but a challenging one. These children have multiple other orthopedic problems such as tight hamstrings,



Duck feet

Fig. 14.—Swivel walker, ORLAU version.

posterior pelvic tilt, lumbar kyphosis, obesity, and decubitus ulcers. These other issues may complicate the management of the spine. These children may be candidates for a modified type of suspension TLSO (see Fig. 20).

### SMA Type 3: Juvenile Spinal Muscular Atrophy

SMA Type 3, also known as Kugelberg-Welander Disease or Mild SMA, is an autosomal recessive inherited disease. It has an onset of one to 15 years of age and is either non-progressive or very slowly progressive. The patient is usually wheelchair bound by 20 to 30 years of age with an almost normal life expectancy.

Type 3 patients differ from the previous two types of SMA in that they are able to stand and walk but have difficulty in running and climbing stairs. Other clinical signs include a wide-based flat foot or waddling gait, Gowers' Tripod Sign (Fig. F18), legs usually weaker than arms, proximal muscles weaker than distal muscles, tongue fasciculations, pseudohypertrophy of calves or thigh atrophy, respiratory weakness and weakness of neck flexors. (Fig. 2.)

The management of these patients includes vigorous treatment of res-

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piratory infections and encourages activity and ambulation. Orthotic intervention is therefore indicated to aid in ambulation and prevent progression of scoliosis and thereby reduce the risk of respiratory compromise.

Because Type 3 SMA is a "flaccid" type neuromuscular disease, continued observation for the onset of scoliosis is critically important. The potential progression of scoliosis in neuromuscular disease of this type is chiefly due to the wheelchair confinement and pull of gravity, coupled with periods of rapid growth. Unlike idiopathic scoliosis, the progression of scoliosis is possible even after skeletal maturity is reached with neuromuscular scoliosis. Because the SMA Type 3 patient has a greater life expectancy and a longer period of wheelchair confinement, the need for TLSO's and custom-molded seating systems is greater. Like their SMA Type 2 counterparts, the SMA Type 3 patients typically exhibit severe hypotonia, lumbar and thoracic kyphosis, tight hamstrings, posterior pelvic tilt, decubitus ulcers, obesity, and respiratory compromise and may either have a tracheotomy or a respirator. If they are poor surgical candidates, their scoliosis usually requires orthotic treatment. Various TLSO designs may be used, but a *neuromuscular* or *total contact TLSO* design is usually the best choice. The *neuromuscular TLSO* utilizes bubbles instead of cutouts, thereby reducing the risk of skin breakdown at the

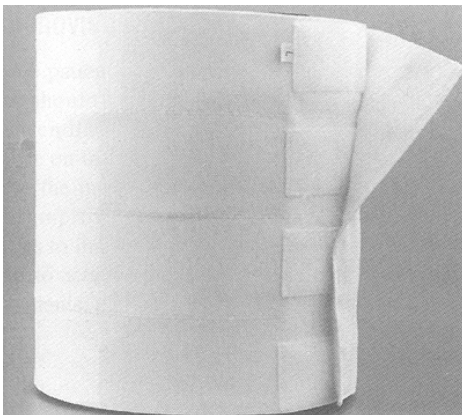


Fig. 16.—Elastic abdominal binder

edges of the cutouts. It can be bivalved or have an anterior opening. The *bivalved TLSO* (Fig. 19) lends itself to modifications such as abdominal or thoracic cutouts for breathing. Equally important is that it is also easier for parents to apply as the child gets older and heavier.

The older suspension TLSO for relief of pelvic obliquity is rarely used due to the difficulties of wearing such a TLSO. However, I have made a modified version of the *suspension TLSO* which can be used to relieve pressure over decubitus ulcers due to pelvic obliquity and to correct for decompensation by posting one side of the posterior shell (Fig. 20). We have on occasion fabricated this modified type of TLSO which provides a degree of unweighting by allowing the longer posterior shell to come into contact with the seat of the chair. This has been used with older children who are not surgical candidates. Because such a TLSO will cause the patient to pitch forward, it is necessary to change the back-to-seat angle of the wheelchair by increasing it by 10 to 15 degrees. Such positioning will accommodate the TLSO and direct the weight line behind the TLSO, thus preventing the patient from falling forward. The pressure can be taken at the axillae and the inferior costal margins.

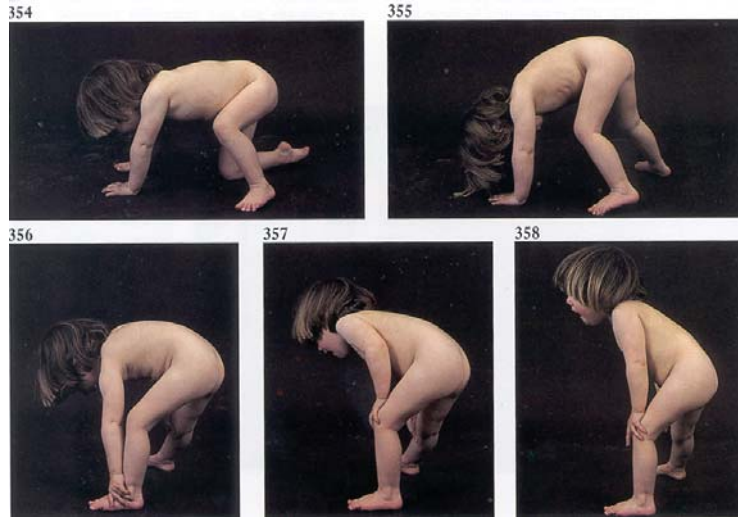


Fig. 18.—This 2 1/2-year-old girl displays Gowers' sign getting up from the floor.

Such treatment aims not to correct their curves but rather to attempt to prevent their progression.

Another variation is a "bucket style" TLSO, which can double as a car seat and a seating system. Such a TLSO is fabricated in a seating position and encompasses both the torso and the upper thighs. It is important to ventilate this TLSO well, especially around the pelvis and peroneal area. This type of TLSO is usually appropriate for SMA Type 2 patients.

The SMA Type 3 patient is better able to ambulate and ambulates longer than the two previous types of SMA. He will consequently utilize both AFO's and KAFO's. The success of these orthoses will depend on trunk strength. The SMA Type 3 pa-

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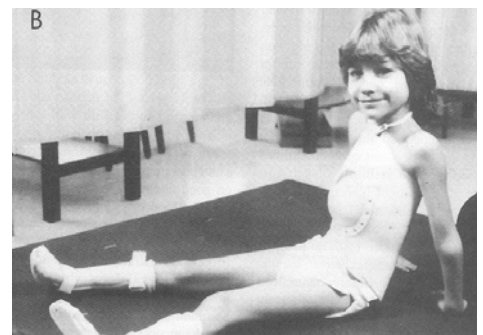


Fig. 17.—Long sitting with body jacket support.

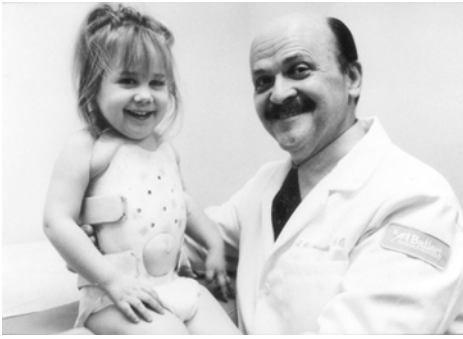


Fig. 19.—Bivalved TLSO

tient uses trunk sway and locked knees to ambulate in the same way as the Duchennes patient. The use of a TLSO when the patient ambulates is therefore probably contraindicated, although it may be needed to provide sitting balance and prevent progression of scoliosis. The patient and caregiver should be taught by physical therapy to perform daily stretching to maintain range of motion, and encouraged to ambulate to maintain available muscles. Ambulation may require the use of *supramalleolar orthoses* (SMO, Fig. 22), *floor reaction AFO's* (Fig. 21), *tone-balancing orthosis* (TBO) (Fig. 22) or *tubular style KAFO's* (Fig. 9), depending on the patient's strength. The SMA Type 3 patient is also more likely to be able to use RGO's (see discussion in SMA Type 2). At

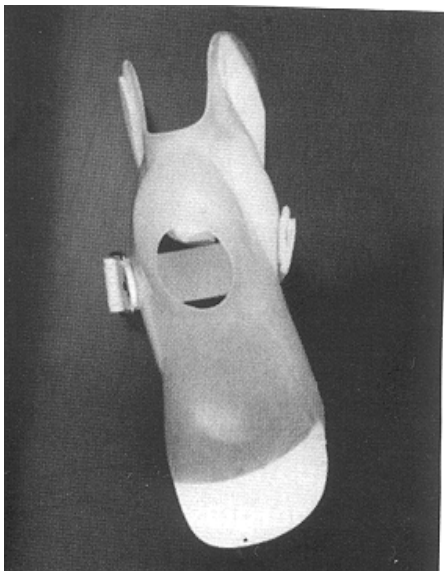


Fig. 22.—Tone-balancing orthosis.

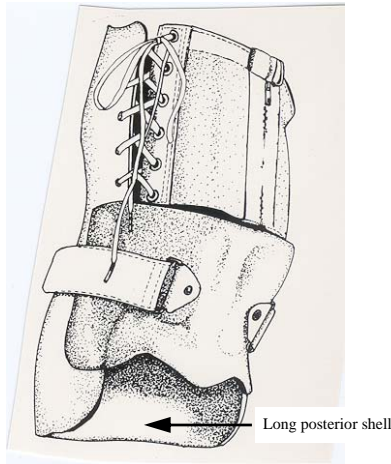


Fig. 20.—Modified version of a suspension TLSO.

least three different types of RGO's are available on the market today. They are the *Steeper*, the *Double or Single Cable Type* and the *Isocentric Type* (Fig. 23). The successful use of an RGO will depend on the available upper arm and trunk strength.

These patients may require *cervical orthoses* of the supportive type for riding in busses or cars. Usually non-custom cervical orthoses such as a *Miami J collar* will suffice. The child who is wheelchair-bound may utilize a headrest attached to the wheelchair and bound with a forehead strap. A custom-molded CTO may be another option. (Fig. 24). This can be used in the car seat and other chairs or seats.



Fig. 23.—Reciprocating Gait Orthosis (RGO)



Fig. 21.—Floor reaction AFO.

#### SMA Type 4: Adult Spinal Muscular Atrophy

This is the mildest form of SMA and is a welcome diagnosis for the patient who was suspected as having ALS. It has an onset of 18 to 50 years of age and is non-progressive or very slowly progressive.

The life expectancy is normal. Some of the clinical signs are mild weakness of hands and feet that may involve other muscles of the body, cervical spondylolisthesis, and weakness of the tongue. The patient is encouraged to do exercise and normal activities. Conventional AFO's and *floor reaction AFO's* are used to assist in ambulation.

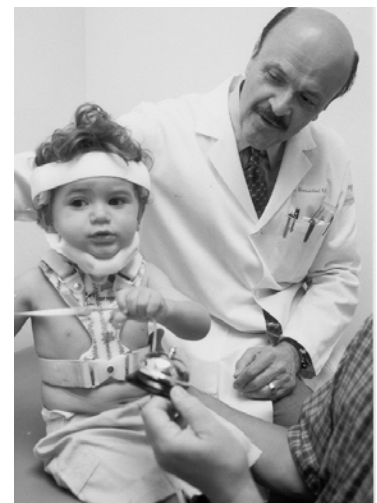


Fig. 24.—Cervical thoracic orthosis (CTO).

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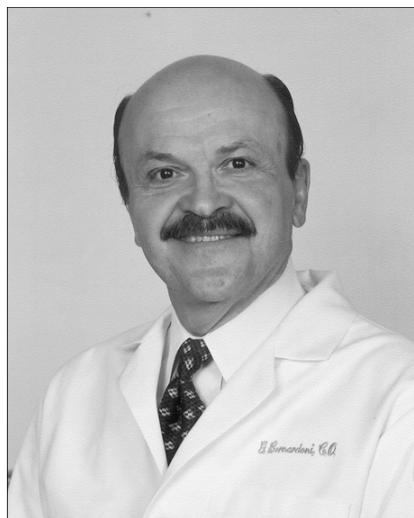
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Bernardoni is on the auxiliary staff at Northwestern University's Orthotics and Prosthetics Program (NUPOC) and has lectured widely in orthotic applications.

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