

A Note to Physical, Occupational and Speech Therapists

Treating Children with Hurler Syndrome

Because Hurler syndrome is such a rare disease, we have provided some basic information to assist you in planning your therapy sessions. If you have any questions about Hurler syndrome or the child's condition, please contact the child's doctor.

About Hurler syndrome

Hurler syndrome is an inherited metabolic storage disease that results from deficiency of a single lysosomal enzyme. Because of this enzyme deficiency, there is a systemic accumulation of the enzyme substrate (glycosaminoglycans) in the body. This affects the functions of the heart, lungs, liver and spleen. It also causes multiple skeletal anomalies, including those in the skull, cervical and thoracic spine, shoulders, hips, knees, rib cage and wrists. Children with Hurler syndrome are often affected by hydrocephalus, decreased vision from corneal clouding and some amount of hearing loss.

Because the affected organ systems and orthopedic anomalies influence development, these children will have delays in mobility, gross motor skills, fine motor skills, speech and language. There is also a strong potential for vision and hearing deficits. Intensive rehabilitation services that include physical therapy, occupational therapy, and speech and language pathology are essential for these children from the time of diagnosis through their course of medical treatment and throughout early childhood. Children affected by Hurler syndrome face extra challenges in learning independent mobility skills, self-care skills

(feeding, bathing, dressing) and speech and language skills. Each of these delays will later affect school readiness.

Children who receive a blood or marrow transplant (BMT) may face neurologic changes, graft-versus-host disease and other complications related to BMT. Because of immunosuppression, these children are at greater risk for common illnesses and infections. Low blood counts during the recovery period may place them at greater risk for muscle tears and bruising. As certain medications are tapered, children may have mood swings and changes in body fluid retention. It is important for the treating therapist to be aware of these possible BMT-related complications.

Precautions

Before setting up a physical, occupational or speech therapy program, consider the following.

Children with Hurler syndrome have potential cardiac and respiratory issues. These are disease-related and may affect activity levels.

Avoid vigorous joint-stretching programs at the risk of causing joint injury. Infants and toddlers with Hurler syndrome have true joint anomalies and rarely have joint contractures. Joint flexibility should be maintained by gentle range-of-motion exercises as well as gross motor activities that include transitional movements, bending, squatting, reaching and climbing.

Children with Hurler syndrome have cervical spine anomalies. Take care to avoid activities and contact sports that might place stress on the cervical spine.

Physical therapy

Gross motor skills

All gross motor skills are affected by decreased joint motion in the neck, spine, shoulders and hips. Typically a gibbus (spinal kyphosis) decreases thoracic extension and spinal rotation; shoulder flexion is limited to 90 to 100 degrees; the hips do not fully extend; and often there is limited range of motion in the wrists, fingers and ankles. This lack of joint flexibility hinders the child's ability to fully extend or rotate the affected joints and makes transitional movements (to sit, to four-point and to stand) much more difficult. Lack of trunk rotation makes it difficult to move from a supine to sitting position. Lack of spinal extension and decreased shoulder flexion make crawling almost impossible and the pull-to-stand from the floor very difficult.

One challenge commonly faced by the PT is a child who has good sitting stability, the ability to stand and the ability to walk, but who cannot transition between these positions independently. Other affected skills include running, jumping, walking up stairs, climbing onto furniture or stairs, squatting in play, squatting to retrieve a dropped object and propelling oneself on a ride-on toy or tricycle.

Goals of PT

The physical therapy program will include maintenance of joint range of motion, progression of gross motor skills with an emphasis on transitional skills and mobility, and the provision of orthotics or gait aids as needed.

In the infant and early childhood years, the physical therapy program should be intensive to maximize the child's potential. Some children may benefit from an adapted stroller or wheelchair for distance mobility.

Home programs for parents and coordination with school services will be essential. (Home program or discharge recommendations specific to this child will be included in the Home Program section of this binder.)

Typically, as children with Hurler syndrome grow older, they will have corrective spinal surgery, hip and knee surgery and carpal tunnel surgery. Physical therapy may be needed post-surgery for balance, strengthening and mobility.

Occupational therapy

Fine motor skills

Children with Hurler syndrome typically present with mild finger flexion contractures and decreased range of motion in wrist movement and supination. Fine motor skills are generally not as significantly affected as gross motor skills, because the joint limitations relate to functional positions of the hands. In infants and toddlers, gross motor skills and language skills are often more delayed than fine motor skills.

Range of motion

Due to a thickening of their connective tissue, children with Hurler syndrome typically present with decreased range of motion in shoulder flexion and abduction. Children compensate for the decrease in shoulder range by laterally flexing their spines, which allows for functional reach.

Range of motion should be addressed by stabilizing the scapula and ranging the shoulder, so the shoulder musculature is not overstretched. The long-term goal for these children is not to achieve full range of motion in the upper extremities, but to maintain functional range of motion.

Goals of OT

Occupational therapy programs should include maintenance of joint range of motion, progression of fine motor skills, age-appropriate activities of daily living and continuous assessment of the need for adaptive equipment. In the infant and early childhood years, occupational therapy should be intensive to maximize the child's potential.

Orthopedic complications

Children with Hurler syndrome generally require orthopedic surgeries, including carpal tunnel releases as well as spine, hip and knee surgeries. Occupational therapy will be involved post–carpal tunnel surgery, as most children are casted after this surgery. After spine, hip and knee surgeries, occupational therapy should assess the child's positioning (in a bed, wheelchair, stroller, car seat). Because the child's mobility will be more limited after surgery, the OT can help the child maximize function with adaptive equipment in order to maintain independence with activities of daily living.

Speech and language pathology

Language

During the first year of life, most children with Hurler syndrome will develop speech and language skills on time unless they have hydrocephalus or problems that compromise oxygen supply to the brain (like sleep apnea). After the first year, development begins to slow. These children will continue to add new speech and language skills, but at a slower rate than other children their age. By age 2, most children with Hurler syndrome are noticeably delayed. By 3 years of age, most are significantly impaired.

Children with Hurler syndrome have greater difficulty with speech and language development than with other abilities. They have more trouble expressing themselves than understanding what is said to them,

even when they have hearing loss. The reason for the speech and language delay is not entirely understood, though the question is currently under study. Additionally, children with Hurler syndrome have fine and gross motor impairments that limit their ability to point or use gestures.

Sometimes parents overlook speech and language problems because their child's physical problems are so severe. It is important to address speech and language problems as early as possible to prevent further delays. The first two years of life are most important in the development of a child's language skills.

Hearing

Hearing problems are common in children with Hurler syndrome. Some are caused by fluid in the middle ear, necessitating PE (pressure equalization) tubes; others are caused by problems with auditory nerves. Although hearing problems are not the sole cause of language problems, hearing loss does play a role in delayed language development.

Mental development

A blood or marrow transplant will prevent or slow the deterioration of the child's mental development. It will not reverse delays that have already occurred. Children generally have a slightly lower level of mental function after transplant, but they continue to develop new skills, and hearing generally improves.

Both before and after transplant, intense speech and language therapy can make a significant difference in the child's cognitive and language development. Children who have daily intervention do better overall than children with once-a-week therapy or no therapy. Parental involvement is also extremely important, not only as an adjunct to speech and language therapy, but also as a source of language stimulation for the child. Children with Hurler syndrome respond positively to a language-rich environment.

Oral motor skills

In addition to a delay in communication skills and a possible decrease in cognitive skills, the speech and language pathologist must address feeding and swallowing problems. Children with Hurler syndrome have large tongues that often have poor range of motion. Due to their specific facial morphology, they also tend to have limited jaw range of motion and weak lip closure. Their narrow airways and increased risk for cardiac anomalies may also impact feeding development or oral feeding recovery after an acute illness. These children may have a short- or long-term need for a feeding tube, because nutrition is critical to sustaining a blood or marrow transplant.

Swallowing problems are not directly related to Hurler syndrome; however, weak lip closure and limited jaw and tongue movement can cause delayed motor skills affecting both feeding and speech.

Goals of speech and language therapy

The goal of speech therapy is to stimulate speech and language by addressing the development of oral motor, cognitive, play and social skills using one-to-one play in a semi-structured setting.

If the child does not speak, the therapist should work on imitating sounds, as well as exploring and developing alternative communication systems (such as gestures, American Sign Language or picture symbols). The therapist must also coordinate with the OT regarding fine motor impairments if these impairments limit progress. The therapist needs to increase the child's understanding of spoken language (receptive language skills).

The therapist must also monitor the child for signs of a change in hearing ability. It is extremely important that children with Hurler syndrome have their hearing checked regularly by an audiologist. If they have hearing loss, it must not be ignored. Treatment may include PE tubes or hearing aids.

The child's brain development depends on the kind and amount of stimulation provided. As the child grows older, the brain is less adaptable and the positive effects of stimulation are less dramatic. Therefore, speech and language therapy should be especially intensive from birth to age 3, when children are in their most sensitive period of brain development. This will help offset delays and counteract the effects of the disease and transplant. Intense language stimulation is needed both before and after transplant to support normal development and to treat children who are delayed.

Sample Letter for Insurance Companies

A note for the treating therapist: You may use the following information as you prepare your letter to the insurance company requesting coverage. Please add the child's personal information to this letter, as well as his or her treatment plan and goals. Because Hurler syndrome is not common, it may be helpful to include additional information about the condition.

Dear third-party payer,

Hurler syndrome is an inherited metabolic storage disease that results from deficiency of a single lysosomal enzyme. Because of this enzyme deficiency, there is a systemic accumulation of the enzyme substrate (glycosaminoglycans) in the body. This affects the functions of the heart, lungs, liver and spleen. It also causes multiple skeletal anomalies, including anomalies in the skull, cervical and thoracic spine, shoulders, hips, knees, rib cage and wrists. Children with Hurler syndrome are often affected by hydrocephalus, decreased vision from corneal clouding, and some amount of hearing loss.

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Due to the multiple organ involvement and orthopedic anomalies, rehabilitation requires the skills of a physical therapist, an occupational therapist, and a speech and language pathologist. The patients—infants and children—are at the stage in their lives when the accumulation of language, motor skills and learning is most rapid. For this reason, rehabilitation services must be intensive. As the children grow older and are able to access services provided by the school system, a combination of medically based and educationally based services would be appropriate.

Please include the following information:

- *The child's functional status/progress toward goals*
- *The child's ongoing therapy goals*
- *Recommendations*

Sincerely,

Treating Therapist