PHYSICAL THERAPY FOR CHILDREN WITH MPS AND ML

Stacey C. Dusing, PT, MS
Angela Rosenberg, DrPH, PT
Sadye Paez, PT, MS
Wendy Westphal, MPT
Reviewed and updated by Nicole Needles, PT, DPT (2021)

Children diagnosed with MPS (mucopolysaccharidosis) or ML (mucolipidosis) have a wide range of bone, joint, and muscle impairments secondary to accumulation of mucopolysaccharides in the connective tissue which contribute to decreased motor abilities. The type of motor impairment and the degree to which it occurs is specific to the child's diagnosed disease. In addition, the rate of progression varies significantly depending on the diagnosed MPS or ML disease and the availability of medical treatments such as Hematopoietic Stem Cell Transplantation (HSCT) and Enzyme Replacement Therapy (ERT) (Kakkis et al.). The purpose of this fact sheet is to provide general guidelines for physical therapists that can be individualized to each child's specific needs.

Children with MPS and ML have multi-system medical problems that may impact physical therapy examinations and interventions. Physical therapists should be in contact with the child's primary care doctors and specialists to identify parameters for exercise and report any problems or changes observed during the course of therapy. Physical therapists should be aware of safety precautions specific to children with MPS or ML. Respiratory compromise may result from airway obstruction due to enlarged adenoids, tonsils and narrowing of the trachea as well as changes in the shape, size and extensibility of the rib cage. Cardiac compromise may occur resulting in decreased endurance and tolerance to exercise (Sifuentes et al.). Given these precautions, heart and respiratory rate, as well as work of breathing, should be monitored in children with MPS or ML throughout examinations and interventions. In addition, selecting appropriate objective tests and measures will guide the plan of care and goal writing.

Hydrocephalus can also be an onset complication of MPS or ML or may develop as the disorder progresses possibly requiring shunt placement. Signs of hydrocephalus or shunt malfunction include: nausea, vomiting, irritability, changes in tone, and sun setting eyes. Children with MPS or ML that have significant skeletal compromise, (MPS I, II, IV, or ML) may also be at risk of atlanto-axial subluxation, spinal cord compression, scoliosis, gibus deformity, and in some cases spinal cord instability thus putting child at risk for myelopathy or quadraparesis. Other skeletal problems that facilitate the need for precaution include carpal tunnel syndrome, hip dysplasia, hip dislocations, coxa valga, and knee flexion contractures. These children need to be identified prior to initiation of

therapy and precautions observed during therapy, in addition to yearly x-rays and nerve conduction studies as recommended by the primary care or orthopedic physicians.

Children diagnosed with MPS or ML should be referred to a physical therapist as soon as possible after diagnosis. The physical therapist must conduct a detailed review of the child's medical history and review medical records prior to the initial evaluation. Obtaining copies of all pertinent medical records, including knowledge of if the child had HSCT or ERT, prior to the initial evaluation will improve the accuracy of the history and documenting safety precautions that must be observed. The physical therapy examination should include a comprehensive assessment of gross and fine motor abilities using standardized measurement tools in addition to passive joint range of motion, strength testing, joint mobility assessment, endurance testing, and in some cases cardio pulmonary assessment.

Children with MPS or ML may have discrepancies in their gross and fine motor abilities warranting the use of assessment tools that have distinct fine and gross motor scales. Standardized measures are beneficial for documenting changes in motor abilities over time or (a lack of regression). Processes should be in place to assess for skill maintenance, especially for children who have not received HSCT or ERT. In addition to standardized measures, parents and children should be included as much as possible to obtain supplementary information regarding the child's functional abilities. If available a quality of life tool may offer valuable insight into the family's present situation as well as their future goals.

Parents and children with MPS or ML are an integral part of the evaluation process and provide the foundation for functional goals and intervention/treatment strategies. Children with MPS and ML may benefit from a variety of treatment options based on the impairments, activity limitations and precautions identified in the examination. Gait training, static and dynamic balance, and activities of daily living should all be addressed in a functional context. Functional skills such as moving between positions or completing age-appropriate daily routines should be emphasized. Strengthening activities are especially important for children who have had a HSCT as multiple medications may contribute to a decline in strength needed for functional activities. Children with decreased chest expansion may benefit from retraining of the inspiratory muscles to decrease episodes of dyspnea (Savci et al.). In addition, breathing activities should encourage diaphragmatic breathing during activities of daily living and functional mobility. One idea of an age-appropriate functional cardio activity is a bicycle; this may need to be adapted or modified to ensure the child is safe.

In addition to approaching therapy through functional skills it is also important to address the impairments present. Thus it is important to encourage prolonged muscular and body stretching in a functional context multiple times each day to preserve range of motion. Examples include long sitting, figure-four sitting, or wedge standing while watching TV or reading to address hamstring and gastroc flexibility. Joint mobilization may be beneficial in improving ROM in adjunct to stretching and strengthening in the

absence of any contraindications. Applying splints for nightwear or during naptime may also be effective at maintaining or regaining range of motion.

Physical therapists may also need to assess the child's need for orthotics and/or braces and make the appropriate referral. In general, active joint motions must be restricted as little as possible while maintaining proper lower extremity alignment. Repeated range of motion over the course of the day is beneficial to maintaining joint motion and should not be restricted unless it is contributing to poor alignment. Children should not be forced into a brace for daily use that does not accommodate their current joint deformity and available range of motion. Thus having a child wear a solid ankle foot orthosis that is set at neutral when their ankle cannot be dorsiflexed to neutral will only damage their foot rather than improve their ankle range of motion.

Aquatic therapy is another way to offer functional stretching, strengthening, balance training, and gait training in a gravity-reduced environment. Also, as therapy pools are warm, this water temperature can allow for capitalizing on stretching to increase joint flexibility. Depending on water depth the water's resistance can also aid in strengthening both inspiratory and expiratory musculature.

Children who have more involved needs may need to be assessed for adaptive equipment such as wheelchairs, special car seats, bath chairs, hand splints, adaptive eating utensils, etc. It is recommended that the physical therapist consider working with other professionals such as occupational therapists, speech therapists, augmentative communication specialists, and psychologists to evaluate children with MPS or ML. Evaluating children with MPS or ML with an interdisciplinary team approach will maximize the skills of all participating therapists and ensure the child receives all needed services.

As with intervention strategies, the frequency of physical therapy intervention should be individualized. As a general guide, children who are being treated to maintain skills and prevent the regression of skills may benefit from 1 to 2 times per week outpatient physical therapy with a function-based home program the family can complete on a daily basis. In contrast, children who have received a HSCT or are receiving ERT may benefit from more intensive therapy to capitalize on their potential for skeletal changes and improving motor patterns. Likewise, if a child has undergone any surgeries secondary to orthopedic issues, such as hip surgery related to hip dysplasia, there is benefit to both prehab and post-operative PT. Prehab is skilled PT offered for a few weeks prior to a child's pending surgery and has anecdotally been shown to speed up post-operative care. Post-op PT should be offered at a higher frequency.

The goal of physical therapy for children with MPS and ML is to maximize the children's functional abilities. Physical therapy will not cure MPS or ML and will not stop the progression of the disease. However, physical therapy can be beneficial to limit the impact that changes in the body structures have on functional abilities. More importantly, physical therapy can facilitate independence and participation in age

appropriate activities as well as provide the parents education on the disease and its impact on the child's function from changes in the musculoskeletal system.

In summary, physical therapy is a vital component of the care plan for children with MPS and ML. Physical therapists must be aware of safety precautions specific to each child and be in frequent communication with the child's medical team, conduct standardized and functional assessments, and develop appropriate treatment strategies to meet functional goals in collaboration with the child and family.

References

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