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CdLS and Physical Therapy

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What is physical therapy? How can it help my child with Cornelia de Lange Syndrome (CdLS)? When should my child start physical therapy? Where do I find a qualified physical therapist? These are among the many questions I receive from parents. This article explores answers to these questions and offers suggestions for parents to help their children get the greatest benefit from physical therapy, as well as guide professionals who work with individuals who have CdLS.

What is Physical Therapy?

Physical therapy is a skilled service provided by a licensed physical therapist (PT) or physical therapist assistant (PTA). Physical therapy helps a person, child or adult, to achieve their optimal physical function and independence. It focuses on improving gross motor abilities (such as rolling, crawling, sitting, standing, walking, and moving from one position to another) as well as strengthening weak muscles, stretching tight muscles, improving endurance and balance. Physical therapists may recommend adaptive equipment, assistive devices, and/or orthotics to help achieve these goals. Additionally, physical therapists also work to prevent the loss of movement following illness, injury, or surgery.

Common Physical Therapy Issues in Children with Cornelia de Lange Syndrome

Children with CdLS can be born with orthopedic problems which may include small hands, clinodactyly (incurved fifth finger), a proximally placed thumb, limited elbow movement, webbing of the toes, missing or shortened limbs, and/or a club foot. While most of these orthopedic conditions are not treated directly by physical therapy, the therapist must be familiar with them to adapt treatment sessions as necessary. A club foot may necessitate serial splinting, which might be performed by a physical therapist.

A very common characteristic of CdLS is developmental delay. As children with CdLS grow, they may develop other orthopedic conditions as the result of delayed development. Developmental conditions commonly seen include tight heelcords, tight hamstrings, hallux valgus (bunions), and arthritis. Children with CdLS typically have low muscle tone (hypotonia) which also impacts development. Occasionally, children with CdLS develop hip dislocation or Legg-Calve-Perthes Disease, for which they may be referred to physical therapy. Physical therapists work with children to treat developmental conditions, whether orthopedic or neurological, as well as to promote development of gross motor skills.

When working with a child who has CdLS, a physical therapist will first perform an evaluation. The purpose of the evaluation is to determine each child's existing issues as well as potential problems that might arise in the future. Once the evaluation is complete, the therapist will develop an individualized treatment plan. The treatment plan may include developmental activities, therapeutic exercises, balance and coordination exercises, gait



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training with/without assistive devices, aquatic therapy, hippotherapy, and recommendation of equipment such as orthotics, braces, and assistive devices. The treatment program should include parent education and a home program, which will be discussed in further detail later in this article.

Specific information about common conditions is outlined below:

Tight heelcords: Tight heelcords result when the gastrocnemius muscle (located in the back of the leg running from behind the knee to the bottom of the foot) is shortened. When the gastrocnemius is not tight, it allows the ankle joint to bend beyond a 90 degree angle when the knee is straight, allowing for a normal walking pattern with a heel to toe pattern. When the heelcord (or gastrocnemius) is tight, this cannot happen. As a result, the child will often walk on his or her toes, or will place the entire foot flat with each step rather than using a mature heel-to-toe pattern. Toe walking may be the result of tight heelcords, but may also be the cause. If a child walks on his or her toes all the time (often due to sensory reasons such as a child not wanting the bottom of his foot touching the floor), then the gastrocnemius is not able to lengthen properly, and over time the muscles will become short. Treatment of tight heelcords should include a stretching program (both in physical therapy and at home), teaching the child to use the longer muscle once more motion is gained, and may include recommendation of ankle foot orthoses (either for use during the day or at night while sleeping). The child's orthopedist may recommend Botox as a treatment. This is effective if the child has the motion but is not using it, combined with intense physical therapy. In my experience, when a child is given Botox but does not have the available range of motion (or close to it), does not wear an ankle foot orthoses after the Botox treatment, or does not have intense physical therapy, then the Botox makes little difference. If these treatments do not work, then surgery to lengthen the heelcord may be recommended by the orthopedist.

Tight hamstrings: Tight hamstrings result when the muscle running along the back of the leg from the buttocks to behind the knee becomes tight. This may be due to the child's growth (especially a sudden spurt) and/or due to spending much of the time in a seated position (especially with knees bent). As a result, knee flexion contractures may develop. Treatment for tight hamstrings includes a stretching program (both in PT and at home), teaching the child to use the hamstrings in a longer position, and may include gait training and/or a standing program.

Hallux valgus: Hallux valgus is also known as bunions. If this condition is not painful to the child, it is typically just observed by the orthopedist. Otherwise, orthotics are recommended. In severe cases, the orthopedist may recommend surgery in order to correct the alignment of the foot and toes.

Hypotonia: Hypotonia means a child has low muscle tone. Children with CdLS commonly have hypotonia. The child's limbs and trunk may feel limp, and it will be difficult for him/her to move against gravity. It will also be difficult for the child to support himself/herself in an



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upright position (sitting or standing), as the muscles will have to work harder. As a result, in sitting or standing, the child may be hunched over or have an increased arch in his/her back. As a result of the difficulty of movement, the child may develop muscle contractures later in life from lack of moving. Physical therapy treatment for hypotonia should include teaching the family good positioning for eating, sleeping, and playing (with or without special equipment such as chairs and standers), improving muscle tone, improving midline positioning, and maintaining proper alignment of the body during developmental activities.

Hip Dislocation: Dislocation of the hip may occur in a child with developmental dysplasia of the hip, otherwise known as congenital hip dysplasia. With dysplasia, the head of the femur (top of the thigh bone) or the acetabulum (the part of the pelvis where the head of the femur attaches) grows abnormally. Typically, with dysplasia, the acetabulum is shallow and does not hold the head of the femur securely. As a result, the head of the femur may sublux (come partially out of the acetabulum) or dislocate (come fully out of the acetabulum). Children with dysplasia will have asymmetrical movement of their hips, with the affected hip not being able to move out to the side (abduct) as fully as the non-affected side. Treatment for an infant includes wearing a Pavlik harness to position the hips. The physical therapist will work with the family to teach application of the harness as well as safe ways to lift and carry the baby. For older children, a spica cast may be used, or the child may require surgery as recommended by the orthopedist. For a child in a spica cast, the PT will teach the family safe ways to lift and carry the child, and depending on the orthopedist and the child, may teach the child ways to move and be functional in the cast. For a child who has had surgery, the physical therapist will work with the child on strengthening, range of motion, and improving function.

Legg-Calve-Perthes Disease: In Legg-Calve-Perthes disease, degeneration of the femoral head results from decreased blood supply. It progresses for 2-4 years, and will eventually heal itself. It occurs in boys more often than girls, boys being affected 4-5 times greater than girls. The child with Legg-Calve-Perthes Disease may initially demonstrate a limp while walking, and have pain in the groin, thigh, or inner aspect of the knee. Physical therapy treatment may include stretching, strengthening, teaching the family exercises, and if necessary, using a walker or crutches.

Early Intervention

Physical therapy should be recommended whenever a child has a movement difficulty that limits his or her ability to perform daily activities. The earlier a child starts to receive physical therapy services, the better it is for both the child and the family. Early intervention, between the ages of birth and three years, may help to reduce the likelihood of a child developing compensatory movement strategies that may cause more problems later in life, as discussed above. Physical therapy through early intervention should focus on helping the young child with CdLS to grow and develop, supporting and teaching families in this process. Developmental activities which may be addressed (depending on what the child is able to do) include rolling, sitting, crawling, and walking. It is important for parents/caregivers to be



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involved in this process. The suggestions below will help parents/caregivers facilitate these activities:

Rolling: Position an infant or young child on his/her side, placing toys just outside reach, encouraging him/her to roll onto the stomach. While a child is on his/her stomach, if this is a difficult position, place a rolled towel or small blanket under the chest to make it easier to lift the head. Use toys that attract the child's interest to encourage rolling. Make sure that his/her arms do not interfere with the roll, moving them out of the way as necessary.

Sitting: Place a child who is not yet sitting on his/her own in a sitting position on the floor. Sit behind the child, giving support at the hips or just under the shoulders, depending on what the child needs. Gradually remove your support as the child is able to tolerate without becoming upset or falling.

Another activity is to place the child seated, facing you, on your thigh, with your hands under the child's arms. Bounce the child on your lap, or gradually rock from side to side. Ask the child's therapist to show you ways to teach your child to move from lying down to sitting.

Crawling: Help the child to move onto hands and knees. If this is difficult, place the child on hands and knees over your thigh while sitting on the floor. This will give the child more support and help him/her feel more secure. Once this is easy for the child, or the child is able to maintain this position independently, encourage crawling by placing toys of interest just outside of reach.

Standing/walking: Encourage a child who does not walk to stand by positioning favorite toys just outside reach on a surface above him/her. Help the child to pull up to standing at the surface, such as a couch or coffee table. Stay behind the child with your hands on the child's buttocks or back, as necessary. Encourage cruising (walking sideways) by moving the toy to the side, helping him/her to step sideways. Make sure to allow time for the child to play before moving the toy again, or frustration will result.

For any of these activities, or for more suggestions, ask the child's physical therapist to provide you with further ideas.

Guidelines for Parents/Caregivers

It is very important for parents/caregivers to be involved with their child's therapy program. The therapist may suggest exercises for stretching and/or strengthening, activities for play, and various positions for play, sleeping, and eating. Parents benefit from observing and/or participating in their child's physical therapy session. By being involved, parents will have opportunities to voice concerns, to learn home programs, and learn ways of interacting with their child to promote development. Parents spend more time with their child than therapists; therefore, they need to take an active role in their child's therapy program. Parents should feel comfortable asking their child's PT any questions. Likewise, therapists rely on parents to share information about how the child is performing at home. The parent, child, and therapist are all part of a team, and need to have open lines of communication.

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Finding a Qualified Physical Therapist

Physical therapy may be provided in a variety of settings, including school, home, outpatient departments, preschool, and day-care centers. The physical therapist should specialize in pediatrics, as he/she will have better knowledge and understanding of development. Regardless, all recently graduated practicing PTs must have a masters or doctoral degree from an accredited college (if they have been practicing for several years, they may only have a bachelors degree), have passed a national licensing examination, and be state certified. Physical therapy services may also be provided by a physical therapist assistant (PTA), under the direction and supervision of a licensed physical therapist. The PTA will have attended a two-year program and have passed an examination.

Physical Therapy and IEPs

The Education for All Handicapped Children Act (PL 94-192) mandates that a “free and appropriate education” be provided for all children aged 6-21 with disabilities, including the right to related services (including physical therapy). The Individualized Education Plan (IEP) outlines the related services the child will receive, including annual goals and objectives. Physical therapy provided under the IEP must be educationally relevant, meaning that without the service, the child cannot participate fully in the educational environment. For example, children need to be able to sit in school. For a child with CdLS, this may be difficult due to low muscle tone and weak muscles. Therefore, the school PT may work on independent sitting, and provide an adaptive chair if needed. Physical therapy goals and objectives in the IEP must be functional and educationally relevant. During the child’s IEP meeting, goals and objectives are discussed with the team, including the parent. It is during this discussion that parents should bring up any questions or concerns relating to PT goals and objectives. Additionally, the level of service is determined at the IEP meeting based upon the child’s needs.

Sometimes, physical therapy services are denied. In this case, parents should question the reason. Often the reason PT is denied is because the service is not educationally relevant. This does not mean that the child does not need PT; rather, it means that the child is functioning in the education environment, but may benefit from physical therapy under the medical model (outpatient services). If services are denied and the parent feels that physical therapy IS educationally relevant, then a physical therapy evaluation should be requested.

In Conclusion

Physical therapy is just one aspect of care for a child with CdLS. It should be personalized to each child, as every child is different. The child should be evaluated or assessed on an ongoing basis, with treatment goals changed as the child progresses. Parents should be involved in physical therapy, as they have the potential to implement programs at home for the child’s maximal benefit.