12. Physical Therapy (PT)

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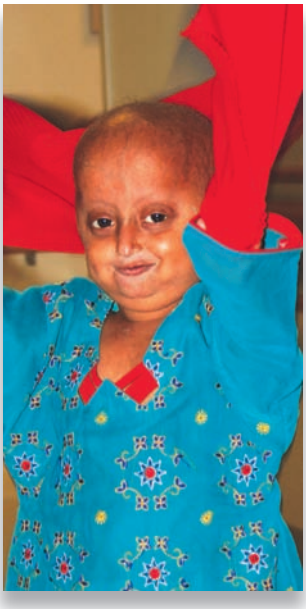


Joint contracture occurs in all children with Progeria as they get older. Physical therapy and activity may positively impact progression.

Generally, physical therapy (PT) promotes health with a focus on gross motor skills.

This chapter presents general recommendations for children with Progeria. Children vary widely in their presentation. Therefore, evaluation by appropriate health care professionals is necessary to address individual needs.

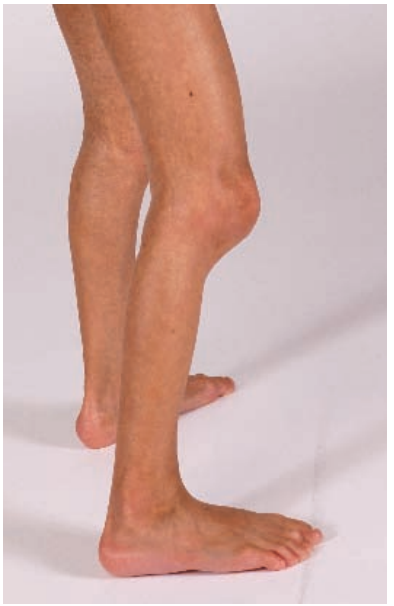
Please also refer to *Living with Progeria*, section 17, for additional advice on physical adaptations from parents of children with Progeria.

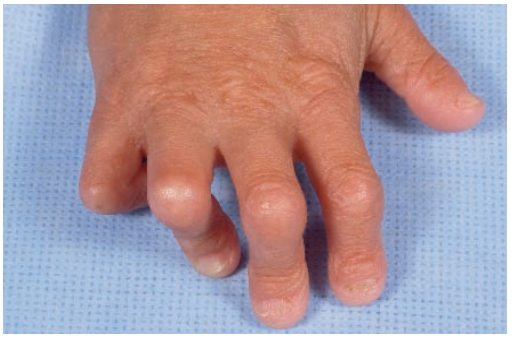
Children with Progeria develop contractures of their joints and associated boney deformities early in life. These impairments are progressive and impact their ability to perform activities of daily living and to fully participate in the typical activities of similarly-aged peers. Rate and degree of progression is highly variable.

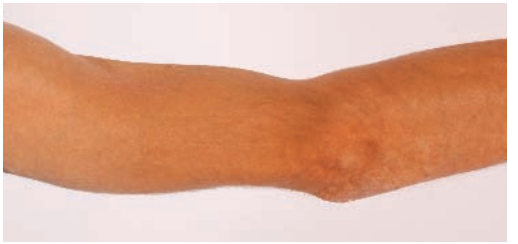
There have been no studies to determine the effectiveness of physical therapy interventions on physical activity with this population. The recommendations in this handbook are based on clinical observations and discussion with patients and their health care providers.

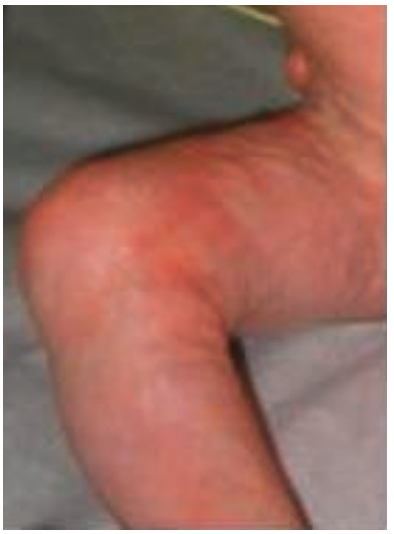
Most children with Progeria should receive physical therapy. Physical therapy includes evaluation, direct and consultative services by a qualified professional, and a home exercise program. All are integral parts of the whole plan of care. A frequency of three times a week is generally recommended for direct treatment. If direct service is not available, home care by care-

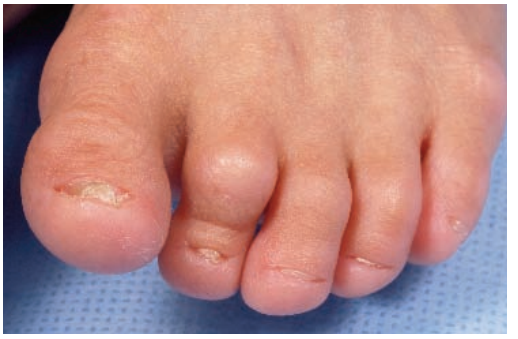
> Global joint contractures











takers – with twice yearly evaluations – is necessary to revise the physical therapy plan of care.

A physical therapy evaluation should include the following assessments: range of motion and muscle length, muscle performance, posture, pain, gait, locomotion, balance, self care and home management, neuromotor development, sensory integrity, community participation, the need for assistive, and adaptive devices, and orthotics.

Interventions include developmental and functional activities, therapeutic exercises, and prescription of adaptive equipment and orthotics. Physical therapists can also assist with locating appropriate programs for physical activity, such as local swimming classes with qualified instructors.

Clinical presentation

Children with Progeria develop contractures in all joints of the body. Additionally, changes to the bones including resorption of the distal clavicles and distal phalanges of both the hands and feet contribute to the children's functional impairments. Coxa valga and acetabular dysplasia are found in virtually all children. Progression to unilateral or bilateral hip dislocation can also occur in later stages.

Characteristic patterns of limited range of motion have been observed in the hip joint, flexion, rotations in both flexion and extension, and abduction. In the knee joint, motion is limited in both flexion and extension. Hamstring length is relatively preserved with popliteal angles not differing significantly from knee extension. In the ankle joint, the subtalar joint becomes fixed in eversion at an early age. Plantar flexion beyond neutral is limited to absent.

Gait is characterized by a crouched appearance in the sagittal plane and significant calcaneal position at the ankle with hindfoot valgus and midfoot pronation. Segmental transverse plane motion during ambulation is very limited.

Hip and foot pain are common features in children with Progeria, but can occur in other areas as well. Hip pain can be sudden or have an insidious onset and may or may not be associated with trauma. Pain in the hip may be a symptom of a serious bony problem and should always be evaluated by a physician.

Foot pain appears to be related to the calcaneovalgus position of the foot and ankle, and the lack of subcutaneous fat under the calcaneus. These factors cause increased weight-bearing on the poorly padded calcaneus. Foot pain

can be significant enough that children cannot walk barefoot and ambulation becomes limited.

Younger children with Progeria have demonstrated delay in their balance responses which may result in injury. Assessment of both dynamic and static balance is indicated. The precise mechanism of the balance dysfunction is unknown, although contractures may play a role, especially in the more severely affected child.

Interventions

> Therapeutic exercise

Range of motion exercises may be of some benefit in preserving joint range. Exercises should be done several times a week and stretches should be maintained at end range. Activities which cause the child to move through the full excursion of joint range of motion are more functional and more enjoyable for the children and should be encouraged.

Aerobic conditioning is not necessarily indicated, as function is limited more often by joint contractures and pain and less by the secondary effects of cardiovascular impairment. However, it appears the more active the children are, the more functional they remain.

Muscle strengthening may be beneficial for strengthening the muscles opposing the areas of most common contractures such as gluteus maximus, quadriceps, and gastrocsoleus complex to help maintain range of motion.

Orthotics may be necessary to provide support or improve alignment. Fabrication of a well-padded orthotic that distributes the child's weight more evenly over the entire plantar surface of the foot is helpful in improving tolerance to ambulation by decreasing pain.

> Functional training in self-care and home management

Functional limitations include the inability to assume certain positions such as side-sitting or perform activities such as squatting or climbing stairs. Transitional movements such as moving through kneeling may also be difficult. Limitations in range of motion appear to be the primary reason for these difficulties. Short stature may also impact their function.

Functional limitations will impact the child's ability to get on a school bus, negotiate playground equipment, and perform many self-care activities.

Assessment and provision of assistive devices to optimize independence is needed to allow the children to function similarly to their age-matched peers. Home modifications may also be necessary (refer to *Occupational Therapy*, section 13).

> Functional training in work (job/school/play), community, and leisure integration

Children with Progeria are generally socially and cognitively intact. Locomotor skills are limited due to contractures and short stature. Therefore, children with Progeria may have difficulty keeping up with their peers. Independent mobility is preferable to dependent forms of mobility such as being carried or using a commercial stroller. Provision of mobility devices to allow the children maximum participation in their environments is often necessary as the disease progresses.

Mobility devices allow children with Progeria independent, as well as more age- and developmentally-appropriate, access to their environment. The devices can be an adjunct to mobility, and be situation specific, such as long distance mobility. Whenever feasible, the child should be encouraged to be as active as possible to maintain overall level of function.

When available, power mobility (electric wheelchair) is preferable to manual wheelchairs due to the limitations in the upper extremities. Walkers may also be of some use particularly in children who have had strokes.

Precautions

Any sudden change in functional status, such as the loss of the ability to walk, or pain or significant change in range of motion should be evaluated by a physician even if there is no traumatic event.

Although gentle stretching is part of PT care, aggressive stretching should be avoided as the risk of fracture as a result of this intervention is unknown.

Due to the tendency towards the development of a calcaneal deformity, heel cord stretches should be avoided.

Activity guidelines

Children with Progeria should be encouraged to participate in physical activities. Participation is important as it enhances peer interaction, contributes to physical fitness, and may minimize impairments and functional limitations as the disease progresses.

Children can engage in a wide variety of physical activities, such as walking, dancing, hiking, and swimming. They may not be able to participate in some team sports as they are significantly shorter and have less body mass than their peers, therefore safety may be an issue. Bony deformities may also be a limiting factor for some physical activities. If in doubt, ask for advice from a physician and/or physical therapist who is familiar with your child.

Children and families may need assistance from a therapist in order to find appropriate physical activities or programs. They may also need assistance finding appropriate sized toys or adapted toys (i.e., tricycles) in order to engage in physical activities.

Swimming

Swimming is great for joint flexibility; however, children with Progeria face several challenges with swimming. Because they have a severe lack of body fat, they are not well insulated. Pool water may feel extremely cold; if the water can be heated to a higher degree then the pool will be better tolerated. The ocean or ponds will be more of a challenge. We recommend a wetsuit, fitted to the child if possible. Standard children's wetsuits are too large in the legs and arms, and will not be able to properly insulate the body. In addition, fat is important for the ability to swim because it floats. Therefore, it is much more difficult for children with Progeria to swim without flotation devices. All swimming activities should be supervised by an adult who is qualified in water safety and rescue.