

Physical rehabilitation guidance for achondroplasia

(Physical rehabilitation guidelines collected from different bone dysplasia renowned professional sources for individuals with achondroplasia).

Introduction

The word achondroplasia comes from the Greek and means “without cartilage formation”; a concept which is far from reality. Cartilaginous tissues become bone during the fetal development and the childhood. In achondroplastic individuals, an anomalous process occurs during growth, especially in the long bones. Cartilage cells of the growth plates become bone tissue too slowly due to the blockage of the production of cartilage tissue inside the growth epiphyses. This process leads to short bones and consequently short stature. By contrast, bones formed from membranous ossification (part of the skull and facial bones) are normal.

Apart from bone longitudinal growth arrest, the remaining of the growth mechanism (spine formation, hypertrophy, calcification, and ossification) occurs normally, although the amount of bone formed is significantly lesser. Therefore, achondroplasia is a bone growth and development disorder.

The cause of achondroplasia is genetic. It is caused by a mutation of the gene of the fibroblast growth factor receptor 3, located in the short arm of the chromosome 4

The frequency of this disorder is one in every 20,000 births. However, the chance of having an achondroplastic child is 50% if one of the parents has the disorder and 75% if both of them have achondroplasia.

Key physical characteristics in achondroplasia

- Adult height: 122-144 cm (male) / 117-137 cm (female).
- Disproportion between an average trunk and short limbs.
- Small, flattened (anteroposterior) thoracic cage.
- Broad skull and small face.
- Prominent forehead and jaw.

- Depressed nasal bridge in its upper part.
- Mild discrepancy between the muscular development and the (bone) skeleton. Usually, there is an excess of soft tissues compared to the bone length.
- Hands in trident shape: increase of the third interdigital space.
- Mildly convex abdomen due to weakness of the abdominal muscles. Usually, this is associated with a lumbar hyperlordosis, which is often compensated by a lower dorsal kyphosis.
- Joints with large articular amplitude, caused by ligament laxity. This pattern is widespread in infants and in adults at the knees (*genu recurvatum*).
- Limited joint extension capability, mainly hoses at the elbows and hips.
- Angulations of the axes of the lower limbs: *genu varo* and *genu valgo*, Genu varu is more frequent due to the tendency of hip external rotation.
- Small spinal canal due to the narrowness of the posterior vertebral arches and to the decrease of the space between pedicles, where the spinal cord is located. This, in turn, can lead to spinal cord compression.
- The foramen magnum may have a narrower diameter, as seen by CT (computed tomography) scan or MRI (magnetic resonance). In an achondroplastic adult its transversal diameter is similar to that of a newborn baby and its sagital diameter is similar to that of a two year old toddler. This narrowing can cause cervico medullary compression, with signs and symptoms of high cervical myelopathy and / or central apnea and can be life threatening.
- In a newborn baby, we find a broad muscular hypotonia, which may imply in motor development delay.

Physical therapy guidelines in achondroplasia

Before starting, we need to clarify that this represents a simple guideline showing fundamental physical therapy techniques useful in acondroplasia. To allow better results, they should be started as early as possible. Each affected individual will have distinct characteristics and needs that will define and differentiate him/her from others; therefore the treatment should be planned in an individual basis.

To make this text more didactical we will divide the physical therapist activities in three periods of the life of the affected individual: 1. infancy; 2. childhood and adolescence and 3. adulthood.

Achondroplasia in infancy

During the follow up of an infant, we must consider the possible existence, or the subsequent development of two common clinical situations: foramen magnum stenosis (narrowness of the upper spinal canal) and hydrocephaly.

We have already commented that these children have a smaller than normal foramen magnum, which could cause medullar compression. As there is also hypermobility at atloaxoideo (first two vertebral bodies) level and the smaller size of the odontoid, the neck is considered a risk zone for trauma and, therefore, we must take maximum care throughout treatment. Wobble of the head should be avoided, especially in this period, in which the child still has not acquired a good head control yet. Parents and caregivers must be educated about the care with the neck. .

Development of hydrocephaly may occur in the first two years of life. Head circumference must be followed up during this period along with frequent screening for related signs and symptoms (fastly disproportionate skull growth, headache, drowsiness, vomiting and leg stiffness). If hydrocephaly is suspected, the child should be referred immediately to a pediatric neurologist or to a neurosurgeon.

When evaluating an infant, we frequently observe a generalized muscular hypotonia, most often mild to moderate, which causes apparent motor skill delay. If the child is left without any intervention, by the time he/she is reaching the sitting position, there is a significant risk of developing a marked spine flexion, with wedging of the first two lumbar vertebrae, causing fixed thoracolumbar kyphosis, which may require surgical intervention for correction. Motor stimulation should be applied as earlier as possible in order to facilitate the achievement of a good trunk control and proper standing and walking.

Nevertheless, it's important not to force the child's motor development. Muscles of head and trunk control should be strengthened prior to sitting. Until this muscle groups' strength is not achieved, the child should not be allowed to sit without support, because this could favor the establishment of the above mentioned thoracolumbar kyphosis. A cushion can be placed on the child's back for proper seating.

To increase the muscle tonus the infant must be included in a stimulation program performed in three fundamental positions: prone and lateral decubitus, sedestation (sitting) and bipedestation (standing).

Exercise should begin in the prone position with maneuvers designed to achieve control of the cephalic pole weight by increasing the strength of the cervical and thoracic musculature, which should include the stimulation of the *rachis erector*.

Once the infant have acquired appropriate muscle tonus, we will perform all exercises designed to improve trunk control in a sitting position (sedestation). The development of head and trunk responses can be stimulated through diagonal inclinations.

Finally, with the child on a kneeling position, exercises are performed for proper achievement of trunk weight control. The goal is to prepare the child for the standing position. This can also be worked out from a sitting position with leg extension. There are two topics that must be taken in account: first, child walking maybe delayed up to two years (age); and a step should be positioned under the child feet when in prolonged sitting to avoid the tendency for hip external rotation.

Finally, also during this period, respiratory exercises should be introduced, in order to start appropriate diaphragm conditioning.

Physical therapy in children and teenagers

This period is characterized by the establishment of a number of orthopedic deformities that may progress, leading to seriously limiting the child. Among these deformities it should be noted the restriction that appears at the elbows. The range extension of this joint is usually limited, resulting in arm flexion.

To keep the flexibility and the range of movement, the child, after previous training and under supervision, must perform exercises, which involve repetitive elbow extension as well as gentle stretching of the flexor muscles of the arm. When there is severe muscle contraction, the physical therapist should apply the appropriate techniques to allow improvement of the member mobility.

The other joints subjected to limiting flexion are the hips, which tend to progress, causing more pelvic anteversion and consequently, lumbar hyperlordosis. To prevent or reduce this complication, or if it has been already established, exercises

in pelvic retroversion should be applied, with emphasis in the training of the transverses abdominis, before other abdominal muscle groups are engaged. Gentle and progressive stretch work on the muscles responsible for the lordotic posture, especially the hamstrings and psoas-. With proper training, the child or teenager can learn to perform the exercises by him/herself..

Postural education is also very important because it will have a significant impact in an adequate control of the sitting (sedestation) and prolonged standing (bipedestation) postures and in handling loading activities. Frequent monitoring of the column axis to detect scoliosis should be in place. Scoliosis may frequently lead to change in lower extremities axes.

Both *genu valgo* and *genu varo* may occur, but *genu varo* is more common. This characteristic leg angulation produces *tibia vara*, also causing foot *varus* with foot pronation restriction. If the deformity caused by the *genu varo* is not too severe, we can work to reduce its progression by training the stretch of internal leg muscle groups while reinforcing the external groups, which are usually weak and distended. For *genu valgo*, the strategy is exactly the opposite. In severe leg deviations, orthopedic surgery should be considered.

Furthermore, during childhood and especially during puberty, tendon reflexes must be under frequent surveillance. The finding of local deficits, asymmetries or hyperreflexia could suggest the presence of lumbar stenosis. We will talk more about this below, as this is a common complication in adulthood.

Sport practice is a concern among children and teenagers with achondroplasia. Sports that have emphasis in force, collisions or repetitive jumps should be avoided to reduce unnecessary stress on the joints. Sports like swimming and cycling are encouraged.

Obesity is a common problem in achondroplasia. Therefore, it is important to prevent overweight as it may cause excessive joint loading stress. Diet is essential.

In summary, physical therapy during this growth period is important to minimize or control the common orthopedic features of achondroplasia, trying to allow the child to reach adulthood in the best possible physical condition.

Physical therapy in adulthood

Lumbosacral spinal stenosis is a very common problem in the achondroplastic adult and may result in severe consequences. The lumbar stenosis may result in spinal cord compression because the space for the spinal cord is too narrow (the bone is

too small for a normal cord). The stenosis may be worsened by the common hyperlordosis found in achondroplasia.

Clinically, the spinal cord compression usually begins with pain in the legs or in the lower back, heavy-footed walk or leg weakness, which can prone to sudden falls. These symptoms may be progressive. If the diagnosis is delayed, there is increased risk of paraplegia and loss of bladder control. Therefore, monitoring tendon reflexes and column lordosis status are important during clinical evaluations.

In addition, as a preventive measure for lumbar spine stenosis, adults are advised to avoid jobs which require long period standing or with repetitive spine extensions; both situations can lead to further narrowing of spinal cord space and consequent development of back pain.

In summary, people with achondroplasia and others with pathological short stature (we have worked on achondroplasia as the most common condition) need and require physical therapy assistance, among other kind of health care throughout his/her life span. Physical therapy is of utmost importance from infancy to puberty, for the muscle-skeletal problems this condition usually brings. Physical therapy could help preventing many complications that can cause several disabilities and compromise the daily lives of the affected individuals. The physical therapy purpose is to improve the quality of life of the achondroplastic patient; a purpose that must be shared by a multidisciplinary team, which comprehensively and integrally addresses the individual social and health care needs.

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