Pseudoachondroplasia

Overview

Pseudoachondroplasia is a disproportionate form of dwarfism that affects an estimated one in 30,000 people. Common features include varus deformities (bowed legs), curvature of the spine, joint instability and/or hypermobility and early-onset osteoarthrosis. Children are not usually diagnosed at birth but are usually tested in the toddler years. It usually presents in an autosomal dominant fashion and is caused by a mutation in the cartilage oligomeric matrix protein (COMP). Treatment for symptoms may include monitoring, physical/occupational therapy and/or surgery. Psuedoachondroplasia does not affect life expectancy.

Heredity and genetics

Pseudoachondroplasia (PSACH) was once thought to be a milder form of achondroplasia, but research has shown they are different disorders with separate genetic mutations. All individuals with pseudoachondroplasia have a mutation in the COMP gene, which is normally responsible for making cartilage oligomeric matrix protein, a protein found between chondrocytes (cartilage-forming cells). When the mutation is present, abnormal amounts of protein accumulate inside the cells, causing the cells to die and slow bone growth. This gene has also been found to have been implicated in multiple epiphyseal dysplasia, a related but milder disease. In both PSACH and MED, severity of symptoms and characteristic features may vary between individuals.

Pseudoachondroplasia almost always occurs from an autosomal dominant mutation, so an affected adult has a 50 percent chance of passing the mutation to each child. It is also possible for two average-statured parents to have a child with the mutation, in which case PSACH resulted from a random, sporadic mutation in the sperm or egg cell. If this occurs, the chance of having another child with pseudoachondroplasia increases with each pregnancy. For an average-statured family with one child with PSACH, the risk of recurrence is approximately 1-2 percent, while the recurrence rate for an average-statured family with more than one affected child may grow to approximately 50 percent. This phenomenon is referred to as germinal mosaicism.

Diagnosis and testing

Compared to other skeletal dysplasias, diagnosing pseudoachondroplasia often takes longer because clinical features do not usually present until 2 to 4 years of age. Growth starts at an average rate but slows as the infant enters the toddler stage. Delayed growth and gait abnormalities usually signals the need for diagnostic testing. A diagnosis of
pseudoachondroplasia may be suspected through radiographic imaging but should be confirmed through genetic testing. If there is a family history of pseudoachondroplasia genetic testing may be performed prior to the child entering the toddler stage. Prenatal DNA analysis is also available for families with a positive family history.

Clinical features:

- Short Stature
  - Average adult height range: 2-foot-11 to 4-foot
- Joint laxity in the hands, knees and ankles
- Curvature of the spine (kyphosis, scoliosis and/or lordosis)
- Leg deformities, including genu valgum (knock-knees), genu varum (bowed legs) or windswept alignment
- Joint pain
- No unique facial involvement

Radiographic features:

- Leg length discrepancy
- Proximal tibia recurvatum
- Distal femur procurvatum
- Shortening of the tubular bones with irregular metaphyses and epiphyses

Orthopaedic problems and treatment

Growth monitoring

Growth should be monitored using pseudoachondroplasia-specific weight, weight-by-height and BMI charts.

Short-statured patients may find it difficult to complete everyday tasks such as hygiene routines, driving, reaching household items, etc. Adaptability products such as extendable reachers, pedal extenders, ergonomic keyboards, etc., may be necessary to aid in accessibility.

Growth hormone treatment is not recommended for pseudoachondroplasia and limb-lengthening surgery should only be discussed when the patient is old enough to consider the possible risks and benefits.

Spine deformities

Outward and/or side-to-side curvature of the spine (kyphosis and scoliosis) is common and affects an estimated 50 percent of patients.
Kyphoscoliosis is monitored with clinical exams and radiologic imaging — AP and lateral spine X-rays. If further treatment is required, it may include bracing and, less frequently, surgical intervention in the form of a spinal fusion.

Lordosis, or inward curvature of the spine, of the lumbosacral region is common but is less of a concern. If low back pain is present because of the curvature, physical therapy may be necessary to strengthen the core muscles and stretch the hip flexors.

Cervical spine instability

About one in six experience cervical spine instability, which increases the risk for cervical cord compression. C-spine instability is not as common as in other spondyloepiphyseal dysplasias but may occur in a small portion of the population. This is usually monitored with lateral cervical spine x-rays in the flexion, neutral and extension positions. If symptoms such as upper extremity numbness, tingling or loss of fine motor control (fixing buttons, writing, sewing, etc.) develop, an MRI may be used to determine if surgery is necessary. However, surgical intervention is not usually indicated.

Joint hypermobility

Hypermobility, or “loose joints,” causes joint instability in the shoulders, wrists, knees, and ankles. When severe hypermobility is present, one might experience difficulty in fine motor activities such as knitting, writing, drawing, etc., and may find the hypermobile joints to fatigue more easily. Joint hypermobility is often noted at a young age then monitored as the child gets older. If symptoms interfere with everyday activities, consider modifying the environment with ergonomic keyboards, modified eating and writing utensils, etc.

Osteoarthritis

Premature osteoarthritis usually begins to develop during late adolescence/early adulthood and will progress with age. Symptoms usually begin in the knees and hips but may later present in the shoulders, elbows, ankles and feet. Osteoarthritis and joint degeneration may be painful and limit daily activities. It is monitored with radiologic images, and progression may be delayed by adapting activities to reduce stress on the joints.

Leg alignment abnormalities

Almost all pseudoachondroplasia patients have some sort of leg alignment abnormality. The most common is genu varum (bowed legs), but genu valgum (knock-knees) or a windswept alignment (one leg varus the other leg valgus) can occur. Treatment varies according to the severity of the deformity and may include clinical monitoring using a series of X-rays called an “extremity alignment series.”
Leg length discrepancy

Leg length discrepancies, or differences in the lengths of each leg, are a result of uneven bone growth between the lower extremities. This is frequently observed in those with pseudoachondroplasia and is treated using a shoe lift or insert.

Other common problems and treatment

Weight management

Weight management is important for everyone, regardless of stature. Going above a healthy weight increases the amount of pressure on the joints and can increase the risk of complications during surgeries or other medical procedures, in addition to cardiovascular symptoms. When extra weight becomes distributed over a smaller surface area like that seen in any skeletal dysplasia, these observations may be more dramatic or more noticeable. However, there may be less incidence of obesity-related type 2 diabetes.

Because body mass index (BMI) takes height and weight into account, dwarfs tend to have a naturally higher BMI but can still be considered “healthy” for their size. Use pseudoachondroplasia-specific BMI charts to monitor appropriate height and weight ratios.

When exercising or engaging in physical activity, avoid high-impact sports such as football, wrestling, etc., unless the activity has been modified for achondroplasia (such as DAAA activities). Low-impact and individualized sports and activities such as tennis, swimming, walking, etc., tend to put less pressure on the joints and are typically more comfortable forms of exercise.

Resources

Pseudoachondroplasia general information


Growth charts  

Little People of America  
Adaptive Products

http://www.lpaonline.org/adaptive-products-