CONDITION

Achondroplasia

Learn how achondroplasia affects growth, how it is diagnosed, and how lifelong monitoring and treatment can support health and independence.

WHAT IT IS
Genetic condition affecting bone growth

BODY SYSTEM
Bones and joints

MAIN SYMPTOMS
Short limbs, larger head, bowed legs, low muscle tone

WHAT CAUSES IT
Genetic FGFR3 change

HOW IT IS DIAGNOSED
Blood or saliva FGFR3 genetic test

MAIN TREATMENT
Lifelong monitoring and complication-specific care

CAN IT BE CURED?
No, treatment cannot remove all effects

IS IT AN EMERGENCY?
Sometimes, severe trouble breathing or sudden paralysis

LONG-TERM OUTLOOK
Many lead active, independent lives with lifelong follow-up

Overview

Achondroplasia is a genetic condition that affects bone growth. It is the most common cause of disproportionate short stature, sometimes called short-limbed dwarfism. "Disproportionate" means that the arms and legs are shorter in relation to the chest and abdomen.

People with achondroplasia have characteristic differences in bone structure. These may include shorter upper arms and thighs, a larger head, a prominent forehead, and a flatter area between the forehead and nose. Adult height is shorter than average, but the exact height varies from person to person.

Achondroplasia does not usually affect intelligence. Children may reach some movement milestones, such as sitting or walking, later than average because of differences in body proportions, muscle tone, and joint flexibility. They usually follow their own developmental

pattern.

The condition matters medically because bone growth differences can narrow the spaces around the brainstem, spinal cord, or nerves. Some people also develop breathing, hearing, dental, orthopedic, or weight-related concerns. Careful monitoring helps identify these problems early.

Many people with achondroplasia lead active, independent lives. Care often includes a primary care clinician, a geneticist, and specialists familiar with skeletal dysplasias, which are conditions that affect bone and cartilage development.

Symptoms

Features of achondroplasia may be seen during pregnancy, at birth, or in early infancy. They vary in severity.

Common physical features include:

Infants often have low muscle tone, meaning their muscles feel less firm and provide less support. This may contribute to later sitting and walking. Development should be assessed using growth and milestone charts designed for children with achondroplasia rather than charts for the general population.

Infants may develop a rounded curve in the middle or lower back while sitting. This is called thoracolumbar kyphosis. It may improve after the child begins walking, although persistent or severe curves need evaluation.

Narrowing at the opening in the base of the skull can press on the brainstem or upper spinal cord. Possible signs in an infant include weak breathing, pauses in breathing, poor feeding, unusual sleepiness, bluish skin, weakness, or loss of previously gained abilities. These symptoms require prompt medical assessment.

Sleep-disordered breathing is also common. A child or adult may snore, gasp, pause while breathing, sleep restlessly, develop morning headaches, or feel tired during the day.

Middle ear fluid and repeated ear infections can cause temporary or lasting hearing loss. Speech development may be affected if hearing problems are not recognized. Crowded teeth, an underbite, and other dental differences may also occur.

Adults may develop back or leg pain, numbness, weakness, or difficulty walking because of spinal stenosis. Spinal stenosis means that the canal containing the spinal cord and nerves has become too narrow.

- Shorter upper arms and thighs, known as rhizomelic limb shortening

- A trunk that is closer to average size
- A larger head with a prominent forehead
- A flatter bridge of the nose
- Short fingers, sometimes with extra space between the middle and ring fingers
- Limited ability to straighten the elbows fully
- Flexible joints in some areas and restricted movement in others
- Bowing of the legs
- An increased curve in the lower back when standing or walking

Causes and risk factors

Achondroplasia is caused by a disease-causing change in the FGFR3 gene. This gene provides instructions for a protein that helps control bone growth. In achondroplasia, the altered protein is overactive and slows the growth of cartilage into bone, especially in the long bones of the arms and legs.

Nothing a parent does during pregnancy causes achondroplasia. Nutrition, exercise, and routine activities do not cause the gene change.

Achondroplasia follows an autosomal dominant inheritance pattern. This means that one altered copy of the FGFR3 gene is enough to cause the condition. A person with achondroplasia has a 50% chance of passing the altered gene to each child.

Most children with achondroplasia are born to parents who do not have the condition. In these cases, the genetic change occurred for the first time in the egg or sperm. This is called a new, or de novo, variant. Older paternal age is associated with a higher chance of this new genetic change, although the overall chance in any pregnancy remains small.

When both parents have achondroplasia, each pregnancy has a chance of resulting in average stature, achondroplasia, or two altered FGFR3 copies. Having two altered copies causes a severe condition called homozygous achondroplasia, which is usually fatal before or soon after birth. Genetic counseling can explain these possibilities without directing a family's reproductive decisions.

Diagnosis

Achondroplasia may be suspected before birth when a prenatal ultrasound shows

shorter-than-expected long bones, a larger head, or other characteristic findings. These changes may not become clear until later in pregnancy. Ultrasound findings alone do not always confirm the diagnosis.

Prenatal genetic testing can look for a disease-causing FGFR3 variant. Testing may use a sample obtained through chorionic villus sampling or amniocentesis. These procedures have benefits and risks that should be reviewed with a maternal-fetal medicine specialist or genetic counselor. In some situations, specialized testing of cell-free fetal DNA may be available, but access and diagnostic reliability vary.

After birth, a clinician may suspect achondroplasia from body proportions, facial features, and physical examination. X-rays can show characteristic changes in the skull, spine, pelvis, and long bones. A blood or saliva genetic test can confirm the diagnosis by identifying an FGFR3 variant.

Evaluation after diagnosis may include measurements of head size, length or height, weight, and body proportions. Clinicians should use achondroplasia-specific growth charts. Depending on age and symptoms, additional assessment may include neurologic examination, hearing testing, sleep studies, or imaging of the skull and spine.

A diagnosis can affect several family members emotionally. Genetic counseling can help families understand test results, inheritance, future pregnancies, and reliable sources of support.

Treatment and management

There is no single treatment that removes all effects of achondroplasia. Care focuses on supporting development and independence, preventing avoidable complications, and treating problems that affect health or daily life.

Infants require careful head and neck support because of low muscle tone and the possible narrowing near the base of the skull. Unsupported sitting devices, curved-back positions, and activities that place sudden force on the head and neck may be discouraged until a specialist confirms that they are safe.

Clinicians monitor head growth, neurologic development, breathing, and signs of pressure near the brainstem. If severe compression is present, surgery may be needed to enlarge the opening at the base of the skull. The decision is based on examination, imaging, sleep or breathing studies, and specialist assessment rather than imaging alone.

Hearing should be checked regularly. Ear infections or persistent middle ear fluid may be treated with hearing support, ear tubes, or other ear, nose, and throat care. Speech therapy can

help when hearing loss or oral structure affects communication.

Obstructive sleep apnea occurs when the airway repeatedly narrows during sleep. Treatment depends on the cause and may include removal of enlarged tonsils or adenoids, positive airway pressure therapy, weight management, or other specialist-directed care.

Physical therapy can support strength, movement, and safe positioning. Therapy should account for joint flexibility, spinal alignment, and body proportions. Forceful manipulation of the neck or spine should be avoided.

Bowed legs, persistent spinal curves, or pain may require orthopedic evaluation. Some problems can be observed, while others may benefit from bracing or surgery. Spinal decompression surgery may be considered for significant nerve pressure.

Vosoritide is a medicine approved in the United States for children with achondroplasia whose growth plates remain open. It is given as an injection and can increase the rate of linear growth while a child is still growing. It does not cure achondroplasia or remove the need for monitoring. Families considering it should discuss expected benefits, treatment burden, possible adverse effects, and personal goals with an experienced clinician.

Limb-lengthening surgery can increase height but involves multiple procedures, long rehabilitation, and risks such as pain, infection, nerve injury, joint problems, and impaired bone healing. It is not routine care. Decisions should be made without pressure and with input from experienced teams and, when possible, the person undergoing treatment.

Practical support also matters. Step stools, lower light switches, adapted furniture, pedal extensions, and school or workplace accommodations can improve safety and independence. Nutrition and enjoyable physical activity can help manage weight without placing unrealistic expectations on body size.

Complications

Not everyone develops every complication. Important concerns include compression near the base of the skull, fluid buildup around the brain, obstructive or central sleep apnea, repeated ear infections, hearing loss, bowed legs, spinal curves, and spinal stenosis.

Hydrocephalus is an abnormal buildup of fluid around the brain. A rapidly increasing head size does not always mean hydrocephalus, because a larger head is typical in achondroplasia. Clinicians look for changes from the child's usual growth pattern and symptoms such as vomiting, unusual sleepiness, irritability, eye movement changes, or a tense, bulging soft spot.

Excess weight can increase pressure on the spine and joints and may worsen breathing problems. Weight should be interpreted using condition-specific charts and body proportions

rather than appearance alone.

Seek emergency care for severe trouble breathing, bluish or gray skin, loss of consciousness, a seizure, sudden inability to move an arm or leg, or new loss of bladder or bowel control.

Prompt medical assessment is also appropriate for new weakness, persistent numbness, worsening walking difficulty, repeated breathing pauses, severe or rapidly worsening headache, unexplained vomiting, or loss of developmental skills. Routine concerns such as stable leg bowing, school accommodations, or planned hearing follow-up can usually be discussed during scheduled visits unless symptoms are changing quickly.

Follow up

Achondroplasia requires lifelong follow-up, but the focus changes with age. Infants need close monitoring of head growth, breathing, muscle tone, development, and the opening at the base of the skull. Children need continued review of hearing, sleep, dental health, spinal alignment, leg bowing, weight, and progress at school.

Adults should report changes in walking, balance, strength, sensation, bladder or bowel control, pain, and sleep quality. These symptoms may suggest spinal stenosis or another treatable problem.

Follow-up may involve genetics, neurosurgery, orthopedics, ear, nose, and throat care, audiology, sleep medicine, dentistry, physical therapy, and rehabilitation. Not every person needs every specialist. A coordinated care plan can reduce unnecessary testing while making sure important concerns are not missed.

Emotional well-being should be part of routine care. Children and adults may encounter stigma, unwanted attention, or barriers in buildings, education, employment, and health care. Connection with peer organizations, counseling, and disability accommodations may be helpful when chosen by the individual or family.

Sources

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