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ACADEMIC MEDICAL CENTRE

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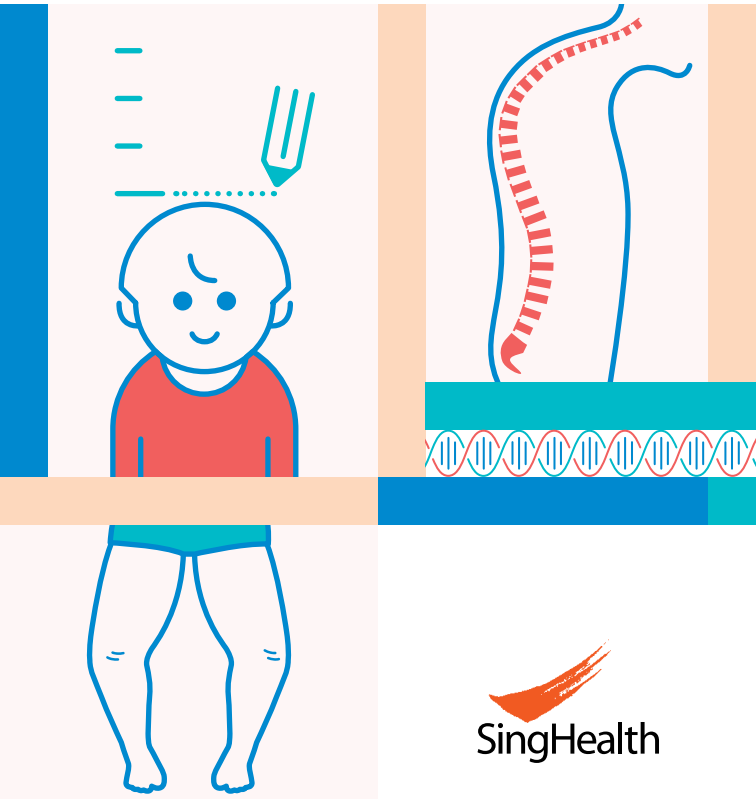
Achondroplasia



SingHealth

What is achondroplasia?

Achondroplasia is the most common type of short-limb short stature. It is a genetic disorder that affects bone growth.

Individuals with achondroplasia have disproportionate short stature and certain characteristic features. Intellect is typically normal.

How common is achondroplasia?

It is estimated that **one in 22,000 people** are born with achondroplasia. Occurrences of achondroplasia are present among all racial groups and affect both males and females equally.

What are the features of achondroplasia?

Achondroplasia is a condition one is born with. It can sometimes be diagnosed before birth by antenatal ultrasound scans in the third trimester.

The severity of symptoms may vary from person to person, even among family members with achondroplasia.

The main features include:

Short stature. The average adult height is approximately 120-135 cm

Facial features. Larger head size with a prominent forehead, and flattened nose bridge

Rhizomelic shortening. Short extremities with shortened bones in upper arms and thighs

Limited elbow extension. Inability to fully straighten elbows

Short fingers with trident appearance of hands and short toes. Short fingers with increased space between the third and fourth fingers

Bow legs. Outward curving of the bones, between the thighs and ankles

Kyphosis. Curving of the upper spine due to reduced muscle tone, typically in infancy and which improves once the child begins walking

Lumbar lordosis. Curving of the lower spine which commonly develops after the child starts walking

Other medical concerns

The following issues can also be present in individuals with achondroplasia, and need to be monitored for and treated if present:

Craniocervical junction constriction. Narrowing of the junction between the skull and neck spinal bone

Spinal stenosis. Due to small vertebral canals (spaces inside the spinal bones) leading to spinal cord compression

Hydrocephalus. Build-up of fluid in the brain

Regular ear infections due to middle ear dysfunction

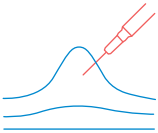
Obstructive sleep apnoea. No breathing for short periods of time when asleep

Restrictive pulmonary disease. Due to small chest causing smaller lung volumes

Obesity

Dental problems

Can achondroplasia be cured?



In the past, there was no known cure for achondroplasia or way to reverse its effects.

Voxzogo (vosoritide) is a drug that has been introduced to treat achondroplasia, which was approved in 2021. It has been shown to improve annual growth rate in children with achondroplasia and promote proportional growth.

How is achondroplasia managed?

The multidisciplinary medical team looking after your child will be able to address specific medical concerns and related health issues.

Below are some related medical issues and how they can be managed:

Medical issue	Treatment
Middle ear dysfunction	• Pressure-equalising (tympanostomy) tubes to reduce infections and avoid hearing loss

Medical issue	Treatment
Obstructive sleep apnoea and/or restrictive pulmonary disease	• Evaluation with a sleep study and/or lung function test • Use of oxygen supplementation and support
Kyphosis and spinal stenosis	• Surgical intervention in severe cases to reduce pressure on the spinal cord
Hydrocephalus	• Ventriculoperitoneal shunting to reduce pressure on the brain
Craniocervical junction constriction	• Decompression surgery to reduce pressure on the spinal cord
Obesity	• Healthy eating and staying active

What causes achondroplasia?

Genes are instructions for cells to make proteins in the body. Achondroplasia is the result of a pathogenic (disease-causing) change in the *FGFR3* gene located on chromosome 4.

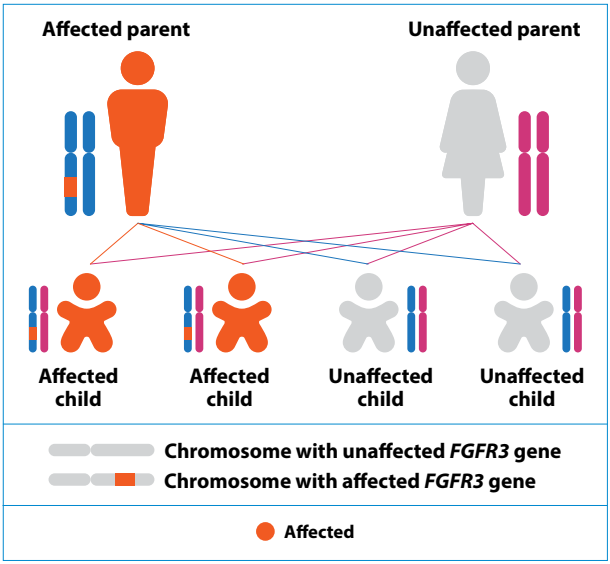
This gene provides instructions for a protein that is important for bone growth. When there is a pathogenic change in the *FGFR3* gene, the protein is affected and bone development is impacted.

This causes affected individuals to be short, and have shortened upper limbs, hands and feet. Bones in the spine and legs are also at risk of curving.

How is achondroplasia inherited?

Everyone carries two copies of each gene, one inherited from each parent.

Achondroplasia follows a **dominant inheritance pattern**, which means that a pathogenic change in one copy of the *FGFR3* gene can cause achondroplasia. If one parent has achondroplasia, a child has a 50% chance of inheriting the disease.



About 80% of people with achondroplasia have parents who are of average height, while the remaining 20% have at least one parent who has achondroplasia.

How is achondroplasia diagnosed?

Achondroplasia is diagnosed after a full evaluation by a doctor familiar with the disorder. This involves a physical examination and radiological evaluation.

Genetic testing for a causative pathogenic (disease-causing) variant in the *FGFR3* gene can also be helpful to establish a diagnosis in some cases.

How likely will I have another child with achondroplasia if neither my spouse nor I have it?

In 80% of cases, achondroplasia is the result of a spontaneous change in the genetic material of the sperm or egg at conception, in families with no previous history of the condition.

In such cases, the risk of having another child with achondroplasia is low (<1%). However, increasing paternal age has been reported to be a contributing risk factor.

Achondroplasia is a lifelong condition. Should you require financial assistance or emotional support, please approach your doctor for referral to a medical social worker.

Support Groups

Little People of America
Little People of America (LPA) supports people of short stature and their families, across the United States and internationally.
www.lpaonline.org

MAGIC Foundation
The MAGIC Foundation provides support for families of children whose growth is affected for a variety of reasons.
www.magicfoundation.org

Club Rainbow Singapore
Club Rainbow Singapore supports and empowers children with chronic illnesses and their families by providing relevant compassionate services in their journey.
Tel: 6377 1789
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www.clubrainbow.org

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