Ask About
Achondroplasia

How will achondroplasia affect his development?

What kinds of doctors will be involved in his care?

Lawson, age 4, living with achondroplasia, and his mom.

The answers you need for the questions ahead

For children living with achondroplasia, new questions can arise at every step. Start here to learn more about the condition—and talk to your healthcare team to better understand your child's journey.

Lilly-Anne, age 10, and her mom, both living with achondroplasia.

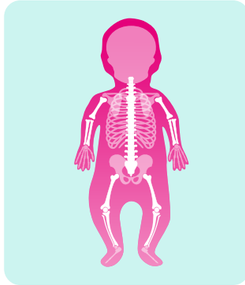
> **Even though we're small, *we've got to stand tall... and stand out.***
>
> *- Leigh-Anne, Lilly-Anne's mom.*

Understanding Achondroplasia

What is achondroplasia?

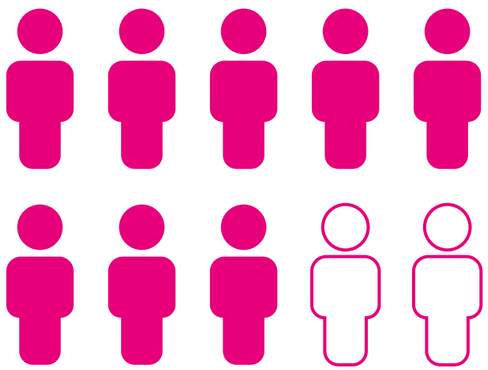
Achondroplasia is a rare genetic condition that affects how cartilage and bones grow and develop.

The condition leads to shorter arms and legs. **It can also affect other parts of the body and may cause health challenges with breathing, movement, or development.** These challenges can impact a child's daily life and overall well-being.



What causes achondroplasia?

Achondroplasia is caused by a genetic variation (or change) in a gene called **FGFR3**. The gene variation is present from birth. Most of the time, this happens by chance. Sometimes, the change is inherited from a parent with achondroplasia.



80%

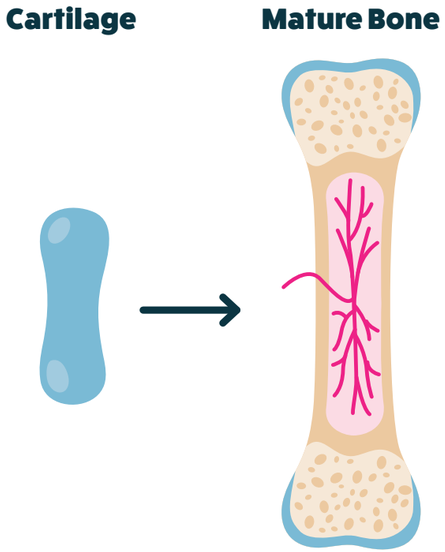
of children with achondroplasia **are born to parents who do not have the condition.**

In the remaining **20%** of cases, achondroplasia is inherited from a parent who has the condition.

FGFR3=fibroblast growth factor receptor 3.

How does FGFR3 affect bone growth?

Usually, FGFR3 and CNP work together to help bones grow from cartilage into mature bone.



FGFR3 works with CNP for balanced bone growth

Typically, FGFR3 works with a natural messenger in the body called CNP to help keep bone growth balanced

CNP sends its signals through a helper protein called NPR-B, and this pathway controls how bones grow

When FGFR3 is working correctly, it helps balance bone growth

In achondroplasia, FGFR3 is too active. This slows cartilage and bone growth and results in shorter bones.

FGFR3 is not only found in the bones; it is also found throughout the body.

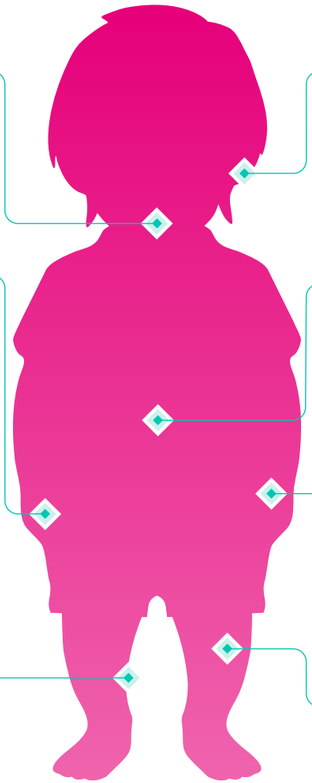
CNP=C-type natriuretic peptide; NPR-B=natriuretic peptide receptor B.

Lilly-Anne, age 10, living with achondroplasia.

How Achondroplasia Affects the Body

Lilly-Anne, age 10, and her mom, both living with achondroplasia.

FGFR3 is found throughout the body and can impact different parts of a growing child.



Breathing complications may lead to poor sleep and daytime tiredness

Chronic ear infections and hearing loss can cause discomfort and affect hearing or speech development

Short fingers and toes can make it harder to grip or handle small objects

Irregular curving of the spine may cause back pain or affect posture

Bowing of legs and joint pain may cause discomfort when walking or standing

Shorter limbs can make it harder to reach or move in certain ways

Short stature may affect daily activities

Lower muscle strength can make some movements more difficult or require extra effort

Achondroplasia has effects throughout the body:

- The **spine, joints, muscles, ears, and airways** can all be affected
- Because some parts of the body like **arms and legs grow differently than the trunk**, achondroplasia can also cause differences in bodily proportions

Cortez, age 10, living with achondroplasia, and his mom.

You may want to seek guidance from a specialized facility called a skeletal dysplasia center.

Skeletal dysplasia=a group of genetic conditions that affect the development of bones and cartilage.

Get the facts

Separate myth from fact as you learn more about living with achondroplasia

Scan to learn more at askaboutachondroplasia.com/understanding-achondroplasia

Growth and Development

Growth over time

Achondroplasia can affect how your child interacts with the world. They may have trouble reaching high places, using the toilet on their own, and joining in sports or active play.

It is common for children with achondroplasia to be delayed in sitting up, crawling, and walking.

Talking with your healthcare team

A healthcare team is made up of different specialists. They will monitor growth, help manage complications, and provide support. **Your child's team may include:**

Pediatrician
General practice doctor that handles day-to-day concerns

Ear, nose, and throat specialist
Can help with problems like sleep apnea, ear infections, and more

Orthopedic surgeon
Explores how surgery may help your child

Endocrinologist
Helps provide treatment and address any hormonal concerns

Geneticist
Can help confirm diagnosis and navigate treatment

Other
You may reach out to different specialists to respond to unique concerns

From infancy through adolescence, parents will work closely with doctors to guide crucial care decisions for their child. A child's care pathway may include:

Infancy (0-12 months)

- Regular checkups to track growth proportions
- Scans to check for fluid buildup in the brain or pressure on the spinal cord
- Sleep study to check for apnea
- Review for frequent ear infections or hearing loss

Childhood (6-12 years)

- Annual reviews with core healthcare team
- Monitoring of leg alignment and spine health
- Repeat sleep study if symptoms return or persist
- Support for emotional and social development
- Neurology follow-up as needed

Early childhood (1-5 years)

- Monitoring of growth, mobility, and development
- Ongoing neurology and orthopedic assessments
- Physical or occupational therapy to support motor skills
- Hearing and dental checks as needed

Adolescence (13-18 years)

- Screening for spinal stenosis and joint pain
- Discussion of treatment options, including surgery
- Final height
- Transition planning to adult care

Your child's healthcare team will be made up of different medical specialists.

Early detection of new or worsening symptoms can help the healthcare team provide the right support at the right time.

Lawson, age 4, living with achondroplasia, and his parents.

I think people see Lawson for his character, but nobody realizes about these issues that affect him... *because he just gets on with it and does as best he can.*

- Chelsea and Thomas, parents of Lawson.

Ask About Achondroplasia

It's important to ask questions and explore new possibilities.

You know your child best, and your healthcare team can help you understand if your child's current care is meeting their needs.

You may want to know:

- What does this diagnosis mean for my child's growth, health, and development?
- Which specialists will be involved in my child's care? What does each one do?
- How often will we need to see the healthcare team? What will those visits involve?
- Where can we find support and connect with other families in similar situations?

Early signs of complications may not always be obvious.
Staying engaged with your child's healthcare team is essential to ensure early detection and the best long-term outcomes.

Find the answers you're looking for at AskAboutAchondroplasia.com

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