



# Achondroplasia

This information sheet from Great Ormond Street Hospital explains the causes, symptoms and treatment of achondroplasia and where to get help.

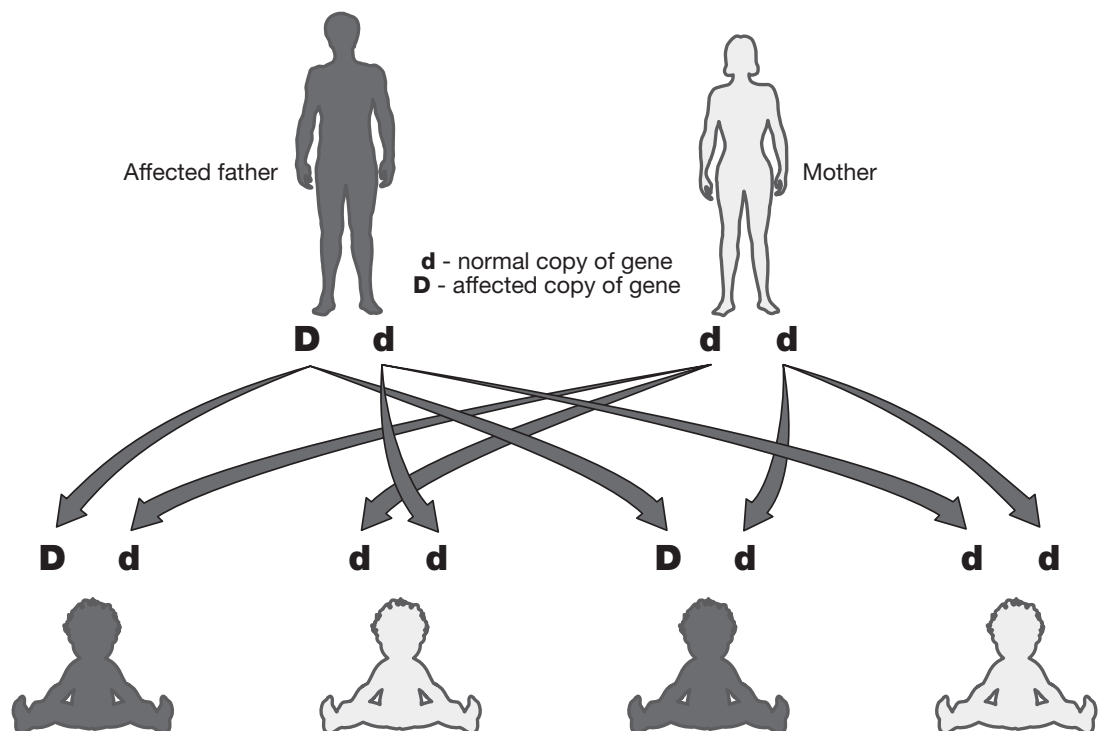
Achondroplasia is the most common type of short limb (or disproportionately short stature). The condition affects how some of the bones develop, particularly the limb bones and specifically the upper arms and thighs. There are obvious problems with how some of the facial and skull bones grow, too. This means the shortness is disproportionate so the spine appears long and the limbs appear short.

About one in 25,000 people are born with achondroplasia.

## What causes achondroplasia?

Achondroplasia is caused by a genetic mutation – a change within a gene. This mutation can occur when the egg or sperm is being formed. It is a random event, but the gene change can also be inherited.

Anybody can be born with achondroplasia but, once a person has the condition, it will run in the family and one in two of that person's children will inherit the condition.





## **What are the signs and symptoms of achondroplasia?**

People with achondroplasia have a normal sized trunk but short legs and arms. The condition mainly affects the growth of the upper arms and thighs.

Other signs include a prominent forehead, a sunken nose, crowded teeth and a protruding jaw. The average height of a person with achondroplasia is around four feet.

## **How is achondroplasia normally diagnosed?**

This condition might be discovered during an ultrasound scan performed before a baby is born. If it hasn't been diagnosed before the birth, doctors and parents are likely to notice the relatively short limbs at birth. Bone measurements on x-rays can be used to help diagnose the condition. DNA testing can help to confirm the condition if necessary.

## **How is achondroplasia normally treated?**

There is no cure for achondroplasia. Infants with achondroplasia often have a curve in the lower spine that might need a brace for the first year or so of life. Some people with achondroplasia develop bow legs. Surgery can straighten them. Others might wish to be considered for leg lengthening treatment. This can add as much as 25-30cms to a child's final height over two to three separate years, but the treatment is difficult and time consuming.

## **What happens next?**

A child with achondroplasia might be more prone to middle-ear infections and breathing problems. Later on, narrowing of the spinal canal can lead to pressure on the spinal cord and problems with walking. Spinal surgery may be required.

However, the child can expect to lead a healthy and independent life.

## **Further help and advice**

Talk to the child's doctor or health visitor.