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## Dwarfism

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### Summary

- Dwarfism is a disorder characterised by shorter than normal skeletal growth. It can be genetic.
  - Over 100 different conditions can cause dwarfism.
  - Achondroplasia is a common form of short-limbed dwarfism.
  - Most children born with achondroplasia have average-sized parents.
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Dwarfism (or conditions of short stature) refers to a group of conditions characterised by shorter than normal skeletal growth. This shortness can be manifested in the arms and legs or trunk.

There are over 100 conditions that cause abnormal skeletal growth and dwarfism. Achondroplasia is the most common type of short-limb dwarfism, occurring in around one in 25,000 children with both sexes at equal risk. This type of skeletal dysplasia (abnormal skeletal growth) is usually diagnosed at birth. This page presents information focussed on achondroplasia, but it may also apply to other conditions.

Most children born with achondroplasia have average-sized parents. Children with achondroplasia may experience delay developing motor skills, such as controlling the movements of the head, but their intellectual development is normal.

The average final height for a person with achondroplasia is 130 cm for men and 125 cm for women. Short-statured people lead normal, fulfilled lives. Achieving higher levels of education and career and personal ambitions is not limited by stature.

### Symptoms of dwarfism

The characteristics of achondroplasia include:

- a trunk of relatively normal length
- disproportionately short arms and legs
- bowed legs
- reduced joint mobility in the elbow
- other joints that seem overly flexible, or 'double jointed', because of loose ligaments
- shortened hands and feet
- a large head
- a flat mid-face
- crowded teeth, because of small upper jaw
- a prominent forehead
- a flattened bridge of the nose.

### Skeletal dysplasias and dwarfism

Dysplasia means 'abnormal growth'. There are around 100 different types of skeletal dysplasias, with achondroplasia being the most common form of disproportionate short stature.

Other skeletal dysplasias can cause short-trunk disorders and proportionate short stature. A skeletal dysplasia is diagnosed with a variety of tests, including physical examination, x-rays, tissue and bone samples, and specific genetic tests.

## Causes of dwarfism

About 80 per cent of people born with achondroplasia have average-sized parents. This means that the genetic mutation that causes achondroplasia occurs during conception, when the mother's egg is fertilised by the father's sperm. It is not known why this genetic mutation occurs, or how the mutation translates into the characteristics of achondroplasia.

Twenty per cent of people born with achondroplasia inherit the faulty gene from an affected parent. If one parent has achondroplasia, then their child has a 50 per cent chance of inheriting the gene for the condition.

If both parents have achondroplasia, their child has:

- a one in four risk of inheriting the faulty gene from both parents, which causes a fatal condition known as 'double dominant' or homozygous achondroplasia. Children born with this variation generally don't live beyond 12 months of age
- a 50 per cent chance of inheriting one copy of the gene for the condition, and therefore having achondroplasia
- a one in four chance of not inheriting the gene, and having normal stature.

## Dwarfism – common problems during infancy and childhood

Children with achondroplasia face a number of difficulties, including:

- breathing difficulties – including snoring and sleep apnoea (the regular cessation of breathing during sleep), caused by narrowed nasal passages
- ear infections – caused by narrowed Eustachian tubes (tubes leading from the ears to the throat) and nasal passages
- bowed legs – the legs are initially straight, but over time (in some cases) they become bowed once the child starts walking
- increased lumbar lordosis – a backward curve in the lower spine
- reduced muscle strength – the child has softer muscle tone than normal, and needs to be adequately supported until the muscle groups are ready to support the neck and spine
- hydrocephalus – the child has an increased risk of hydrocephalus (one in 100), which is an accumulation of cerebrospinal fluid inside the skull that can lead to head enlargement
- narrow foramen magnum – the child has a smaller than normal opening at the base of the skull (foramen magnum), where the spinal cord begins. This can sometimes press against the brain stem and cause symptoms including apnoea (cessation of breathing) and neurological signs.

## Dwarfism – common problems during adulthood

Problems faced by adults with achondroplasia can include:

- nerve compression – the nerves in the lower back or lumbar region are squashed, which can cause symptoms such as numbness or tingling in the legs
- obesity – most adults experience difficulties in maintaining a healthy weight for their height
- crowded teeth – the upper jaw is typically small, which causes the teeth to overcrowd
- higher risk pregnancies – pregnant women with achondroplasia need expert antenatal care. Caesarean section is the usual mode of delivery.

## Treatment for dwarfism

There is no cure for achondroplasia. Human growth hormone has no place in its management, as the condition is not caused by a lack of growth hormone. Treatment focuses on the prevention, management and treatment of medical complications as well as social and family support. This may include:

- surgery – may be advised to relieve pressure on the nervous system, generally at the base of the skull and lower back, or to open obstructed airways by removing the adenoids
- dental and orthodontic work – to correct malocclusion and ensure dental health
- support from other health care providers – including geneticists, neurologists and paediatricians.

There are currently preliminary trials on a medication called vosoritide to treat the symptoms of achondroplasia, but these are only in the initial stages.

### **Genetic counselling and dwarfism**

If your child or another family member has been diagnosed with dwarfism, or if dwarfism runs in your family, it can be helpful to speak to a genetic counsellor. Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand dwarfism and what causes it, how it is inherited, and what a diagnosis means for your child's health and development, and for your family. They can also explain what genetic testing options are available to you and other family members. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

You may choose to visit a genetic counsellor if you are planning a family – to find out your risk of passing the condition on to your child, or to arrange for prenatal tests.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by dwarfism.

### **Where to get help**

- **Victorian Clinical Genetics Services (VCGS)**
- Southern Cross Bone Dysplasia Centre (Director, Dr Ravi Savarirayan) Tel. **(03) 8341 6201** for appointments
- **Short Statured People of Australia**
- Your **GP (doctor)** can refer you to VCGS bone dysplasia clinics
- **Genetic Support Network of Victoria (GSNV)** Tel. **(03) 8341 6315**

**This page has been produced in consultation with and approved by:**

Short Statured People of Australia

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