

## Dwarfism

Dwarfism 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 300 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.

The majority of children born with the disorder have average-sized parents. The child may experience delay developing motor skills, such as controlling the movements of the head, but intellectual development is normal in children with achondroplasia. The average final height for a person with this condition is 130cm for men and 125cm for women. Short-statured people lead normal, fulfilled lives. Achieving higher levels of education and career and personal ambitions is not limited by stature.

### Signs

The characteristics of achondroplasia include:

- Trunk of relatively normal length
- Disproportionately short arms and legs
- Bowed legs
- Reduced joint mobility in the elbow
- Other joints seem overly flexible, or 'double jointed', because of loose ligaments
- Shortened hands and feet
- Large head
- Flat mid-face
- Crowded teeth, because of small upper jaw
- Prominent forehead
- Flattened bridge of the nose.

### Skeletal dysplasias

Dysplasia means 'abnormal growth'. There are around 300 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.

### Genetic mutation

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, then 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.

### Common problems during infancy and childhood

The child with achondroplasia faces 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 become bow-legged once the child starts walking. Over time, in some cases, the weight of the child's body causes the legs to bow.
- **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.

## Common health problems during adulthood

Problems faced by the adult 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 options

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.

- **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.

## 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 (Victorian branch) Tel. (03) 9560 6895
- Your local doctor (can refer you to VCGS bone dysplasia clinics).

## Things to remember

- Dwarfism is a disorder characterised by shorter than normal skeletal growth that can be genetic.
- Achondroplasia is a common form of short-limbed dwarfism.
- The majority of children born with the disorder have average-sized parents.

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

Short Statured People of Australia

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