Frequently Asked Questions

1. **If no members of my family are affected by Achondroplasia, can I still have a child with the condition?**
   Yes. The majority of cases of Achondroplasia occur in families where there has been no history of the condition previously reported. The genetic change involved occurs randomly.

2. **Since Achondroplasia is a genetic condition, is it possible to have more than one child with the condition?**
   It is extremely unlikely for this to happen if the parents do not have the condition themselves, as the vast majority of cases are not inherited but rather occur by chance.

3. **Do people with Achondroplasia have a shortened lifespan?**
   No. With proper treatment and management people with the condition are able to live a normal lifespan.

4. **Can people with Achondroplasia marry and have children who are average statured?**
   Yes. Due to the autosomal dominant inheritance, the recurrence risk is 50% (1in 2) in each pregnancy.

5. **Are there medical complications associated with the disorder?**
   The main medical issue to be considered is the narrowing of the lower spine canal resulting in the compression of nerves. This may require surgery.

6. **What are other challenges faced by people with Achondroplasia?**
   Among the challenges they face include reaching for higher objects and finding suitably sized clothes. There may also be some difficulties in moving around in public places and accessing public transportation.

References

Fact Sheet 38: Achondroplasia and Other Short Statured Syndromes [online]
Available from: http://www.genetics.com.au/pdf/factSheets/FS38.pdf [Accessed March 2010]

Gene Reviews - Achondroplasia [online]
Available from: http://www.geneclinics.org/profiles/achondroplasia/details.html [Accessed March 2010]

Genetics Home Reference. [online]
Available from: http://ghr.nlm.nih.gov/condition=achondroplasia [Accessed March 2010)

For further Information, please contact us at:

Persatuan Penyakit Jarang Jumpa Malaysia
(Malaysian Rare Disorders Society)
16, Lorong 5/10D
46 000 Petaling Jaya
Selangor Darul Ehsan

Telephone: 019-382 7386/019-771 4543
Fax: 03-7958 8459
E-mail: info@mrds.org.my Website: www.mrds.org.my

MALAYSIAN RARE DISORDERS SOCIETY

Rare Disorders Series:

Achondroplasia

What is Achondroplasia?

Achondroplasia is a genetic disorder characterised by short stature due to abnormal bone formation. It is the most common type of dwarfism.

The abnormal bone formation is caused by a change (mutation) in the *FGFR3* gene, located on the fourth chromosome pair.

Genes are located on chromosomes and contain 'recipes' to make proteins.

The protein made by the *FGFR3* gene functions to limit the formation of bone from cartilage. Mutations cause the protein to be overly active, resulting in disturbances in bone growth.

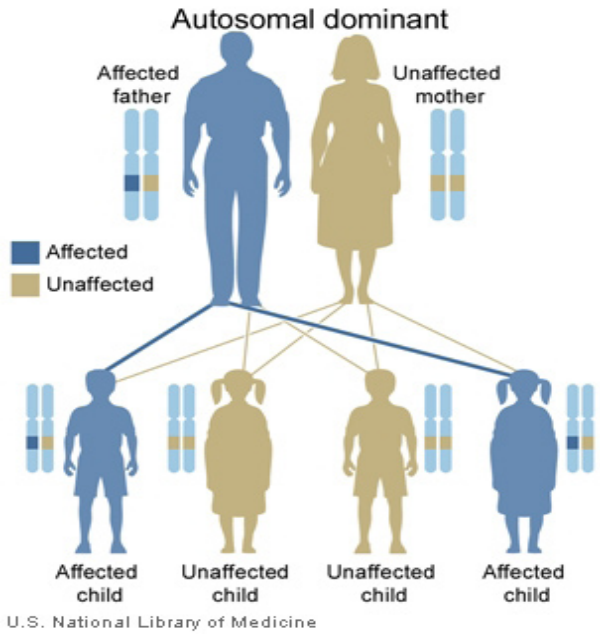
The disease follows an autosomal dominant pattern of inheritance. This means only one copy of the changed gene is needed to result in the condition.

The large majority of cases are due to a new mutation in the family. These happen by chance and are not due to anything either parent did or did not do before or during pregnancy.

Chances of Achondroplasia recurrence are very low in families where both parents do not have the condition. However, it is advised that couples who have a child with the condition seek genetic counselling to ascertain the risks for future pregnancies.

When both parents have the condition, there is a 25% risk with each pregnancy that they will have a child severely affected with Achondroplasia which does not survive the pregnancy.

Patients are advised to consult a geneticist to obtain further information on disease progression.



Inheritance pattern of Achondroplasia in cases where one parent has the condition

Signs and symptoms

- Shortened arms and legs
- Bowed lower legs
- Larger head size with prominent forehead and a flattened nasal bridge
- Curved lower spine
- Poor muscle tone at infancy
- Frequent middle ear infections
- Delayed developmental milestones

Other ways Achondroplasia may affect the body

- Compression of spinal cord and/or upper airway obstruction may lead to infant paralysis and respiratory problems
- Accumulation of spinal/brain fluid
- Speech delay due to hearing impairment
- Psychosocial difficulties

Testing

Genetic counselling by qualified personnel is advised if there is a family history of the condition and before any genetic testing is done.

Diagnosis is made before birth by foetal ultrasound or after birth by complete physical examination.

Genetic testing is available to confirm foetal ultrasound findings.

Treatment

There is currently no known cure for Achondroplasia.

Treatments are given to manage symptoms.

Height and weight need to be regularly monitored to avoid obesity.

No significant success has been recorded so far on the use of growth hormone.

Surgeries may be performed to correct bowing of legs and spinal deformities. Limb lengthening procedures have been described.

Ear infections must be treated immediately to avoid risk of hearing loss.

Speech evaluation by two years of age to check for delay attributed to hearing loss.

Sleep studies may be performed to monitor sleep apnoea (slowing or stopping of breathing for short periods).