BHARATI VIDYAPEETH'S
BHARATI HOSPITAL
AND RESEARCH CENTRE
PUNE

ACHONDROPLASIA

Frequency is 1:26,000-1:28,000 live birth affecting both males and females

About Achondroplasia

Achondroplasia is a genetic condition that affects bone growth. People with achondroplasia are shorter in height. The bones of limbs are predominantly involved. The head is well developed and hence appears large relative to the body. Average height is around 4 feet.

Signs

A person with achondroplasia may have

- Short stature
- Short limbs
- A slightly larger head with a prominent forehead
- Bowed legs as the child starts walking
- Changes in the curvature of back
- Possible overcrowding of teeth
- Intelligence is normal

Treatment & Management

There is no cure, but associated problems can be managed with regular medical care. Regular check-ups to monitor growth, evaluation of spine, sleep apnea, obesity, and ear health. Physiotherapy and exercises to improve posture, strength, and weight management. Treatment of complications such as ear infections, breathing problems, or back pain. Surgery may be needed in some people for severe spine or leg problems.

Other therapies

Limb length surgery is complex, invasive and has limited utility in gain of height. Growth hormone therapy results in limited gain in final adult height. Vosoritide injection is a new drug treatment, its long term efficacy is not known.

Cause

It happens because of a small change (of 1 nucleotide) in a gene called *FGFR3* that controls bone growth. It doesn't usually run in the family. In 80 out of 100 cases the child is the first in the family to have it. Usually parents are of normal height.

Diagnosis

- Before birth (during last 3 months of pregnancy): Achondroplasia may be suspected on an ultrasound scan when the baby's arms and thigh bones look shorter than expected and the head appears larger.
- At birth: Doctors can often diagnose achondroplasia due to characteristic facial features and short stature.
- X-rays, which show characteristic bone changes
- Genetic testing: A blood test can confirm the diagnosis by detecting a change in a specific gene (*FGFR3*).

Genetic counselling

Most cases happen by chance, and parents are usually of normal height. If both parents are unaffected, the chance for another child to have achondroplasia is very low (less than 1%). If one parent has achondroplasia, each pregnancy has a 50% chance of being affected. Each family should consult a genetic specialist before planning next pregnancy.

Patient support group

Below organization is dedicated to supporting and empowering people with dwarfism

1. The Little People of India (Mumbai): https://www.facebook.com/littlepeopleindia/
2. ORDI: https://ordindia.in/

Credit: Dr. Shubha Phadke (Medical Geneticist)
Ms. Yashavi Gupta (Genetic Counsellor)