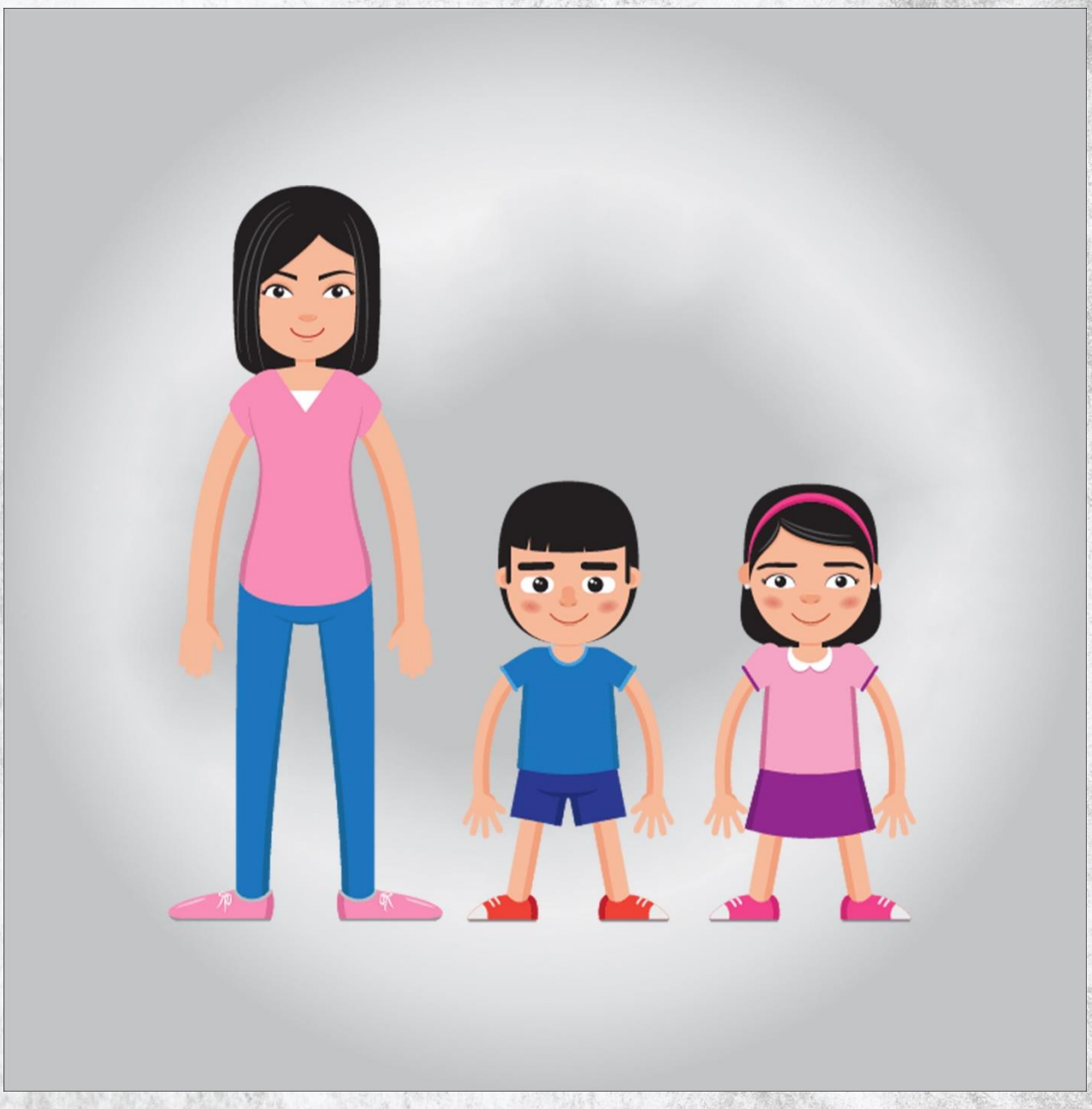

My Health Digest

Achondroplasia

Achondroplasia is a bone growth disorder that causes disproportionate dwarfismcaused by a genetic condition affecting a protein in the body called the fibroblast growth factor receptor.

It's the most common type of disproportionate dwarfism.

People with achondroplasia are short in stature with a normal sized torso and short limbs.

Causes:

During early fetal development, the skeleton is made up of cartilage. Normally, most cartilage eventually converts to bone. However, if the baby has achondroplasia, a lot of the cartilage doesn't convert to bone. This is caused by mutations in the FGFR3 gene. The FGFR3 gene instructs your body to make a protein necessary for bone growth and maintenance. Mutations in the FGFR3 gene cause the protein to be overactive. This interferes with normal skeletal development.

Symptoms:

- Bones are shortened (thigh, upper arm).
- Short hands and feet.
- Large separation between third and fourth fingers.
- Short stature.
- A large head with a prominent forehead and a flat nose
- Delayed development in infants (sitting, crawling, and walking).

Diagnosis:

Achondroplasia can be diagnosed before or after birth.

Before birth: by fetal ultrasound

After birth: by complete medical history and physical examination.

DNA testing is now available before birth to confirm fetal ultrasound findings for parents who are at increased risk of having a child with achondroplasia.

Treatment:

There is no treatment or cure for achondroplasia other than managing symptoms. Monitoring height, weight and head circumference is recommended during early diagnosis in infants to track growth progress.

Is achondroplasia inherited?

In more than 80 percent of cases, achondroplasia isn't inherited, but if one parent has the condition, the child has a 50 percent chance of getting it.