Achondroplasia

Patient Information Series – What you should know, what you should ask.

What is achondroplasia?

Achondroplasia is a genetic condition related to abnormal growth of the long bones in the body. In this condition, there is stunted growth of the long bones of the arms and legs that results in significantly short stature. Other features of the condition include an enlarged head, a flattened nose, abnormal curvature of the back and spine, and bowing of the legs.

How does achondroplasia happen?

Achondroplasia is the result of a genetic mutation that impacts the growth of long bones in the body. The affected gene is the fibroblast growth receptor factor 3 (FGFR3) gene. It is a rare condition that can happen in about 1 in 20,000 to 30,000 live births. While sometimes the mutations are passed along in certain families when one or both parents are affected by achondroplasia, most of the mutations are spontaneous and a result of genetic accidents and not due to factors that can be controlled.

When is it detected?

Achondroplasia is typically detected during the third trimester of pregnancy when the long bones fail to demonstrate adequate serial growth or it may be detected postnatally, The most common finding that is first noticed on ultrasound are shortened arm and leg bones in the fetus.

Should I have more tests done?

Many patients will choose to have more tests done to know more about the condition of the fetus. Tests to ask about include:

- A **fetal ultrasound; this** is typically done in all pregnancies and is part of routine prenatal care to look at the full anatomy of the fetus. This technology uses sound waves to identify structures without posing any risks of radiation to the mother or fetus.
- An **amniocentesis**; this is done by removing a small amount of amniotic fluid surrounding the fetus with a needle and syringe. This fluid contains cells and genetic material from the fetus and may be tested to see if the genetic mutation associated with achondroplasia is present for confirmatory testing.

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What are the things to watch for during the pregnancy?

Patients carrying fetuses affected by achondroplasia tend to have normal pregnancies without complications. Cesarean delivery may be recommended if your baby has achondroplasia and has an especially enlarged head that may not be compatible with a vaginal delivery.

What does it mean for my baby after it is born?

Most infants with achondroplasia do well. Limited pharmacologic treatment options exist and can be discussed with a pediatrician. Some children with achondroplasia have a minor motor delay that can be treated with physical therapy. Some lifestyle modifications and accommodations can be made for patients with achondroplasia to assist them given their short stature. Occupational therapy may also be helpful. Some possible complications of achondroplasia include recurrent ear infections, sleep apnea, and obesity, which can be treated as needed.

Will it happen again?

The risk of achondroplasia recurring in a future pregnancy is low given that most cases are due to a spontaneous gene mutation. If one or both parents have a known diagnosis of achondroplasia, there is a 50% chance that each child will be affected by achondroplasia.

What other questions should I ask?

- How often will I have ultrasound examinations done?
- Can diagnostic testing be performed?
- Where and when should I deliver?
- Is a cesarean delivery necessary?
- Where will the baby receive the best care after it is born?
- Can I meet the team of doctors that will be assisting my baby when it is born in advance?

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