MY ACHON JOURNEY
Pathways to Navigating Achondroplasia

This guide is not intended to serve as medical advice or substitute for consultation with a US health care professional. Always talk to your doctor if you have any questions or concerns.

Questions to ask about prenatal testing

If your unborn baby has shown signs of achondroplasia on an ultrasound exam and/or there is a family history of achondroplasia, then your doctor may advise you to meet with a genetic counselor to talk about prenatal genetic testing. Deciding whether to undergo prenatal testing is a personal decision that can be difficult. Below are some questions that you may consider asking your health care provider to help you determine whether prenatal genetic testing is the right decision for you and your family.

1. What are the pros and cons of doing genetic testing before versus after birth?
2. Should I get prenatal testing even though achondroplasia does not run in our family?
3. What are the potential risks of genetic testing procedures?
4. How will a diagnosis of achondroplasia affect my delivery?
5. Will my insurance cover genetic testing? Will I need to pay anything out of pocket?
6. Where and when will the procedures take place? Can I bring a support person?
7. Is the procedure painful? Will it hurt me or the baby?
8. What does the DNA test look for? Will we be testing for other conditions, too (besides achondroplasia)?
9. How long will it take to get the test results? How (and by whom) will I be informed about the results?
10. Is it still possible that my child has achondroplasia, even if the test comes back negative?
11. What are the next steps if the test gives a positive result? What about if the test is negative?

Notes: