

How to Talk with Your Healthcare Provider About Genetic Testing

If you or a loved one are considering genetic testing, we have developed questions to ask your provider when you request testing as well as when you receive the results of testing.

1. What to Ask Your Provider When Genetic Testing Hasn't Been Mentioned

- Would you recommend genetic testing for me/my family member?
 - Why, or why not?
 - If genetic testing is not ordered now, is it something that can be considered later?
- I learned that the National Society of Genetic Counselors and American Epilepsy Society are recommending exome or genome sequencing for unexplained epilepsy and other professional organizations recommend it for other neurodevelopmental conditions. Are you familiar with these guideline?
- It sounds like the practice guideline recommends genome or exome as the best test to start with, which would you recommend for me/my family member?
 - If the provider is recommending another test, ask what benefits they see to starting with that test and if there is an option to start with the recommended test.
- What test/s can your practice access?
 - If your provider cannot access genetic testing, consider asking to be referred to a genetics provider or a level 4 epilepsy center for testing.
 - Visit the REN website to find a genetic counselor or a level 4 epilepsy center.

2. How to Ask About Genetic Testing and What To Expect

- What we can learn from this test?
 - What are some of the potential benefits of the test?
 - Will it lead to any changes in treatment plan?
 - What are some limitations of the test?
 - Are there any risks to be aware of?
- What are the costs and is the test covered by insurance?
- How long will it take to get results? How will I be informed?
- Will I be able to access genetic test results in my electronic medical record?
- If this genetic test does not provide us with an answer, what would be the next step?
- Thank you so much for all of this information. I feel like it might be helpful for me to talk with a genetic counselor. Can you to refer me to someone?

3. How to Talk with your Provider about Genetic Test Results

- What is the name of the genetic test(s)?
- Was it exome or genome sequencing? A multi-gene panel? Some other test?
- What is the name and contact information for the laboratory that performed the genetic test? (This may be important at a later time)
- Why did I /my family member have this genetic test?
- When was the test completed?
- What was the result(s) of the genetic testing? What gene(s) were involved?
- Were any variants identified? How was the variant/s classified by the laboratory?

4. If Results Were “Positive” or Showed a “Variant of Uncertain Significance (VUS),” Ask:

- Do these results explain why I or my family member has epilepsy or seizures?
- Do these results explain any other medical conditions?
- Do these results affect my or my family member’s risk for another medical condition in the future?
- Do these results change my or my family member’s treatment plan?
- Did I or my family member inherit the variant from a parent?
- Are any other family members at risk of developing epilepsy or another condition?
- If the testing showed a VUS and was done at least one year ago, could you ask the testing lab if they could re-analyze the VUS?

5. If the Results were “Negative” or “Normal”, Ask:

- Are there any further genetic testing options available?
- Will our insurance company cover this additional testing?
- Is it possible for the laboratory to re-analyze the data from the testing?

6. No Matter the Results, Ask:

- Could you send me a copy of the genetic testing report(s)?
- Is there an epilepsy genetics specialist that may be able to help further?

Learn more at rareepilepsynetwork.org/genetic-testing | rareepilepsynetwork.org/info@rareepilepsynetwork.org