

What to Expect After Genetic Testing

Understanding and managing your child's medical testing can be challenging. This fact sheet covers information on how to prepare for your child's genetic testing appointment. For more information on genetic testing, see our Fact Sheets: ***Genetic Testing Basics*** and ***Preparation for Genetic Testing***.

Understanding Your Child's Genetic Testing Results

Genetic testing can be a complicated process for families, and genetic testing results can often be difficult to understand.

Genetic testing is sometimes referred to as a "diagnostic odyssey" due to its complexity. The scientific understanding of genes is continually changing and improving, and future or follow-up genetic testing can sometimes be needed for proper diagnosis as medical understanding of certain genes or genetic conditions improves. Because genetic testing and diagnosis can be a long journey for some children and families, it's important that your Geneticist and Genetics Counselor are supportive and understanding.

Remember that the Genetics Counselor is educated in both genetics and counseling, and they will likely include your child in the conversation to the extent that they are able and within your family's comfort level. The Genetics Counselor is trained to help explain complex medical terms to your family in a way that you can understand, even if you have no previous medical knowledge or experience. They are also trained to provide your family with emotional support during this often complex and complicated process.

Understanding a Negative Genetic Testing Result

If your child has a negative finding (no result found), your family will likely be contacted by your Geneticist, Genetics Counselor, or a nurse in your genetics provider's office to let you know the results of testing.

Negative genetic results can lead to a wide range of emotions for families. Some families are relieved to find that their child does not have a suspected genetic condition while other families are frustrated to not receive information on or answers about their child's medical status that may have helped guide and plan treatment options moving forward.

If no results are found, it may not be clear what your next steps forward should be. Your Geneticist may not schedule a follow-up appointment when no results are found; however, when this happens, you may request a follow-up appointment if you feel it would be most beneficial for your child. You may also discuss with your provider if your child is eligible for reanalysis in the future or if more in-depth genetic testing may help your child get properly diagnosed.

Understanding a Positive or Uncertain Genetic Testing Result

If your child has a positive finding (result found) or an uncertain finding (result found, but limited information available on its impact), your family will likely be contacted by your Geneticist or Genetic Counselor, and you may have an appointment with either or both providers for "genetics counseling" to discuss your child's results, diagnosis, and treatment plan.

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During the post-test counseling, the providers may discuss:

- What specific genetic result was found.
- If there is a known diagnosis attached to the finding.
- What research and information are available for this specific genetic diagnosis.
- If the findings impact your child's treatment plan.
- If anyone else in the family may need to consider testing for this diagnosis or to find out if they are carriers for the specific genetic change.
- If further or more in-depth testing is needed.
- What referrals that may be beneficial to your child's care moving forward.

It may also be helpful to come prepared with questions for your genetic providers to ensure that you fully understand the information being given to your family.

Even with a genetic diagnosis, information is sometimes limited and answers may not be readily available, which can lead to feelings of frustration for families. It's also important to remember that even with a diagnosis, every child is different and it can be difficult for providers to make predictions on how your child will be impacted by their genetic finding.

Genetic Testing Reanalysis

Because the scientific understanding of genes is continually changing and improving, some types of genetic testing allow for 1 free reanalysis to be done at a later time. This follow-up testing is typically done between 1-5 years after the initial genetics testing is complete; however, you should talk with your genetic providers to discuss the timeline that would be best for your child if they are eligible for this type of reanalysis.

Communicating With Your Providers

Remember, it is so important to communicate with your provider and advocate for your child before, during, and after the genetic testing process. Your child's Geneticist and Genetics Counselor are a team that should help you through your child's genetics testing process. Families should not be afraid to ask questions, ask for clarification, or ask for additional resources and information from their genetics providers. Many families have a wide range of emotions during the genetics testing process, including feelings of frustration and grief. Genetics counselors are trained to not only help families through the genetic testing process, but also through the feelings that come along with the process.

Where to find more information

For more information about genetics testing and diagnosis, see our other genetic testing fact sheets or the following resources:

- [Genetics Counseling](#) (Kids Health, 2021)
- [Understanding Genetic Test Results](#) (Mayo Clinic, 2022)
- [Genetic Disease Foundation: GDF](#) (GDF, 2022)
- [National Organization for Rare Disorders: NORD](#) (NORD, 2022)
- [The Journey Through Diagnosis](#) (Midwest Genetics Network, 2022)

If you have additional questions about the genetic testing process or would like help finding more information, contact Indiana Family to Family at 1-844-323-4636 or inf2f.org

Programs and systems change often. It is important to ensure that you are using the most current information. This fact sheet was updated July 2022. Please check <https://www.inf2f.org/fact-sheets.html> for the most recent edition.
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