

Genetic Testing for Cancer Predisposition in Childhood

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Learning Objectives

After reviewing this Guide you will be able to:

- Understand why genetic testing may be recommended for your child
- Describe the possible results of genetic testing and what each result may mean for your child
- Identify considerations for genetic testing decision-making

Introduction

In the past, very little was known about why children get cancer. Advances in science have revealed that some pediatric cancers may be associated with underlying genetic conditions, most of which are undiscovered until an individual undergoes genetic testing. You may wonder why your providers are recommending genetic testing when you already know your child has cancer. Therefore, it's important to understand how the results from genetic testing may be used.

Why Genetic Testing Might be Recommended for Your Child

Your child's care team may consider and recommend one or more types of testing:

- Somatic testing (also called tumor testing) looks only at genetic changes within the cancer cells and not throughout the body. Somatic testing can help guide diagnosis and, in some cases, treatment—but how useful it is depends on the type of cancer. For some cancers, it's a key part of determining diagnosis and treatment, while for other cancers testing may not provide as much useful information. Ask your care team what tumor sequencing might offer for your child's specific cancer type.

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- Germline genetic testing (also called germline sequencing) looks for genetic changes in the cells of your child's body that could raise their risk of developing another cancer. You might be offered different levels of testing:
 - A single gene test, if your family has a known history of a specific cancer predisposition syndrome; if there is a known genetic variant in the family, or if your child exhibits certain physical features
 - A small panel of genes, chosen based on your child's type of cancer, physical features noted during an exam, or family history
 - A larger panel, or even whole genome/whole exome sequencing, which looks at many more genes at once, including ones linked to a wide range of childhood and adult cancers

The larger the germline panel, the more likely it is that the tests will find something unrelated to your child's current cancer. For example, testing might reveal a gene change linked to a higher risk of breast or colon cancer later in adulthood—even if that has nothing to do with the cancer your child has now.

Germline test results usually fall into one of three categories:

- Negative—No genetic changes were found that are known to raise cancer risk
- Positive—A genetic change was found that is known to raise cancer risk
- Uncertain/Variant of Uncertain Significance (VUS)—A genetic change was found, but it is not yet clear whether it raises the risk of cancer or not

Over time, as researchers learn more, VUS results are often being reclassified as either negative or positive. Most turn out to be negative, but this can take many years to determine. Depending on how much is currently known, a VUS may be treated like a negative result (VUS-low), monitored over time (VUS-mid), or treated more like a positive result, with possible changes to your child's care (VUS-high).

Sometimes doctors do somatic and germline testing at the same time. Testing the cancer cells (somatic) and the rest of the body (germline) at the same time can have benefits, but is not required. Some parents prefer to talk through germline testing first — with a genetic counselor or family member—before deciding. In that case, tumor testing can be done on its own.

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Potential Benefits of Understanding Genetic Predisposition

Many cancer predisposition syndromes come with their own set of recommendations for managing risk. These might include:

- Regular checkups (scans, bloodwork, physical exams) to catch new cancers early, when they're most treatable
- Visiting specialists who monitor organs that may be at higher risk of developing cancer
- Preventative steps to lower the risk of developing cancer (like extra sun protection, or considering surgery to remove a body part) that may be more prone to cancer

Many parents say that learning the genetic cause behind their child's cancer brings some peace of mind; it gives them a proactive plan for monitoring and reducing future risk. Identifying a cancer predisposition can also lead to genetic testing for other family members, so parents, siblings, or other relatives can learn about their own risk too. For parents who worry about being blindsided by another serious diagnosis, having this information can offer real reassurance.

Potential Downsides to Consider

Germline genetic testing isn't without drawbacks, and it's worth talking these through with your care team:

- Not every genetic variant linked to cancer risk has clear guidelines yet for monitoring or treatment. The findings may add uncertainty rather than resolving it.
- Larger testing panels may reveal risks for cancers that wouldn't show up until adulthood, with no clear next steps recommended until then. Some families choose to hold off on testing for these genes until their child is older.
- Test results can come back negative or uncertain (VUS), which may leave you without a clear answer for why your child developed cancer.
- If a result comes back uncertain, there's no set timeline for when or if the variant is updated. It is important to stay in touch with your care team who ordered testing.
- Some parents think about how results might affect health or life insurance.
- Testing your child can sometimes reveal genetic information about you, the parent, as well. This may or may not be desirable, or tolerable.

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Questions to Ask

As you think through whether to move forward with germline testing, it may help to consider how comfortable you are with uncertainty. Consider how you will feel if Uncertain (VUS) or Positive results are found, and what kind of information you actually want to know. Some questions worth asking a genetic counselor or oncologist:

- How many genes are included in this test?
- What could a result for each of these genes actually mean for us?
- Is there a panel option specific to my child's cancer type?
- Will this include genes that wouldn't affect my child until adulthood?
- Will uncertain (VUS) results be shared with us?
- If we get an uncertain result, who will tell us if new research findings are discovered? How will this impact follow-up or monitoring?
- What should we know about genetic testing for siblings?
- Do we need to test right now, or could results still shape my child's treatment plan later?
- Can I talk to a genetic counselor before deciding?
- Who will support us after we find out the results?



Conclusion

If your child's care team is recommending genetic testing, it likely means that they believe that the results could have a benefit to your child's health now or later. But as with other recommendations, the choice to move forward is yours. Asking questions about genetic testing, your options, and what the implications of the results are for your child will help you make a more informed decision. Then, if you do consent to genetic testing, you feel more prepared to understand the results.

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