Genetic Testing for Celiac Disease

Nearly all people who have celiac disease carry a gene that predisposes them to develop the disease. If a child’s first-degree relative has celiac disease, genetic testing can help figure out if a child can possibly develop celiac disease in the future. In this handout, you will learn more about genetic testing for celiac disease and whether your patient should have testing for the disease.

CAN CELIAC DISEASE BE PASSED ON TO CHILDREN?

Yes, celiac disease can be passed on to children if a first-degree relative (parent or sibling) carries a gene for the disease. But, carrying a gene doesn’t mean a child will definitely have celiac disease. It means he/she is at risk of developing celiac disease.

WHAT ARE THE GENES FOR CELIAC DISEASE?

The two genes for celiac disease are human leukocyte antigen (HLA) DQ2 and DQ8. Most people who have celiac disease have at least 1 of these genes. In very rare cases, a person won’t have these genes, but might develop celiac disease anyway.

HOW DO I DO GENETIC TESTING?

You can test a child for the genes through a blood test.

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HOW CAN GENETIC TESTING FOR CELIAC DISEASE HELP A CHILD?

Genetic testing can help a child by figuring out if he/she:

- Is at risk of developing celiac disease at any point in his/her lifetime
- If he/she carries the genes for celiac disease, which could be passed to his/her children in the future

WHAT DO THE TEST RESULTS MEAN?

- If the test results are positive for HLA DQ2 or DQ8, a child is at risk of developing celiac disease. It doesn’t mean he/she will definitely have celiac disease and the chance of developing celiac disease is still low. But, he/she should have regular blood tests to see if/she has blood markers (signs in the blood) for celiac disease.
- If the test results are negative for HLA DQ2 or DQ8, a child does not carry the genes for celiac disease. He/she cannot develop celiac disease or pass it on to his/her children.

Sometimes, the test results can be hard to interpret. You should talk with a gastroenterologist who specializes in celiac disease if you have questions about the results. He/she can review results if you have a pediatric patient who has been genetically tested for celiac disease.

IS GENETIC TESTING COVERED BY INSURANCE?

In many cases, no, genetic testing for celiac disease is not covered by insurance. The testing can be expensive. If you want your patient to have genetic testing for celiac disease, tell the parents or family to call their insurance company to ask about the cost and coverage of the test.

Only 3 out of every 100 people who carry a gene for celiac disease will go on to develop the disease.