BOSTON MEDICAL CENTER

INTRODUCTION TO GENETIC TESTING

How It Works and Who Should Consider Testing

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WHAT IS GENETIC TESTING?

- Genes are the instructions that tell our bodies how to function. We have over 20,000 genes and each one has a specific function. When a gene is not working properly, it can cause the body to develop or function differently than expected.
- Genetic testing is a way to look at the code of genes related to a specific condition a person is experiencing to see if those genes are working or not working properly. Testing can be done with a blood, saliva, or cheek cell sample.
- If the genetic code has a difference that makes the gene not work properly, we say that it has a 'pathogenic variant' or a 'mutation'.

WHAT IS GENETIC TESTING USED FOR?

- If genetic testing finds an abnormality in a gene relevant to a person's condition, it might help explain why that person developed the condition they have.
- This genetic result might help determine what specialists someone should see or suggest changes in medical management or screening. For example, if a person with intellectual disability is diagnosed with a genetic condition that affects intellect and heart function, it can be recommended that the person be seen by a heart doctor (cardiologist).

WHO SHOULD CONSIDER GENETIC TESTING?

Genetic testing may be recommended due to someone's personal or family history of a condition. One might consider genetic testing if they or a family member has seizures, autism, or intellectual disability for example. One might also consider genetic testing if they or a family member is seeking explanations for medical issues affecting the heart, vision, hearing, kidneys, lungs, or endocrine system.

TYPES OF RESULTS

There are four possible types genetic testing results:

- **Positive:** This means a "pathogenic variant" or a "mutation" was found in a specific gene that explains a person's condition. The gene is not working properly.
    - Other family members may have this abnormality, or the genetic abnormality could be new to the person who had genetic testing. Further testing may be recommended for family members, such as parents, siblings, and children.
    - The person may be referred to see specialists or change their current care plan.
    - There may be a support group to connect with for individuals diagnosed with the same genetic condition.
- **Negative:** This means with our current understanding of genetics and the specific genetic testing done, no abnormality was found. There is still a possibility that a person's symptoms could be genetic. Further testing may be available in the future.
- **Uncertain:** This means a variation in the genetic code was found, but there is not enough evidence to determine if the variant causes a health problem. People with uncertain variants should check back in with the provider that ordered the test over time for updates.
- **Unexpected:** Genetic testing can occasionally reveal that a person has a condition they did not realize. Also, with family testing, genetic testing can occasionally reveal that family members are related in an unexpected way.

GENETIC DISCRIMINATION

The Genetic Information Nondiscrimination Act of 2008 (GINA) is a federal law that protects individuals from genetic discrimination for most health insurance policies and employment status; the cost of medical insurance premiums cannot be raised and employers cannot change your employment status based on genetic testing results. GINA does not protect those with medical insurance through a small business and for military personnel, VA patients, federal employees, or Indian Health Services. Also, GINA does not protect from increased cost or denial of life insurance coverage, disability insurance, and long-term care insurance; this aspect is more concerning to individuals who are mildly affected or who do not yet have symptoms at the time of testing, but are found through testing to have an increased risk for a given condition such as cancer or heart disease. Visit this website for more information about GINA: http://ginahelp.org.

I WOULD LIKE TO DISCUSS GENETIC TESTING MORE IN DETAIL WITH A GENETIC SPECIALIST - WHAT DO I DO NEXT?

- Talk to a member of your healthcare team and let them know that you are interested in seeing a genetics professional.
- A referral is needed to see a member of the Boston Medical Center's Pediatrics/Adult Genetics Team. If your healthcare provider has questions on how to refer, they can call 617-414-4841 or fax a referral to 617-638-6756. Local genetics providers can also be located by zip codes at the following links:
    - *https://findageneticcounselor.nsgc.org*
    - *https://clinics.acmg.net/*