Genetic Testing: Consent

JOHNS HOPKINS GENOMICS

WHAT IS INFORMED CONSENT?

Before genetic testing is ordered for a patient, it is important that they understand the test, including the benefits and limitations and the possible consequences that can come with results. The process of educating a person about this information and obtaining permission to perform the test is called informed consent. "Informed" means that a person has enough information in order to make an educated decision on the matter.

Informed consent can only be obtained from adults who are able to make medical decisions. For individuals who are unable to make their own medical decisions (such as children and individuals with an impaired mental status), informed consent can be obtained from a parent, guardian, or legal representative of that individual.

TOPICS TO INCLUDE:

- Description of the test (purpose, condition being tested, etc.)
- How a sample will be obtained
- Description of the possible results and what they mean
- Physical or emotional risks associated
- Whether the sample/results can be used for research (if applicable)
- Whether the results might provide information about family members' risks
- How results will be disclosed

BENEFITS OF GENETIC TESTING

Genetic testing can have benefits whether the results are positive or negative. These results can help provide a patient with a sense of relief from uncertainty. They can also aid patients and providers in making informed decisions about managing one's healthcare. A positive result has the potential to direct a person towards available prevention, monitoring, and treatment options that may be available. Depending on the disorder, these results can also help an individual make decisions about having children. In some cases, a negative result has the potential to eliminate unnecessary checkups and screening tests.

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RISKS OF GENETIC TESTING

Some of the risks associated with genetic testing involve emotional, social, or financial consequences that may arise. Individuals may feel emotions such as anger, guilt, or anxiety relating to their results. Sometimes genetic test results can cause tension within a family if they reveal information about other family members in addition to the person who was tested originally. Genetic discrimination can also be a concern for individuals in the case of employment or insurance (discussed below).

GINA

Genetic discrimination is when an individual is treated differently by their employer or insurance company based on a genetic test result that causes an increased risk of an inherited disorder. This is a common fear among individuals considering genetic testing.

A law called the Genetic Information Nondiscrimination Act (GINA) is designed to protect people from this type of discrimination. This law makes it illegal for health insurance companies to use genetic information/results to make decisions about a person's insurance eligibility or coverage. It also makes it illegal for employers to use genetic information/results when making decisions about terms of employment (ex. hiring).

It is important to know that GINA does not protect individuals from genetic discrimination in all cases. For example, it does not cover people in the U.S. military and does not apply to employers with fewer than 15 employees.

POSSIBLE RESULTS

Positive

A positive result means that a change or "variant" was found in the gene(s) tested that may confirm a diagnosis or identify an increased risk of developing a certain disease. These variants are known to contribute to disease.

Negative

A negative result means that a change or "variant" contributing to disease was not identified in the genes tested. A negative result does not mean that there is not a genetic cause for the individual's health concerns.

Uncertain

A variant of uncertain clinical significance (VUS) is a change found in the gene(s) tested in which the lab does not have enough information to determine if it is contributing to disease. Some changes in our DNA are harmless, like the changes that make our eye or hair color different from those around us. A doctor or patient can check back with the lab at a later time to see if more information is known about this variant.

Unexpected Results

In some cases, a test result may uncover information that you were not expecting. Genetic testing analyzes DNA, which people share with their blood relatives. Because of this, sometimes labs can detect things like non-paternity and unexpected familial relationships.

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