Information about genetic testing

Genetic testing may have been discussed with you today. Here is some information about this testing:

What is genetic testing?

- Genes are made up of DNA and provide the instructions for our body to function.
- We are all born with genetic differences, called variants, that make us different from the next person. Sometimes, a person can have a variant in their DNA (also called a "mutation") that increases their chance to develop a medical condition.
- Genetic testing is done on a blood or saliva sample and works by looking for these variants in the DNA.

Why have genetic testing? Genetic testing may:

- Help your doctors better understand your diagnosis or provide targeted treatment options or better ways to monitor your health.
- Give family members information about their chances to develop a medical condition.

What is being tested?

- A panel including genes associated with your health condition.
- This includes testing of genes that are related to your medical condition. Sometimes, the lab can find a change in a gene that causes your known health condition, but also other health conditions.

The results of genetic testing could be:

- **Positive:** means that a genetic variant was found. This can increase your risk for a medical condition or help to determine a course of treatment or method to monitor your health.
- **Negative:** means that no genetic variant that affects your health was found. This can also help determine treatment options and how you are monitored.
- **Uncertain:** means that a genetic variant was found that the lab is not sure causes an increased chance to have a medical condition.

Risks and limits of genetic testing:

- There is a small risk for errors, like errors in processing a sample and technical problems.
- If a genetic diagnosis is not found, we cannot completely rule it out. You might have a condition that was not tested for, not discovered yet, or that cannot be found with current technology.

Privacy and Protections for genetic test results:

- Genetic test results are protected health information. Penn Medicine privacy practices apply to genetic test results.
- The Affordable Care Act (ACA) does not allow the use of pre-existing conditions (like cancer or heart disease), to deny health insurance coverage or raise premiums. If you would like to learn more, visit About the ACA | HHS.gov.
- The Genetic Information Non-discrimination Act (GINA) is another law with more protections.
    - GINA does not allow the use of genetic information by employers and health insurance companies.
    - GINA does NOT apply to employers smaller than 15 employees. GINA does NOT apply for Life, Long-term Care, or Disability insurance.
    - If you would like to learn more, visit http://www.ginahelp.org

Cost of testing:

- Genetic testing is usually billed to your insurance, just like any other medical lab or procedure.
- In many cases insurance fully covers the test. If there is a cost, it is usually less than $100.
- You will be contacted by the lab if you owe more than $100. The lab's billing department will discuss payment options with you.

Testing process:

- The lab will ship a saliva kit to your home address with instructions and supplies.
- The office that ordered your testing will contact you to review the results.
- You may have the option to review these results in the patient portal before your healthcare provider has a chance to talk about them with you.
- You should still talk with your healthcare provider about the results, even if you see the results first. They will let you know if there are any next steps for your medical care, or for your family.