**Supplement C: Patient Education Sheet**

## What are Genes?

Genes are a biological material that provide instructions for how our bodies look and function. Genes are found in every cell of our bodies. Our genes are made up of a chemical called DNA (deoxyribonucleic acid).

## What are Gene Variants?

The DNA of genes are made up of four chemicals labeled with letters from the alphabet A, T, C, G. The letters need to be in a particular order for our genes to work properly. Gene variants are when the letters are out of order, added or missing. A gene variant is also called a gene mutation. Gene variants may or may not affect the function of the gene. There are certain types of gene variants that can lead to benign and cancerous tumors.

There are a few ways that gene variants can occur. Everyone is born with gene variants that are inherited from our parents. Gene variants can also occur by chance at conception. Variants that we inherit from our parents and variants that happen at conception can be passed on to children. We also can acquire gene variants after we are born over our lifetime from the environment we live in, our lifestyle, and as a natural part of aging. Acquired gene variants cannot be passed on to children.

## How do Gene Variants Lead to Tumors?

Tumors are caused by certain types of gene variants that are in one cell. These variants cause a cell to grow out of control and become a tumor. Tumors can be liquid tumors such as leukemia and solid tumors such as colon cancer.

## What is Tumor/Normal Whole Exome Gene Sequencing?

This is a test that looks for variants by determining the order or sequence of the DNA letters in your genes. The test looks for gene variants in your tumor and also for variants in over 150 cancer genes you were born with. The lab will then compare the variants in your tumor genes to the variants in the genes you were born with. It is important to determine the variants that are only in the tumor because these variants may help guide your treatment.

## What Kind of Sample is Needed to do Identify Gene Variants?

Tumor cells are used to test for tumor gene variants. Blood or saliva or skin cells are used to identify variants in genes you were born with.

## What are the Risks of this Test?

There may not be enough tumor to complete the test. To do this test, you must agree to also receive the results from the variants in the genes you were born with. Those results could have information about your health and/or your family's health which can be upsetting.

**What are the Limitations of this Test?**

Not all gene variants can be found with this test.

**What are the Benefits of this Test?**

Tumor gene variants may help guide your treatment. Variants in the genes you were born with may impact you and your family's healthcare options.

**What kinds of Results can I Expect?**

There are two separate results from this test. You will receive results from the test on your tumor genes and results from the test on the genes you were born with.

Results from the tumor gene test will include information about the variants that your doctors may be able to use to guide your treatment.

Results from the test evaluating the genes you were born with will include only variants in genes that increase the risk to develop tumors. These variants may impact you and your family's healthcare.

**When will my Results be Available?**

Your results will be available in about 1 month from the date that **both** your tumor and blood, saliva, or skin sample has arrived in the laboratory.

**Who will Give me my Results?**

One of your treating providers at the National Cancer Institute (NCI) will give you the results about the gene variants in your tumor and discuss your treatment options based on the findings.

A genetic healthcare provider at NCI will inform you if there are any variants in the genes you were born with.

**Will my Results be in my Electronic Medical Record?**

Yes, your results will be in your electronic medical record at NCI, which is private. The results can only be released with your consent.

**Are there other Resources on this Testing?**

Yes, the NCI has developed information about this kind of testing.
https://www.cancer.gov/about-cancer/treatment/types/precision-medicine/tumor-dna-sequencing

**What if I have More Questions?**

Call 240-760-7350 OR Email TumorNormalWES@mail.nih.gov