**Somatic Disease/Germline Comparator Sequencing**

You or your child will be having a genetic test called somatic disease/germline comparator sequencing. This test looks for changes in DNA that could cause or contribute to a
medical condition.

**About your genetics**

Our bodies are made up of billions of cells. Each cell contains DNA. DNA contains genetic information we inherit from parents. Small parts of DNA are called genes. Genes tell our body how to grow, develop and function. There are about 22,000 genes in human DNA. Our complete set of genetic material is called our genome.

Genetic changes in our DNA, called ***variants***, can have an impact on our health. Everyone is born with their own DNA changes that are found in every cell in the body. These are called ***germline variants***. Changes can also happen after birth and may only be found in certain cells in the body. These are called ***somatic*** or ***acquired variants***. Some DNA variants cause or contribute to diseases while other changes do not.

Most of the variants known to cause or contribute to disease are found in the exome portion of the DNA. The exome is the parts of all genes that tell our body how to work. In rare cases, a variant in DNA outside the exome can cause the disease.

**The test**

For this test, your healthcare provider will order exome sequencing (ES) or genome sequencing (GS). Exome sequencing and genome sequencing are types of genetic tests that look for variants in a person’s DNA that may cause or contribute to their medical condition.

* ES - tests for variants in the exome.
* GS - tests for variants in all genetic material (includes the exome and areas outside of the exome).

**How the test is done**

For this sequencing test, two samples are needed:

* One sample from the area of the body affected by the medical condition. Examples include a tumor or skin. Your clinicians will talk to you about the kind of sample needed.
* One sample from an area that is not affected. Examples include blood or saliva.

This test compares results from the two samples. This helps us find and understand DNA variants that may be related to the disease. By testing unaffected and affected areas at the same time, we can find both ***somatic*** and ***germline*** variants to give you and your healthcare provider more information about how to best care for you and your family.

**Test results**

Test results may give helpful information about:

|  |  |
| --- | --- |
| * your specific diagnosis
 | * the cause or part of the cause for your condition
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| * how your condition may progress
 | * the best way to manage or treat your condition
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There are different kinds of findings you can get from your test results:

* **Primary findings** refer to the variants found in your DNA related to your medical condition. There are three types of primary findings:
* Positive for disease association - Somatic or germline DNA variant that is known to cause or contribute to your current medical condition.
* No disease-associated findings - No DNA variants related to your medical condition were found.
* Uncertain findings - It is not known if the genetic variant causes or contributes to your medical condition.
* **Secondary findings** refer to germline variants found in your DNA that are NOT related to your current medical condition. You can choose if you want this information included in your ES or GS report. These changes could be important to know for these reasons:
* causing or contributing to other serious medical conditions
* giving information that may be important for family planning or future health choices

Your health care provider will talk to you about the test results. More genetic testing may or may not be recommended. If you have questions, ask your genetic counselor or the health care provider who ordered this test.