What you need to know about
Genetic Testing Panels
BEFORE you go to the laboratory

GENETIC TESTING PANELS
Genetic testing panels are ordered when many different genes cause the same or a similar medical condition. Testing a panel of genes can improve our ability to find the genetic cause for the medical condition in the family. There can be anywhere from a few genes to several hundred genes on a panel, depending on the panel and the condition being tested. A single blood test evaluates all of the genes at the same time. However, testing more genes does not always mean better information. A genetic testing panel is carefully selected by your Genetics provider based on your medical history and your family history. This handout includes details that may be helpful to know before you have testing.

GENES INCLUDED ON A TESTING PANEL
- Testing panels do not include every possible gene that could be related to your medical condition. We are still discovering which genes are related to medical conditions that run in families. There may be genes related to your condition that are not yet known or included on the genetic testing panels currently available.

- Panels can include genes that have only a moderate risk to cause medical problems.
  Not all genes have the same potential to cause medical problems. There may be some genes that are more likely to cause problems than other genes on the same panel.

- Testing panels may include genes that are not very well understood.
  Some genes have been reported in lots of families and have well-defined risks. However, some genes included on a panel may have very limited medical information available.

RESULTS FROM A TESTING PANEL
- You may get a negative result (no mutation detected), even when you have a genetic condition.
  Genetic testing panels do not detect all mutations and only test the genes on the panel. That means not all causes for a genetic condition can be detected with our current testing.

- Pathogenic (disease-causing) mutations are not all the same.
  There are many mutations that can interfere with the normal function of a gene. Some mutations have a greater impact and cause a higher risk for related medical problems.

- The more genes we test, the more likely we are to find a variant of uncertain significance.
  Genetic variants are differences in the DNA code. Genetic testing panels sometimes detect a genetic variant that is not well understood. This type of result is called a variant of uncertain or unknown significance (VUS or VOUS).

- It is possible to have more than one genetic variant and/or mutation detected.
VARIANTS OF UNCERTAIN SIGNIFICANCE

- Genetic changes (variants) do not always cause problems.
  Variants that are common are often known to be benign and not cause medical problems. Benign variants may not be reported by all laboratories.

- We do not know the disease-causing potential of a variant of uncertain significance.
  Variants of uncertain significance may or may not be benign. Most reported VUS are rare and may be unique to one family. In most cases there is very little information available to help clarify how a particular change in a gene will affect the way the gene works. That means we are unable to predict whether or not that variant poses a risk.

- Variants of uncertain significance DO NOT help guide a person’s medical care or treatment plans. This result does not help clarify your medical risk, so decisions about your medical care should continue to be made based on your personal medical history and your family history.

- Medical information about variants of uncertain significance can change.
  Some VUS eventually get reclassified as benign or disease-causing as we learn more about a gene or a particular variant. Families are encouraged to check back with Genetics from time to time.

More About:
GENETIC VARIANTS

Genetic variants are changes in the genetic code (DNA). Variants are classified as “benign”, “disease-causing (pathogenic)”, or “of uncertain significance” based on how likely they are to change the way a gene works. Genetic variants may also be called mutations, especially disease-causing variants.

Pathogenic or Disease-Causing Mutation
- Evidence indicates that the variant has the potential to cause medical problems
- Usually not found in the general population
- Confirms that the condition in the family is genetic and can be inherited. Family members can be tested

Variant of Unknown or Uncertain Significance (VUS or VoUS)
- There is not enough evidence to determine if the variant will cause medical problems
- Usually not found in the general population
- Testing family members may help to clarify the potential risk

Benign Variant (Polymorphism)
- Evidence shows that the variant does not cause disease
- Can usually be found in the general population
- There is no reason to test family members for benign variants