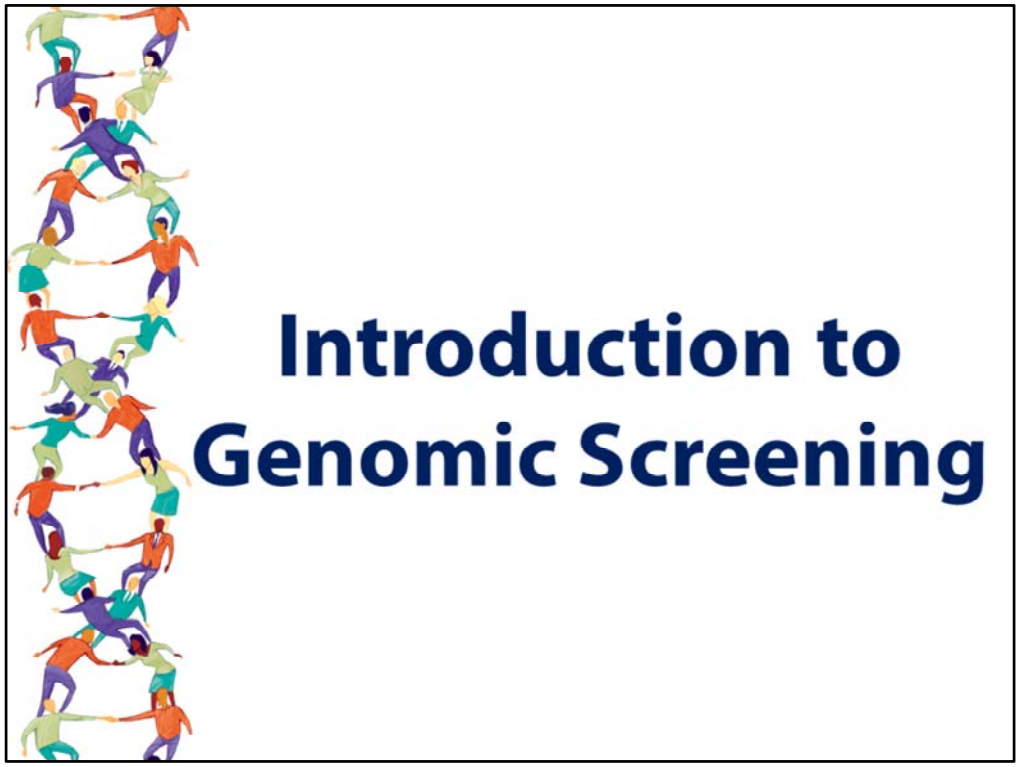


This information sheet will provide an introduction to genomic screening.

The words underlined are explained in more detail in the genomics terms sheet.

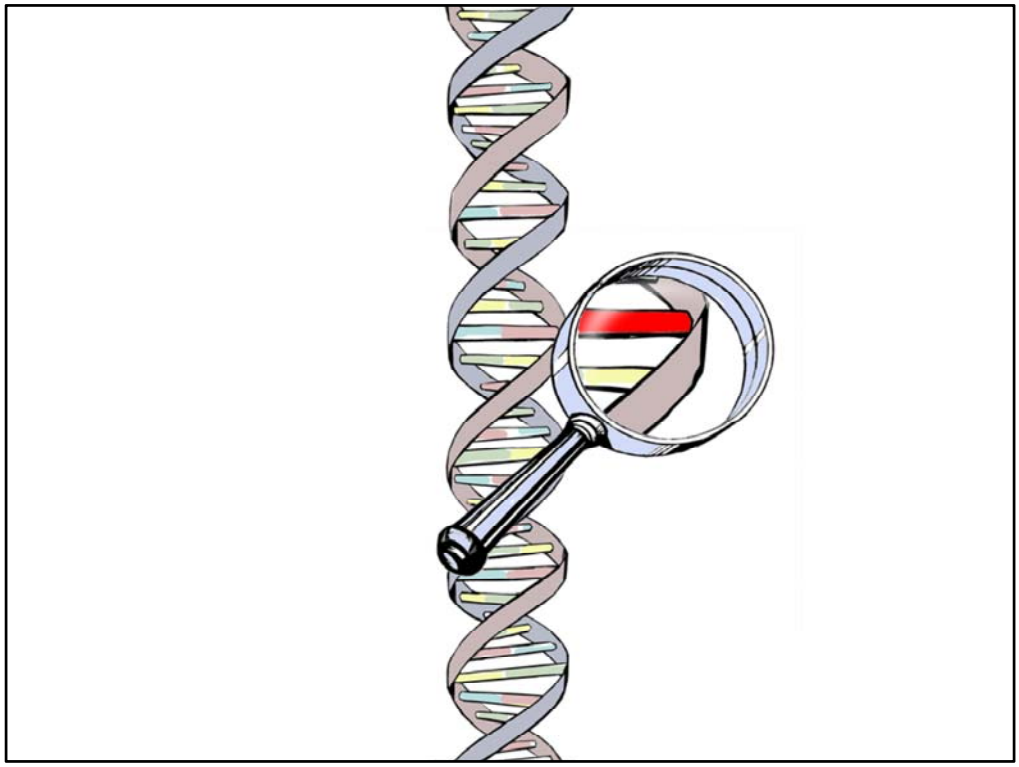
All people have variants or differences in their genes give them individual physical features.

Genetic Variant: A change in your gene that may or may not affect how your body functions.

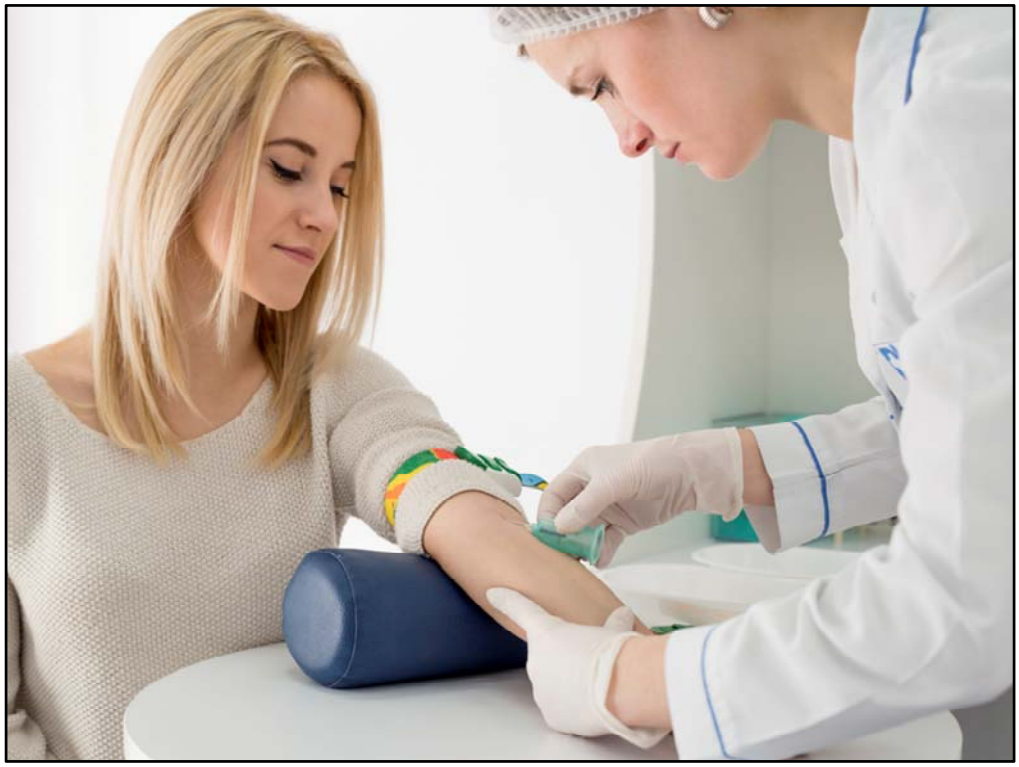
A genetic variant is a change in the gene that may or may NOT affect the function of the gene.

Genetic Mutation:
A change in your gene that causes it not to function properly and causes a genetic condition.

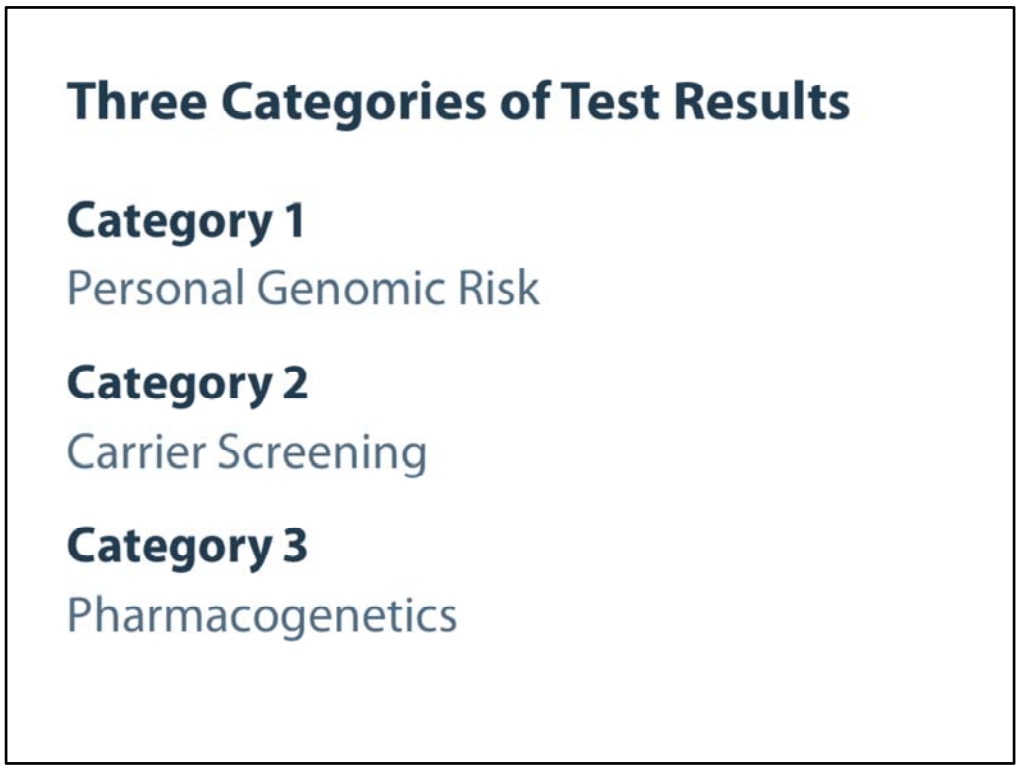
A mutation is a change in the gene that cause it not to function properly and cause a genetic condition.



Through genomic screening, it is possible for scientists and doctors to identify genetic variants and mutations in your genes that could be relevant to your health and the health of your family.



Genomic screening is done by collecting a small sample of blood that is mailed to a laboratory, where it is analyzed for specific genetic mutations.



If you choose to have genomic screening, there are three categories of test results that you will have the option to learn about.

Personal Genomic Risk

Genomic screening that tells you about your own personal risk of developing a genetic condition

The first category, Personal Genomic Risk, tells you about your own risk of developing a genetic condition. Genetic mutations identified in this category indicate that you have a higher risk of developing certain genetic conditions.

Carrier Screening

Genomic screening that provides information about the risk of your children or future children developing a condition

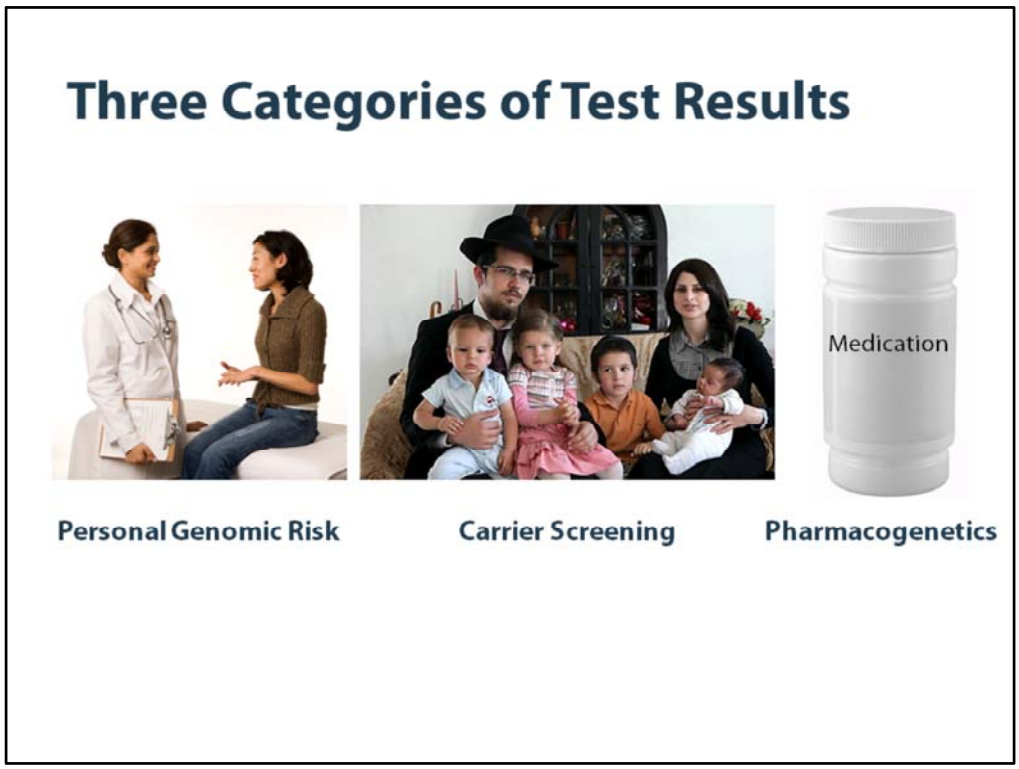
The second category is carrier screening. It provides information about your carrier status for genetic mutations that typically do not cause a condition in you but increase the chance that your children or future children could have a genetic condition.

Pharmcogenetics
Genetic variation that affects the way that your body processes certain medications

The third category can tell you about genetic variants that affect the way your body processes certain medications. This is called pharmacogenetics.

“Pharma” refers to medications and “genetics” refers to your genes.

These genetic variants do not cause disease, but rather they affect the way that your body breaks down or responds to certain medications.



These categories are all discussed in further detail in the videos and information sheets on Personal Genomic risk , Carrier Screening, and pharmacogenetics.

Again, if you choose to have genomic screening, you will have the opportunity to learn about results from all of these categories or only some of these categories. It is your choice.

We understand this is complex information. If you'd like to discuss this information with a genetic expert, please click on the option on the website

Email: **jw2500@columbia.edu**

Call: (212) 305-6987

The following Information Sheets or Videos may also be helpful

- Introduction to Genetics
- Personal Genomic Risk
- Carrier Screening
- Pharmacogenetics