Understanding your results:
Low penetrance alleles

CARRIER SCREENING identifies patients who may be at increased risk of having a child affected with a genetic disorder by looking at changes (called variants or mutations) in specific genes.

- The same DNA variant does not always cause disease or the same symptoms in every person who carries it.

PENETRANCE refers to the probability of a gene or trait being expressed among people with the same variant or set of variants.

- Complete penetrance: everyone who inherits an autosomal recessive condition and receives two copies of the disease-causing variants will have clinical symptoms of the disease.
- Incomplete or reduced penetrance: some people who inherit an autosomal recessive condition and receive two copies of the disease-causing variants will have clinical symptoms of the disease. Incomplete or reduced penetrance is a common phenomenon in genetics and can apply to dominant conditions as well.

EXPRESSIVITY refers to the set of possible signs and symptoms expressed in a particular condition.

- Variable expressivity occurs when people with the same genetic condition experience different symptoms and severity levels. For example, two people have a condition that causes headaches and joint pain. One may experience symptoms of joint pain while the other only experiences headaches. Many conditions have a wide range of symptoms and severity, and it is possible to have variable expressivity within the same family.

LOW-PENETRANCE ALLELES are specific genetic variants that are less likely to cause the signs or symptoms of a particular disease. When signs or symptoms are present, they often tend to be milder than other variants with higher penetrance in the same gene.

For conditions or variants with incomplete or reduced penetrance, it is not always possible to predict whether an individual who inherits these variants will have any signs or symptoms of the condition or how severe these symptoms may be. Other genetic and non-genetic factors including diet, lifestyle, and environmental exposures can impact the overall penetrance and expressivity of a variant in any one person.

Examples of conditions that are impacted by the presence of low-penetrance alleles are CFTR-related disorders, galactosemia, hereditary hemochromatosis, biotinidase deficiency, and alpha-1-antitrypsin deficiency.

It is important for carriers of a genetic disease to look at all of the information provided to see if there is any information regarding low-penetrance or impact on disease severity.

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