Variant Classification for Carrier Screening

Variant Classification

Variant classification is based on a Mendelian perspective and employs the recommended five-tier classification system recommended by the American College of Medical Genetics, ACMG.

A sequence change can be classified as:

Positive - Pathogenic Variant(s) Detected

This result means that a mutation or a genetic change that increases the risk of being a carrier for a particular disease was identified. This variant directly contributes to the risk of being a carrier. Additional evidence is not expected to alter the classification of this variant. The final report contains detailed risk information specific to the mutation(s). Follow up with physicians, healthcare providers and genetic counseling is recommended.

Variants(s) Likely to be Pathogenic (VLP) Detected

The result means that a mutation or a genetic change that is likely to increase the risk of being a carrier for a particular disease was identified. However current scientific evidence is insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of pathogenicity, but we cannot fully rule out the possibility that new evidence may demonstrate that this variant has little or no clinical significance. The final report contains detailed risk information specific to the mutation identified. Follow up with physicians, healthcare providers and genetic counseling is recommended.

Variant(s) of Unknown Significance (VUS) Detected

Variant(s) of Unknown Significance (VUS) are variants that do not fit into pathogenic, likely pathogenic, or benign classifications according to ACMG and/or other relevant Professional standards, or the criteria for pathogenic or likely pathogenic and benign are contradictory for the variant. There is not enough information at this time to support a more definitive classification of this variant on cancer risk.

Presumed Negative - Likely Benign and/or Benign Variant(s) Detected

The variant identified is not expected to have a major effect on disease; however the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion, but we cannot fully rule out the possibility that new evidence may demonstrate that this variant can contribute to an increased risk.

Negative - No Reportable Variant(s) Detected

The result means that no variant or genetic change known to be associated with or predicted to be associated with, based on current knowledge and test methods, disease(s) was identified. No known pathogenic variant(s) was identified. A benign variant does not cause disease.

Test Inconclusive or Test Failed

Test evaluation was inconclusive. Possible reasons are not enough DNA present in test sample, sample failed quality assurance measures. A new sample submission is recommended.