athena insight™

Family Insight Program

Collecting Family Information for Variant Classification

A Variant(s) of Uncertain Clinical Significance (VUS) has been identified in your patient.

What does a VUS mean?

When a variant's contribution to disease is unknown and cannot be identified as either benign or pathogenic, the genetic variant is reported as a VUS. When this occurs, Athena Insight™ performs an in-depth investigation to evaluate the likelihood that the VUS causes disease. The details of this investigation are included in your clinical result report.

What is the Family Insight Program?

We use numerous methods and techniques to gather information about VUS. One of these tools is the Family Insight Program, which allows for segregation analysis, or determining whether the VUS is tracking with the disease in the family. Through this program, we may offer to test one or more family members for the VUS identified in your patient. Testing is offered when a family study is likely to lead to a better interpretation of the VUS.

It is important to note, participation in the Family Insight Program does not guarantee VUS reclassification. Participation of family members in the Family Insight Program should also not replace genetic counseling and/or clinically indicated genetic testing.

Of note, incidental findings identified through this program will not be evaluated. Family members tested under this program will not be receiving a report.

What is the next step?

If your patient is interested in participating in our Family Insight Program, please visit AthenaDiagnostics.com to access the forms required to start this process. Upon receipt of completed information, we will notify you about whether or not your patient has been approved for the family studies program.

What happens if a VUS is not reclassified?

If the VUS is not reclassified as a result of the family testing, it is possible new information (external and internal) will become available in the future that may cause the VUS classification to change. Since we cannot predict if or when this reclassification will occur, it is important for your patient to remain in contact with your office to ensure they can also be notified of any change in the interpretation of the VUS.

What are the exclusions to this program?

Sometimes, a family study will not be informative for interpreting a VUS. In these cases, testing additional family members may not be offered. These situations may include, but are not limited to:

- The VUS has since been classified as benign, likely benign, likely pathogenic, or pathogenic
- The VUS is found at a high frequency in the general population
- A pathogenic or likely pathogenic variant(s) explains the phenotype of the patient, and an additional VUS is likely noncontributory
- Your patient's results are not consistent with the inheritance pattern associated with the gene(s) or phenotype(s) involved Some of examples of this may include, but are not limited to: a single variant identified in a recessive gene; a variant identified in a mitochondrial gene; a variant in a gene associated with both dominant and recessive inheritance; a gene associated with reduced penetrance
- The clinical presentation of your patient or family member(s) is not consistent with the reported phenotype associated with the gene(s) or phenotype(s) involved

A reminder

Medical management recommendations and reproductive decisions should not be based on the presence or absence of a VUS. Instead, recommendations should be made based on your patient's personal and/or family history. In some cases, additional genetic testing may be warranted.

If you have any questions about the Family Insight Program, please contact us at 1.800.394.4493 x 2031 or Genetics@AthenaDiagnostics.com, and ask to speak with a genetic counselor.

We are committed to providing the very best diagnostic tools and services to our customers. We hope you find our services exceed your expectations and provide the most clinically useful results to you and your patients. We look forward to hearing from you.

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