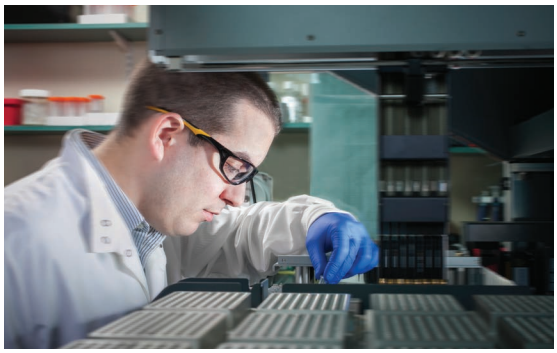
athena insight™

Family Insight Program

Collecting Family Information for Variant Classification



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

What does a VUS mean?

When a variant's contribution to disease is uncertain and cannot be identified as either benign or pathogenic, the genetic variant is reported as a VUS. In that case, 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 next step?

In order for us to interpret the meaning of the VUS in your patient, we are encouraging clinicians to participate in our Family Insight Program. This program is focused on offering free genetic testing in cases where genetic information is likely to lead to a better interpretation of a VUS which would be otherwise inconclusive without family member participation. Please note, this does not guarantee a variant interpretation change at the time of your testing. Please answer the following questions to help us assess if our Family Insight Program would be beneficial to you and your patient:

Is there a family history of the disease for which you are testing? ☐ Yes ☐ No

How many family members are available for testing?

Diagnosed Affected _____

Diagnosed Unaffected _____

Proband Accession # _____

Clinician Contact Information

Name _____

Phone _____

Email _____

If you would like to determine if your patient and their family members qualify for participation in our Family Insight Program, please contact one of our Genetic Counselors at genetics@athenadiagnostics.com or fax this form to them at 1-774-849-3207.

What are the exclusions to this program?

We will not be offering testing to family members if:

1) The VUS is determined to be inconsequential or benign because of its high frequency in the general population (based on internal data, dbSNP, public allele frequency databases, and published literature).
2) The variant is determined to be benign, predicted pathogenic, or pathogenic.
3) The VUS is associated with a specific inheritance pattern and further family genetic information will not clarify the significance of the VUS.

Incidental findings identified through this program will not be evaluated. Family members tested under this program will not be receiving a report.

What determines classification of a variant?

Athena Insight (AI) Scientists use the following criteria to classify variants as benign or pathogenic: frequency in the general population, co-occurrence with pathogenic mutations, functional studies, clinical presentation, and segregation of symptoms with disease in families. This information is collected from internal data, public databases and peer-reviewed literature.

Athena Diagnostics is 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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