

Family Insight Program

Collecting Family Information for Variant Classification



What is the goal of the Family Insight Program?

The goal of this program is to clarify whether a genetic finding is associated with disease or not. This helps patients and their healthcare providers know more about the cause of disease in a family.

What is a variant of uncertain clinical significance (VUS)?

Each person's genetic makeup is different from the next. These differences are called *variants* and are what make us unique. Most variants are normal findings and are harmless.

In some cases, a specific variant has not been seen often enough to know if it is associated with disease or not. We call this a *variant of uncertain clinical significance (VUS)*.

How could my participation contribute to learning more about the VUS my family member carries?

One way to learn more about a specific VUS is to see whether it pairs with disease in a family. Since one of your family members carries a VUS, we are interested to know more about their relatives' health history to determine if family testing is likely to be useful.

If I'm interested in participating, what are the next steps?

If you are interested in participating, you would sign a consent form and provide a blood sample. Your healthcare provider can arrange sample collection locally.

What kind of information might I get if I participate?

This testing may be able to clarify if the VUS in your relative is associated with disease or not. Identity testing to confirm biological relationships, including parentage, may be performed as a quality measure. If family testing **does** clarify the VUS, we will update your relative's laboratory report and share the new information with their healthcare provider. If family testing **does not** clarify the VUS, we would inform their healthcare provider and encourage that person to check in annually for updates. It is important to know that you will not be provided with any results. In addition, your information may be included in the updated report to your relative.

Is there a cost for this test?

There is no additional cost to participate in the Family Insight Program.

Patient Attestation of Informed Consent:

My signature below indicates that I have received information about the Family Insight Program, and that I have read and understood the material in this document. I have been given a full opportunity to ask questions that I may have about the testing procedure and related issues.

Signature of Patient

Date

Signature of Guardian

Date

For the Physician:

As the referring physician, I understand the benefits and limitations of this study. I attest to the fact that I have provided the patient with the information contained above and fully answered any questions. I believe that the patient understands the information and is voluntarily signing this informed consent. I understand that neither I, nor this family member, will get any results from this test.

Signature of Physician/Health Care Professional

Date

