



VUS CLARIFICATION SERVICE INFORMED CONSENT

This consent form includes important information about genetic testing through our VUS Clarification Service to help you decide if participating is right for you and your family. Please go through this form carefully with your healthcare provider or genetic counselor.

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 genetic variants are not associated with disease. Sometimes, we do not have enough information to know whether a variant causes disease (referred to as pathogenic or likely pathogenic) or does not cause disease (referred to as benign or likely benign). In this case, the variant is called a variant of uncertain significance (VUS).

What is the goal of the VUS Clarification Service?

The goal of this program is to clarify whether the genetic change(s) in your relative is associated with disease or not.

How could my sample help?

One way to learn whether a VUS is the cause of an individual's symptoms is to see if it is present in other family members with the same symptoms and is absent in family members without symptoms. Since one of your family members has a VUS, learning if you also have the VUS and about your health history may help us determine whether the VUS is disease-causing (likely pathogenic). Once testing is complete, your family member's report will be updated to indicate whether their VUS has been reclassified to disease-causing, based on the VUS Clarification Service analysis. Neither your results nor your health status will be shared with your family member or written on their report.

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

If you would like to participate, please sign this consent. Then, your healthcare provider can arrange for a blood sample to be collected at a location near you or can arrange for a buccal swab kit to be sent directly to your home. The sample will then be sent to our laboratory along with information about your health status.

If I participate in the VUS Clarification Service, will I learn if I have the same VUS as my relative?

The purpose of the VUS Clarification Service is to try to make a diagnosis for your relative. **If you choose to participate in the VUS clarification service, you will not receive any results.** If you think you might like to receive a copy of your results, you can discuss the pros and cons of genetic testing with your healthcare provider. If you and your provider decide to pursue further genetic testing in order to receive a report, then your provider can order Family Variant Testing.

Is there a cost to participate in the VUS Clarification Service?

There is no cost to you or your family members to participate in the VUS Clarification Service. We can provide the sample collection kit and will also cover the cost of return shipping.



What are you consenting to?

By signing below, you are choosing to participate in the VUS Clarification Service as described above.

As part of its normal operating procedures, Blueprint Genetics will retain your sample or the DNA extracted from your sample for 12 months and it is disposed of after that unless earlier disposal is required by applicable laws. During that time, if you and your healthcare provider decide to order a diagnostic test from Blueprint Genetics, we can use the stored sample at your healthcare provider's request instead of collecting a new sample from you.

In addition, you may have Blueprint Genetics store your sample collected for the VUS Clarification Service (or the DNA extracted from it) for longer than 12 months, in which case Blueprint Genetics may only use the sample or DNA for the purposes described below during the related extended storage period. Consent for long term storage and/or research are separate and unrelated to the VUS Clarification Service. You are under no obligation to choose one or either of them and your decision does not affect your ability to participate in the VUS Clarification Service.

By checking this box, you consent to Blueprint Genetics retaining your sample or the DNA extracted from it for up to three (3) years at the Blueprint Genetics diagnostic laboratory. During this period, if you and your healthcare provider decide to order a diagnostic test from Blueprint Genetics, Blueprint Genetics may use the stored sample or DNA for the testing if your healthcare provider requests, instead of collecting another sample from you. **You may revoke this consent at any time by contacting Blueprint Genetics or the healthcare provider to whom you provided this consent. If you or your healthcare provider provides Blueprint Genetics written notice of such revocation, Blueprint Genetics will destroy any sample or DNA being stored for you; you will need to provide a new sample if you and your healthcare provider decide to order a diagnostic test from Blueprint Genetics in the future.**

By checking this box, you consent to Blueprint Genetics retaining your sample or the DNA extracted from it for up to fifty (50) years at at the Blueprint Genetics diagnostic laboratory. During this period, Blueprint Genetics may use the sample or DNA for research into hereditary diseases and the efforts to improve their diagnosis and treatment. The research may be for a disease unrelated to the condition or disease that underlies the VUS Clarification Service. Blueprint Genetics may collaborate with one or more other organizations or companies on the research. Blueprint Genetics will treat information about you as confidential by coding it in such a way that your identity cannot reasonably be discovered without the Blueprint Genetics key code. Where necessary, other organizations or companies may process and use such coded research data. **You may revoke this consent at any time by written notice to Blueprint Genetics. Upon receipt of such revocation, Blueprint Genetics will no longer use your sample or DNA for any further research. However, the data collected up to the date of your revocation will be used as part of any ongoing research.**

PATIENT SIGNATURE

By signing this form, I acknowledge that I have read the Informed Consent and understand its content. I have had the opportunity to ask questions about this form and my questions have been answered.	
Patient name (please print):	Patient DOB:
Patient signature:	Date:
Name and relationship of Legal Representative, if patient is a minor (please print):	Signature of Legal Representative, if patient is a minor:

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Read more at blueprintgenetics.com/vus-clarification