

Produced by:



Questions to ask for Research Study:

What genes are included in the test? CSF1R only, whole exome?

Are there any key test limitations? The testing done as part of a research study is designed to meet the needs of the research project but may not be comprehensive. Be sure to ask how the test differs from a clinical test (it may not differ at all) and what the test limitations are.

Is the test done in a clinical lab? Sometimes genetic testing done for research purposes is not performed in a lab that meets clinical standards. Labs that want to return results to the participants must have CLIA certification (CLIA is the Clinical Laboratory Improvement Amendment). In some cases, the initial testing is not performed in a CLIA-certified lab, but any positive result is validated in a CLIA-certified lab. Ask whether the test is performed or validated in a CLIA-certified lab.

Who gets the results? Research studies are not obligated to return the results of all tests to you. In some cases, they may not be allowed to do so. Ask who gets the results of research genetic testing. Do you get a copy of the results? Does your physician? Is there anyone else the results are shared with?

Are there any specific privacy considerations? Genetic testing is unique because it can provide information about what may happen in the future (when testing is performed on someone without symptoms); it also provides some information about other relatives and may identify others at risk of developing ALS. However, it is important to confirm who will have access to test results as part of the study records. Will the data have your name or other identifiers attached to it? How is your privacy maintained throughout the research study and

once the study ends? Researchers are almost always required to carefully protect the identity of participants and their genetic data, but it does not hurt to ask and confirm.

Questions to ask for Sponsored Testing:

Why is the sponsor paying for testing? A pharmaceutical company might sponsor genetic testing to increase the number of people with a confirmed genetic diagnosis, the number eligible for a clinical trial investigating a potential treatment, and/or the number of people eligible to use this treatment once a drug has FDA approval. In some cases, the sponsor's primary motivation may be simply to raise awareness or provide a service.

What does the sponsor get in return for paying for testing? Sponsors need to be clear and transparent about what information they are getting back in exchange for paying for your testing. If the sponsor is receiving any identifying information (such as your name), then they must have your written consent before testing. Usually, your identifying information will not be shared with the sponsor, although there are exceptions. The sponsor may be given the name of the physicians who ordered the testing, which allows them to speak with these physicians about their study or potentially approved treatment. Other sponsors may require that you enroll in their registry or sign up for their mailing list to be eligible for free testing. By enrolling in a registry, you are giving the sponsor permission to contact you in the future. You can usually opt out of a registry or mailing list later, but it is important to understand what the obligations are before agreeing.

What genes are included in the test? Consider whether this is the right test for you. If your family history includes other neurodegenerative illnesses, genetic testing that includes additional genes may be recommended. Some sponsors will only pay for genetic testing for the gene or genes that are directly related to the drug they are developing.

Is genetic counseling included with testing? Many, but not all, sponsored programs include pre-test genetic counseling at no charge. Genetic counselors

are healthcare professionals with special training in genetics who can help you make an informed decision about genetic testing.

Are there any specific privacy considerations? Genetic testing is unique because it can provide information about what may happen in the future (when testing is performed on someone without symptoms); it also provides some information about other relatives and may identify others who are at risk of developing ALSP. The power and potential impact of this unique information makes it important to understand who will have access to your test results, and how they will be used. In some cases, your de-identified data (data without your name or other personal identifiers) may be used by the lab or the pharmaceutical company for research purposes or to develop new products that they will then sell. They may also sell your deidentified data.

How do I choose a sponsored testing program? Consider differences in the programs such as; is the program available only in the United States, is the program available to asymptomatic ALSP patients or only those with symptoms, is pre-and-post genetic counseling available at no cost.