

Informed Consent for Carrier Screening

I, _____ First and Last Name, hereby request genetic carrier screening testing which may include molecular and/or biochemical analyses. I have received verbal and written information (please see sema4.com/test-catalog for test-specific information sheet) from my physician or from a genetic counselor that described, in words that I understood, the nature of the genetic testing that I am about to undergo.

I understand that specimen(s), such as peripheral blood or saliva will be taken from me. I understand that the samples will be used for determining if I am a carrier of a genetic variant that may affect the health of my future potential offspring. I also understand that the results of this test may inform me about my own health and my susceptibility to develop a genetic disease or medical condition.

The nature of the carrier screening options available to me, also available at sema4.com/products/expandedcarrierscreen, have been explained to me, including details regarding the accuracy of such test and its risks and limitations. Several technologies are used to perform carrier screening, which may include:

- High-throughput, next-generation sequencing
- Sanger sequencing
- Targeted mutation analysis
- Capillary electrophoresis
- Southern blot analysis
- Multiplex ligation-dependent probe amplification
- Enzymatic analysis

I understand that although the likelihood of an incorrect diagnosis or a misinterpretation of the result is extremely small, infrequent errors may occur. The likelihood of this occurring has been estimated to be less than 1%. I understand that a negative result reduces, but does not eliminate, the possibility that I carry a mutation or mutations in the genes analyzed. I understand that carrier screening includes assessment of a particular set of genes and that no determination may be made regarding other genes that are not included in this test. I understand that this testing may yield incidental findings that indicate that I am affected with one of the diseases on the panel or with another genetic abnormality.

No test will be performed on my sample other than the one(s) authorized by this consent. I hereby give consent to have my specimen tested for:

- Sema4 Expanded Carrier Screen (ECS) - 283 gene panel, OR
- The following panel/genes _____

De-identified research

Sema4 may also give the de-identified information to its research partners and may submit this de-identified information to research databases for use in scientific and medical research, including scientific databases that are maintained by the federal government, such as a database kept by the National Institutes of Health ("NIH") (an agency of the federal government that funds research). Researchers have to apply to the NIH to see the information in the database. Any information that could directly identify you (such your name or address) will not be provided to a scientific database.

If I prefer not to have any of my de-identified health information used in research consistent with this consent, I may initial here _____ Initials or request this by contacting Sema4, including by emailing privacy@sema4.com.



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Permission to contact

I understand that Sema4 may wish to contact me in the future, including for the following reasons: research purposes, the provision of general information about research findings, and/or the provision of information about the results of tests on my sample(s). I understand that I may notify Sema4 to opt out of such future contact, including by emailing privacy@sema4.com.

I understand that, because of the nature of carrier screening tests, my results may be shared with my reproductive partner, and I hereby authorize Sema4 and my physician to disclose and discuss my results with my reproductive partner to the extent necessary to provide the testing and genetic counseling services that I have requested.

I understand that the Laboratory may wish to contact me in the future for the following reasons: Research purposes, the provision of general information about research findings, and/or the provision of information about the results of tests on my sample(s). I understand that I may notify the Laboratory to opt out of such future contact and doing such will not affect my clinical care.

The results of my test will be explained to me by a genetic counselor or by my physician who will have the opportunity to discuss my results with a geneticist. I have had the opportunity to have all of my questions answered.

I understand that this consent is being obtained in order to protect my right to have all of my questions answered before testing. I understand that the results of this testing will become part of my medical record and may only be disclosed to individuals who have legal access to this record or to individuals who I designate to receive this information.

Sample storage

Initials

By initialing here, I agree that Sema4 may store, de-identify, and use my sample and information to support medical and academic research relating to health, disease prevention, drug development, and other scientific purposes, and that I will receive no compensation in connection with such research. If I do not initial here, my sample will be destroyed at the end of the testing process or not more than 60 days after collection. I understand that I may withdraw this consent by contacting Sema4 (including by emailing privacy@sema4.com).

Signature of person being tested
(or guardian)

Date

Revised 10/4/2018