

## Informed Consent for Prenatal Testing

By signing below, I hereby request and authorize Sema4 to determine the chromosome constitution, copy number status, biochemical or molecular status of my unborn fetus.

I, \_\_\_\_\_, First and Last Name, hereby request the following genetic testing:

- Chromosome analysis
- AFP (amniotic fluid)
- Aneuploidy FISH (chromosomes 13, 18, 21, X, and Y)
- Microdeletion FISH panel (13 common microdeletion syndromes)
- Chromosomal microarray with or without SNP (whole genome chromosome microarray)
- Noonan syndrome and related disorders panel (14 genes)
- FGFR3* hotspot with or without reflex to full gene sequencing
- Limb defects panel (8 genes)
- Molecular testing for \_\_\_\_\_ Disease and Gene
- Biochemical testing for \_\_\_\_\_ Disease and Test

On the sample specified below:

- Chorionic villus sampling (CVS)
- Amniotic fluid
- Other \_\_\_\_\_

It has been explained to me and I understand that:

- An attempt to obtain a viable tissue culture from cells of any particular sample of chorionic villi or amniocytes may be unsuccessful or the chromosome preparation may be of poor quality and unusable, or the biochemical study may be unsuccessful due to technical difficulties. A second prenatal diagnostic procedure or fetal blood sampling may be offered to provide cells to make the diagnosis.
- Amniocentesis is available for the verification of chromosomal and/or biochemical and/or molecular test results or for the provision of information to aid in interpretation of results from the CVS studies, which might be difficult to interpret, due to factors including placental mosaicism and/or maternal cell contamination.
- I have received verbal and written information (please see [sema4.com/testcatalog](http://sema4.com/testcatalog) for test-specific information sheet) on this testing from my physician or genetic counselor. The nature of chromosome, chromosomal microarray (with or without SNP information), biochemical and/or DNA analyses has been explained to me and the accuracy of the test and its limitations have been detailed. I understand that while results obtained from this testing are usually highly accurate, infrequent errors may occur. There is the possibility that the cell culture or direct tissue specimen may not accurately reflect the

status of the fetus due to mosaicism or maternal cell contamination. It is also possible that the results of the genetic testing performed may not accurately reflect the status of the fetus due to inherent limitations of the testing performed.

- In addition, due to insufficient specimen size or cell growth, testing may fail to yield results. The likelihood of this occurring has been estimated to be less than 1%. Additionally, an error in diagnosis may occur if the true biological relationships in the family involved in this study are not as I have stated. My results will be explained to me by a genetic counselor or by my physician who will have the opportunity to discuss my results with a clinical geneticist.
- I understand that if the results of this test are normal it is possible that the fetus/baby may still be affected with the disorder/syndrome that was tested for due to other causes than were tested for, or another disorder/syndrome that was not tested for.
- I also understand that maternal cell contamination studies will be performed on all specimens that have molecular or biochemical analysis as part of the testing process, which may result in discovery of non-maternity and/or invalidate the test results due to the presence of maternal DNA.
- I understand that this testing may yield results that are of unknown clinical significance and that parental blood samples will be tested to better interpret the results. As a result of parental studies, non-maternity and/or non-paternity may be detected. In addition, I understand that I may receive a result for which no clinical information can be provided. I also understand that I may receive a result relating to an adult onset condition or infertility regarding my fetus. Finally, incidental findings that are not related to the primary diagnosis may be identified in me/my fetus.
- No test will be performed and reported on my fetal sample other than the one(s) authorized by my doctor.

### 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](mailto:privacy@sema4.com).

### 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](mailto:privacy@sema4.com).

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I have had the opportunity to have all of my questions answered and undertake professional genetic counseling prior to signing the form. I understand that this consent is being obtained in order to protect my right to have all of my questions answered before testing. I also understand that the results of this testing



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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](mailto:privacy@sema4.com)).

\_\_\_\_\_  
Signature of person being tested  
(or guardian)

\_\_\_\_\_  
Date

*Revised 10/4/2018*