## Parental Carrier Screening

### Carrier Screening Clinical Information:

<table>
<thead>
<tr>
<th>Patient ancestry:</th>
<th>Preferred Language:</th>
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History of BMT or recent blood transfusion in the last 4 weeks?  
Yes  No

Is the patient or their partner pregnant?  
Yes  No

Is the patient currently using birth control medication?  
Yes  No

Previous carrier screening?  
Yes  No

If yes, what gene variant:  

Family History of:  

<table>
<thead>
<tr>
<th>Gene</th>
<th>Disorder</th>
<th>CHROMOSOME ANALYSIS</th>
<th>CYTOGENETICS AND CYTOGENOMICS</th>
<th>INFECTION/PREGNANCY LOSS</th>
<th>FLUORESCENT IN SITU HYBRIDIZATION (FISH)</th>
<th>PRENATAL DIAGNOSTIC TESTING</th>
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### Laboratory Test(s) Ordered

**Specimen Required:**

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**Infertility/Pregnancy Loss**

- Test for Microdeletions of Y Chromosome (male)
- Cystic Fibrosis with CFTR Intron 9 Poly(T) (male)
- Chromatina Test (2 variants below)

**Cytogenetics and Cytochromosomes**

- Chromosome Analysis (includes aPP with amniocentesis fluid)
- Includes repeat if no growth for POC specimens
- Reflux to array if normal results are available

**CHROMOSOME ANALYSIS**

- Chromosome Analysis (includes aPP with amniocentesis fluid)
- Includes repeat if no growth for POC specimens
- Reflux to array if normal results are available

**CHROMOSOMAL MICROARRAY: Array Comparative Genomic Hybridization (aCGH)**

- 180k + SNP
- For prenatal specimens, submit maternal blood for Maternal Cell Contamination (MCC)
- Prenatal chromosomal microarray (lower resolution)
- High Resolution Chromosomal Microarray

**INFECTION/PREGNANCY LOSS**

- Includes high resolution microarray analysis, triploid detection, UPA analysis, molar pregnancy analysis and MCC studies with submission of maternal blood or saliva sample. Include blood (1 EDTA purple top, 1 Sodium heparin green top) from the parents of the proband/pregnancy if available.

**Cytogenetic Studies**

- Include blood (1 EDTA purple top, 1 Sodium heparin green top) from the parents of the proband/pregnancy if available.

**FLUORESCENT IN SITU HYBRIDIZATION (FISH)**

- Aneuploidy FISH (chromosomes 13, 18, 21, X,Y)
- Microdeletion FISH Panel (13 disorders)
- Angelman Syndrome (15q11.2)
- CHARGE (4q21.1 - c12.2)
- Chi-defect Syndrome (5p15.2)
- DiGeorge/Velo Cardio-Facial Syndrome (22q11.2)
- DeLong-Giedion (8q23.3 - 8q24.11)
- Miller-Dieker Syndrome (17p13.3)
- Prader-Willi Syndrome (15q11.2)
- FISH other:
  - Fish for Kartlan Syndrome
  - Fish for STS Deficiency
  - Fish for SRY deletion

**Prenatal Diagnostic Testing**

- FISH for sequencing if negative
- Lamb Defects Next Gen Sequencing Panel (7 genes)
- Noonan Syndrome Next Generation Sequencing Panel (19 genes)
- Single gene/Diagnostic testing

**Bacterial Infections**

- Maternal Cell Contamination
- Other

**Biochemical Infections**

- Tay-Sachs enzyme analysis
- Sandhoff enzyme analysis

**Limitations:**

- As this assay is screening test and not diagnostic, false positive and false negatives can occur. Positive results need diagnostic confirmation by alternative testing methods. Negative results do not fully exclude the diagnosis of any of the syndromes or the possibility of other chromosomal abnormalities or birth defects. Potential sources of inaccurate results include, but are not limited to, mosaicism, low fetal weight, limitations of current diagnostic techniques, or misidentification of samples. Results should be interpreted by a clinician in the context of clinical and familial data, and the patient should receive genetic counseling.
Informed Consent for Genetic Testing

I, _________________________________, hereby request genetic testing, which may include molecular, cytogenetic and/or biochemical analyses, for

☐ Myself
☐ My child _________________________________

I have received verbal and written information (please see sema4.com/testcatalog 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/my child am about to undergo.

I understand that specimen(s), such as a peripheral blood, saliva, cheek swab, dried blood spot, skin biopsy, amniotic fluid, chorionic villi and/or urine sample, will be taken from me/my child. I understand that the samples will be used for determining if I/my child have a genetic disease, are carriers of a genetic disease, or are more likely to develop a genetic disease or condition.

The nature of the genetic test, _________________________________, has been explained to me and the accuracy of the test and its risks and limitations have been detailed. I understand that infrequent errors may occur, even though the likelihood of an incorrect diagnosis or a misinterpretation of the result is extremely small. 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/my child carry a mutation(s) in the gene(s) analyzed or in other gene(s) that are not included in the test.

I understand that no test will be performed on my sample other than the one(s) authorized by this consent and 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/my child’s de-identified health information used in research consistent with this consent, I may initial here or request this by contacting Sema4, including by emailing privacy@sema4.com.

Permission to contact
I understand that Sema4 may wish to contact me/my child 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/my child’s 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 this testing may yield results that are of unknown clinical significance and that parental or other relative’s specimens may also be tested to determine whether a specific finding was inherited. In addition, incidental findings that are not related to the primary diagnosis may be identified in me/my child. An error in the diagnosis may occur if the true biological relationships of the family members involved are not as I have stated and this test may detect non-paternity.

The results of my/my child’s 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. If I am signing this form on behalf of a minor for whom I am the legal guardian, I am satisfied that I have received enough information to sign on his or her behalf.

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

By initialing here, I agree that Sema4 may store, de-identify, and use my/my child’s sample and information to support medical and academic research relating to health, disease prevention, drug development, and other scientific purposes, and that I/my child will receive no compensation in connection with such research. If I do not initial here, my/my child’s 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 _________________________________

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