Parental Carrier Screening

Carrier Screening Clinical Information:
Patient ancestry: ____________________________ Preferred Language: ____________________________

History of BMI 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:

- Standard Pan-ethnic Panel (41 genes)
- High Frequency Pan-ethnic Panel (11 genes)
- ECS 39 (39 genes)
- ECS 152 (152 genes)
- Comprehensive Jewish Carrier Screen (101 genes)
- Ashkenazi Jewish Disorders (47+17 genes)
- Sephardi-Mizrachi Jewish Disorders (37+17 genes)

Partner Carrier of:

- X-Linked Supplemental Panel (21 genes)
- Single Gene/Variant(s)
- Phase analysis
- PGT set-up
- Test only for AR disorders partner screened positive for - hold sample pending partner results (mark on reverse side or list here)
- MSS re-analysis of ECS 283 + enhancements included in ECS 283. Previous test order date:

Pre-Authorization #: ____________________________ Please include a copy of all insurance paperwork.

ASSIGNMENT AND RELEASE: I hereby authorize my insurance benefits be paid directly to the provider and understand that I am financially responsible for services. I also authorize the release of any information required to process the claim. Billing inquiries, please call 800-298-6470, Option 3.

Physician Signature of Consent (Required): I certify that this patient (and/or their legal guardian, as necessary) has been informed of the benefits, risks, and limitations of the laboratory tests(s) requested. I have answered this person’s questions. I have obtained a signed informed consent from this patient or their legal guardian for this testing in accordance with applicable laws and regulations, including N.Y. Civil Rights Law Section 79-L, and will retain this consent in the patient’s medical record.

Indications for Testing:

- CHROMOSOMAL MICROARRAY: Array Comparative Genomic Hybridization (aCGH) 180K + SNP

For prenatal specimens, please submit maternal blood for Maternal Cell Contamination (MCC) if maternal cell contamination is an issue.

- Prenatal Chromosomal Microarray (lower resolution)
- High Resolution Chromosomal Microarray prenatal/postnatal/POC

- POC Microarray Plus: Includes high resolution microarray analysis, triploidy detection, UPD analysis, maternal pregnancy analysis and MCC studies with submission of maternal blood or saliva sample.

- Targeted testing: Maternal variant: ____________________________ Paternal variant: ____________________________

- Additional Cell Culture: Hold Grow Mosaicism study

FLUORESCENT in situ HYBRIDIZATION (FISH)

- Aneuploidy FISH (chromosomes 13, 18, 21, X & Y, Triploidy, and 22q11.2 deletion syndrome)
- Cri-du-chat syndrome, Angelman syndrome, and Prader-Willi syndrome

- Prader-Willi/Angelman (15q11.2)
- Rubenstein-Taybi syndrome (16p13.3)
- Smith-Magenis syndrome (17p11.2)
- Sotos syndrome (4q34)
- Kallman syndrome (Xp22.3)
- Williams syndrome (7q11.23)
- Wolf-Hirschhorn syndrome (4p16.3)
- FSH other:

Prenatal Diagnostic Testing

- Fetal Testing:

- Targeted testing: Maternal variant: ____________________________ Paternal variant: ____________________________

- Biochemical testing:

- Tay-Sachs enzyme analysis
- Sandhoff enzyme analysis

- Maternal blood is required for all prenatal specimens for maternal cell contamination

- If patient/partner was NOT tested at Sem4, parental bloods are required (5-10ml EDTA) to confirm the variant in-house. Please also provide a copy of any previous results.

- Please contact the laboratory for all prenatal molecular/biochemical testing.
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(s) that have been ordered in connection with this consent 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 me and my healthcare provider. I have reviewed the test order made in connection with this consent, and I hereby give consent to have my specimen tested as set forth in the order.

De-identified research

Sema4 may de-identify and use all data and information generated and received in connection with this test to support medical and academic research relating to health, disease prevention, drug development, and other scientific purposes, and I will receive no compensation in connection with such research. Data and information are “de-identified” by removing any information that could be used to identify a specific person, such as a name, email address, or date of birth. Sema4 may also give the de-identified data and information to its research partners and may submit it 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.

If I do not want to have any of my de-identified data and information used in research consistent with this consent, I may initial here__________, or I may withdraw this consent 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.

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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.

Signature of person being tested (or guardian) _____________________________ Date _____________________________

Rev.04/17/2020