Informed Consent for NY Clients – Fragile X Syndrome

Instructions: Please obtain patient signature on consent form below. All samples collected in the State of New York must be accompanied by a signed consent form. EGL Genetics (EGL Genetic Diagnostics LLC) is unable to proceed with testing in the absence of a signed consent from the patient. Once completed with signatures of patient/parent and clinician, forward the signed consent to EGL Genetics, either with the transport of the specimen or by fax (see above).

I, ________________________________, voluntarily request of EGL Genetics to perform DNA-based testing for Fragile X in myself/my child (child’s name ______________________________________________________________________________________) in an attempt to determine whether I/my child am a carrier of a disease gene or at increased risk to be affected by a genetic condition. The following points were explained and I understand that:

1. The purpose of this analysis is to test for Fragile X syndrome, a hereditary (X-linked) form of intellectual disability. This test can identify affected individuals, female premutation carriers and females at risk for Premature Ovarian Insufficiency, and individuals at risk for Fragile X-associated tremor/ataxia syndrome.
2. This is a genetic (DNA-based) test performed by PCR and Southern Blotting. These methods are used to quantify the size of the CGG repeats in the 5’ untranslated region of the FMR1 gene.
3. I (or the person for whom I am signing) may want genetic counseling before consenting to this test. If the test is positive, you or other family members may wish to have further testing, consult your physician or receive genetic counseling.
4. This analysis can have the following outcomes:
   a. Positive:
      • When a full mutation (>200 repeats) is found in a woman, there is a 50% chance of her having an affected male offspring.
      • When a full mutation is found in a male, there is an almost 100% chance of Fragile X syndrome.
      • A woman with an allele with 55-200 repeats (“premutation”) has an increased risk for having affected male children with greater than 200 repeats.
      • Women with “premutation” alleles have a higher risk for premature ovarian insufficiency.
   b. Negative:
      • A woman with an allele in the intermediate region (45-54 repeats), has an increased risk for having children with a premutation allele. These children are not at risk for Fragile X syndrome.
      • A female or male with <45 repeats is considered to not be at risk and is negative for the disease.
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5. Approximately 1% of cases of FMR1-associated intellectual disability are due to mutations that cannot be detected by this test. Other testing such as sequencing and deletion/duplication of the FMR1 gene might be warranted.

6. The results of the above test will be reported to the ordering physician/genetic counselor/medical provider/institution and will become a part of the patient’s medical record. Results may be made available to individuals/organizations with legal access to the patient’s medical record, on a strict ‘need-to-know’ basis, including, but not limited to the physicians and nursing staff directly involved in the patient’s care, the patient’s current and future insurance carriers, and others specifically authorized by the patient/authorized representative to gain access to the patient’s medical records.

7. The laboratory does not return the remaining tissue/DNA sample to individuals or physicians; however, in some cases, it may be possible to perform additional studies on the remaining sample. The request for additional studies must be made by the referring physician or other authorized healthcare professional and there will be an additional charge.

8. Remaining DNA samples will be retained in the laboratory in accordance with the laboratory retention policy. Remaining DNA samples may be de-identified and used for internal laboratory purposes with the consent of the patient (see below). The de-identified portion of the sample will not be available for future clinical studies. All original samples (blood) will be destroyed after 60 days of receipt according to laboratory retention policy.

9. I consent to my DNA sample being stored indefinitely and be used for other laboratory purposes in the future, **PLEASE INITIAL HERE:_________________________**, I have the right to withdraw this consent at any time, in writing with registered receipt, and any remaining DNA sample will be destroyed.

My signature below acknowledges my voluntary participation in this test and I state that I have been appropriately counseled about the testing process and the different possible outcomes.

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<th>Patient/Parent Signature</th>
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