Informed Consent for NY Clients – Pan-Ethnic and Ashkenazi Jewish Carrier Screens

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, (name)________________________________________________________________________________________, voluntarily request of EGL Genetics to perform DNA-based testing for Pan-Ethnic or Ashkenazi Jewish Carrier Screen 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 Pan-Ethnic Carrier Screen and the Ashkenazi Jewish Carrier Screen are tests that analyze a specific set of genes that cause autosomal recessive and X-linked disorders to identify carriers of disease-causing changes. Carriers are typically healthy individuals but can pass on these changes to any children they have. If two carriers for the same genetic condition have children, they are at a higher risk to have an affected child.

2. This test is indicated for individuals or couples seeking to assess reproductive risk for a variety of conditions and individuals or couples of high-risk ethnic backgrounds.

3. This is a genetic (DNA-based) test. All the specified genes are analyzed to identify reportable disease-causing changes. Depending on the patient choice, some additional genes can be included in the analysis. The targeted testing option involves analysis of specific disease-causing changes listed on the EGL Genetics website, while full gene sequencing will analyze the entire gene sequence for potentially disease-causing changes.

4. I (or the person for whom I am signing) may want genetic counseling before consenting to this test. If the test is positive, I or other family members may wish to have further testing, consult my physician or receive genetic counseling.

5. This analysis can have the following outcomes:
   a. Positive:
      A pathogenic variant (disease-causing) could be identified in one or more of the genes being tested for and the person is identified as a carrier which increases their risk of having a child with the condition if their partner is also identified as a carrier for the same condition.
   
   b. Negative:
      No pathogenic variant is identified. This reduces the risk of being a carrier for the specified diseases considerably but does not eliminate it completely. The reduced risks for each condition are listed in the report.
   
   c. Inconclusive:
      Due to technical issues the results are inclusive and the test might need to be repeated.
6. Possible diagnostic errors include sample mix-ups, genotyping errors, rare genetic variants that interfere with analysis and other sources. SMA analyses will not detect pathogenic variants other than the exon 7 and 8 deletion in the SMN1 gene. The targeted mutation panel will not identify other possible variants in the genes. Only pathogenic variants are reported when the full gene sequencing panel is ordered. This analysis will not detect some pathogenic variants in the promoter or other regulatory regions. Some intronic pathogenic variants will not be detected by this assay.

7. The results of the above test will be report 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.

8. 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.

9. 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, tissue, etc.) will be destroyed after 60 days of receipt according to laboratory retention policy.

10. 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.

Patient/Parent Signature __________________________________________________________________________ Date __________ Printed Name __________________________________________________________________________

Healthcare/Clinician Signature __________________________________________________________________________ Date __________ Printed Name __________________________________________________________________________