Informed Consent for NY Clients – DMD Deletion/Duplication Analysis

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 Duchenne/Becker Muscular Dystrophy in myself/my child (child’s name______________________________________________________________) in an attempt to determine whether I/my child am a carrier of a Duchenne/Becker muscular dystrophy disease gene or are at increased risk to be affected by the condition. The following points were explained and I understand that:

1. Duchenne/Becker muscular dystrophy testing analyzes the DMD gene to diagnose affected individuals and identify carriers of disease-causing changes. Duchenne and Becker muscular dystrophies (DMD, BMD) are allelic X-linked muscular diseases that result from abnormalities of the dystrophin protein. Pathogenic variants in the DMD gene located on the X chromosome cause DMD and BMD. In males, a pathogenic variant of the single copy of the DMD gene causes disease. The pathogenic variant spectrum in the DMD gene is ~60% deletions, ~5% duplications, and ~35% point pathogenic variants. Carrier females are typically healthy individuals, but may develop symptoms of the disease, and can pass on these changes to any children they have. If a female carrier has children, they are at a higher risk to have an affected child.
2. This test is indicated for the following individuals: males or females with a clinical diagnosis or symptoms of Duchenne or Becker muscular dystrophy and females who are at risk to be a carrier or have a family history of Duchenne or Becker muscular dystrophy.
3. This is a genetic (DNA-based) test. The DMD gene is analyzed to identify reportable disease-causing variants.
4. You (or the person for whom you are 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.
5. This analysis can have the following outcomes:
   a. Positive: A pathogenic variant (disease-causing) could be identified in the DMD gene and the person is identified as being affected by Duchenne or Becker muscular dystrophy, or as a carrier which increases their risk of having a child with the condition.
   b. Negative: No pathogenic variant is identified. This reduces the risk of being affected by, or a carrier for, Duchenne/Becker muscular dystrophy, but does not eliminate it completely. Duchenne/Becker muscular dystrophy can also result from pathogenic variants which are not detected by this test.
   c. Inconclusive: Due to technical issues the results were inconclusive and the test might need repeating. Results may also be inconclusive due to the identification of a variant of unknown significance.
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d. Incidental Finding (for deletion/duplication testing): A "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported unless relevant to the individual's clinical presentation.

6. Possible diagnostic errors include sample mix-ups, genotyping errors, rare genetic variants that interfere with analysis, and other sources. These analyses may not detect pathogenic variants in the promoter or other regulatory regions. Deletion/duplication analysis will not detect point mutations or some intronic mutations. Sequence analysis will not detect large deletions and duplications.

7. 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 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. Identifying information may be removed and remaining DNA samples may be used for internal laboratory purposes with the consent of the patient (see below). These samples will not be available for future clinical studies. All samples will be destroyed after 60 days, unless consent is given.

   I consent to my DNA sample being stored indefinitely and to 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