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 Chromosome Y Microdeletion 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 microdeletions in the Y chromosome. This test is indicated for men with azoospermia or oligospermia. Approximately 15-20% of men with azoospermia and 5-10% of men with oligospermia have microdeletions in the q11 region of the Y chromosome. This is a genetic (DNA-based) test performed by PCR, targeting four AZF regions on the long arm of the Y chromosome and the SRY gene on the short arm of the Y chromosome.

2. 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 independent testing, consult my physician or have genetic counseling.

3. This analysis can have the following outcomes:
   a. Positive: A microdeletion of one or more regions tested for is identified.
   b. Negative: No deletion is identified and all the target regions are present.
   c. Inconclusive: Due to technical issues the results were inconclusive and the test might need to be repeated.

4. Direct detection of Y-chromosome microdeletions is highly accurate. Possible diagnostic errors include sample mix-ups, genotyping errors, rare genetic variants that interfere with analysis and other sources.

5. 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. The 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.
Informed Consent for NY Clients – Chromosome Y Microdeletion

6. 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.

7. 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 (e.g. tissue, blood, etc.) will be destroyed after 60 days from date of receipt in accordance with the laboratory retention policy.

8. 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