

## Informed Consent – PRENATAL MOLECULAR TESTING

**Instructions:** Please obtain patient signature on consent form and provide a signed copy to EGL Genetics to permit testing and processing. If a signed consent is not submitted, EGL Genetics assumes that the ordering clinician has discussed testing with the patient and obtained the patient's informed consent.

I, \_\_\_\_\_, voluntarily request of EGL Genetics to perform fetal DNA-based testing, in an attempt to determine whether my fetus is at increased risk for: (condition) \_\_\_\_\_.  
I understand that the cells required for such an analysis are obtained from an amniotic fluid or chorionic villus sampling (CVS) by a transabdominal or transcervical procedure.

The following points were explained and I understand that:

- Viable fetal cells are required to complete the test(s) requested. Additional samples may be needed if the sample is damaged in shipment or inaccurately submitted. Occasional attempts to obtain a viable culture may be unsuccessful or the sample preparations may be of poor quality. In these cases, the prenatal procedure may need to be repeated to complete the requested genetic studies.
- In order to perform accurate prenatal testing, additional cytogenetic, molecular or biochemical studies, or samples from the affected proband, parents, or additional family members, may be required to interpret the results. Testing of other persons may be required prior to the laboratory accepting the prenatal sample.
- DNA-based studies performed are specific to the condition indicated above. The accuracy of genetic testing is limited by the methods employed, the clinical diagnosis, and the nature of the specific condition for which testing is requested. In some cases, the test will detect an abnormality, called a pathogenic variant, in the gene. In other cases the test is unable to identify an abnormality although an abnormality may still exist. This event may be due to the current lack of knowledge of the complete gene structure or an inability of the current technology to identify certain types of changes (pathogenic variant) in a gene. These tests are currently available for clinical laboratory testing, however, improvements will be made as scientific knowledge advances. As with any complex genetic test, there is always a small possibility of a failure or error in sample analysis. Extensive measures are taken to try to avoid these errors. The methods are not 100% accurate due to the possibility of rare genetic variations in the DNA of an individual or due to the complexity of the testing itself. A low error rate, approximately 1 in 1000 samples, is generally estimated to exist in a laboratory.
- It is the responsibility of the referring physician or health care provider to understand the specific use and limitations of the testing ordered, and to educate the patient regarding these limitations. Additional information describing indications, methodology and detection can be found on the EGL website at: <https://www.egl-eurofins.com/>
- Accurate interpretation of test results is dependent upon the patient's clinical diagnosis or family medical history and upon reported family relationships being true biological relationships. An erroneous clinical diagnosis in the patient or family member can lead to an incorrect interpretation in the laboratory result. Genetic testing in family members can sometimes reveal that true biological relationships are not consistent with the reported biological relationships. For example, non-paternity may be detected, which means that the stated or assumed father of an individual is not the true biological father.
- The results in no way guarantee the health of my baby. Approximately 3-5% of all pregnancies have birth defects which cannot be detected by testing amniotic fluid/CVS.

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- Due to the complexity of DNA testing and potential implications of test results, results will be reported directly to my ordering provider, who will then review and discuss the test results with me. Patient-identifying results and information at EGL will remain confidential and may only be released to other parties with my expressed written consent or as permitted or required by applicable law.
- EGL Genetics is not a DNA banking facility and does not guarantee the future availability of isolated DNA. Any requests for additional studies must be ordered by the referring provider and charges will be incurred. Once the test is complete, identifying information may be removed and remaining DNA samples may be used for de-identified laboratory purposes. These samples will not be available for future clinical studies. Any results obtained cannot be related back to the original source, so no results can be reported.
- I can request that remaining DNA not be used for research purposes by initialing here: \_\_\_\_\_

The risks, benefits and limitations of prenatal testing have been explained to me. I have read and will receive a copy of this consent form.

Patient: \_\_\_\_\_ Date \_\_\_\_\_

Spouse/Partner (optional): \_\_\_\_\_ Date \_\_\_\_\_

**Physician/Counselor/Clinician Statement:**

I have explained DNA testing to the patient/parent/guardian. The consent form and limitations of genetic testing were reviewed with the patient/parent/guardian. I accept responsibility for pre- and post-test genetic counseling. I will use my independent professional judgment and the patient's best interests in advising the patient/parent/guardian regarding DNA test results, the use and limitations of same, and any research study, clinical trial, drug, treatment or device brought to my attention by EGL or others.

\_\_\_\_\_  
Clinician Signature

\_\_\_\_\_  
Date