EGL Genetics | Eurofins Clinical Diagnostics

2460 Mountain Industrial Boulevard •Tucker, GA 30084
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Informed Consent – PRENATAL CYTOGENETIC 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, (name)_____, voluntarily request of EGL Genetics to perform DNA-based testing on a prenatal specimen from my current pregnancy in an attempt to determine whether the fetus is a carrier of a disease gene or are at increased risk to be affected by a genetic condition. The following points were explained and I understand that:

- Prenatal cytogenetic testing requires cells from the fetus which has risks associated with obtaining the sample. Additional samples may be needed if the sample is damaged in shipment or inaccurately submitted. In order to perform accurate prenatal testing, samples from the affected individual, parents, or additional family members may be required.
- This test may be ordered in case of abnormal ultrasound findings, abnormal fetal chromosome findings, history of recurrent miscarriage or stillbirth of unknown cause, previous pregnancy/child with a microarray abnormality, family history of a genetic imbalance, advanced maternal age or parental concern.
- In some cases, additional studies (including parental studies) will be recommended to determine whether or not a detected chromosome imbalance is clinically meaningful and/or was inherited. Most inherited changes are benign. Rarely, however, an inherited change which causes minimal or no issues in a parent may result in significant physical or developmental problems in a child. Submission of parental samples with the prenatal sample can expedite results.
- 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.
- This testing may identify a chromosomal abnormality. Identifying this abnormality may be useful in directing additional care, evaluation, or monitoring for your pregnancy or for you child after delivery.
- The rate of birth defects and intellectual disability in the general population ranges from 3-5%. Therefore, a normal testing result does not guarantee a healthy child.
- This analysis can have the following outcomes:

  Positive:
  - This means that a loss or gain of genetic material that may be clinically significant has been detected.

  Negative:
  - This means that prenatal microarray analysis did not detect an extra or missing piece of chromosome material that is associated with a known genetic syndrome or has been reported in the literature to be associated with physical or developmental problems. A normal result does not exclude all genetic conditions.

  Indeterminate result:
  - This means that a loss or gain of material of unclear significance has been detected.
  - Please be aware that some such losses or gains of genetic material may be benign, with no impact on fetal development. Other such losses or gains may cause birth defects or developmental disabilities, the extent of which cannot be determined until after delivery.

- Possible diagnostic errors include sample mix-ups, genotyping errors, rare genetic variants that interfere with analysis and other sources. In addition, due to insufficient specimen size or cell growth, testing may fail to yield results. This testing may yield results that are of unknown clinical significance and that parental blood samples may be also be tested to determine whether the changes were inherited. As a result of parental studies, non-maternity and/or non-paternity may be detected. One may receive a result for which no clinical information exists. One may receive a result relating to an adult onset condition or infertility regarding my fetus.

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- 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/
- Due to the complexity of DNA testing and potential implications of test results, results will be reported directly to the patient's 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: _____

Chromosome Analysis:

- Chromosome analysis will detect abnormalities in chromosome number and large deletions/duplications of chromosome material, as well as balanced chromosomal rearrangements. These abnormalities cause different conditions due to the loss or gain of genetic material.
- The test can be performed on a sample of chorionic villi or amniotic fluid. Cells from the sample are grown in culture, harvested and stained so that the chromosomes can be viewed under a microscope. The chromosomes are arranged to form a karyotype to identify number and structure of the chromosomes.

Microarray Analysis:

- Prenatal microarray analysis is designed to identify very small (submicroscopic) pieces of genetic material (DNA) that are extra and/or missing and cannot be detected with standard chromosome analysis. These submicroscopic chromosome imbalances may cause birth defects, developmental disabilities, and/or behavioral issues. Prenatal microarray can identify more than 180 known genetic syndromes.
- Prenatal microarray cannot detect balanced chromosome rearrangements, point mutations, or imbalances of regions not included on the microarray.
- Maternal cell contamination studies are required with all prenatal microarray testing.
- This is a genetic (DNA-based) test. DNA isolated from the prenatal sample (chorionic villi, amniotic fluid or cord blood) is hybridized to a custom array containing probes across the genome to detect copy number imbalances. FISH analysis or another method, such as G-banding, is used to confirm abnormal findings.

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

_____

Patient Signature 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