EGL Genetics | Eurofins Clinical Diagnostics

2460 Mountain Industrial Boulevard •Tucker, GA 30084
Tel 470.378.2200 • Toll Free 1.855.831.7447 • Fax: 470.378.2250

NY Informed Consent – Chromosome Analysis

NOTE: Please obtain patient signature on consent form and provide a signed copy to EGL Genetics to permit testing and processing.

I, (name)_____, voluntarily request of EGL Genetics to perform DNA-based testing for Chromosome Analysis 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:

- 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.
- 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.
- This test may be ordered in case of a known or suspected family history of a chromosome abnormality, recurrent miscarriage or infertility, suspected trisomy 13, 18, or 21, congenital abnormalities and/or developmental delay present. Prenatal indications include abnormal ultrasound findings, abnormal serum screening, advanced maternal age, family history of a genetic imbalance, parental concern and/or a prior pregnancy with chromosomal abnormality.
- The test can be performed on a sample of blood or tissue. 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.
- 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.
- This analysis can have the following outcomes:
    - **Positive:** This means a loss or gain of genetic material that may be clinically significant has been detected.
    - **Negative:** This means that the chromosome 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. A microarray should be considered in individuals with an abnormal phenotype.
    - **Indeterminate Result:** This means that a loss or gain of chromosome 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 development. Other such losses or gains may cause birth defects or developmental disabilities, the extent of which cannot be clearly determined.
- Possible diagnostic errors include sample mix-ups, genotyping errors, rare genetic variants that interfere with analysis and other sources. This testing may yield results that are of unknown clinical significance and that parental blood samples may 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. In addition, 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 the patient.

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- 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.
- Samples received from the State of New York for genetic testing will not be stored indefinitely or used for research purposes unless otherwise specified. Extracted DNA for NY samples and other clients with opt out policies will be discarded according to laboratory policy. Please contact EGL Genetics for details.
- I can request that remaining DNA be retained and used for research purposes by initialing here: _____
    - EGL is not a DNA banking facility and does not guarantee the future availability of isolated DNA. 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 as permitted by the Health Insurance Portability and Accountability Act ("HIPAA"). Remaining DNA samples may also be used for EGL's laboratory/internal purposes as permitted by HIPAA; these samples will not be available for future clinical studies.
    - Scientific knowledge is continuing to advance in this area. I request and authorize EGL to notify the ordering provider if there is a research study or clinical trial studying the abovementioned condition or a drug, treatment or device which might assist in the patient's care. I understand EGL may receive remuneration for those activities. The ordering provider will discuss the information that he/she receives with me, and it is my voluntary decision whether to participate. My decision will not affect the testing or results in any way.

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/Guardian Signature Printed Name 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.

_____ _____ _____

Healthcare Provider Signature Printed Name Date