

## Informed Consent for Chromosomal Analysis (required by New York State)

**What is chromosome analysis?** Chromosomes contain the genetic material or DNA which is present in every cell of our bodies. A test called chromosome karyotyping is used to identify chromosomal abnormalities. This test checks the number and structure of the chromosomes which may be altered.

**What is the purpose of this test and what are its limitations?** A blood test for chromosome karyotyping is usually performed on children to evaluate developmental delay, on adults to evaluate couples with a history of two or more spontaneous miscarriages, or on infants to evaluate an abnormal appearance that suggests a genetic abnormality. The amniotic fluid test is done to evaluate a developing fetus for chromosome abnormalities.

**What is required to perform this test?** The test can be performed on a sample of blood, tissue, amniotic fluid or bone marrow. 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.

**How will I obtain results from this test?** Chromosome analysis testing and proper interpretation of these results is complex. The information from this test will be provided in the form of a written report to your physician who will inform you of the results and discuss with you the clinical implications of any findings if abnormal. The laboratory will not provide results directly to patients as we cannot provide the additional detailed clinical evaluation or interpretation which may be required. Your physician may recommend follow-up genetic counseling. To the extent permitted by law, all of your laboratory records and results are confidential and shall not be disclosed without your written authorization.

**How reliable is this test?** This test is very reliable and time-tested. It is performed by specially trained and licensed professionals in cytogenetics. Because the testing is complex and uses specialized methods including sterile cell culture there is always a small possibility that an error will occur or that the test might not be completed, for example if the cell culture fails. The laboratory may then request a repeat sample.

**What happens to my sample after testing?** The only testing that will be performed on this sample is the specified chromosome analysis. In most cases, the original sample will be discarded at the end of the testing process or stored not more than 60 days. It may be possible for the laboratory to perform additional studies on the sample upon request. The request for additional testing must be ordered by my referring physician/counselor and there will be an additional fee. In some circumstances, a patient's specimen may be used as a control sample in future testing, but, in this event, all identifiers will be removed and the sample will be anonymous.

My signature below indicates that I have received information about this test and that I have read and understood this document. I have been given a full opportunity to ask questions that I may have about the testing procedure and related issues. I agree to undergo this testing.

\_\_\_\_\_  
Date Patient Signature

\_\_\_\_\_  
Name of Parent/Guardian (if Patient is a minor) Signature of Parent/Guardian

**For the Physician:** As the referring physician, I understand the benefits and limitations of this study and have requested that the above-named patient be tested. I attest to the fact that I have provided the patient with the information contained above and fully answered any questions. I believe that the patient understands the information and is voluntarily signing this informed consent.

\_\_\_\_\_  
Printed Name of Physician/ Health Care Professional Signature of Physician/ Health Care Professional

I am aware that these test results may be difficult to understand or may require follow-up genetic counseling. I therefore request that my test results be provided to the following healthcare professional:

Name: \_\_\_\_\_ Phone: \_\_\_\_\_ Fax: \_\_\_\_\_

Address: \_\_\_\_\_ City/State/Zip: \_\_\_\_\_