



Department of Pathology
PATIENT INFORMED CONSENT FOR
CYTOGENETICS TESTING

Patient Name: MR#:
Account #: DOB: Date:

The New York State Civil Rights Act, Section 79-1 requires that all individuals be informed of the nature of the genetic testing being requested. You may also wish to obtain genetic counseling prior to signing this form.

- 1. What is karyotype analysis? Karyotype analysis is the study of the chromosomes that are present in human cells. The chromosomes are structures on which the genes are located. Genes encode the hereditary material (DNA) that provides the blueprint for an individual.
2. What is the purpose of karyotype analysis and what are its limitations? This test examines the chromosomes to determine if there is any change in total number or structure that might be associated with the patient's clinical history or clinical abnormalities. This information could lead to a specific diagnosis. Occasionally, a structural defect may not be detected because it is too small to be seen visually.
3. What is fluorescence in situ hybridization (FISH)? FISH is specialized technology that uses fluorescently labeled DNA fragments of known composition that can bind to a patient's DNA.
4. What is the purpose FISH and what are its limitations? The goal of the test is to provide information regarding genetic abnormalities that will aid in confirmation of a diagnosis. FISH can provide clinical information on chromosome anomalies that are too small to be detected by standard cytogenetic analysis. It is particularly powerful in identifying microdeletions and unbalanced chromosome rearrangements. This test is greater than 97% accurate in appropriate applications, but, since some disorders may have more than one cause, FISH may not provide a definitive diagnosis.
5. What will happen to the cells once the test is complete? No tests other than those authorized will be performed. The cells will be discarded within 60 days after testing is complete. If there is a medical necessity and authorization is provided, cells may be stored up to 10 years.
6. What will happen to the test results? The results of the genetic testing will be sent to the health care professionals who requested the testing. Furthermore, if you are a patient of University Hospital, University Hospital clinics, or Golisano Children's Hospital, your test results will be included in your medical record and the University Hospital electronic database.
7. How will I obtain results from the test? Please contact your physician to obtain test results and interpretation of the findings.
8. I have been provided with a full opportunity to ask any questions or express any concerns I may have. My questions have been answered and my concerns addressed to my satisfaction. I understand that I may ask for further information and it will be given to me.
9. I have read this entire document and understand its contents. In addition, I have been told that I am free to withdraw any portion of my consent.
10. I have either completed or crossed off and initialed any unacceptable statements above prior to my signing.

If you have any questions about the test which will be performed, you may contact the Cytogenetics Laboratory at 315 464-4716.

Please indicate the name and address of any other physicians to whom you wish a copy of the report to be sent.

Name: Address:

My signature below indicates that the above information has been explained to me and that I give consent for [] karyotype analysis and/or [] FISH analysis (check one or both).

Date:

Signature of Patient or Parent/Guardian if patient is a minor:

Patient's name (printed):