DT82875

UPSTATE
UNIVERSITY HOSPITAL

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 told of the nature of the genetic testing being called for. You may also wish to get genetic counseling before 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 found. Genes encode the hereditary material (DNA) that gives the blueprint for a person.
2. **What is the purpose of karyotype analysis and what are its limits?** This test looks at the chromosomes to find out if there is any change in total number or structure that might be linked to the patient's clinical past or clinical abnormalities. This information could lead to a specific diagnosis. Sometimes, a structural change may not be found 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 make up that can bind to a patient's DNA.
4. **What is the purpose of FISH and what are its limits?** The goal of the test is to give facts about genetic abnormalities that will aid in proof of a diagnosis. FISH can give clinical information on chromosome changes that are too small to be detected by standard karyotype analysis. It is particularly powerful in finding microdeletions and unbalanced chromosome rearrangements. This test is greater than 97% accurate in proper applications but since some medical conditions may have more than one cause, FISH may not give a certain answer.
   a. Identifying certain chromosome changes may facilitate a specific diagnosis. Syndromes that can be found by FISH include: Deletion 22q (VCFS) and Duplication 22q syndromes; Prader-Willi (PWS) and Angelman (AS) syndromes; Steroid Sulfatase Deficiency (SSD) (also known as X-linked Ichthyosis); trisomies 13, 18, and 21; Turner Syndrome; Klinefelter Syndrome.
   b. Before signing this consent, you may wish to discuss the testing with your doctor and/or genetic counselor.
5. **What will happen to the cells once the test is complete?** No tests other than those approved will be performed. The cells will be discarded within 60 days after testing is complete unless clearly approved in writing for a longer period of time.
6. **What will happen to the test results?** The results of the genetic testing will be sent to the health care professionals who called for the testing. Also, 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 records. If you have any questions about the test results, you may also seek genetic counseling.
7. **How will I obtain results from the test?** Please contact your doctor to get test results and interpretation of the findings.
8. I have been provided with a full chance 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 whole document and understand its contents. Also, I have been told that I am free to withdraw any part of my consent.
10. I have either completed or crossed off and initialed any unacceptable statements above before 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: _____

Patient's Name: _____ Account #: _____ MR#: _____

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

Sample retention: I agree to having my sample (fixed cells, amniocytes/fibroblasts) retained for greater than 60 days after the end of testing.

_____ (Initials) Date: _____

Date	Time	Signature of Patient	Print Name

Parent/Guardian:

Date	Time	Signature of Parent/Guardian (if patient is a minor)	Relationship to Patient

Consent Form Witness:

Date	Time	Signature of Witness	Print Name

Physician/Healthcare Professional:

Date	Time	Signature/Title of Physician/Healthcare Professional	Print Name