

INFORMED CONSENT FOR CYTOGENETICS TESTING

Patients (or their authorized representative) must provide their written informed consent prior to receiving genetic testing, and the ordering physician must maintain documentation of the informed consent in the patient's medical record. If warranted, patients (or their authorized representative) may wish to obtain professional genetic counseling prior to giving consent in order to fully understand what the risks and benefits are to having testing completed.

General Description of the Disease or Condition Related to the Test (to be filled in by the ordering health care provider)

Karyotype Analysis
Description: Karyotype analysis is a test that evaluates the number, size, and condition of chromosomes, the structures that contain the body's genetic blueprint.
Purpose: This test can help identify cytogenetic problems as the cause of a disorder or disease.
Limitations: Very small or cryptic abnormalities, complex aberrations, and marker chromosome may not be accurately identified or fully characterized.

Fluorescence In Situ Hybridization (FISH)
Description: FISH is a specialized technique that uses fluorescently labeled DNA fragments that bind to a person's DNA to detect and locate the presence, number, or structure of a specific DNA sequence on a chromosome.
Purpose: This test is utilized to diagnose genetic diseases and identify chromosomal abnormalities. FISH can detect abnormalities that are too small to be detected by standard cytogenetic analysis.
Limitations: FISH can only detect deletions or duplication of regions specifically targeted by the probe used and which are larger than the probe used. It is recommended that FISH be used as an adjunct to chromosome analysis or chromosomal microarray.

Test Results
A positive test result is an indication that I (or the individual being tested) may be predisposed to or have the specific disease(s) or condition(s) tested for and may wish to consider further independent testing, consult my or his/her/their physician or pursue genetic counseling.
A normal test results does not exclude the possibility that I (or the individual being tested) may have a genetic condition which is not evaluable by conventional Cytogenetics or FISH.
There may be the possibility that the laboratory findings will be uninterpretable or of unknown significance. In rare circumstances, findings may be suggestive of a condition different than the diagnosis that was originally considered.

DISCLOSURE OF TEST RESULTS
All tests are confidential and will be disclosed only to the ordering health care provider (or their designated representative) unless otherwise authorized by the patient or patient's authorized representative in writing or required by law.

Retention and Additional Testing
No tests other than those authorized shall be performed on my (or the individual being tested) sample. The sample will be destroyed at the end of the testing process or not more than 60 days after the sample was collected unless I (or my authorized representative) expressly authorize a longer period of retention in writing. If authorization is granted on the specimen requisition, any remaining sample may be retained longer than 60 days after the completion of testing and be used as a de-identified sample for test development and improvement, internal validation, quality assurance, and training purposes.

Patient and Ordering Provider Information	
Patient's Name:	Patient's ID Number:
Patient's Date of Birth:	Ordering Provider's Name:

Patient Signature	
My signature below indicates that the above information has been explained to me and that I understand and give consent for <input type="checkbox"/> Karyotype analysis and/or <input type="checkbox"/> FISH analysis (check one or both).	
Patient or Patient's Authorized Representative	Date (Month,DD,YYYY)
Ordering Health Care Provider	Date (Month,DD,YYYY)

Ordering Health Care Provider Signature	
I have explained this genetic test, its risks, benefits and alternative to the patient or patient's authorized representative and addressed all questions.	
Ordering Health Care Provider	Date (Month,DD,YYY)

Form is available on Genetics Associates, Inc.'s website: <https://geneticsassociates.com>