Cytogenetics

Cytogenetics	
Standard	***Guidance***
Cytogenetics Standard of Practice 1 (CG S1): Informed Consent The laboratory must notify requestors that informed consent is required for genetic testing. The laboratory must make available to requestors a model consent form and test-specific information that includes: a) general description and statement of purpose for the test; b) indication that the individual may wish to obtain professional genetic counseling prior to giving consent; c) a statement that a positive result is an indication that the individual may be predisposed to or have the specific disease or condition tested for and may want to consider further independent testing, consult their physician or pursue genetic counseling; d) a general description of the disease or condition related to the test; e) the level of certainty that a positive test result serves as a predictor of the disease; f) the persons or organizations to whom the test result or other test related information may be disclosed; g) a statement that no tests other than those authorized shall be performed on the biological sample and that the sample shall be destroyed at the end of the testing process or not more than sixty days after the sample was taken, unless a longer period of retention is	Informed consent is not required for cancer cytogenetic testing. While patient consent forms are recommended to be on file in the laboratory, the referring physician may sign the test requisition or other form indicating that she or he conveyed the required information to the patient and obtained consent. Genetic testing is covered by Section 79-L of the Civil Rights Law, available at: www.wadsworth.org/regulatory/clep/laws. Additional information related to genetic testing is provided in Section 79-L of the Civil Rights Law, including provisions for court ordered genetic testing, consent for genetic testing on a deceased individual, and research related genetic testing.

Cytogenetics	
Standard	***Guidance***
expressly authorized in the consent; and h) provision for the signature of the individual subject of the test or if the individual lacks the capacity to consent, the signature of the person authorized to consent for the individual. The laboratory must have a system to document the informed consent status for each specimen.	
Cytogenetics Standard of Practice 2 (CG S2): Clinical Information In addition to the requirements in Test Request Standard of Practice 3, the laboratory must request: a) gestational dating for prenatal analysis; and b) any other clinical information necessary to guide testing and result interpretation.	The laboratory may include a section on the requisition for this information. The laboratory should document any missing information and note on the report any limitations on result interpretation as required by Reporting Standard of Practice 2.
Cytogenetics Standard of Practice 3 (CG S3): Specimen Identification The laboratory must have standard operating procedures and policies that ensure accurate and reliable patient specimen identification during all phases of testing, including: a) accessioning; b) culture, if performed, or other processing; c) imaging; d) reporting; and e) storage of documentation, results, karyotypes, and images.	

Cytogenetics	
Standard	***Guidance***
Cytogenetics Standard of Practice 4 (CG S4): Turnaround Times The laboratory must establish critical limits for turnaround times for all clinical tests, including standard methods, fluorescent in-situ hybridization (FISH), and chromosomal microarray analysis (CMA).	Turnaround time targets should be based on criteria that include specimen type and indication/reason for referral.
Cytogenetics Standard of Practice 5 (CG S5): Replicate Cultures The laboratory must prepare replicate independently established cultures for each specimen, including: a) a minimum three (3) cultures if sufficient specimen is available for prenatal, tissue or fibroblast cultures; and b) duplicate cultures for all others.	
Cytogenetics Standard of Practice 6 (CG S6): Redundant Incubation The laboratory must have standard operating procedures and policies that protect against loss of prenatal cultures. These must include culturing replicates in at least two (2) incubators supported by independent power and gas sources.	Power can be independent circuits and/or emergency back-up.
Cytogenetics Standard of Practice 7 (CG S7): Karyotyping The laboratory must prepare a minimum of two (2) karyotypes per specimen that are traceable to the patient, specimen and culture when performing standard metaphase chromosome analysis, including: a) a minimum one (1) karyotype per cell line; and	

Cytogenetics	
Standard	***Guidance***
b) banding quality and resolution that meet the laboratory's specifications.	
Cytogenetics Standard of Practice 8 (CG S8): Metaphase Analysis Standard metaphase chromosome testing must include: a) analysis of a minimum of twenty (20) metaphases, except for prenatal, in situ, which requires fifteen (15) metaphases; and b) analysis and/or counting of at least two (2) cultures, except peripheral blood analyzed for constitutional aberrations.	When mosaic chromosome anomalies are suspected, an analysis of at least fifty (50) cells is recommended. The technologist should analyze cells from at least two (2) independent cultures, except for routine blood cultures that yield adequate numbers and quality of cells with consistent results from a single culture.
Cytogenetics Standard of Practice 9 (CG S9): Laboratory Developed Fluorescence in situ Hybridization (FISH) Analysis For laboratory developed tests (LDT) for fluorescence in situ hybridization (FISH) analysis, the laboratory must analyze a number of cells appropriate to the specimen type, reason for referral, and aberrations expected. At a minimum, the laboratory must analyze: a) for metaphase FISH: i. ten (10) cells to detect nonmosaic microdeletion; ii. five (5) cells to characterize abnormal chromosome(s);	Information on Departmental approval of a laboratory developed test (LDT) is available at: https://www.wadsworth.org/regulatory/clep/clinical-labs/obtain-permit/test-approval. Unexpected results may require analysis of more cells. FDA-approved/cleared tests should be analyzed as described in the package insert or its equivalent. FISH for microduplications should include analysis of interphase nuclei. Lab should have policies for "borderline" results near cutoff values. A pathologist must guide identification of tumor cells in tissue sections.

Cytogenetics	
Standard	***Guidance***
iii. twenty (20) cells for mosaic constitutional aberrations or samples expected to be mosaic based on indications; and b) interphase FISH: i. constitutional disease – fifty (50) nuclei; ii. acquired disease: a. suspension culture – one hundred (100) cells; and b. tissue section – twenty-five (25) tumor cells.	
Cytogenetics Standard of Practice 10 (CG S10): Metaphase Preparation Acceptability Laboratories must establish criteria to determine the acceptability of standard metaphase chromosome preparations and document acceptability of each preparation prior to reporting.	Criteria may describe circumstances (for example, irreplaceable sample) under which a preparation not meeting acceptability criteria might be reported.
Cytogenetics Standard of Practice 11 (CG S11): Fluorescence in situ Hybridization (FISH) Acceptability Laboratories must establish acceptability criteria for FISH hybridization and document the acceptability of each hybridization prior to reporting. Such criteria must include: a) signal intensity; b) background/noise; and c) results of appropriate internal (normal homolog and/or control probe) and/or external controls, as required in	

Cytogenetics	
Standard	***Guidance***
Quality Control Standard of Practice 1, or established according to Quality Control Standards of Practice 2, 3 and 4.	
Cytogenetics Standard of Practice 12 (CG S12): Reporting In addition to the requirements of 10 NYCRR Part 58-1.11 and Reporting Standard of Practice 2, the final report must include: a) use of the current International System for Human Cytogenetic Nomenclature (ISCN); b) the number of cells analyzed and, when applicable, the number of karyotypes; c) band resolution; d) suggestions for additional testing when appropriate; e) suggestions for the physician and/or patient to obtain genetic counseling; f) reports that include FISH results must also include: i. probe target and vendor; ii. cutoff values for interphase FISH; and g) reports that include chromosomal microarray analysis (CMA) must include: i. platform description, including number and distribution of probes; and ii. genome build used for analysis and interpretation.	A summary and interpretation of the results are recommended. Results may be reported in other formats *in addition to* ISCN.

Cytogenetics	
Standard	***Guidance***
Cytogenetics Standard of Practice 13 (CG S13): Required Records Records for each case must include: media used; reactions observed; culture conditions including incubation times; adverse observations; subculturing information (if any); number of cells analyzed and additional cells counted; type of banding; the number of cells from which karyotypes were prepared and karyotypes prepared.	
Cytogenetics Standard of Practice 14 (CG S14): Records Retention The laboratory must have a system for maintaining and retrieving the entire case record according to Document and Specimen Retention Standard of Practice 9 for the required twenty-five (25) years, including, when applicable, the original: a) metaphase and interphase images and karyotypes; b) metaphase and interphase fluorescence in situ hybridization (FISH) images representative of results; and c) chromosomal microarray analysis (CMA) analysis file(s) that include relative copy number and genotype, as applicable, and values for data quality metrics.	The laboratory must have mechanisms to ensure that data is retrievable when the electronic reporting systems are upgraded or replaced according to Laboratory Information Systems Standard of Practice 2. Report retention is required under 10 NYCRR paragraph 58-1.11(c)(5).