

Cytogenetics		
Tag #	Standard	Guidance
	The following specialty sustaining standards of practices shall be incorporated into the laboratory's quality management system, where applicable to the scope of services provided.	
CG1	<p>Cytogenetics Standard 1</p> <p>The laboratory shall request clinical information necessary for proper initiation of test procedures and interpretation of test results, including, for prenatal analysis, the gestational dating.</p>	This may be accomplished by including an appropriate section on the test requisition. If the clinical information is not included with the specimen, the laboratory should request this information. If the clinical information is not received, the laboratory record should be so noted and the report should state that the clinical information was not provided and should include any limitations of the result due to this omission.
CG2	<p>Cytogenetics Standard 2</p> <p>The laboratory shall have a system to distinguish specimen types to assure proper processing, handling and analysis, to facilitate quality assurance review, and to segregate data for reporting.</p>	The identification system should be part of the accession system in order to identify the specimen type.
CG3	<p>Cytogenetics Standard 3</p> <p>Patient identification using at least two unique means of identification shall be maintained through all phases of specimen processing and analysis.</p>	This applies to retained specimens, culture vessels, cell preparations, microscope slides, cell images and karyotypes.
CG4	<p>Cytogenetics Standard 4</p> <p>The laboratory shall use a biological safety cabinet for processing all specimens and cultures.</p>	<p>The use of a biological safety cabinet is imperative to reduce environmental contamination.</p> <p>Refer to Safety Sustaining Standard of Practice 9 (SSP9) for operational guidelines.</p>

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<p>CG5a</p> <p>CG5b</p> <p>CG5c</p>	<p>Cytogenetics Standard 5</p> <p>The laboratory shall establish and implement a procedure for:</p> <p>a) contamination control in media;</p> <p>b) monitoring bacterial, viral, fungal and mycoplasma contamination; and,</p> <p>c) in-house growth support testing of tissue culture media.</p>	<p>a) Laboratories that choose not to routinely use antibiotics in cultures should document that individual cultures are routinely checked for signs of contamination.</p> <p>b) Laboratories that use commercially prepared media should retain the manufacturer's documentation that each shipment or lot of media has been subjected to appropriate quality control procedures. The user should visually examine each shipment for contamination, appearance, or evidence of exposure to extremes of temperature, and notify the media manufacturer of problems related to the quality of the media, including failure to support growth or provide expected colony size, or evidence of contamination.</p> <p>c) In-house growth support tests may include parallel testing of the mitotic index or cell doubling time of cultures and criteria for acceptance for growth support should be established. This may include growth support studies performed by the manufacturer if available.</p>
<p>CG6a</p> <p>CG6b</p> <p>CG6c</p>	<p>Cytogenetics Standard 6</p> <p>The laboratory shall prepare replicate independently established cultures:</p> <p>a) for prenatal specimens, a minimum of three cultures shall be set up for each specimen;</p> <p>b) for other tissue or fibroblast cultures, a minimum of three cultures shall be set up for each specimen; and,</p> <p>c) for all other specimens, duplicate cultures shall be set up.</p>	<p>Analyzed cells should be selected from at least two independently established cultures, except for routine blood cultures when the laboratory has pre-determined that adequate numbers and quality of cells with consistent results are obtained from a single culture.</p>

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CG7	<p>Cytogenetics Standard 7</p> <p>Prenatal cultures shall be split between two incubators used exclusively for prenatal cultures with independent electrical circuits and emergency alarms.</p>	<p>If such arrangements are not feasible, the laboratory should establish a written protocol for prompt handling of prenatal cultures in the event of an equipment failure that might adversely affect viability and test outcome.</p>
CG8	<p>Cytogenetics Standard 8</p> <p>Independently established prenatal cultures shall be processed so as to maintain individual culture integrity.</p>	<p>Processing includes setting up, feeding, and harvesting cultures, and labeling slides.</p>
CG9	<p>Cytogenetics Standard 9</p> <p>The laboratory shall establish and implement procedures to ensure utilization of accepted intervals of culture to optimize cell division.</p>	<p>Approximate processing times vary for each diagnostic area, but generally should fall within the following time frames:</p> <p>Blood: 48-72 hours; 96 hours for special methods Amnio: 6-14 days Tissue: 1-6 weeks Bone Marrow - Direct: 72 hours Others: As established by the laboratory.</p>

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<p>CG10a</p> <p>CG10b</p> <p>CG10c</p> <p>CG10d</p>	<p>Cytogenetics Standard 10</p> <p>The laboratory shall prepare a minimum of two karyotypes per specimen:</p> <p>a) if more than one cell line is detected, a minimum of one karyotype per cell line;</p> <p>b) using photographic or other image reproduction techniques;</p> <p>c) using banded cells which meet the laboratory's pre-established criteria for banding quality and resolution; and,</p> <p>d) identified with the metaphase source and specimen identifiers.</p>	<p>c) The laboratory shall identify individual chromosomes by banding methods, including G, Q or R or other methods that allow identification of all homologs.</p> <p>The laboratory shall document policy and review procedures to ensure that the intended chromosome band resolution, or other appropriate measure for non-banded preparations, is attained and is appropriate to the specimen and clinical information provided in order to rule out the cytogenetic abnormality(ies) reasonably expected based on the clinical information provided.</p> <p>The average band resolution attained should be included in the result report (Cytogenetics Standard 21c).</p> <p>If the band resolution attained is not optimal for the clinical indications for testing, appropriate comments and recommendations should be included in the result report (Cytogenetics Standard 21d,e,f).</p> <p>d) The metaphase may be identified by vernier location, and/or film and frame number of photograph.</p>
<p>CG11a</p> <p>CG11b</p>	<p>Cytogenetics Standard 11</p> <p>For sex chromatin studies (or Barr bodies):</p> <p>a) a minimum of 100 cells shall be examined; and,</p> <p>b) atypical results shall be considered preliminary and shall be confirmed with chromosome analysis.</p>	<p>This assay is considered outmoded and inconclusive; however, it may be useful in very limited circumstances. Performance of this test is limited to laboratories holding a cytogenetics permit.</p>

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<p>CG12a</p> <p>CG12b</p>	<p>Cytogenetics Standard 12</p> <p>The laboratory shall analyze a minimum number of metaphases as indicated below:</p> <p>a) a minimum of 20 metaphases, except for prenatal, in situ, which requires 15 metaphases; and,</p> <p>b) count cells from at least two cultures for all specimens except peripheral blood for constitutional chromosome abnormality analysis.</p>	<p>Analyzed means to establish the number of centric chromosomes in a metaphase AND evaluate individual chromosomes in their entirety, i.e., each metaphase is critically analyzed, including chromosome count, sex chromosome complement, cytogenetic aberrations and vernier location.</p> <p>The minimum count will often be exceeded when multiple cell lines are observed. Based on a laboratory's pre-established criteria, cells from replicate cultures may be analyzed.</p> <p>When mosaicism is suspected on the basis of a phenotype that does not fit with the karyotype, when sex chromosome abnormalities are suspected, or when single trisomic cells are found during a study, an analysis of at least 50 cells is recommended.</p>
<p>CG13a</p> <p>CG13b</p> <p>CG13c</p> <p>CG13d</p> <p>CG13e</p>	<p>Cytogenetics Standard 13</p> <p>For fragile X determinations, the laboratory shall:</p> <p>a) use two different culture systems;</p> <p>b) include an inducing agent in at least one culture;</p> <p>c) monitor folate sensitive sites to determine whether the induction is working;</p> <p>d) use banded chromosome preparations; and,</p> <p>e) analyze 50 male or 100 female cells.</p>	<p>Molecular testing for Fragile X syndrome is recommended over the less sensitive cytogenetic assay.</p> <p>Records should indicate which inducing agent(s) were used.</p>

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CG14	<p>Cytogenetics Standard 14</p> <p>Routine blood cell analysis for constitutional chromosome abnormalities shall be performed on all specimens submitted for fragile X analysis.</p>	
CG15	<p>Cytogenetics Standard 15</p> <p>For laboratories conducting spontaneous breakage studies a normal (negative control) shall be included with each culture event.</p>	
CG16	<p>Cytogenetics Standard 16</p> <p>For laboratories conducting breakage studies on presumed positive specimens, a normal (negative control) and if possible an abnormal control for the condition in question shall be included with each culture event.</p>	
CG17	<p>Cytogenetics Standard 17</p> <p>The laboratory shall establish and implement a protocol for checking microscope stage vernier readings, and making corrections as necessary.</p>	
CG18	<p>Cytogenetics Standard 18</p> <p>Records for each case shall 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 utilized, the number of cells from which karyotypes were prepared and karyotypes prepared.</p>	

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CG19	Cytogenetics Standard 19 The laboratory shall have a system for maintaining and retrieving the entire case record, including the original metaphase and interphase images and karyotypes, when applicable, for the required 25 years.	This applies to image analysis software as well.
CG20	Cytogenetics Standard 20 Preliminary reports shall include a statement regarding the preliminary nature of the result(s). Their issuance shall be documented on the final report.	This standard applies to verbal, electronic or written preliminary reports.

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	<p>Cytogenetics Standard 21</p> <p>In addition to the requirements of Part 58, the final report shall include:</p>	<p>A summary and interpretation of the results are recommended.</p>
CG21a	a) use of the current International System for Human Cytogenetic Nomenclature (ISCN);	a) Refer to ISCN-95
CG21b	b) the number of cells analyzed and, when applicable, the number from which karyotypes were prepared;	
CG21c	c) band resolution attained;	
CG21d	d) in cases of culture failure or where a definitive diagnosis is not possible, suggestions for additional testing;	
CG21e	e) an interpretation of findings;	
CG21f	f) a statement on limitations of the test, including possible inaccuracies;	
CG21g	g) suggestions as to whom the physician and/or patient may consult for discussion of prognosis implications of abnormal results (e.g., genetic counseling); and,	
CG21h	<p>h) reports for fragile X analysis shall include a statement on the limitations of the test in cases with:</p> <p style="margin-left: 40px;">i) low positive (less than 4%) fragile X results;</p> <p style="margin-left: 40px;">ii) negative fragile X results; and,</p> <p style="margin-left: 40px;">iii) low folate-sensitive site expression.</p>	<p>h) The report should indicate that, for the most part, this test has been clinically superseded by molecular methods.</p>

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CG22	<p>Cytogenetics Standard 22</p> <p>Reports shall contain the signature of the qualified person who reviewed, approved and/or diagnosed the case.</p>	<p>For purposes of this standard, a qualified person is a director or assistant director who holds a valid New York State certificate of qualification in Cytogenetics. Laboratories using electronic signatures should have a procedure in place that ensures and documents the qualified person's authorization for each signature occurrence (such as access limited by password).</p>
CG23	<p>Cytogenetics Standard 23</p> <p>Information relevant to a particular case shall be readily trackable and retrievable within 24 hours of request.</p>	
CG24	<p>Cytogenetics Standard 24</p> <p>The laboratory shall have the capability to track a specimen from accession number to karyotypes, when applicable, and report, to microscope slide and conversely.</p>	

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<p>CG25a</p> <p>CG25b</p> <p>CG25c</p> <p>CG25d</p>	<p>Cytogenetics Standard 25</p> <p>The laboratory shall establish critical limits for turn-around-times of certain tests important for prompt patient management decisions, which minimally include:</p> <p>a) final reports of prenatal analyses are provided within 15 days in at least 95% of cases;</p> <p>b) final reports of peripheral lymphocyte and bone marrow analyses are provided within 28 days in at least 90% of cases;</p> <p>c) <u>preliminary</u> reports for STAT fetal blood or STAT neonatal blood samples are available within 72 hours in at least 90% of cases; and,</p> <p>d) final reports for non-neoplastic fibroblast analysis are provided within six weeks in at least 90% of cases.</p>	<p>The laboratory should have a policy to ensure that later gestational age specimens are given priority so that results are released prior to the 25th week of gestation in order to allow patient decisions regarding pregnancy termination.</p>
<p>CG26</p>	<p>Cytogenetics Standard 26</p> <p>The laboratory shall monitor and document the nature and rate of cultures that fail to yield metaphases, and take remedial action in all cases.</p>	<p>This should be an ongoing quality assurance monitor.</p>
<p>CG27</p>	<p>Cytogenetics Standard 27</p> <p>The laboratory shall establish and implement procedures to obtain follow-up information for confirmation of all prenatal diagnosis.</p>	<p>The responsibility of obtaining this information cannot be delegated.</p> <p>Discrepancies of phenotypic sex and abnormal outcome should be fully evaluated. This is the only means a laboratory has to obtain predictive value of the analysis.</p>

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	<p>Cytogenetics Standard 28</p> <p>The laboratory shall notify practitioners wishing to order a cytogenetic test that informed consent is required and shall make available to the practitioner test-specific information for patient use in decision-making and the informed consent process. These materials shall include:</p>	<p>Informed consent is not required for cancer cytogenetic testing.</p>
CG28a	a) general description and statement of purpose for the test;	Laboratories should be aware that cytogenetic testing is also covered by Section 79-l of the Civil Rights Law.
CG28b	b) indication that the individual may wish to obtain professional genetic counseling prior to giving consent;	Reasonable effort should be made to obtain patient consent and document the process.
CG28c	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;	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.
CG28d	d) a general description of the disease or condition related to the test;	
CG28e	e) the level of certainty that a positive test result serves as a predictor of the disease;	
CG28f	f) the persons or organizations to whom the test result may be disclosed;	g) Research testing may be performed on residual specimen pursuant to a research protocol approved by an institutional review board provided that:
CG28g	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 expressly authorized in the consent; and,	1) the subject, or the subject's authorized representative, has provided written informed consent for the specific research;
CG28h	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.	2) the sample has been permanently stripped of identifying information; and 3) the research participant has consented to the de-identification.

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CG29	Cytogenetics Standard 29 Laboratories must obtain the subject's written consent, or if the individual lacks the capacity to consent, the signature of the person authorized to consent for the individual, before records, findings or results may be re-disclosed to any individual or organization other than those authorized on the test requisition to receive the result.	

Genetic Testing		
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	The following specialty sustaining standards of practices shall be incorporated into the laboratory's quality management system, where applicable to the scope of services provided.	Laboratories must submit full validation packages for all test using molecular techniques except for assays labeled by the FDA as "For <i>in vitro</i> diagnostic testing." Copies of the Submission Guidelines can be downloaded from www.wadsworth.org/labcert/clep/clep.html or call 518-473-5378.
GT1	<p>Genetic Testing Standard 1</p> <p>Identification of the patient shall be maintained through all phases of specimen processing and analysis.</p>	It should be possible to go readily from accession number to patient files and report and conversely.
GT2	<p>Genetic Testing Standard 2</p> <p>For linkage analysis-based tests, each family studied shall be assigned a unique code to monitor relatedness between core families.</p>	There should be a system in place to link family identifiers with individual patient identifiers.
GT4a GT4b GT4c	<p>Genetic Testing Standard 4</p> <p>The SOPM shall include up-to-date references which document:</p> <p>a) linkage relationships for each disorder offered by indirect linkage methods, which minimally address:</p> <p>i) proximal or distal to disease gene; and,</p> <p>ii) recombination fractions and/or zero values at 95% confidence intervals; and,</p> <p>b) loci, probes, and/or primers and conditions of their use.</p> <p>c) clinical validity and utility if applicable and detection of variants in disease populations.</p>	<p>These may be literature references or, for in-house generated probes, the reference may be the laboratory's validation studies.</p> <p>Refer to Standard Operating Procedures and Compliance Standards for additional SOPM requirements.</p>

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	<p>Genetic Testing Standard 5</p> <p>The laboratory shall notify practitioners wishing to order a genetic test that informed consent is required and shall make available to the practitioner test-specific information for patient use in decision-making and the informed consent process. These materials shall include:</p>	<p>Laboratories should be aware that genetic testing is also covered by Section 79-I of the Civil Rights Law.</p> <p>Reasonable effort should be made to obtain patient consent and document the process.</p> <p>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.</p> <p>g) Research testing may be performed on residual specimen pursuant to a research protocol approved by an institutional review board provided that:</p> <ol style="list-style-type: none"> 1) the subject or the subject's authorized representative, has provided written informed consent for the specific research; 2) the sample has been permanently stripped of identifying information; and 3) the subject has consented to the de-identification.
GT5a	a) general description and statement of purpose for the test;	
GT5b	b) indication that the individual may wish to obtain professional genetic counseling prior to giving consent;	
GT5c	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;	
GT5d	d) a general description of the disease or condition related to the test;	
GT5e	e) the level of certainty that a positive test result serves as a predictor of the disease;	
GT5f	f) the persons or organizations to whom the test result may be disclosed;	
GT5g	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 expressly authorized in the consent; and,	
GT5h	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.	

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<p>GT6a</p> <p>GT6b</p> <p>GT6c</p> <p>GT6d</p> <p>GT6e</p> <p>GT6f</p>	<p>Genetic Testing Standard 6</p> <p>Reports shall include:</p> <p>a) a statement of and an interpretation of findings;</p> <p>b) a statement on technical limitations of the test, including possible inaccuracies;</p> <p>c) suggestions for additional or alternative testing, if applicable;</p> <p>d) recommendations for referral to a genetic provider when appropriate;</p> <p>e) methodology used for the test; and,</p> <p>f) a list of all of the variants examined in the assay if applicable.</p>	<p>a) A summary and interpretation of the results directly applicable to the patient are recommended. The laboratory should also provide a voice or FAX number of a person qualified to assist practitioners with the interpretation of the results.</p> <p>b) Technical limitations should include the possibility of laboratory error. Literature references applicable to the analysis should be included.</p>
<p>GT7</p>	<p>Genetic Testing Standard 7</p> <p>Reports shall contain the signature of the qualified person who reviewed, approved, and interpreted the test results, unless the test is defined as a screening test.</p>	<p>For purposes of this standard, a qualified person is a director or assistant director who holds a valid New York State certificate of qualification in the appropriate Genetic Testing subcategory.</p> <p>Laboratories using electronic signatures should have a procedure in place that ensures and documents the qualified person's authorization for each signature occurrence (such as access limited by password).</p> <p>A screening test is characterized by one or both of the following:</p> <p>a) the package insert indicates that the assay is for screening purposes only; and/or</p> <p>b) there is no pre-existing indication that the individual is at high risk for a genetic disease prior to testing, for example, newborn screening tests.</p>

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GT8	<p>Genetic Testing Standard 8</p> <p>The laboratory shall establish critical limits for turn-around-times of certain tests important for prompt patient management decisions.</p>	<p>The laboratory should have a policy to ensure that later gestational age specimens are given priority so that results are released prior to the 25th week of gestation in order to allow patient decisions regarding pregnancy termination.</p>
GT9	<p>Genetic Testing Standard 9</p> <p>The laboratory shall establish and implement procedures to obtain follow-up information for prenatal diagnosis confirmation.</p>	<p>Discrepancies should be fully evaluated.</p>
GT10	<p>Genetic Testing Standard 10</p> <p>The laboratory shall run appropriate controls with each run of patient specimens.</p>	<p>Controls should be selected based on the patient population and should be as comprehensive as possible based on the rarity of the disease. For example, a heterozygous sample or a normal and a homozygous mutant sample is sufficient for single mutation assays. Cases of rare variants should be verified, e.g. bi-directional sequence or repeat of the sample.</p>
GT11	<p>Genetic Testing Standard 11</p> <p>The laboratory shall keep up-to-date records of DNA probe documentation that minimally includes chromosome/band, and restriction enzyme(s) needed to visualize the RFLP.</p>	

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GT12a GT12b GT12c GT12d	<p>Genetic Testing Standard 12</p> <p>For each applicable analysis, the laboratory reports shall contain:</p> <p>a) name of the test DNA locus as defined by the International Human Gene Mapping Workshop;</p> <p>b) name of the probe;</p> <p>c) name of the restriction endonuclease; and,</p> <p>d) size or alphanumeric description of all detected alleles.</p>	<p>b) This is relevant when performing Southern blot techniques.</p> <p>c) This is relevant when performing PCR/RFLP and Southern blot techniques.</p> <p>d) Any variant detected must be reported regardless of clinical implication. Alternatively, it must be clearly noted that the information can be made available to the physician.</p>
GT13	<p>Genetic Testing Standard 13</p> <p>Conditions of time, temperature and concentration which achieve desired amplification results shall be empirically determined, periodically verified and documented for each set of primers using known controls.</p>	<p>There should be equal amplification of normal and mutant alleles. Proper reaction conditions should be documented on worksheets.</p>
GT14	<p>Genetic Testing Standard 14</p> <p>Laboratories must obtain the subject's written consent, or if the individual lacks the capacity to consent, the signature of the person authorized to consent for the individual, before records, findings, or results may be re-disclosed to any individual or organization other than those authorized on the test requisition to receive the result.</p>	