1) Claussen U, *et al.* Exclusion of chromosomal mosaicism in perinatal diagnosis. *Human Genetics.* 1984;67:23-28
2) Hsu LYF, *et al.* Proposed guidelines for diagnosis of chromosome mosaicism in amniocytes based on data derived from chromosome mosaicism and pseudomosaicism studies. *Prenatal Diag*. 1992;12:555-573
3) American College of Medical Genetics, Standards and Guidelines for Clinical Genetics Laboratories, 2021 edition.
4) Ing PS, *et al*. Detection of mosaicism in amniotic fluid cultures: a CYTO2000 collaborative study. *Genet Med.* 1999 Mar-Apr;1(3):94-7

CYG.40700 Counting - Amniotic Fluid or Chorionic Villus Culture (Non-In Situ) Phase II

For any non-in situ amniotic fluid cell or chorionic villus culture (ie, trypsinized culture), at least 20 cells are counted, with cells from at least two cultures (this may include any combination of in situ and non-in situ cultures), with the exception of abbreviated studies.

NOTE: The number of cells counted should be distributed as equally as possible between independently established cultures. Under special clinical circumstances, fewer than 20 cells may be counted. The laboratory must have written criteria for the circumstances under which an abbreviated study can be performed. Criteria should address the rationale for such studies, the clinical reason for referral, and the minimum number of cells counted.

Evidence of Compliance:
- ✓ Patient records/worksheets

REFERENCES
1) Claussen U, *et al.* Exclusion of chromosomal mosaicism in perinatal diagnosis. *Human Genetics.* 1984;67:23-28
2) Hsu LYF, *et al.* Proposed guidelines for diagnosis of chromosome mosaicism in amniocytes based on data derived from chromosome mosaicism and pseudomosaicism studies. *Prenatal Diag.* 1992;12:555-573
3) American College of Medical Genetics, Standards and Guidelines for Clinical Genetics Laboratories, 2021 edition.

CYG.40900 Counting - Non-Neoplastic Solid Tissue Samples Phase II

For non-neoplastic solid tissue samples, at least 20 cells are counted with the exception of abbreviated studies.

NOTE: Under specific clinical circumstances, fewer than 20 cells may be counted (eg, for confirmation of an abnormal prenatal chromosome result). The laboratory must have written criteria for the circumstances under which an abbreviated study can be performed. Criteria should address the rationale for such studies, the clinical reason, and the minimum number of cells counted.

Evidence of Compliance:
- ✓ Patient records/worksheets

REFERENCES
1) American College of Medical Genetics, Standards and Guidelines for Clinical Genetics Laboratories, 2021 edition.

NUMBER OF CELLS ANALYZED

Inspector Instructions:

READ

- Policy for number of cells analyzed
- Sampling of patient records/worksheets

Analyses should be performed from two independent cultures, if possible.

CYG.41100 Analysis - Non-neoplastic Samples Phase II

A minimum of five cells, with the exception of abbreviated studies, are analyzed.

NOTE 1: Under special clinical circumstances, fewer than five cells may be analyzed. Examples of such circumstances are confirmation of an abnormal prenatal chromosome result, in conjunction with chromosomal microarray analysis, or peripheral blood chromosome studies on family members to exclude a previously identified chromosome rearrangement. The laboratory must have written criteria for the circumstances under which an abbreviated study can be performed. Criteria should address the rationale for such studies, the clinical reason for referral, and the minimum number of cells analyzed.

NOTE 2: Under other special clinical circumstances (eg, rule out mosaicism of a numerical or structural abnormality, including sex chromosome mosaicism, which may involve both numerical and structural abnormalities), additional cells may need to be analyzed. The laboratory must have written criteria for the circumstances in which additional cells should be analyzed.

Evidence of Compliance:

✓ Patient records/worksheets

REFERENCES

1) Wiktor AE, Bender G, Van Dyke DL. Identification of sex chromosome mosaicism: Is analysis of 20 metaphase cells sufficient? *Am J Med Genet* Part A 2009; 149A:257-259.
2) Wolff DJ, Van Dyke DL. Laboratory guideline for Turner syndrome. *Genet Med* 2010:12(1):52-55.
3) American College of Medical Genetics, Standards and Guidelines for Clinical Genetics Laboratories, 2021 edition, Section E5.1.2.

CYG.41500 Analysis - Neoplastic Samples — Phase II

For neoplastic disorders studied in marrow, blood or solid tumor specimens, at least 20 cells are analyzed, if possible.

NOTE: Under special clinical circumstances, fewer than 20 cells may be analyzed in lymphomas, solid tumors, and metastatic neoplasms with complex karyotypes. A sufficient number of metaphase cells (generally at least 10) should be analyzed to permit characterization of the abnormal clone(s). The circumstances under which abbreviated studies may be performed must be stated in the laboratory procedure.

Evidence of Compliance:

✓ Patient records/worksheets

REFERENCES

1) American College of Medical Genetics, Standards and Guidelines for Clinical Genetics Laboratories, 2021 edition.

CYG.41550 Analysis - Neoplastic Samples — Phase I

For neoplastic bone marrow/blood/solid tumor specimens, two or more cultures are analyzed, when possible.

NOTE: For neoplastic bone marrow/blood/solid tumor specimens, cells from two or more culture conditions are analyzed, when possible.

Evidence of Compliance:

✓ Patient records/worksheets

REFERENCES

1) Lebeau M. A.C.T. cytogenetic analysis of hematological malignant diseases, In Cytogenetics laboratory manual, MJ Barch (ed), 2nd ed. Raven Press, 1991:chap 9