Duplicate amniotic fluid and chorionic villus flasks or dishes are harvested independently.

NOTE: To prevent failures due to contamination or technical error, all cultures from a patient should not be harvested in the same batch.

Evidence of Compliance:
- ✓ Patient records/worksheets

NUMBER OF CELLS COUNTED

Inspector Instructions:

READ

- Sampling of test procedures for cells counted
- Sampling of patient records/worksheets

CYG.40500 Counting - Stimulated Blood Samples Phase II

For stimulated blood samples (non-neoplastic disorders), at least 20 cells are counted with the exception of abbreviated studies.

NOTE 1: Under special clinical circumstances (eg, rule out sex chromosome mosaicism or mosaicism of another numerical or structural abnormality), additional cells may need to be counted. The laboratory must have written criteria for these circumstances in which additional cells should be counted.

NOTE 2: Under other 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) 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.40600 Counting - Amniotic Fluid or Chorionic Villus (In Situ) Samples Phase II

For amniotic fluid or chorionic villus (in situ) samples, at least 15 cells from 15 different colonies are counted, with cells from at least two 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 circumstances, fewer than 15 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.
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