- *Refer to CYG.43200 for in situ hybridization*

Evidence of Compliance:

✓ Records of alternative control procedures

REFERENCES

1) Department of Health and Human Services, Centers for Medicare and Medicaid Services. Clinical laboratory improvement amendments of 1988; final rule. *Fed Register*. 2003(Jan 24): [42CFR493.1256(h)].

REPORTS

Reporting requirements for use of analyte-specific reagents and other reagents used in laboratory-developed tests are included in the All Common Checklist (COM.40850).

Inspector Instructions:

	• Sampling of reporting policies and procedures • Sampling of patient preliminary and final reports for completeness, appropriate use of ISCN edition and recommendations for genetic consultation or additional studies • Sampling of TAT statistics
	• How does your laboratory maintain records of verbal/telephone preliminary reports? • What is your course of action when turnaround times exceed limits?
	• Search for reporting errors. Determine whether the investigation was thorough and appropriate corrective action was taken.

CYG.31825 Preliminary Reports **Phase I**

Provision of preliminary reports (especially verbal, telephone reports) is recorded on the final report.

CYG.31875 Final Report Elements **Phase II**

The final reports contain all of the following required elements:

1. **Name and address of testing laboratory**
2. **Patient name**
3. **Unique identifying number**
4. **Patient date of birth**
5. **Name of physician, or authorized person ordering test**
6. **Specimen source**
7. **Date specimen received in the laboratory**
8. **Date of report**
9. **Clinical indication(s) for the test**
10. **Number of cells counted, analyzed, and karyograms prepared, as applicable**
11. **Band resolution (required only for constitutional cases), as applicable**
12. **Banding methods, as applicable**
13. **Comment on adequacy of specimen, if indicated**

NOTE: Items 10, 11, and 12 above apply to conventional cytogenetics (G-banded) analyses.

REFERENCES
1) Department of Health and Human Services, Centers for Medicare and Medicaid Services. Clinical laboratory improvement amendments of 1988; final rule. *Fed Register.* 2003(Jan 24):1043-1044[42CFR493.1276], 1047-1048 [42CFR493.1291]

CYG.31880 Report Review **Phase II**

The final report for conventional cytogenetics (G-banding) and metaphase FISH analyses are reviewed and signed by the cytogenetics section director (or qualified cytogeneticist designated by the section director).

NOTE: A qualified designee must be: 1) a doctor of medicine or doctor of osteopathy licensed to practice medicine in the state in which the laboratory is located; or 2) hold an earned doctoral degree in a biological science or clinical laboratory science from an accredited institution.

In addition, each qualified cytogeneticist must have either a) successfully completed an accredited fellowship with an emphasis on clinical cytogenetics, or b) in the absence of fellowship training, have four years of training or experience or both in human medical genetics or pathology, two of which have been in clinical cytogenetics.

CYG.31903 Turnaround Time **Phase II**

The laboratory has defined sample turnaround times that are appropriate for the intended purpose of the test and performs ongoing monitoring for compliance.

NOTE: Appropriate turnaround times will vary by test type and clinical application. There are certain clinical situations in which rapid completion is essential. For example, inappropriate delays in completing a prenatal diagnosis test can cause unacceptable emotional stress for the parents, make ultimate pregnancy termination (if chosen) much more difficult, or even render the results of the test unusable.

Evidence of Compliance:

✓ Records showing that defined turnaround times are routinely met

REFERENCES
1) American College of Medical Genetics, Standards and Guidelines for Clinical Genetics Laboratories, 2021 edition.
2) Shao L, Akkari Y, Cooley LD, et al. Chromosomal microarray analysis, including constitutional and neoplastic disease applications, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). *Genet Med.* 2021;23(10):1818-1829.

CYG.32071 Final Report Contents **Phase II**

The final report includes a summary of the results and an interpretation that includes correlation of the cytogenetic results with clinical information and previous studies, when appropriate.

NOTE: The interpretation must be written to facilitate understanding by a non-geneticist.

CYG.32100 Nomenclature **Phase II**

For conventional cytogenetic studies, the current International System for Human Cytogenetic Nomenclature (ISCN) is used correctly in the final report.

NOTE: The purpose is to provide universal interpretation of cytogenetic results without pictures of the karyogram.

REFERENCES
1) McGowan-Jordan J, Hastings R, Moore S, eds; International Standing Committee on Human Cytogenomic Nomenclature. *ISCN: An International System for Human Cytogenomic Nomenclature (2020).* Basel, New York: Karger; 2020.