GENOMIC COPY NUMBER ANALYSIS USING ARRAYS

This technology is used to assess copy number of genomic regions. Regardless of platform used (eg, CGH, SNP), reagents for hybridization and detection, or analytic components for evaluation, the laboratory is responsible for assuring that appropriate controls are performed and records retained for all aspects of analysis. This technology may also include a variety of reverse and forward hybridization formats. Reverse hybridization arrays use multiple unlabeled probes on a solid support to investigate a patient sample that carries a label, either direct (fluorescent or radioactive) or indirect (affinity labels such as biotin, digoxigenin, etc.). Another form of array involves multiple real-time amplification assays to measure multiple targets simultaneously. Controls for arrays monitor those steps carried out by the laboratory (sample preparation and labeling, hybridization and detection) and by the manufacturer (assay preparation, detection and hybridization reagents). Manufacturers also contribute to QC by producing products under good manufacturing practices, providing control material for each analyte, and by providing sequence information or confirmatory tests to resolve ambiguous results.

Inspector Instructions:

READ	• Sampling of array procedures, including nucleic acid extraction and analytical wet bench and bioinformatics processes • Sampling of array validation studies • Sampling of array performance monitoring records • Sampling of patient test reports
ASK	• How does your laboratory validate assay performance prior to test implementation? • What processes are used to monitor ongoing assay performance?
DISCOVER	• Review records of assay performance monitoring. If any problems are found during review of performance monitoring records, or when asking questions, further evaluate the laboratory's investigation and resolution.

CYG.49525 Nucleic Acid Extraction/Isolation/Purification Phase II

Nucleic acids are extracted, isolated, and purified by methods reported in the literature, by an established commercially available kit or instrument, or by a validated method developed by the laboratory.

NOTE: Extraction procedures may combine purification or isolation of nucleic acids according to the level of purity needed for downstream applications.

Evidence of Compliance:

✓ Records to support nucleic acid extraction/isolation/purification is performed by a validated method

REFERENCES

1) South ST, Lee C, Lamb AN, Higgins AW, Kearney HM, Working Group for the American College of Medical Genetics and Genomics Laboratory Quality Assurance Committee. ACMG standards and guidelines for constitutional cytogenomic microarray analysis, including postnatal and prenatal applications: revision 2013. *Genet Med.* 2013; 15(11):901-9.
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.