- *Written procedure for any portion of the wet bench process performed by a referral laboratory, if applicable.*

***REVISED** 12/26/2024*

CYG.49585 Array Analytical Bioinformatics **Phase II**

The laboratory defines the steps in the bioinformatics process (also termed pipeline) used to analyze, interpret, and report array findings.

NOTE: A bioinformatics pipeline includes all algorithms, software, scripts, parameters, reference sequences, and databases, whether in-house, vendor-developed, or open source.

The written procedure must describe the bioinformatics process(es) including, where applicable:

- *Individual software applications (open source, proprietary, and custom scripts) and versioning*
- *Description of input and output data files for each step of the pipeline, including in silico control files or sources*
- *Annotations and their sources (eg, public or private databases, with versions used)*
- *Criteria and thresholds for detection of array findings (eg, minimum number of probes or genomic size for copy number variants)*
- *Determination of the limits of detection*
- *Additional scripts or steps used to connect discrete applications in the pipeline*
- *Quality control metrics, including batch or sample-specific metrics and acceptance and rejection criteria for the results generated by the analytical bioinformatics process. Criteria must be based on metrics and quality control parameters established during test optimization and utilized during validation*
- *Required corrective actions when results fail to meet the laboratory's acceptance criteria*
- *Limitations in the test methodology*
- *Written procedures for any portion of the array bioinformatics process performed by a referral laboratory or a commercial service provider, if applicable. This should include a written description of how the security of identifiable patient information (eg, HIPAA compliance) is ensured during transmission and storage of data by the referral laboratory or commercial service provider.*

CYG.49590 Interpretation and Reporting of Array Findings **Phase I**

The laboratory follows defined criteria for classification, interpretation, and reporting of array findings.

NOTE: The laboratory must have a written algorithm for classifying and interpreting the clinical significance of identified findings. The ACMG guidelines can be used for classification and interpretation of copy number variants in inherited disorders.

Genome-wide array analysis may yield genetic findings unrelated to the clinical presentation for which the patient is undergoing testing. The laboratory policy must describe which, if any, and for what reasons, findings unrelated to the clinical purpose for testing are reported and the method of communication to the ordering physicians and patients, as applicable.

The written policy must include indications for confirmatory testing. The laboratory must determine by confirmation studies during validation if and when confirmatory testing of identified findings should be performed.

Evidence of Compliance:

- ✓ Records of compliance with procedure for classification, interpretation, and reporting of findings **AND**
- ✓ Laboratory database of findings identified and/or reported

REFERENCES