QSA.15.02.01

The laboratory’s verification studies for molecular testing include representatives from each specimen type expected to be tested in the assay and specimens representing the scope of reportable results.

The laboratory performs validation studies for molecular testing.

Elements of Performance (EPs) for QSA.15.02.01

3. The laboratory performs verification studies for molecular testing. The verification studies are documented.

3. The laboratory’s validation studies for molecular testing cover all steps from extraction to final results.

Note: When a laboratory does not perform all or part of the testing process, the laboratory performing the test is responsible for documenting proper validation of the entire testing process (for example, next-generation sequencing and bioinformatics).

QSA.15.04.01

The laboratory validates next-generation sequencing bioinformatics pipelines.

Elements of Performance (EPs) for QSA.15.04.01

1. A qualified medical professional with appropriate training in next-generation sequencing interpretation reviews all validation components and oversees the validation process. (See also HR.01.02.03, EP 8)

2. Validation is only performed after completion of design, development, and optimization of a bioinformatics pipeline and includes all components used in the analysis.

3. Bioinformatics pipeline validation matches and aligns with the laboratory environment where the test is performed.

4. Bioinformatics pipeline validation meets its intended clinical use, specimen, and variant types detected by the next-generation sequencing test.

5. The identity of the sample is preserved throughout each step of the next-generation sequencing bioinformatics pipeline.

6. Supplemental validation is performed when a significant change is made to any component of the bioinformatics pipeline.

QSA.16.01.01
The laboratory uses policies and procedures for molecular genetic testing.

The laboratory follows its policies and procedures for molecular genetic testing.

**Elements of Performance (EPs) for QSA.16.01.01**

4. The laboratory follows its policies and procedures for molecular genetic testing.

4. The laboratory’s policies and procedures for molecular genetic testing address the clinical validity and clinical utility of each individually requested test based on published literature and professional recommendations (for example, Clinical and Laboratory Standards Institute document MM20 [Quality Management for Molecular Genetic Testing]).

Note: For molecular genetic tests, clinical validity refers to the ability of a test to detect the presence or absence of a disease and corresponds to associations between genotype and phenotype. Clinical utility refers to identifying the outcome associated with specific test results.