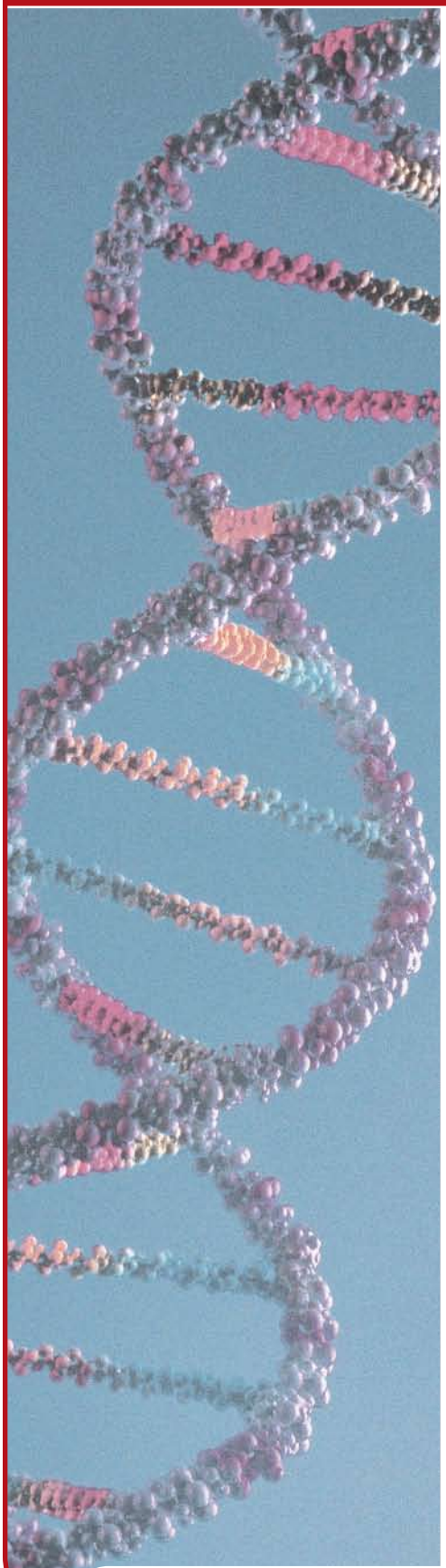
Top 10 Recommendations for Laboratories Performing Molecular Genetic Testing



Top 10 Recommendations for Laboratories

1. **Provide information regarding the laboratory's molecular genetic tests to users to facilitate appropriate test selection and requests, specimen handling and submission, informed decisions, and patient care. Test information to be provided should include:**
   - Information necessary for selecting and requesting appropriate tests;
   - Information on appropriate collection, handling, transport, and submission of specimens;
   - Patient information that the laboratory needs to perform the test and report test results;
   - Implications of test results for the patient's family members, when applicable;
   - Laboratory and/or genetic consultation regarding test selection and ordering, specimen submission, result interpretation and implications; and
   - Cost of testing, whenever possible.

2. **Ensure test requests solicit information needed for selecting appropriate test methods, determining the mutations or variants to be tested, interpreting test results, and timely reporting of test results. Information to be solicited includes:**
   - All information required by CLIA; and
   - Indications for testing, relevant clinical and laboratory information, patient race/ethnicity, family history, and pedigree when applicable.

3. **Ensure adequate establishment and verification of analytic performance specifications before introducing any new molecular genetic test for patient testing, and document available information on clinical validity.**

4. **Implement specific quality control (QC) practices in addition to meeting the applicable general CLIA requirements:**
   - Ensure control materials are comprehensive and resemble patient specimens when possible;
   - Include an extraction control in any test that has a nucleic acid extraction step;
   - Perform control procedures each time patient specimens are tested;
   - Ensure appropriate alternative control procedures when control materials are not available; and
   - Ensure adequate procedures to monitor unidirectional workflow for amplification procedures and prevent cross-contamination.

5. **Participate in available proficiency testing for each molecular genetic test that the laboratory performs, and perform alternative performance assessments for those molecular genetic tests for which no proficiency testing program is available.**

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6. Molecular genetic test reports should provide information necessary for accurate understanding and interpretation of test results. In addition to meeting CLIA test report requirements, laboratories should:

- Include the recommended additional information (e.g., test method, performance specifications and limitations, result interpretation and implications for relatives); and
- Maintain an up-to-date database of the laboratory's molecular genetic tests and provide updates to users when knowledge advancement affects performance specifications or result interpretation.

7. Ensure adequate retention of test reports, records, and tested specimens for quality assurance and quality assessment:

- Molecular genetic test reports should be retained for a minimum of 25 years after the date reported.
- Previously tested specimens that are stable should be retained until the next proficiency testing event or the next alternative performance assessment is performed.

8. Ensure confidentiality of all patient information, including information regarding family members. This is especially important when patient test results are used for the assessment and care of family members.

9. Comply with applicable federal, state, and local requirements regarding direct-to-consumer genetic testing:

- Verify that test requests are from authorized persons;
- Release test results only to persons authorized by applicable laws and regulations to receive test results, persons responsible for using the test results, and/or the referring laboratory; and
- Follow accepted professional guidelines.

10. Ensure personnel have appropriate qualifications needed to fulfill their responsibilities for the high complexity molecular genetic testing they perform:

- Laboratory Directors must, at a minimum, meet the CLIA requirements for laboratory directors of high complexity testing.
- Technical Supervisors should have qualifications equivalent to the CLIA qualification requirements for clinical cytogenetics technical supervisors or current certification in molecular genetic testing.
- Clinical Consultants must meet the minimum qualifications required by CLIA and should have relevant training, experience, or both with the testing for which they provide consultation.
- General Supervisors must fulfill the CLIA requirements for high complexity testing and should have specific training and/or experience with the molecular genetic testing the laboratory performs.
- Testing Personnel must meet the CLIA qualification requirements for high complexity testing and have training/competency with the molecular genetic testing they perform.

For additional information go to:
http://www.cdc.gov/dls/moleculartesting

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