MAP Post-Analytic Checklist

Contents

Part 7: Results Reporting (Post-Analytic)

Procedures for the reporting of results should be defined to ensure the clear understanding of the screening results to a non-expert physician. Discrepancies between preliminary and final reports should be investigated and documented.

7A: Summary of Methods, Loci or Mutations Tested, and Interpretation

7A.1: Does the laboratory have guidelines for reporting of molecular test results?
N/A ☐ YES ☐ NO ☐

Comments:

7A.2: Does the results report clearly describe the gene/loci target of the molecular assay?
N/A ☐ YES ☐ NO ☐

Comments:

7A.3: Does the results report include a summary of the molecular test method?
N/A ☐ YES ☐ NO ☐

Comments:

7A.4: For a targeted genotyping assay, does the results report list all mutations or variations that are detected by the assay?
N/A ☐ YES ☐ NO ☐

Comments:

7A.5: For quantitative molecular assays, does the results report define the normal reference range of the analytes targeted?
N/A ☐ YES ☐ NO ☐

Comments:

7A.6: Does the results report contain an analytic interpretation of the results that is readily interpretable by a non-expert physician?

N/A ☐ YES ☐ NO ☐

Comments:

The report should contain the specific information delineated in the standard operating procedure manual. This should include an appropriate summary of the methods, the loci and mutations, as well as the analytic and clinical interpretation, if appropriate.

7A.7: Does the report include an estimation of mutation detection rate and the residual risk of being a carrier for one of the mutations not tested?

N/A YES NO

Comments:

7B: Technical Limitations

7B.1: Does the results report contain a statement of the technical limitations of the molecular assay?

N/A ☐ YES ☐ NO ☐

Comments:

The technical limitations of an assay defined in the standard operating procedure manual should be included in a statement in the results report. Technical limitations of test procedures should include interfering substances, detection limits of the test (e.g., specific DNA sequences assayed, gene deletions detected) as well as the possibility of laboratory error.

7C: Limitations of Findings

7C.1: Does the results report contain a statement that a screen-negative result does not rule out the possibility that the test subject may be a carrier of a mutation in the tested gene/loci?

N/A ☐ YES ☐ NO ☐

Comments:

Testing may not be able to identify all mutations for the disorder. Laboratories should provide the list of mutations in the testing panel and the clinical sensitivities for selected racial/ethnic groups when available and appropriate.

7D: Final Report Review

7D.1: For a screen-positive result, is the final results report reviewed and signed by the laboratory director or designee?

N/A ☐ YES ☐ NO ☐

Comments:

The final report should be reviewed and signed by the laboratory director or designee if there is a subjective or interpretive component to the test.

7D.2: Does the report include a recommendation that patients receive appropriate genetic counseling for screen positives?

N/A ☐ YES ☐ NO ☐

Comments:

The final report should contain a statement that a positive result is an indication that the individual may be predisposed to or have the specific disease or condition tested for and may want to consider further genetic counseling to explain the implications of the test results.

7E: Standard Nomenclature Used to Designate Genes and Mutations

7E.1: Does the results report include standard gene/loci and mutation nomenclature as defined by the Human Genome Variation Society?

N/A ☐ YES ☐ NO ☐

Comments:

Gene names are defined by the HUGO gene nomenclature committee (http://www.genenames.org/), and for mutations nomenclature, refer to den Dunnen and Antonarakis (Hum Mutat 15:7-12, 2000) and den Dunnen and Paalman (Hum Mutat 22:181-182, 2003) or the Human Genome Variation society (http://www.hgvs.org/mutnomen/) for specific guidance. Alleles that have established names in the literature should have both the recommended nomenclature from HGVS and the traditional allele designations.

7F: Critical Alert Procedures

7F.1: Does the results report clearly define a prompt communication process to report test results that require immediate attention?

N/A ☐ YES ☐ NO ☐

Comments:

A protocol for reporting imminently life-threatening results, or panic/alert values should be established.

7G: Literature References

7G.1: Do the results contain references and literature citations applicable to test analysis?

N/A ☐ YES ☐ NO ☐

Comments: