Biomarker Ontology

| Alteration Type | Description |
|---|---|
| Mutation | A genomic alteration resulting in the substitution, insertion, or deletion of nucleotides |
| Amplification | Copy Number Amplification |
| Deletion | Copy Number Deletion |
| Fusion | Rearrangement of chromosomal material that results in complete or partial sequences of two or more separate genes fused to create a novel transcript |
| Overexpression | Protein Overexpression |
| Loss | Protein Loss |
| Any | Any Type of Altered Status |
| Wildtype | Wildtype |
| Positive | Positive for expression of the protein. Dependent on the biomarker, a certain threshold of detection may be required for classification of the sample as "positive" |
| Negative | Lack of protein expression |
| Non-Amplified | Lack of copy number amplification |
| Instability Intermediate | Instability of microsatellite loci was detected at an intermediate level |
| High | Level of the biomarker is considered "high" by the testing platform. Currently used to describe the level of tumor mutation burden |
| Intermediate | Level of the biomarker is considered "intermediate" by the testing platform. Currently used to describe the level of tumor mutation burden |
| Low | Level of the biomarker is considered "low" by the testing platform. Currently used to describe the level of tumor mutation burden |
| Instability High | Instability of microsatellite loci was detected at a high level |
| Instability Low | Instability of microsatellite loci was detected at a low level |
| Stable | Instability of microsatellite loci was not detected |
| Tumor Positive | Expression of the biomarker was detected specifically within tumor cells |
| Immunocyte Positive | Expression of the biomarker was detected specifically within immunoctyes |
| Tumor Negative | Expression of the biomarker was not detected specifically within tumor cells |
| Immunocyte Negative | Expression of the biomarker was not detected specifically within immunocytes |
| Unknown | Biomarker Status is Unknown |
| Uninterpretable | Biomarker Status was tested but results are uninterpretable |
| Clonal | Expression of the biomarker was clonal |
| Equivocal | Biomarker status could not be unequivocally determined |
| Promoter Methylation Positive | DNA methylation was detected at the promoter region of the gene |
| Promoter Methylation Negative | DNA methylation was not detected at the promoter region of the gene |
| Alternative Transcript | Expression of an alternative transcript lacking official nomenclature was detected |
| Phospho Overexpression | Overexpression of a phosphorylated form of the protein was detected |