

CATS
(Cancer Genomic Test Standardized)
Format

Synopsis

By Section of Genomic Data Management,
C-CAT

v1.4~~3~~.0~~2~~~~1~~

2024~~3~~/0~~3~~~~1~~~~0~~/1~~9~~~~1~~~~7~~

I. Synopsis

[Example1. Only shortVariants](#)

[Example2. Only copyNumberAlterations](#)

[Example3. Only rearrangements](#)

[Example4. shortVariants/copyNumberAlterations/rearrangements/otherBiomarkers](#)

Example1. Only shortVariants

```
{
  "metaData": {
    "schemaVersion": "1.43.021",
    "referenceGenome": {
      "grcRelease": "GRCh37.p13"
    }
  },
  "testInfo": {
    "testId": "12345678901231900001",
    "testType": "tumor and matched-normal",
    "panelName": "Multi-gene Panel A",
    "panelVersion": "v1.3.1"
  },
  "sequencingSamples": [
    {
      "itemId": "sample01-tumor-dna",
      "tumorOrNormal": "tumor",
      "nucleicAcid": "DNA",
    }
  ],
  "variants": {
    "shortVariants": [
      {
        "itemId": "shortVariant-1",
        "chromosome": "2",
        "position": 29445258,
        "referenceAllele": "C",
        "alternateAllele": "T",
      }
    ]
  }
}
```

Note: Describe the version of CATS Format used.

Note: Describe "position" and "referenceAllele", "alternateAllele" according to the rules of VCF v4.3.

```
"alternateAlleleFrequency": 0.54,
"transcripts": [
  {
    "transcriptId": "ENST00000389048.4",
    "transcriptDatabaseName": "Ensembl",
    "geneSymbol": "ALK",
    "cdsChange": "c.3467G>A",
    "aminoAcidsChange": "p.C1156Y"
  }
],
"sampleItemId": "sample01-tumor-dna",
Note: Describe the "itemId" of a relevant sequencingSamples instance.
"variantOrigin": "somatic",
"reported": true
}
]
}
}
```

Example2. Only copyNumberAlterations

```
{
  "metaData": {
    "schemaVersion": "1.3.1",
    "referenceGenome": {
      "grcRelease": "GRCh37.p13"
    }
  },
  "testInfo": {
    "testId": "12345678901231900001",
    "testType": "tumor and matched-normal",
    "panelName": "Multi-gene Panel A",
    "panelVersion": "v1.3.1"
  },
  "sequencingSamples": [
    {
      "itemId": "sample01-tumor-dna",
      "tumorOrNormal": "tumor",
      "nucleicAcid": "DNA"
    }
  ],
  "variants": {
    "copyNumberAlterations": [
      {
        "itemId": "copyNumberAlteration-1",
        "chromosome": "8",
        "startPosition": 128706589,
        "endPosition": 128801451,
        "copyNumberMetrics": [
          {
            "value": 11,
            "unit": "copy number"
          }
        ],
        "copyNumberAlterationType": "amplification",
        "transcripts": [
          {
            "geneSymbol": "MYC"
          }
        ]
      }
    ]
  }
}
```

```
    ],  
    "sampleItemId": "sample01-tumor-dna",  
    "variantOrigin": "somatic",  
    "reported": true  
  }  
]  
}  
}
```

Example3. Only rearrangements

```
{
  "metaData": {
    "schemaVersion": "1.3.1",
    "referenceGenome": {
      "grcRelease": "GRCh37.p13"
    }
  },
  "testInfo": {
    "testId": "12345678901231900001",
    "testType": "tumor and matched-normal",
    "panelName": "Multi-gene Panel A",
    "panelVersion": "v1.3.1"
  },
  "sequencingSamples": [
    {
      "itemId": "sample01-tumor-dna",
      "tumorOrNormal": "tumor",
      "nucleicAcid": "DNA"
    }
  ],
  "variants": {
    "rearrangements": [
      {
        "itemId": "rearrangement-1",
        "breakends": [
          Note: "breakends" has information on two breakpoints.
          {
            "chromosome": "2",
            "startPosition": 42510050,
            "endPosition": 42510050,
            "transcripts": [
              {
                "geneSymbol": "EML4"
              }
            ]
          }
        ],
        {
          "chromosome": "2",
```

```

        "startPosition": 29445240,
        "endPosition": 29445240,
        "transcripts": [
            {
                "geneSymbol": "ALK"
            }
        ]
    },
    "orderedGenePairs": [
        [
            "EML4",
            "ALK"
        ]
    ],
    "rearrangementType": "gene fusion",
    "sampleItemId": "sample01-tumor-dna",
    "variantOrigin": "somatic",
    "reported": true
}
]
}
}

```

Example4. shortVariants/copyNumberAlterations/rearrangements/otherBiomarkers

```
{
  "metaData": {
    "schemaVersion": "1.3.1",
    "referenceGenome": {
      "grcRelease": "GRCh37.p13"
    }
  },
  "testInfo": {
    "testId": "12345678901231900001",
    "testType": "tumor and matched-normal",
    "panelName": "Multi-gene Panel A",
    "panelVersion": "v1.3.1"
  },
  "sequencingSamples": [
    {
      "itemId": "sample01-tumor-dna",
      "tumorOrNormal": "tumor",
      "nucleicAcid": "DNA"
    }
  ],
  "variants": {
    "shortVariants": [
      {
        "itemId": "shortVariant-1",
        "chromosome": "2",
        "position": 29445258,
        "referenceAllele": "C",
        "alternateAllele": "T",
        "alternateAlleleFrequency": 0.54,
        "transcripts": [
          {
            "transcriptId": "ENST00000389048.4",
            "transcriptDatabaseName": "Ensembl",
            "geneSymbol": "ALK",
            "cdsChange": "c.3467G>A",
            "aminoAcidsChange": "p.C1156Y"
          }
        ]
      }
    ],
  },
}
```



```

        "sampleItemId": "sample01-tumor-dna",
        "variantOrigin": "somatic",
        "reported": true
    }
],
"copyNumberAlterations": [
    {
        "itemId": "copyNumberAlteration-1",
        "chromosome": "8",
        "startPosition": 128706589,
        "endPosition": 128801451,
        "copyNumberMetrics": [
            {
                "value": 11,
                "unit": "copy number"
            }
        ],
        "copyNumberAlterationType": "amplification",
        "transcripts": [
            {
                "geneSymbol": "MYC"
            }
        ],
        "sampleItemId": "sample01-tumor-dna",
        "variantOrigin": "somatic",
        "reported": true
    }
],
"rearrangements": [
    {
        "itemId": "rearrangement-1",
        "breakends": [
            {
                "chromosome": "2",
                "startPosition": 42510050,
                "endPosition": 42510050,
                "transcripts": [
                    {
                        "geneSymbol": "EML4"
                    }
                ]
            }
        ]
    }
]

```

```

        }
      ]
    },
    {
      "chromosome": "2",
      "startPosition": 29445240,
      "endPosition": 29445240,
      "transcripts": [
        {
          "geneSymbol": "ALK"
        }
      ]
    }
  ],
  "orderedGenePairs": [
    [
      "EML4",
      "ALK"
    ]
  ],
  "rearrangementType": "gene fusion",
  "sampleItemId": "sample01-tumor-dna",
  "variantOrigin": "somatic",
  "reported": true
}
]
},
"otherBiomarkers": [
  {
    "itemId": "biomarker-1",
    "biomarkerType": "MSI",
    "biomarkerMetrics": [
      {
        "value": 5.15,
        "unit": "%",
        "type": "percentage of MSI sites"
      }
    ]
  },
  "state": "stable",

```

```
    "sampleItemId": "sequence-1-tumor-dna",  
    "reported": true  
  },  
  {  
    "itemId": "biomarker-2",  
    "biomarkerType": "TMB",  
    "biomarkerMetrics": [  
      {  
        "value": 34.5680122,  
        "unit": "Muts/Mb",  
        "type": "Mutations per megabase"  
      }  
    ],  
    "state": "high",  
    "sampleItemId": "sequence-1-tumor-dna",  
    "reported": true  
  }  
]  
}
```