

CATS
(Cancer Genomic Test Standardized)
Format

Synopsis

By Section of Genomic Data Management,
C-CAT

v1.3.2

2024/01/19

I. Synopsis

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Example1. Only shortVariants

```
{  
    "metaData": {  
        "schemaVersion": "1.3.2",  
        Note: Describe the version of CATS Format used.  
        "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 "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": 42510050,  
                        "endPosition": 42510050,  
                        "transcripts": [  
                            {  
                                "geneSymbol": "EML4"  
                            }  
                        ]  
                    }  
                ]  
            }  
        ]  
    }  
}
```

```
"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
}
]
```