CATS
(Cancer Genomic Test Standardized) Format

Synopsis

By Section of Genomic Data Management, C-CAT

v1.3.1

2023/10/17

I. Synopsis

Example1. Only shortVariants

```
{
    "metaData": {
        "schemaVersion": "1.3.1",
        Note: Describe the version of CATS Format used.
        "referenceGenome": {
            "grcRelease": "GRCh37.p13"
        }
    },
    "testInfo": {
        "testId": "123456789012311900001",
        "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"
          }
```

```json
      ],
      "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",
```

```json
          "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

```json
{
    "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"
                    }
                ],
```

```json
        "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"
```

```json
              }
            ]
          },
          {
            "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",
```

```json
        "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
      }
    ]
}
```