

Variant Data Model Example

Variant model example

```
{
  "id": "1:69511:A:G",
  "names": ["rs75062661"],
  "chromosome": "1",
  "start": 69511,
  "end": 69511,
  "strand": "+",
  "length": 1,
  "type": "SNV",
  "reference": "A",
  "alternate": "G",
  "studies": [
    {
      "studyId": "demo@family:corpasome",
      "secondaryAlternates": [],
      "files": [
        {
          "fileId": "quartet.variants.annotated.vcf.gz"
          "call" : {

          },
          "data": {
            "ABHom": "0.982",
            "AC": "8",
            "AF": "1.00",
            "AN": "8",
            "BaseQRankSum": "2.089",
            "DB": "true",
            "DP": "331",
            "Dels": "0.00",
            "EFF": "NON_SYNONYMOUS_CODING(MODERATE|MISSENSE|Aca
/Gca|T141A|305|OR4F5|CODING|NM_001005484.1|1|1)",
            "FILTER": "VQSRTrancheSNP99.90to100.00",
            "FS": "8.817",
            "HaplotypeScore": "2.4399",
            "MLEAC": "8",
            "MLEAF": "1.00",
            "MQ": "15.47",
            "MQ0": "145",
            "MQRankSum": "-0.047",
            "OND": "0.018",
            "QD": "12.97",
            "QUAL": "4293.01",
            "ReadPosRankSum": "1.662",
            "SB": "-1.450e+03",
            "VCF_ID": "rs75062661",
            "VQSLOD": "-14.4975",
            "culprit": "MQ",
            "set": "FilteredInAll"
          }
        }
      ],
      "sampleDataKeys": ["GT", "AD", "DP", "GQ", "PL"],
      "samples": [
        {
          "sampleId": "",
          "fileIndex": 0,
          "data": ["1/1", "2,171", "173", "99", "2218,228,0"]
        },
        {
          "sampleId": "",
          "fileIndex": 0,
          "data": ["1/1", "0,33", "34", "60", "508,60,0"]
        }
      ]
    }
  ]
}
```

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        "sampleId": "",
        "fileIndex": 0,
        "data": ["1/1", "0,61", "63", "93", "777,93,0"]
    },
    {
        "sampleId": "",
        "fileIndex": 0,
        "data": ["1/1", "0,61", "61", "96", "790,96,0"]
    }
],
"stats": [
    {
        "cohortId": "ALL",
        "alleleCount": 8,
        "altAlleleCount": 8,
        "altAlleleFreq": 1.0,
        "filterCount": {
            "PASS": 0,
            "VQSRTTrancheSNP99.90to100.00": 1
        },
        "filterFreq": {
            "PASS": 0.0,
            "VQSRTTrancheSNP99.90to100.00": 1.0
        },
        "genotypeCount": {
            "0/0": 0,
            "0/1": 0,
            "1/1": 4
        },
        "genotypeFreq": {
            "0/0": 0.0,
            "0/1": 0.0,
            "1/1": 1.0
        },
        "maf": 0.0,
        "mafAllele": "A",
        "mgf": 0.0,
        "mgfGenotype": "0/0",
        "missingAlleleCount": 0,
        "missingGenotypeCount": 0,
        "qualityAvg": 4293.01,
        "refAlleleCount": 0,
        "refAlleleFreq": 0.0
    }
],
"scores": [],
"issues": []
}
],
"annotation": {
    "id": "rs2691305",
    "chromosome": "1",
    "start": 69511,
    "reference": "A",
    "alternate": "G",
    "hgvs": ["ENST00000335137(ENSG00000186092):c.421A>G"],
    "displayConsequenceType": "missense_variant",
    "consequenceTypes": [
        {
            "geneName": "OR4F5",
            "ensemblGeneId": "ENSG00000186092",
            "ensemblTranscriptId": "ENST00000335137",
            "biotype": "protein_coding",
            "cdnaPosition": 421,
            "cdsPosition": 421,
            "codon": "Aca/Gca",
            "exonOverlap": [
                {
                    "number": "1/1",
                    "percentage": 0.108932465
                }
            ]
        }
    ]
}

```

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],
"proteinVariantAnnotation": {
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"rhodopsin-like, "

"7TM",

"disulfide "

"Helical; "

"Name=4",

"transmembrane "

"Olfactory "

"receptor "

"4F5",

"PRO_0000150547",

"end": 280,
"id": "IPR017452",
"start": 34},
{"end": 182,
"start": 90,
"type":

"bond"},
{"description":

"end": 151,
"start": 133,
"type":

"region"},
{"description":

"end": 305,
"id":

"start": 1,
"type": "chain"}],
"keywords": ["Cell "
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"Complete "
"proteome",
"Disulfide "
"bond",
"G-protein "
"coupled "
"receptor",
"Membrane",
"Olfaction",
"Receptor",
"Reference "
"proteome",
"Sensory "
"transduction",
"Transducer",
"Transmembrane",
"Transmembrane "
"helix"],
"position": 141,
"reference": "THR",
"substitutionScores":

{"description": "tolerated",

"score": 0.63,

"source": "sift"},

{"description": "benign",

"score": 0.003,

"source": "polyphen"}},

"uniprotAccession": "Q8NH21"},

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"sequenceOntologyTerms": [{"accession": "SO:0001583",
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"strand": "+",
"transcriptAnnotationFlags": [{"CCDS",
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{"sequenceOntologyTerms": [{"accession": "SO:0001566",
                             "name":
"regulatory_region_variant"}]}],
"conservation": [{"score": 1.149999976158142,
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{"score": 0.1289999932050705,
 "source": "phastCons"},
{"score": -0.527999997138977,
 "source": "phyloP"}],
"cytoBand": [{"chromosome": "1",
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              "name": "p36.33",
              "stain": "GNEG",
              "start": 1}],

"functionalScore": [{"score": -0.7899999618530273,
                    "source": "cadd_raw"},
{"score": 0.03999999910593033,
 "source": "cadd_scaled"}],
"geneDrugInteraction": [],
"geneTraitAssociation": [],

"populationFrequencies": [{"altAllele": "G",
                          "altAlleleFreq": 0.95061594,
                          "altHomGenotypeFreq": 0.93263996,
                          "hetGenotypeFreq": 0.03595196,
                          "population": "ALL",
                          "refAllele": "A",
                          "refAlleleFreq": 0.049384065,
                          "refHomGenotypeFreq": 0.031408086,
                          "study": "GNOMAD_EXOMES"},
{"altAllele": "G",
 "altAlleleFreq": 0.9499386,
 "altHomGenotypeFreq": 0.92997545,
 "hetGenotypeFreq": 0.03992629,
 "population": "OTH",
 "refAllele": "A",
 "refAlleleFreq": 0.050061423,
 "refHomGenotypeFreq": 0.03009828,
 "study": "GNOMAD_EXOMES"},
{"altAllele": "G",
 "altAlleleFreq": 0.999461,
 "altHomGenotypeFreq": 0.99892205,
 "hetGenotypeFreq": 0.0010779734,
 "population": "EAS",
 "refAllele": "A",
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 "refHomGenotypeFreq": 0.0,
 "study": "GNOMAD_EXOMES"},
{"altAllele": "G",
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 "hetGenotypeFreq": 0.040606,
 "population": "AMR",
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 "refAlleleFreq": 0.049160052,
 "refHomGenotypeFreq": 0.028857054,
 "study": "GNOMAD_EXOMES"},
{"altAllele": "G",
 "altAlleleFreq": 0.97795016,
 "altHomGenotypeFreq": 0.9710086,
 "hetGenotypeFreq": 0.013883217,
 "population": "ASJ",
 "refAllele": "A",
 "refAlleleFreq": 0.022049816,
 "refHomGenotypeFreq": 0.015108207,

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"study": "GNOMAD_EXOMES"},
{"altAllele": "G",
"altAlleleFreq": 0.99145377,
"altHomGenotypeFreq": 0.98848504,
"hetGenotypeFreq": 0.0059373877,
"population": "FIN",
"refAllele": "A",
"refAlleleFreq": 0.00854624,
"refHomGenotypeFreq": 0.005577546,
"study": "GNOMAD_EXOMES"},
{"altAllele": "G",
"altAlleleFreq": 0.9727796,
"altHomGenotypeFreq": 0.96255124,
"hetGenotypeFreq": 0.020456737,
"population": "NFE",
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"refAlleleFreq": 0.027220415,
"refHomGenotypeFreq": 0.016992046,
"study": "GNOMAD_EXOMES"},
{"altAllele": "G",
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"altHomGenotypeFreq": 0.47664425,
"hetGenotypeFreq": 0.26158446,
"population": "AFR",
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"refAlleleFreq": 0.39256352,
"refHomGenotypeFreq": 0.2617713,
"study": "GNOMAD_EXOMES"},
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"altHomGenotypeFreq": 0.77478045,
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"population": "ALL",
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"refAlleleFreq": 0.15777917,
"refHomGenotypeFreq": 0.090338774,
"study": "GNOMAD_GENOMES"},
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"altAlleleFreq": 0.9404255,
"altHomGenotypeFreq": 0.9191489,
"hetGenotypeFreq": 0.04255319,
"population": "OTH",
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"refAlleleFreq": 0.05957447,
"refHomGenotypeFreq": 0.038297873,
"study": "GNOMAD_GENOMES"},
{"altAllele": "G",
"altAlleleFreq": 1.0,
"altHomGenotypeFreq": 1.0,
"hetGenotypeFreq": 0.0,
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"refAlleleFreq": 0.0,
"refHomGenotypeFreq": 0.0,
"study": "GNOMAD_GENOMES"},
{"altAllele": "G",
"altAlleleFreq": 0.9410377,
"altHomGenotypeFreq": 0.9103774,
"hetGenotypeFreq": 0.061320756,
"population": "AMR",
"refAllele": "A",
"refAlleleFreq": 0.058962263,
"refHomGenotypeFreq": 0.028301887,
"study": "GNOMAD_GENOMES"},
{"altAllele": "G",
"altAlleleFreq": 0.9672131,
"altHomGenotypeFreq": 0.9508197,
"hetGenotypeFreq": 0.032786883,
"population": "ASJ",
"refAllele": "A",
"refAlleleFreq": 0.032786883,

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        "refHomGenotypeFreq": 0.016393442,
        "study": "GNOMAD_GENOMES"},
    {"altAllele": "G",
     "altAlleleFreq": 0.9918478,
     "altHomGenotypeFreq": 0.98913044,
     "hetGenotypeFreq": 0.0054347827,
     "population": "FIN",
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     "refAlleleFreq": 0.008152174,
     "refHomGenotypeFreq": 0.0054347827,
     "study": "GNOMAD_GENOMES"},
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     "altHomGenotypeFreq": 0.94847214,
     "hetGenotypeFreq": 0.03055722,
     "population": "NFE",
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     "refHomGenotypeFreq": 0.02097064,
     "study": "GNOMAD_GENOMES"},
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     "altHomGenotypeFreq": 0.41246733,
     "hetGenotypeFreq": 0.3523703,
     "population": "AFR",
     "refAllele": "A",
     "refAlleleFreq": 0.4113475,
     "refHomGenotypeFreq": 0.23516238,
     "study": "GNOMAD_GENOMES"}],

    "repeat": [{"chromosome": "1",
                  "copyNumber": 2.0,
                  "end": 87112,
                  "id": "9119",
                  "percentageMatch": 0.992904,
                  "source": "genomicSuperDup",
                  "start": 10001},
               {"chromosome": "1",
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                  "end": 87112,
                  "id": "14903",
                  "percentageMatch": 0.995437,
                  "source": "genomicSuperDup",
                  "start": 18393}],

    "traitAssociation": [{"additionalProperties": [{"name":
"mutationSomaticStatus_in_source_file",
                                                              "value": "Confirmed "
                                                              "somatic "
                                                              "variant"}],
                        "alleleOrigin": [],
                        "bibliography": [],
                        "ethnicity": "Z",
                        "genomicFeatures": [{"featureType": "gene",
                                              "xrefs": {"symbol": "OR4F5"}},
                                             {"featureType": "gene",
                                              "xrefs": {"symbol": "8301"}}],
                        "heritableTraits": [],
                        "id": "COSM4144171",
                        "somaticInformation": {"histologySubtype": "neoplasm",
                                                "primaryHistology": "other",
                                                "primarySite": "thyroid",
                                                "sampleSource": "",
                                                "tumourOrigin": ""},
                        "source": {"name": "cosmic"},
                        "submissions": []}]

    "additionalAttributes": {
"opencga": {
  "attribute": {
    "annotationId": "CURRENT",
    "release": "1"
  }
}

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}
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