VCF with Allele Frequencies

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| chr | pos | ref | alt | ancestral | AF | DAF | Scenario |
| chr22 | 1 | A | G | . | 0.07 | . | ancestral is missing |
| chr22 | 32972923 | C | T | C | 0.1 | 0.1 | ancestral == reference |
| chr22 | 33260795 | G | A | A | 0.05 | 0.95 | ancestral == alternate |
| chr22 | 35423388 | T | C | G | 0.12 | 1 | ancestral != reference & ancestral != alternate |
| chr22 | 36598290 | A | G, T | A | 0.01, 0.03 | 0.04 | N alternate alleles > 1 & ancestral == reference |
| chr22 | 38959105 | C | A, G | G | 0.08, 0.09 | 0.91 | N alternate alleles > 1 & ancestral == any(alternate) |
| chr22 | 40000000 | A | C, T | G | 0.01,0.01 | 1 | N alternate alleles > 1 & ancestral != any(alternate) |

VCF with Genotypes

|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| chr | pos | ref | alt | ancestral | Sample1 | Sample2 | Sample3 | Sample4 | AF | DAF | Scenario |
| chr22 | 1 | A | G | . | 0/0 | 0/0 | 0/0 | 0/1 | 0.125 | . | ancestral is missing |
| chr22 | 32972923 | C | T | C | 1/1 | 0/0 | 0/0 | 0/0 | 0.25 | 0.25 | ancestral == reference |
| chr22 | 33260795 | G | A | A | 0/1 | 0/1 | 0/0 | ./. | 0.33 | 0.66 | ancestral == alternate |
| chr22 | 35423388 | T | C | G | 0|0 | 0|1 | 1|1 | 0|1 | 0.5 | 1 | ancestral != reference & ancestral != alternate |
| chr22 | 36598290 | A | G, T | A | ./. | 0/0 | 0/0 | 1/2 | 0.167, 0.167 | 0.33 | N alternate alleles > 1 & ancestral == reference |
| chr22 | 38959105 | C | A, G | G | 0/2 | 2/0 | 1/2 | 0/0 | 0.125,0.375 | 0.625 | N alternate alleles > 1 & ancestral == any(alternate) |
| chr22 | 40000000 | A | C, T | G | 0/0 | 0/1 | 0/2 | 0/0 | 0.125,0.125 | 1 | N alternate alleles > 1 & ancestral != any(alternate) |