

Allele Frequency Data Specification

The below allele frequency fields are already included in the built_with_change_types.tsv file. None need to be removed.

- 24. Variant_frequency_LOVD
- 25. Minor_allele_frequency_ESP
- 26. EUR_Allele_frequency_1000_Genomes
- 27. AFR_Allele_frequency_1000_Genomes
- 28. AMR_Allele_frequency_1000_Genomes
- 29. EAS_Allele_frequency_1000_Genomes
- 30. Allele_frequency_1000_Genomes
- 31. SAS_Allele_Frequency_1000_Genomes
- 32. Allele_frequency_ExAC

For field number 25, the ESP data will be converted from percentage to decimal format for consistency with other frequency columns.

All fields from 1000 Genomes necessary for visualization are present and correctly formatted.

There are additional fields from ExAC that will be pulled through the pipeline:

AC_AFR	AN_AFR	Het_AFR	Hom_AFR
AC_AMR	AN_AMR	Het_AMR	Hom_AMR
AC_EAS	AN_EAS	Het_EAS	Hom_EAS
AC_FIN	AN_FIN	Het_FIN	Hom_FIN
AC_NFE	AN_NFE	Het_NFE	Hom_NFE
AC_OTH	AN_OTH	Het_OTH	Hom_OTH
AC_SAS	AN_SAS	Het_SAS	Hom_SAS

This data can be found in the "exec.brca12.sorted.hg38.vcf" file present in the output. AC is short for "Allele Count," which is the number of alleles found in a population. AN is short for "Allele Number," which is the total number of sample alleles. "Het" and "Hom" are short for homozygosity, which gives the user the ability to understand the allele counts in terms of individuals in the population.

Allele frequency is calculated by dividing AC/AN, which can be done on the fly for the Allele Frequency Tile. Accordingly, ACs and ANs are sufficient for the pipeline. Overall AC can also be calculated by summing together all ACs of the populations. Overall AN can also be calculated this way.

Regarding AF discrepancies, the overall Allele Frequency calculated by ExAC sometimes uses data that BRCA Exchange does not pull through its pipeline.

Further information about ExAC data can be found at:
<<http://exac.broadinstitute.org/faq>>