

| File extension                         | Description                                                                                                                                                                                                                                                                                                                            | Links                                                                                                                                                                                                                                                           | Programs commonly used in/produced by                    |
|----------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------|
| .fa                                    | Fasta files are files that contain a nucleotide or protein sequence, generally without quality scores. This will typically be a high confidence nucleotide sequence or reference assembly. The name of each sequence included is preceded by the > symbol, followed by the sequence.                                                   | <a href="https://zhanglab.ccmb.med.umich.edu/FAS TA/">https://zhanglab.ccmb.med.umich.edu/FAS TA/</a>                                                                                                                                                           | Many!                                                    |
| .fq/.fastq (also .fq.gz and .fastq.gz) | Read data from next-generation sequencing. There are four lines per read, including information such as the read name, the nucleotide sequences read and relevant quality scores                                                                                                                                                       | <a href="https://support.illumina.com/bulletins/2016/04/fastq-files-explained.html">https://support.illumina.com/bulletins/2016/04/fastq-files-explained.html</a>                                                                                               | Mapping programs, de novo assembly programs, among other |
| .sam/.bam                              | Format for reads mapped to a reference genome including important information such as the read, mapping location, and mapping quality. The bam file is binary while the sam file is uncompressed.                                                                                                                                      | <a href="https://genome.sph.umich.edu/wiki/SAM">https://genome.sph.umich.edu/wiki/SAM</a>                                                                                                                                                                       | bwa, STAR, stampy, and others                            |
| .vcf/.bcf                              | Variant file containing information on variant base pairs, mapping quality, depth, and a large number of base quality metrics relative to the reference genome used for mapping. The vcf file is uncompressed while the bcf file is the binary version. <b>Note: the specifics of the vcf format will vary from program to program</b> | <a href="https://www.ebi.ac.uk/training/online/course/human-genetic-variation-introduction/exercise-title/want-know-how-we-did-it">https://www.ebi.ac.uk/training/online/course/human-genetic-variation-introduction/exercise-title/want-know-how-we-did-it</a> | samtools/bcftools, GATK, and others                      |

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|---------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------|
| .g.vcf  | Similar to a vcf but also containing information for invariant sites. <b>Note: the specifics of the vcf format will vary from program to program</b>                                                                                                                                                                                                          |                                                                                                                                                                   | samtools/bcftools, GATK, and others                                                                           |
| ped map | Compact file format containing pedigree (optional), phenotype (optional), and genotype data that is used by the plink program and several other programs. The ped file must be paired with a map file which gives the locations of SNPs                                                                                                                       | <a href="http://www.gwaspi.org/?page_id=145">http://www.gwaspi.org/?page_id=145</a>                                                                               | plink                                                                                                         |
| bed     | bed files are useful file formats for storing interval information for a particular genome assembly. The first column contains the linkage group/scaffold/chromosome name, the second column contains the feature start position, the third column contains the feature end position. Additional columns after these first three are allowed by most programs | <a href="https://www.genomatix.de/online_help/help_regionminer/bedformat_help.html">https://www.genomatix.de/online_help/help_regionminer/bedformat_help.html</a> | Can be included to incorporate known features (e.g. repetitive regions, exons) in many programs               |
| gff/gtf | Annotation files that contain information on specific features in the genome. A gtf is a special case of a gff file that contains information on gene structure (i.e. exon locations).                                                                                                                                                                        | <a href="http://genomewiki.ucsc.edu/index.php/Genes_in_gtf_or_gff_format">http://genomewiki.ucsc.edu/index.php/Genes_in_gtf_or_gff_format</a>                     | Can be included as input with many programs, usually will be provided by a database (i.e. ensembl, ncbi, etc) |
| insnp   | A file in the format: chromosome, position, basepair current, basepair to change to for use with                                                                                                                                                                                                                                                              |                                                                                                                                                                   | seqtk                                                                                                         |

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|--|-----------------------------------------------|--|--|
|  | the seqtk program for<br>updating fasta files |  |  |
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### Lab-specific file formats:

Ancestry-tsv format: posterior probabilities of ancestry for each individual and ancestry informative site in the genome

genotype format: hard-calls for ancestry in the same format as Ancestry-tsv files (2: homozygous parent 2 – *malinche*, 1: heterozygous ancestry, 0: homozygous parent 1 – *birchmanni*). We typically use a posterior probability cutoff of 0.9.

hybrid index file format: file including the estimated proportion of the genome from *malinche* and estimated ancestry heterozygosity