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Converting from 23 and Me to VCF

The raw 23 and Me results can be downloaded as a tab-delimited file with four columns, the marker ID, chromosome name, position and the genotype:

rs6139074	20	63244	AA
rs1418258	20	63799	CC
rs6086616	20	68749	TT
rs6039403	20	69094	AG

The dot "." can be used for a marker ID in case it is not known. The conversion command is then:

bcftools convert --tsv2vcf input.tab.gz -f ref.fa -s Sampl



It is important to check the output printed on the screen, which may look for example like this:

Rows total: 612647
Rows skipped: 4751
Missing GTs: 20525
Hom RR: 318339
Het RA: 165598
Hom AA: 103420
Het AA: 14

Here the program converted more than 95% of the rows, in the example 20525 genotypes are missing ("--") and 4751 sites were skipped because the tool only considers SNPs and ignores deletions and insertions.

One should also check the number of non-reference heterozygous genotypes, which was 14 in this example. It should be small like this, because large number of heterozygous alts (Het AA) indicates

that the input alleles are not on the forward strand. We can check this explicitly using the <u>fixref plugin</u>.

Merging into multi-sample VCFs

Single-sample VCFs created in the previous step can be merged into one multi-sample VCF using the following commands. The input files must be indexed, then can be merged:

```
bcftools index sampleA.bcf
bcftools index sampleB.bcf
bcftools merge -Ob -o output.bcf sampleA.bcf sampleB.bcf
```

Feedback

We welcome your feedback, please help us improve this page by either opening an <u>issue on github</u>

(https://github.com/samtools/bcftools/issues) or <u>editing it directly</u> (https://github.com/samtools/bcftools/tree/gh-pages/howtos) and sending a pull request.