

# SNP calling and visualization of vcf files

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# SNP calling

There are plenty of different ways to call genotypes

- Samtools
- SOAPsnp
- GATK
- GotCloud
- Stacks
- UNEAK
- CRoPS
- ...

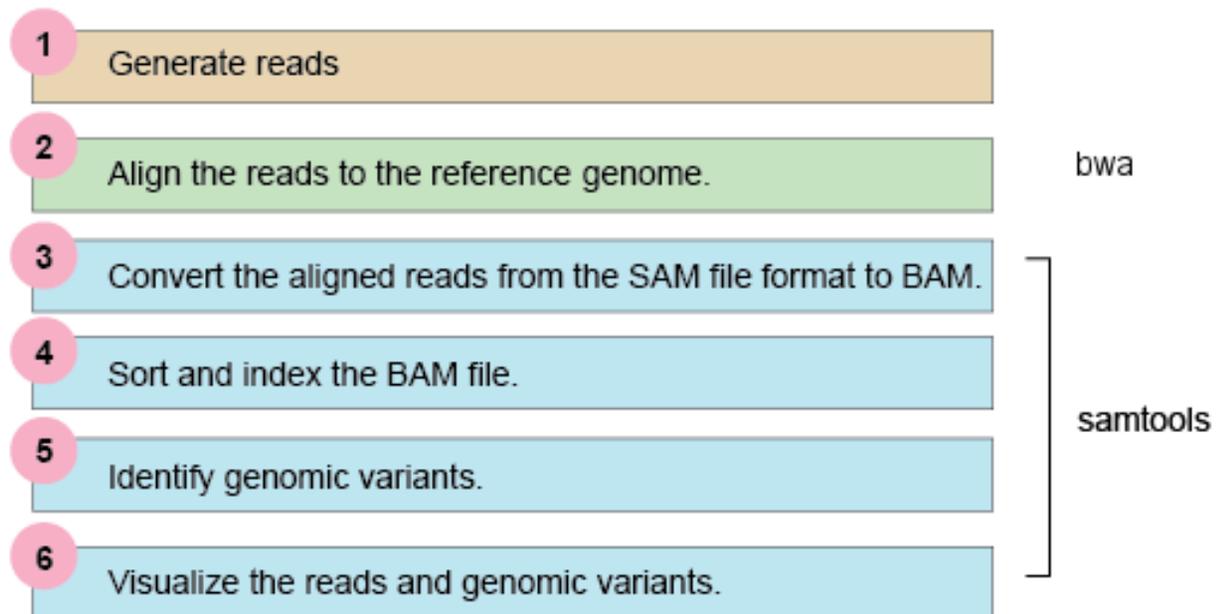
# SNP calling

There are plenty of different ways to call genotypes  
When deciding which one to use, consider

- Which organism am I working on?
- How big is my data set?
- What am I going to do with these SNPs?
- How geeky/nerdy do I want to be?

# Samtools - recap

Why so many steps?



mod. from [biobits.org/samtools\\_primer.html](http://biobits.org/samtools_primer.html)

# Visualization of Polymorphisms

- tview

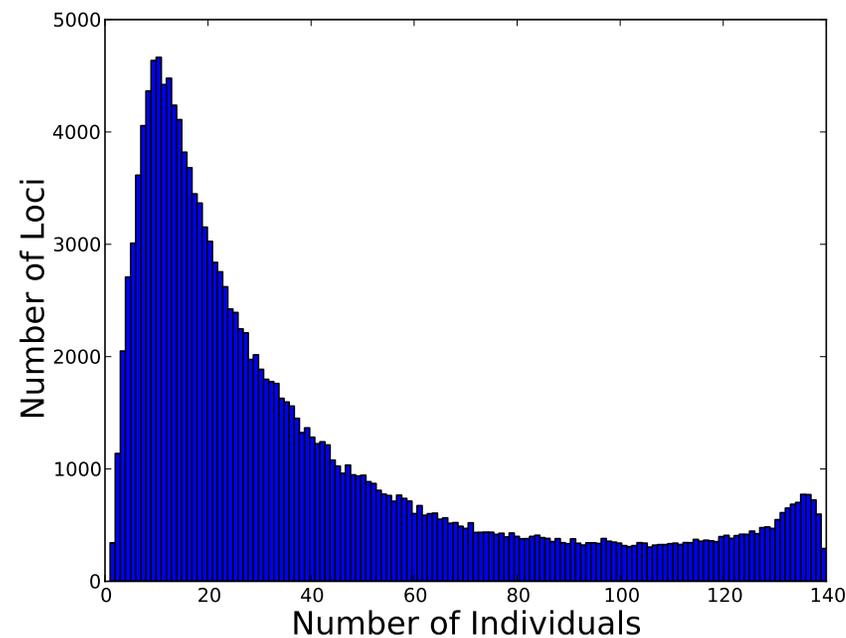
```
711 721 731 741 751 761 771 781 791
GTGGCGAGAAAATGTCGATCGCCATTATGGCCGGCGTGTAGAAAGCCGCTGGTCACAACGTTACCGTTATCGATCCGGTCGAAAAACTGCTGGCAGT
|.....G.....
|.....g.....c.....
|.....c,t.....g.....
|.....t.....g.....
|.....g.....
|.....tg.....t.....
|.....c.....g.....g.....
|.....c.....g,a.....t.....
|.....gC.....
|.....g,g.....
|.....g,g.....g.....
|.....g.....g.....a,c.....
|.....g.....g.....
|.....g,g.....c.....
|.....g.....
|.....g.....g.....t.....
|.....g.....t.....t.....
|.....g.....c.....a,g.....
|.....g.....c.....a.....t.....
|.....c.....tg.....g.....g.....
|.....t.....g.....t.....a.....
```

## Visualization of Polymorphisms

- tview
- Tablet
- IGV (Integrative Genomics Viewer) - kitchen sink of formats
- inPHAP (Haplotypes and SNPs)
- VARB (VARiation Browser)
- Savant Genome Browser (still maintained?)

## A note about Minor Allele Frequencies

- Most nucleotides are invariant or have a rare (MAF  $< 0.05$ ) segregating minor allele. (66% rare with 530k total variants for *Lycaeides* (Gompert et al., 2013))



## A note about Minor Allele Frequencies

- Most nucleotides are invariant or have a rare ( $\text{MAF} < 0.05$ ) segregating minor allele. (66% rare with 530k total variants for *Lycaeides* (Gompert et al., 2013))
- These rare variants could be associated with local adaptation/differentiation
- Given this complexity, are we discarding most of the variation?

## \*References

Gompert, Z., Lucas, L. K., Buerkle, C. A., Forister, M. L., Fordyce, J. A., and Nice, C. C. (2013). Admixture and the organization of genetic diversity in a butterfly species complex revealed through common and rare genetic variants. *Molecular Ecology*.