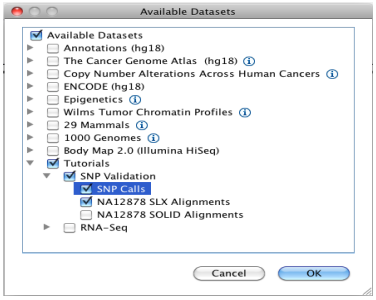
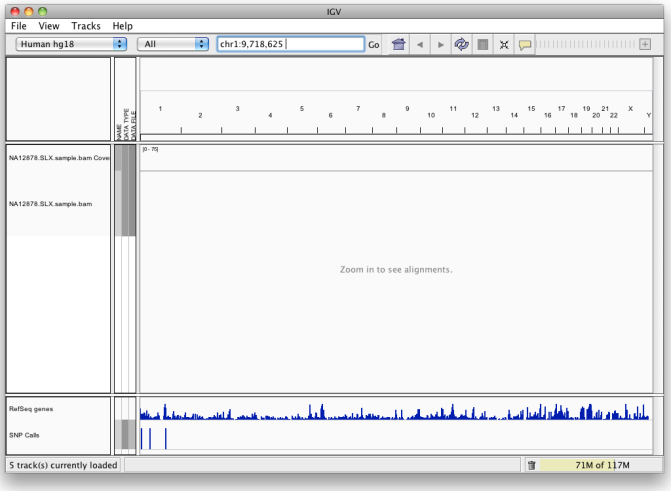
Viewing SNPs

1. Select genome "Human hg18"
2. Load the following files from the server (File > Load from Server..
   - NA12878 SLX Alignments
   - SNP Calls



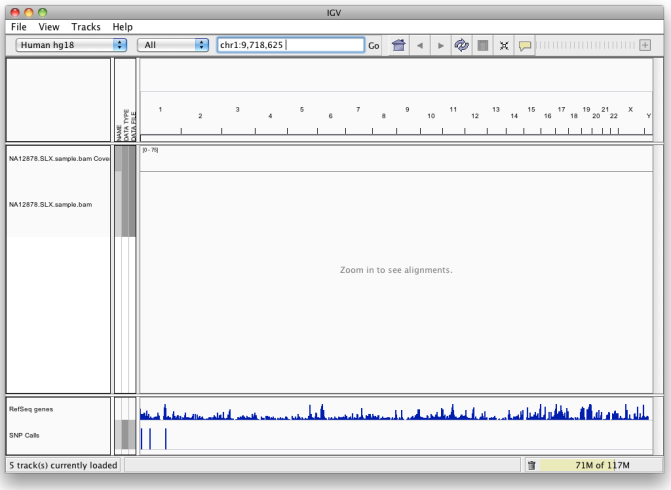
Viewing SNPs

3. Enter the following into the search box and hit "Go": snp1



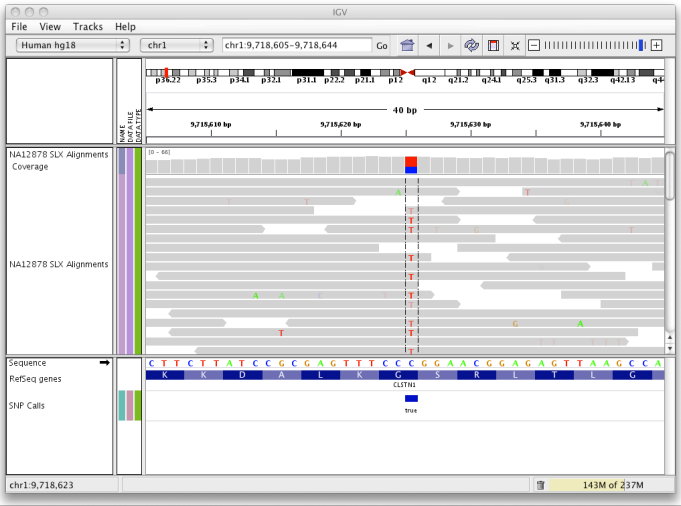
Viewing SNPs

4. Enter the following into the search box and hit "Go": snp1



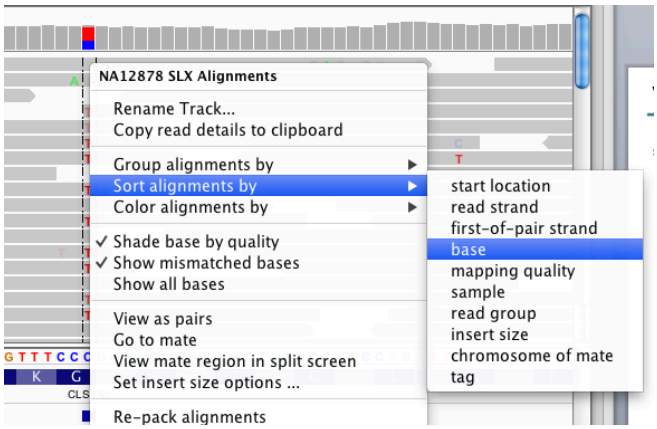
Viewing SNPs

5. Zoom in on the SNP and center it between the vertical dashed lines.



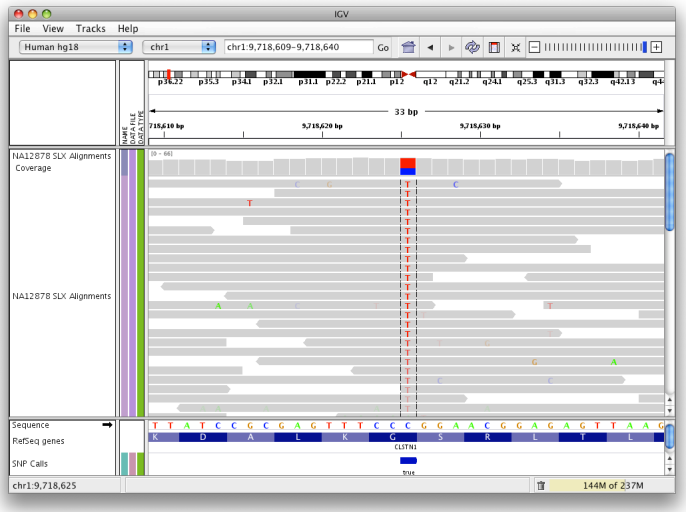
Viewing SNPs

6. Right-click over alignment panel (NOTE: not Coverage track) and select "Sort alignments > by base"



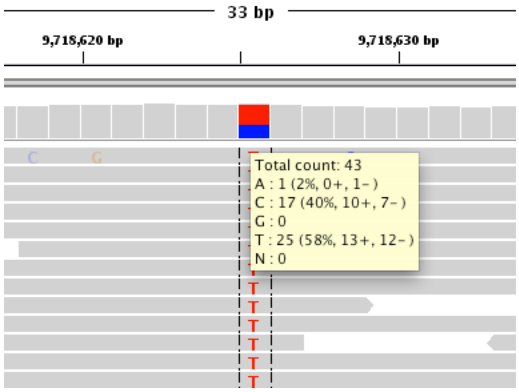
Viewing SNPs

Alignments sorted by base



Viewing SNPs

7. Mouse over red & blue colored bar in coverage track. Note allele counts and frequencies.

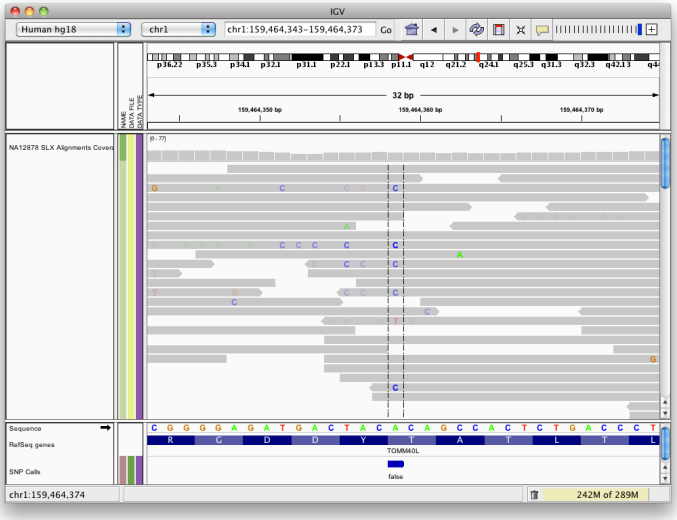


Viewing SNPs

8. Jump to next SNP call: Select "SNP Calls" track by clicking on the name. Hold "ctrl" key down and press "f".

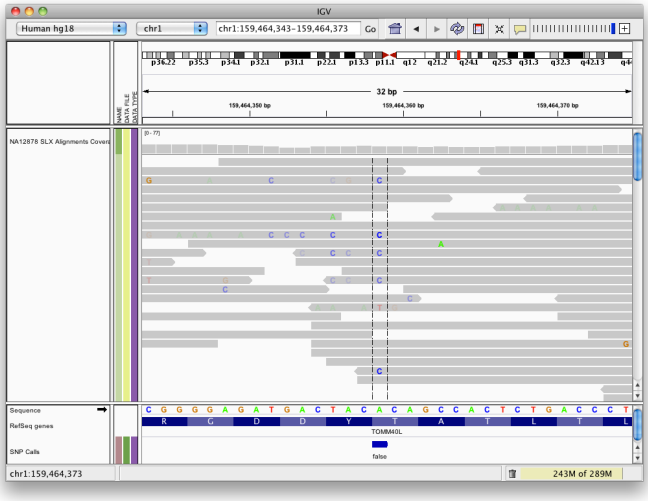
Viewing SNPs

9. Reposition if necessary to center SNP



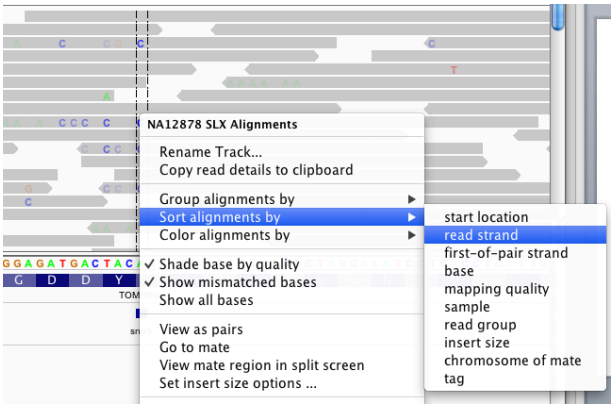
Viewing SNPs

10. Note the large % of low quality base-calls (faint, semi-transparent), and also the scatter of “C” mismatches.



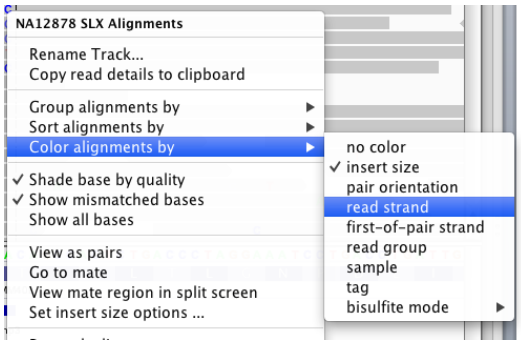
Viewing SNPs

11. Right click on alignments and sort by read strand.



Viewing SNPs

12. Right click on alignments and color by read strand



SNPs

Note that the mismatches are all on one strand (the negative strand).