Track Data and Bedtools Adam m France afrance3@uncc.edu

### Part 3:

## What format is the output that you get?

```
$ bedtools intersect -a genome_Gibas.vcf -b common snp.BED | head -5
      1025301 rs9442400
                           Т
                                C
                                                   GT
                                                         1/1
chr1
      1113121 rs12092254
                            G
chr1
                                                   GT
                                                         0/0
chr1
      1366830 rs873927
                                                  GT
                                                        0/0
                           Α
chr1
      1715011 rs742359
                           C
                                                  GT
                                                         0/1
                                Α
chr1
      1875267 rs2262190
                                G
                                                   GT
                                                         0/1
                           а
```

This looks like VCF format

What happens if you switch things up so that the -a file is the BED file and the -b file is the VCF?

```
$ bedtools intersect -a common snp.BED -b genome Gibas.vcf | head -5
chr1
      1025300 1025301 rs144619388
                                    0
                                         +
     1113120 1113121 rs7518814
chr1
                                   0
                                        +
chr1
     1366829 1366830 rs75051470
                                    0
                                        +
     1715010 1715011 rs3820004
                                   0
chr1
                                        +
chr1
      1875266 1875267 rs141122345
                                    0
```

This looks like BED format

Which human autosome has the highest per-base-pair representation of SNPs in the 23andMe assay?

I used the command below, but repeated for chr 1 - 23, with the highest WC match being for chromosome 9 with 7,053.

\$ bedtools intersect -b genome\_Gibas.vcf -a common\_snp\_wg.BED | grep chr9 | wc 7053

Which SNPs in the 23andMe assay intersect with SNPs in the GWAS (Genome Wide Association Study) Catalog (in the Phenotype and Literature track category)? Do you think the number that you get from a simple intersection is correct? What happens when you include flanking sequence, either via bedtools window or by selecting different options at track download time?

\$ bedtools intersect -a genome\_Gibas.vcf -b common\_snp\_wg.BED >
overlaps.output

Using wc -I on the output file, we see how many snps overlap: \$ wc -I overlaps.output 14915 overlaps.output

## Problem 1: get the coordinate intervals for 100 bases flanking each feature in the GFF.

\$ bedtools flank -i NC\_007898.gff -g NC\_007898.genome -b 100 | head -5

```
RefSeg gene 71536 71635 .
NC 007898.3
ID=gene0;Dbxref=GeneID:3950426;Name=rps12;exception=trans-splicing;gbkey=Gene;gene=r
ps12;locus tag=LyesC2p022;part=1/2
NC 007898.3
             RefSeq gene 71750 71849 .
ID=gene0;Dbxref=GeneID:3950426;Name=rps12;exception=trans-splicing;gbkey=Gene;gene=r
ps12;locus tag=LyesC2p022;part=1/2
NC 007898.3
              RefSeg gene 99139 99238 .
ID=gene0;Dbxref=GeneID:3950426;Name=rps12;exception=trans-splicing;gbkey=Gene;gene=r
ps12;locus tag=LyesC2p022;part=2/2
             RefSeg gene 100033 100132.
NC 007898.3
ID=gene0;Dbxref=GeneID:3950426;Name=rps12;exception=trans-splicing;gbkey=Gene;gene=r
ps12;locus_tag=LyesC2p022;part=2/2
NC 007898.3
              RefSeq CDS
                            71536 71635 .
ID=cds0;Parent=gene0;Dbxref=Genbank:YP_008563067.1;Name=YP_008563067.1;exception=
```

# Problem 2: for each feature in a file, find its nearest physical neighbors on the same strand (say, within 200 bp) in the chloroplast

\$ cat \*.gff | grep CDS > output.cds.gff

```
$ bedtools window -abam BC30.bam -b output.cds.gff -w 200 > prob22.BED -bed
$ head -5 prob22.BED
                          SC998:02260:01254
NC 007898.3 67
                    342
                                                         NC 007898.3
                                                                        RefSea
                                               24
      537
CDS
            1598 .
                                   -0
ID=cds1;Parent=gene2;Dbxref=Genbank:YP_008563068.1;Name=YP_008563068.1;gbkey=CD
S;gene=psbA;product=photosystem II protein D1;protein_id=YP_008563068.1;transl_table=11
              103 385 SC998:01683:02712
NC 007898.3
                                               23
                                                         NC 007898.3
                                                                        RefSea
CDS
      537
            1598 .
ID=cds1;Parent=gene2;Dbxref=Genbank:YP_008563068.1;Name=YP_008563068.1;gbkey=CD
S;gene=psbA;product=photosystem II protein D1;protein id=YP 008563068.1;transl table=11
                          SC998:01587:02448
NC 007898.3
              118
                    343
                                               40
                                                         NC 007898.3
                                                                        RefSeq
CDS
     537
            1598 .
ID=cds1;Parent=gene2;Dbxref=Genbank:YP_008563068.1;Name=YP_008563068.1;gbkey=CD
S;gene=psbA;product=photosystem II protein D1;protein_id=YP_008563068.1;transl_table=11
                    355
                          SC998:02278:01285
NC 007898.3
              118
                                               24
                                                         NC 007898.3
                                                                       RefSeq
CDS
      537
            1598 .
ID=cds1;Parent=gene2;Dbxref=Genbank:YP 008563068.1;Name=YP 008563068.1;gbkey=CD
S;gene=psbA;product=photosystem II protein D1;protein id=YP 008563068.1;transl table=11
              123
                    363 SC998:01312:00848
NC_007898.3
                                               24
                                                         NC_007898.3
CDS
      537
            1598 .
ID=cds1;Parent=gene2;Dbxref=Genbank:YP 008563068.1;Name=YP 008563068.1;gbkey=CD
S;gene=psbA;product=photosystem II protein D1;protein id=YP 008563068.1;transl table=11
```

## Problem 3: for each feature in the GFF, report only the features that have NO reads overlapping them in the BAM file

\$ bedtools window -abam BC30.bam -b output.cds.gff -v > prob33.BED -bed

\$ head prob33.BED					
NC_007898.3	0	102	SC998:00281:01611	3	+
NC_007898.3	0	149	SC998:00509:00586	0	+
NC_007898.3	0	112	SC998:00707:01173	0	+
NC_007898.3	0	82	SC998:00815:01895	0	+
NC_007898.3	0	131	SC998:01085:02291	0	+
NC_007898.3	0	147	SC998:01175:00421	0	+
NC_007898.3	0	206	SC998:01279:02241	0	+
NC_007898.3	0	192	SC998:01286:02666	8	+
NC_007898.3	0	155	SC998:01311:01763	3	+
NC_007898.3	0	148	SC998:01408:01722	0	+
\$ wc -l prob33.BED					
39022 prob33.BED					