Programming for Bioinformatics | BIOL7200

Week 12 Exercise

November 15, 2022

Assume that the user gives you correct inputs all the time. Your script will be graded on the output produced and not on how the errors are handled.

For this assignment, the module list is "sys", "re", "os", and "argparse". Do not use input() for any input.

Instructions for submission

- This assignment is due Monday, November 21, 2022 at 11:59pm. Late submissions will not be graded
- Name your script as overlapBed.py
- Your code should run as ./overlapBed.py -i1 <Input file 1> -i2 <Input file 2> -m <INT: minimal overlap> [-j Optional: join the two entries] -o <Output file>
- **DO NOT HARDCODE** any file name!
- Use #!/usr/bin/env python3 as your shebang
- Your script should finish within 30min, partial credit will be awarded up to 3 hours of run time. Bonus: If your code finishes the overlap on the provided files and produces the correct output within 3 min, we will award you an additional 20% bonus points.

This is a very common task in genome analysis. Often, we want to find functional elements that overlap with other elements, *e.g.* transcription start sites contained in transposable elements, predicted transcription factor binding sites within DNase hypersensitivity sites, or even some set of genes or horizontally transferred regions. Maybe you found some peaks in your ChIP-seq data and you want to see if those overlap with known enhancers, or perhaps you have a set of SNPs and you would like to know what genes they fall in. If you do not know what those words mean, go look them up!

Comparing ALL coordinates of the first set versus ALL coordinates of the second set will work for small sets only. That method scales with the multiplication of the sizes of the two sets. If you have 10^6 members in each set (not at all unrealistic) do you want to make 10^{12} comparisons? You need to find a better way.

What your script should do:

1. Take in two files that contain sets of coordinates in BED format. You can assume that the contents of both files are sorted by chromosome, start and stop. Having the data sorted allows you to greatly speed up the process of the overlap.

Definitions for BED file format can be found: https://genome.ucsc.edu/FAQ/FAQformat#format1

- 2. Take in an option for the minimum percent overlap, *i.e.* percent of bases of any given member of the first set that must be in a member of the second set to be counted as overlapping.
- 3. Print to a file the members of the first set which overlap with the second set and meet the minimum overlap and other conditions specified. Each overlap should only be printed once in this manner.
- 4. Allow an option to print both the member of the first set and the member of the second set that it overlaps with on the same line, in effect 'joining' the rows.

Input files: Two BED files (TE.bed, Intron.bed) can be found at http://jordan.biology.gatech.edu/biol7200/

Examples:

Overlap of the following two sets with a minimum of 100% overlap would yield one row.

Set 1:

chr1	1500 4500	5000	2	0	++
CNTI	8500	9500	3	0	+

Set 2:

```
chr1
1000
2000
a
0
-

chr1
3000
4000
b
0
-

chr1
9000
11000
c
0
-
```

Output:

```
chr1 1500 1750 1 0 +
```

If the joining option (from #4) was given:

```
chr1 1500 1750 1 0 + chr1 1000 2000 a 0 -
```

If the minimum overlap was 50% instead of 100% you would get two rows:

```
chr1 1500 1750 1 0 + chr1 8500 9500 3 0 +
```

Notes:

The overlap of two coordinates can be calculated as

```
MIN(stop one, stop two) - MAX(start one, start two)
```

Anything greater than 0 is the number of overlapping bases, otherwise it indicates no overlap.

Deliverables:

- Code: overlapBed.py
- A README file: this file should contain the <u>number of lines</u> in your output when you run your script with: ./overlapBed.py -i1 TE.bed -i2 Intron.bed -m 80 -o output

<u>Syntax</u>: ./overlapBed.py -i1 <Input file 1> -i2 <Input file 2> -m <INT: minimal overlap> [-j Optional: join the two entries] -o <Output file>

Example command: ./overlapBed.py -i1 TE.bed -i2 Intron.bed -m 50 -j -o testOutput

This command finds overlaps between TE.bed and Intron.bed, prints out the pairs of overlapping entries from both bed files that have minimum percent overlap of 50%.

Additional note:

• This is a great problem that demonstrates how a simple solution can often get you a pretty fast code. We advise you to sketch out a solution (*i.e.*, pseudocode) before you start implementing the actual code. Some of the best solutions we have seen in the past used a nested loop and a handful of if statements.