

# Genome 540 Discussion

February 15th, 2024

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# Assignment 6

# Assignment 6 Questions?

$O(N)$  algorithm to find all maximal D-segs:

```
cumul = max = 0; start = 1;
for (i = 1; i ≤ N; i++) {
    cumul += s[i];
    if (cumul ≥ max)
        {max = cumul; end = i;}
    if (cumul ≤ 0 or cumul ≤ max + D or i == N) {
        if (max ≥ S)
            {print start, end, max; }
        max = cumul = 0; start = end = i + 1; /* NO BACKTRACKING
        NEEDED! */
    }
}
```



# Assignment 7

# Overview

- Part 1: Use your predicted D-segments from hw6 to
  - Generate a new scoring scheme
  - Simulate background sequence
- Part 2: Run your D-segment program on the background and compare to the real data
- Part 3: Answer some questions

# Part 1: New scoring scheme

Read start histogram for non-elevated copy-number segments:

0=331908 - **8422401 (# Ns, don't forget this)**

1=19439

2=4272

>=3=1332

Read start histogram for elevated copy-number segments:

0=1656

1=542

2=352

>=3=499

$\log_2(\text{target freq.}/\text{background freq.})$

Background frequencies:

0={#.#####}

1={#.#####}

2={#.#####}

>=3={#.#####}

Target frequencies:

0={#.#####}

1={#.#####}

2={#.#####}

>=3={#.#####}

Scoring scheme:

0={#.#####}

1={#.#####}

2={#.#####}

>=3={#.#####}

# Part 1: Simulate new background sequence

$N$  = length of sequence to be simulated (length of seq. In HW6 - 8,422,401)

$\text{bkgd}[r]$  = frequency of background sites with  $r$  read starts ( $r = 0, 1, 2, 3$ )

for each  $i = 1 \dots N$

$x$  = random number between 0 and 1 (uniform distribution)

    if  $x < \text{bkgd}[0]$

$\text{sim\_seq}[i] = 0$

    else if  $x < \text{bkgd}[0] + \text{bkgd}[1]$

$\text{sim\_seq}[i] = 1$

    else if  $x < \text{bkgd}[0] + \text{bkgd}[1] + \text{bkgd}[2]$

$\text{sim\_seq}[i] = 2$

    else

$\text{sim\_seq}[i] = 3$

# Part 2: Run D-seg and compare

Real data:

```
5 {# of segments with score >= 5}
6 {# of segments with score >= 6}
7 {# of segments with score >= 7}
```

.  
.  
.

list all the segment score counts for scores  
between 5 and 30  
(only first/last 3 shown here)

.  
.  
.

```
28 {# of segments with score >= 28}
29 {# of segments with score >= 29}
30 {# of segments with score >= 30}
```

Simulated data:

```
5 {# of segments with score >= 5}
6 {# of segments with score >= 6}
7 {# of segments with score >= 7}
```

.  
.  
.

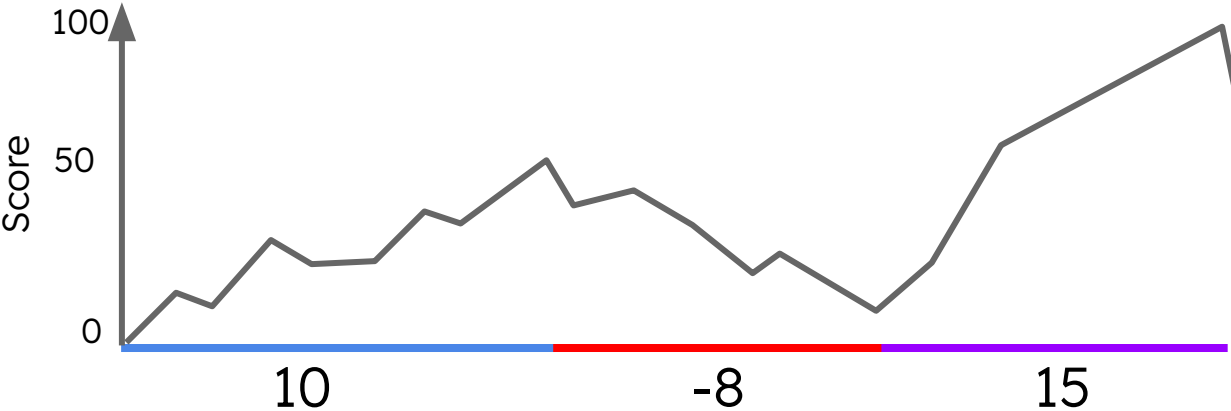
list all the segment score counts for scores  
between 5 and 30  
(only first/last 3 shown here)

.  
.  
.

```
28 {# of segments with score >= 28}
29 {# of segments with score >= 29}
30 {# of segments with score >= 30}
```



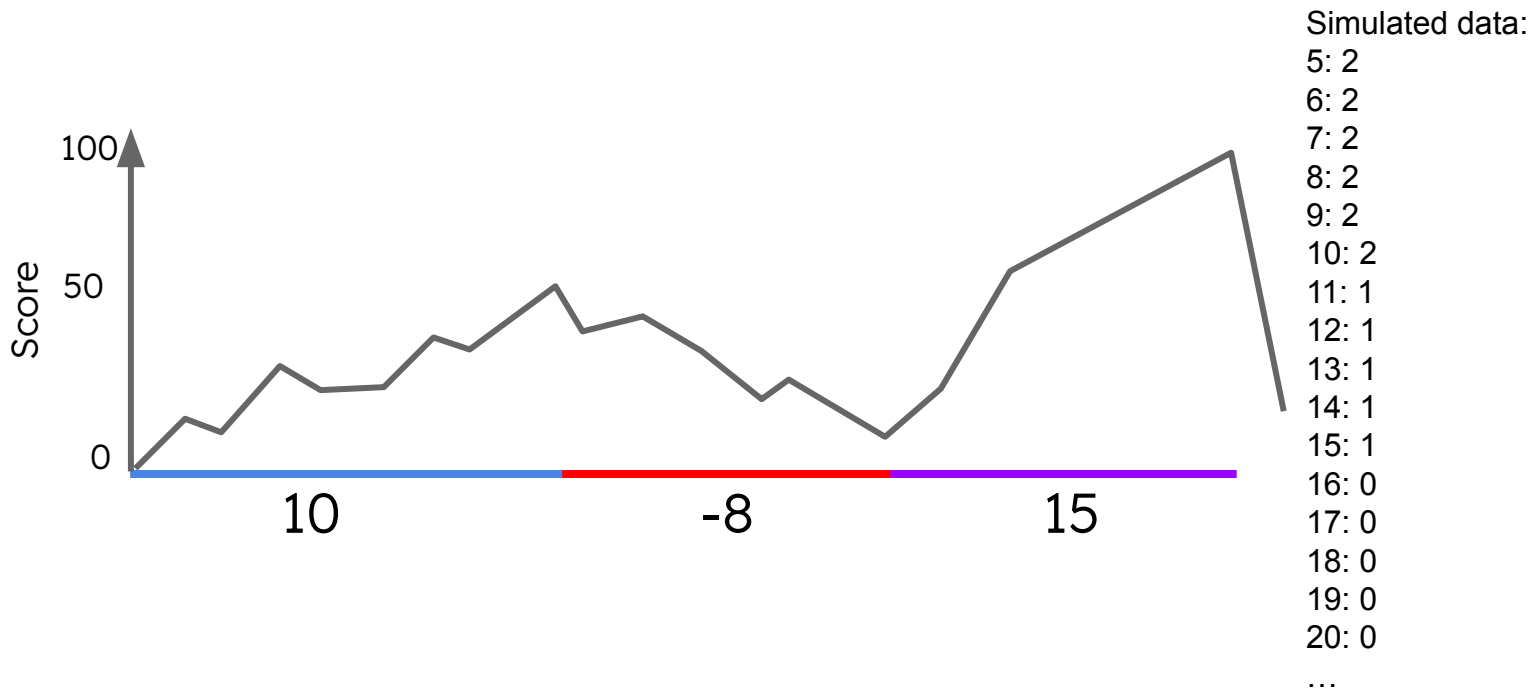
# Example



Simulated data:

- 5: ?
- 6: ?
- 7: ?
- 8: ?
- 9: ?
- 10: ?
- 11: ?
- 12: ?
- 13: ?
- 14: ?
- 15: ?
- 16: ?
- 17: ?
- 18: ?
- 19: ?
- 20: ?
- ...

# Example



# Part 2: Run D-seg and compare

Ratios of simulated data:

$N\_seg(5)/N\_seg(6)$  {# of segments with score  $\geq 5$  / # of segments with score  $\geq 6$ }

$N\_seg(6)/N\_seg(7)$  {# of segments with score  $\geq 6$  / # of segments with score  $\geq 7$ }

$N\_seg(7)/N\_seg(8)$  {# of segments with score  $\geq 7$  / # of segments with score  $\geq 8$ }

.

.

.

list all ratios

(only first/last 3 shown here)

.

.

.

$N\_seg(27)/N\_seg(28)$  {# of segments with score  $\geq 27$  / # of segments with score  $\geq 28$ }

$N\_seg(28)/N\_seg(29)$  {# of segments with score  $\geq 28$  / # of segments with score  $\geq 29$ }

$N\_seg(29)/N\_seg(30)$  {# of segments with score  $\geq 29$  / # of segments with score  $\geq 30$ }

# Reminders

- HW6 due this Sunday, 11:59pm
- Please have your name in the filename of your homework assignment and match the template