

# RLPBWT

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# Outline

- 1 RLPBWT
- 2 Example
- 3 A new Idea

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# Some definitions

The permutation, panel  $M$ ,  $n \times m$

In *RLPBWT* we have a permutation  $\pi_j$ ,  $\forall 1 \leq j \leq m$  that stably sorts the bits of the  $j$ -th column of the PBWT.

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The positions in the columns of the PBWT of the bits in the  $i$ -th row of  $M$  are:

$$i, \pi_1(i), \pi_2(\pi_1(i)), \dots, \pi_{m-1}(\dots(\pi_2(\pi_1(i)))\dots)$$

Extracting the bits of the  $i$ -th row of  $M$  reduces to iteratively applying the  $\pi_{m-1}$  permutations, corresponding to iteratively apply LF in a standard BWT

# The permutation

## Computing the permutation

$$\pi_j(p) = \begin{cases} p - \text{count}_1 & \text{if } \text{column}[\text{pref}[p]] = 0 \\ \text{count}_0 + \text{count}_1 - 1 & \text{if } \text{column}[\text{pref}[p]] = 1 \end{cases}$$

- $\text{count}_0$ : total number of zeros in the PBWT column
- $\text{count}_1$ : number of ones in the PBWT column as far as index  $p$

## “LF-mapping” in Durbin’s algorithm

$$w(i, \sigma) = \begin{cases} u[i] & \text{if } \sigma = 0 \\ c + v[i] & \text{if } \sigma = 1 \end{cases}$$

- $c$ : total number of zeros in the column
- $u[i]$ : number of zeros in the column as far as index  $i$
- $v[i]$ : number of ones in the column as far as index  $i$

# Travis's example

	1	2	3	4	5	6	7	8	9	10	11	12
0	0 1	0 1	0 0	0 0	0 0	0 1	0 0	0 0	0 1	0 1	0 1	0 1
1	0 1	0 1	0 0	0 0	0 0	0 1	0 0	0 0	0 1	0 1	0 1	0 1
2	0 1	0 1	0 1	0 0	0 0	0 0	0 1	0 1	0 1	0 0	0 1	0 1
3	0 1	0 1	0 0	0 0	0 0	0 1	0 0	0 0	0 1	0 1	0 0	0 1
4	0 1	0 0	0 1	0 0	0 0	0 1	0 0	0 0	0 1	0 1	0 0	0 1
5	0 1	0 0	0 1	0 0	0 0	0 0	0 0	0 0	0 1	0 0	0 0	0 1
6	0 1	0 0	0 1	0 0	0 0	0 0	0 0	0 0	0 1	0 0	0 0	0 0
7	0 1	0 1	0 1	0 0	0 0	0 0	0 0	0 0	0 0	1 1	0 0	0 1
8	0 0	0 1	0 0	0 0	0 0	0 1	0 0	0 0	0 0	1 1	0 0	0 1
9	0 1	1 0	0 0	0 0	0 0	0 1	0 0	0 0	0 0	1 0	0 0	0 1
10	0 1	1 1	0 0	0 0	0 0	0 0	0 0	0 0	0 1	1 1	0 0	0 1
11	0 0	1 1	1 0	0 1	0 1	0 0	0 0	0 0	0 1	1 0	0 0	0 1
12	0 0	1 1	1 0	0 0	0 1	0 0	0 0	0 0	0 0	1 0	0 0	0 1
13	0 0	1 0	1 0	0 0	0 1	0 0	0 0	0 0	0 0	1 0	1 0	0 1
14	0 0	1 0	1 0	0 0	0 0	0 0	1 0	0 0	0 0	1 0	1 0	0 1
15	0 0	1 0	1 0	1 0	0 0	0 0	1 0	0 0	0 0	1 0	1 0	0 1
16	0 1	1 0	1 0	1 0	0 0	0 0	1 0	0 0	1 1	1 0	1 0	0 1
17	0 0	1 0	1 0	1 0	0 0	1 0	1 0	0 1	1 1	1 0	1 0	1 1
18	1 0	1 0	1 0	1 0	0 0	1 0	1 0	0 1	1 1	1 0	1 0	1 1
19	1 0	1 0	1 0	1 0	1 0	1 0	1 0	1 1	1 1	1 0	1 0	1 1



# The compressed data structure

## The tables

- a set of  $m$  tables in which the  $m$ -th table stores only the positions of the run-heads in the  $m$ -th column and a bool to check the first symbol: 0 or 1
- the  $i$ -th row of the  $j$ -th table stores a quadruple

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## The quadruple

- 1 the position  $p$  of the  $i$ -th run-head in the  $j$ -th column of the PBWT
- 2 the permutation  $\pi_j(p)$
- 3 the index of the run containing bit  $\pi_j(p)$  in the  $(j + 1)$ -st column of the PBWT
- 4 the threshold, that's the index of the minimum *LCP value* (current column minus divergence array value) in the run

# Row extraction

## First step

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## First step

We start by finding the row of the first table that starts with the position  $p$  of the head of the run containing bit  $i$  in first column of the PBWT, computing:

$$\pi_1(i) = \pi_1(p) + i - p$$

looking up the row for the run containing bit  $\pi_1(p)$  in the the second table and scanning down the table until we find the row for the run containing bit  $\pi_1(i)$

## Next step

We continue repeating this procedure for each column

# Travis's example I

	table 1	table 2	table 3	table 4	table 5	table 6
0	0 9 3	0 11 4	0 0 0	0 0 0	0 0 0	0 14 2
1	8 0 0	4 0 0	2 15 2	11 19 2	11 17 5	2 0 0
2	9 17 5	7 15 4	3 2 0	12 11 1	14 11 5	3 16 2
3	11 1 0	9 3 2	4 16 2			5 1 0
4	16 19 5	10 17 4	8 3 0			8 18 2
5	17 6 1	13 4 3				10 4 2

	table 7	table 8	table 9	table 10	table 11	table 12
0	0 0 0	0 0 0	0 7 4	0 13 1	0 17 2	0
1	2 19 3	2 16 4	7 0 0	2 0 0	3 0 0	6
2	3 2 1	3 2 0	10 14 7	3 15 1		7
3		17 17 4	12 3 2	5 1 0		
4			16 16 7	7 17 1		
5				9 3 1		
6				10 19 1		
7				11 4 1		

# Travis's example II

Extraction of row 9,  $\pi_j(i) = \pi_j(p) + i - p$

$$\blacksquare \pi_1(9) = 17 + 9 - 9 = 17$$

$$\blacksquare \pi_2(17) = 4 + 17 - 13 = 8$$

$$\blacksquare \pi_3(8) = 4 + 8 - 8 = 3$$

$$\blacksquare \pi_4(3) = 0 + 3 - 0 = 3$$

$$\blacksquare \pi_5(3) = 0 + 3 - 0 = 3$$

$$\blacksquare \pi_6(3) = 16 + 3 - 3 = 16$$

$$\blacksquare \pi_7(16) = 2 + 16 - 3 = 15$$

$$\blacksquare \pi_8(15) = 2 + 15 - 3 = 14$$

$$\blacksquare \pi_9(14) = 3 + 14 - 12 = 5$$

$$\blacksquare \pi_{10}(5) = 1 + 5 - 5 = 1$$

$$\blacksquare \pi_{11}(1) = 17 + 1 - 0 = 18$$

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# The matrixes

## Panel and query

0	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19
1	1	0	1	0	0	1	1	1	1	1	0	0	1	0	0	1	0	0	1
0	1	0	0	0	0	1	1	1	1	1	0	0	1	1	1	0	0	1	0
0	0	0	1	0	0	0	0	0	1	1	1	1	0	0	0	1	0	1	0
1	0	0	1	1	0	1	0	1	0	0	0	1	1	1	0	0	0	1	0
0	1	1	0	1	1	1	1	1	0	0	1	0	0	1	1	1	1	0	0
1	1	0	0	1	0	1	0	1	0	1	0	1	0	0	0	1	1	1	1
0	0	0	1	0	1	1	1	1	1	1	1	0	0	1	0	0	0	1	1
0	0	1	0	1	1	1	1	1	1	1	0	0	1	0	0	0	1	1	1

## PBWT Matrix

0	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19
1	1	0	1	0	0	1	0	0	1	1	0	1	1	1	0	1	0	1	1
0	0	0	1	1	0	0	1	1	0	0	1	1	1	1	0	1	0	1	0
0	1	0	1	1	1	1	1	1	0	0	0	0	0	0	0	1	0	1	1
1	0	0	0	0	0	1	0	1	1	1	1	0	0	0	1	0	1	1	0
0	1	1	1	0	0	1	0	1	1	1	0	0	1	1	0	0	0	0	0
1	0	0	0	1	1	1	1	1	1	1	0	1	0	0	1	1	0	1	0
0	1	0	0	0	0	1	1	1	0	1	1	0	0	1	0	0	1	0	1

# Prefix and Divergence Arrays

## Prefix Arrays

0	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19
0	1	2	2	1	1	1	2	2	2	5	3	3	1	4	5	5	6	6	0
1	2	6	6	5	2	2	1	5	5	3	4	5	0	6	2	2	3	3	4
2	4	3	3	4	6	0	0	3	3	4	5	1	4	5	0	0	1	1	6
3	6	1	1	2	0	5	5	1	1	2	2	0	6	2	4	6	5	2	3
4	0	4	0	6	5	3	3	0	0	1	1	4	3	1	6	3	2	0	1
5	3	0	5	3	4	6	6	6	6	0	0	2	5	0	1	4	0	5	2
6	5	5	4	0	3	4	4	4	4	6	6	6	2	3	3	1	4	4	5

## LCP Arrays: current $k$ minus the original Durbin's divergence arrays

0	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19
0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
0	1	2	3	3	1	2	0	1	0	6	3	1	9	3	3	4	3	4	1
0	1	1	2	1	5	4	3	4	5	2	0	2	1	1	1	2	1	2	0
0	1	0	1	0	3	1	2	0	1	0	1	8	2	2	0	1	0	1	5
0	0	2	2	4	0	2	3	4	5	1	2	0	0	0	4	2	5	4	3
0	1	1	3	3	2	0	1	2	3	6	7	1	2	10	1	0	3	0	2
0	1	2	0	2	1	1	2	3	4	4	5	3	1	1	2	2	1	2	1

0 4 4 0 0 3 0 0  
1 0 0 1 1 0 0 1  
3 5 5 3 2 4 1 2  
4 2 2 4 3 1 0 3  
5 6 6 5 4 5 2 4  
6 3 3 6 5 2 0 5  
6 6 2 6

0 0 0 0  
0 3 2 0  
3 0 0 3  
4 6 4 4  
5 1 1 6

0 0 0 0  
1 4 2 2  
3 1 0 3  
5 6 4 5  
6 3 2 6

$$\begin{array}{cccc}
 0 & 0 & 0 & 0 \\
 2 & 5 & 2 & 2 \\
 \color{red}{3} & \color{red}{2} & \color{red}{2} & \color{red}{4} \\
 \color{green}{5} & \color{green}{6} & \color{green}{2} & \color{green}{5} \\
 6 & 4 & 2 & 6
 \end{array}
 \Rightarrow
 \begin{array}{cccc}
 0 & 1 & 1 & 0 \\
 1 & 0 & 0 & 1 \\
 \color{green}{2} & \color{green}{2} & \color{green}{1} & \color{green}{5}
 \end{array}
 \Rightarrow
 \begin{array}{cccc}
 0 & 0 & 0 & 0 \\
 \color{red}{1} & \color{red}{3} & \color{red}{1} & \color{red}{1} \\
 3 & 1 & 1 & 3 \\
 \color{green}{5} & \color{green}{5} & \color{green}{1} & \color{green}{5}
 \end{array}
 \Rightarrow
 \begin{array}{cccc}
 0 & 0 & 0 & 0 \\
 1 & 1 & 1 & 3 \\
 \color{green}{1} & \color{green}{1} & \color{green}{1} & \color{green}{3}
 \end{array}
 \Rightarrow
 \begin{array}{cccc}
 0 & 3 & 2 & 0 \\
 \color{red}{1} & \color{red}{0} & \color{red}{0} & \color{red}{1} \\
 3 & 4 & 2 & 3 \\
 \color{green}{6} & \color{green}{2} & \color{green}{1} & \color{green}{6}
 \end{array}$$

0 2 2 0  
1 0 0 2  
3 3 3 3

⇒

0 0 0 0  
1 4 1 1  
2 1 0 2  
3 5 2 3  
4 2 1 4  
6 6 3 6

⇒

0 4 2 0  
2 0 0 4  
5 6 3 5  
6 3 1 6

⇒

0 4 2 0  
2 0 0 2  
4 6 4 4  
5 2 1 6

⇒

0 3 1 0  
2 0 0 2  
4 5 3 4  
5 2 0 5  
6 6 4 6

0 0 0 0    0 3 1 0    0 0 0 0    0 2 2 0    0 4 0 0  
 3 5 2 3    3 0 0 3    3 5 2 3    4 0 0 4    1 0 0 1  
 4 3 1 4  $\Rightarrow$  5 6 3 5  $\Rightarrow$  4 3 0 5  $\Rightarrow$  5 6 4 5  $\Rightarrow$  2 5 0 2  
 5 6 3 5    6 2 0 6    6 6 3 6    6 1 1 6    3 1 0 5  
 6 4 1 6       6 6 3 6    6 1 1 6    6 6 0 6

# Match with external haplotype I

## First case, bits matches at column $j$ -th

- we are looking at  $d$ -th bit of the  $k$ -th run, that come from the  $i$ -th row of the panel
- if this bit match the next bit of the pattern we can go to column  $j + 1$  and we figure out which bit to look at in that column
- the next bit we look at is still from row  $i$ -th

# Match with external haplotype II

## Second case, bits doesn't matches at column $j$ -th

- we are looking at  $d$ -th bit of the  $k$ -th run and that bit doesn't match the next bit in the pattern
- we look at the threshold for the  $k$ -th run:
  - if  $d$  is at most the threshold (check this "at most") then we move to the last bit of the  $(k - 1)$ -st run in the  $j$ -th column and then we proceed as in *case 1*
  - if  $d$  is greater than the threshold then we move to the first bit of the  $(k - 1)$ -st run in the  $j$ -th column and then we proceed as in *case 1*

# Travis's New Version

A column  $C$ 's representation consists of a bitvector  $B[0..m-1]$  and a sequence of thresholds  $T$ . It supports the query *CANDIDATE\_STEP*, which takes a single integer  $i$  and a bit  $b$  and returns a boolean flag  $f$  and a single integer  $i'$ . If  $B[i] = b$ , then  $f = \text{TRUE}$  and  $i'$  is the position of  $B[i]$  after  $B$  is stably sorted. If  $B[i] \neq b$  but there is some copy of  $b$  in  $B$ , the  $f = \text{FALSE}$  and  $i'$  is the position after  $B$  is stably sorted of either of the last copy of  $b$  before  $B[i]$  or of the first copy of  $b$  after  $B[i]$ , depending on whether  $B[i]$  is before or after the threshold for the run containing  $B[i]$ . If there is no copy of  $b$  in  $B$ , then  $f = \text{FALSE}$  and  $i' = -1$ .

We store  $B$  and  $T$  run-length compressed, so they take  $O(r_c)$  words of space and *CANDIDATE\_STEP* takes  $O(\log \log m)$  time. We start a search with  $i = 0$ ; we go from one column to the next setting  $i = i'$  when  $i' \geq 0$ , with  $f$  telling us whether we've jumped or not (**but not telling us whether we've hit the end of a MEM**); when  $i' = -1$ , we were unable to match a column and we start over at the next column with  $i = 0$ .

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# New Idea I

Let's take a step back and get closer to Durbin's original idea.

At the moment we save:

- the position of every head of a run
- a boolean to mark if the first run is composed by zeros or ones
- the  $c$  value of the column
- a single value for  $u$  and  $v$  (that are as in Durbin)
- the whole *divergence array*, actually the *LCP array*, (**WIP**)

# New Idea I

```
column: 5
start with 0? yes, c: 15
0    0
2    2
3    1
4    3
8    5
0 5 4 1 3 5 5 5 5 5 5 0 5 5 5 2 5 1 5 5
```

Figura: Example, column 5: 00101111000000000000

# New Idea II

## $uv$ values trick

Values  $u$  and  $v$  increase alternately in the biallelic case so we save every time the only value that increase at the head of a run.

The, with a simple *If/Else* selection based on the first element of the column and the index of the run and on being even or odd of the index we can extract both  $u$  and  $v$  values. Infact the two values are, alternatively, saved in the current index and in the previous one.

## $w(i, \sigma)$ function

We can use the same *LF-mapping* as in Durbin but we have to consider every time an *offset* between the position of the head of the run, that's  $i$ , which contains the “virtual” index, and the index itself.

$$w(i, \sigma) = \begin{cases} u[i] + \text{offset} & \text{if } \sigma = 0 \\ c + v[i] + \text{offset} & \text{if } \sigma = 1 \end{cases}$$

# New Idea III

## External haplotype matches

Then we proceed as in Durbin, updating  $f$  and  $g$  using  $w(i, \sigma)$ . Every time we “virtually” use indexes over the whole column but actually run heads plus offsets are used.

In case we update  $e$  using  $f$  and the *divergence/LCP array* (**WIP**). In order to update  $e$  we should in theory follow the line indicated in  $i + 1$  by  $f$  in the original panel which we have not memorized. So, at most at a cost of  $O(r)$  for every column, we proceed to reverse the use of  $u$  and  $v$  to move backwards between the columns virtually following a row of the original panel.

Then we use the *divergence/LCP array* (**WIP**) to update  $f$  and  $g$  depending on the case.

After detect a match, we can know the cardinality of the lines that match but not what they are,

# New Idea IV

## WIP

At the moment *divergence array* is saved as an `sds1::int_vector<>` on which it's used `sds1::util::bit_compress()` in order to save space. The original idea of thresholds seems to me absolutely not applicable but maybe we can think of storing only a subset of the *divergence/LCP array* and I'm thinking how to do it.