

# RLPBWT

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# Outline

- 1 The Old Idea
- 2 The New Idea
- 3 BitVector Version

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# Durbin's Algorithm

## Algorithm 1 Algorithm 5 from Durbin's paper

**function** FIND\_SET\_MAXIMAL\_MATCHES\_FROM\_Z( $z$ )

**for**  $k \leftarrow 0$  **to**  $N$  **do**

$e, f, g \leftarrow \text{Update\_Z\_Matches}(k, z, e, f, g)$

**function** UPDATE\_Z\_MATCHES( $k, z, e, f, g$ )

$f' \leftarrow w(k, f, z[k])$

▷  $a_k$ ,  $d_k$  and  $y_i^k$  as in Durbin's paper

$g' \leftarrow w(k, g, z[k])$

**if**  $f' < g'$  **then**

▷ if  $k$  is  $N - 1$  report matches from  $e_k$  to  $N - 1$

$e' \leftarrow e_k$

**else**

▷ report matches from  $e_k$  to  $k$

$e' \leftarrow d_{k+1}[f'] - 1$

**if**  $z[e'] = 0$  **and**  $f' > 0$  **then**

$f' \leftarrow g' - 1$

**while**  $z[e' - 1] = y_{f'}^{k+1}[e' - 1]$  **do**  $e' \leftarrow e' - 1$

**while**  $d_{k+1}[f'] \leq e'$  **do**  $f' \leftarrow f' - 1$

**else**

$g' \leftarrow f' + 1$

**while**  $z[e' - 1] = y_{f'}^{k+1}[e' - 1]$  **do**  $e' \leftarrow e' - 1$

**while**  $g' < M$  **and**  $d_{k+1}[g'] \leq e'$  **do**  $g' \leftarrow g' + 1$

**return**  $e', f', g'$

# Durbin's Algorithm Example

X	00	01	02	03	04	05	06	07	08	09	10	11	12	13	14
00	1	1	0	1	1	0	0	0	1	0	0	1	1	1	1
01	1	1	0	1	1	0	0	0	1	0	0	1	1	1	1
02	1	1	0	1	1	1	0	0	0	1	1	1	0	1	1
03	1	1	0	1	1	0	0	0	1	0	0	1	1	0	1
04	0	1	0	1	0	1	0	0	1	0	0	1	1	0	1
05	0	1	0	1	0	1	0	0	0	0	0	1	0	0	1
06	0	1	0	1	0	1	0	0	0	0	0	1	0	0	0
07	0	1	0	1	1	1	0	0	0	0	0	0	1	0	1
08	0	1	0	0	1	0	0	0	1	0	0	0	1	0	1
09	0	1	0	1	0	0	0	0	1	0	0	0	0	0	1
10	0	1	0	1	1	0	0	0	0	0	0	1	1	0	1
11	0	1	0	0	1	0	1	1	0	0	0	1	0	0	1
12	0	1	0	0	1	0	0	1	0	0	0	0	0	0	1
13	0	1	0	0	0	0	0	1	0	0	0	0	0	0	1
14	0	1	0	0	0	0	0	0	0	0	0	0	0	0	1
15	0	0	0	0	0	0	0	0	0	0	0	0	0	0	1
16	0	0	0	1	0	0	0	0	0	0	0	0	0	0	1
17	1	0	1	0	0	0	0	0	0	0	1	1	0	0	1
18	0	0	1	0	0	0	0	0	0	0	1	1	0	0	1
19	0	1	0	0	0	0	0	0	0	0	1	1	0	0	1
z	0	1	0	0	1	0	1	0	0	0	1	1	1	0	1

# Durbin's Algorithm Example

	$e = 0$				$e = 3$				$e = 7$				$e = 11$			
X	00	01	02	03	04	05	06	07	08	09	10	11	12	13	14	15
00	0	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15
01	0	0	0	0	0	0	0	0	0	2	2	2	7	9	6	15
02	0	0	0	0	0	1	4	4	4	5	5	5	8	0	0	6
03	0	0	0	0	0	4	2	2	2	0	3	6	9	12	0	0
04	0	0	2	2	0	2	0	0	0	3	6	4	0	6	8	0
05	0	0	0	0	0	0	0	0	0	6	4	0	4	0	0	8
06	0	0	0	0	1	0	5	5	5	4	0	0	0	0	11	0
07	0	0	0	0	3	0	0	0	0	0	0	0	12	8	9	11
08	0	0	0	0	0	0	0	0	0	0	0	7	2	0	0	0
09	0	0	0	0	4	0	0	0	0	0	7	8	5	11	10	10
10	0	0	0	0	0	0	3	3	3	7	8	0	6	9	13	13
11	0	0	0	0	0	5	0	4	6	8	0	0	4	0	7	7
12	0	0	0	0	0	0	4	0	4	0	0	9	0	10	9	9
13	0	0	0	0	2	0	0	0	0	0	9	0	0	13	0	0
14	0	0	0	0	0	0	0	6	0	9	0	4	8	7	12	12
15	0	1	0	0	0	3	6	4	0	0	4	0	0	9	2	2
16	0	0	0	0	0	0	4	0	7	4	0	11	11	0	6	6
17	0	0	0	1	0	4	0	0	8	0	5	9	9	12	14	14
18	0	0	0	3	0	0	0	0	0	5	0	0	0	2	9	9
19	0	0	1	0	0	0	0	7	0	0	10	10	10	6	0	0

Diagram illustrating the Durbin's Algorithm example. The table shows the values of  $X$  (rows) and  $e$  (columns) for various indices. Red circles and arrows highlight specific values and their relationships:

- Red circles around  $X=00, e=00$  and  $X=15, e=00$ .
- Red circles around  $X=16, e=10$  (value 0) and  $X=17, e=11$  (value 9).
- Red circles around  $X=18, e=10$  (value 5) and  $X=19, e=10$  (value 7).
- Red circles around  $X=19, e=06$  (value 0) and  $X=19, e=10$  (value 10).
- Red circles around  $X=19, e=12$  (value 10) and  $X=19, e=14$  (value 14).
- Red arrows pointing from  $X=00, e=00$  to  $X=15, e=00$  and from  $X=15, e=00$  to  $X=16, e=10$ .
- Red arrows pointing from  $X=16, e=10$  to  $X=17, e=11$  and from  $X=17, e=11$  to  $X=18, e=10$ .
- Red arrows pointing from  $X=18, e=10$  to  $X=19, e=10$  and from  $X=19, e=10$  to  $X=19, e=12$ .
- Red arrows pointing from  $X=19, e=12$  to  $X=19, e=14$  and from  $X=19, e=14$  to  $X=19, e=16$ .

# Durbin's Algorithm Example

X	00	01	02	03	04	05	06	07	08	09	10	11	12	13	14	15
00	0	4	0	0	8	14	14	14	14	0	0	0	7	1	18	11
01	1	5	1	1	11	15	15	15	15	16	16	16	19	9	4	18
02	2	6	2	2	12	17	0	0	0	8	11	18	1	10	5	4
03	3	7	3	3	13	0	9	9	9	11	18	17	14	18	6	5
04	4	8	4	4	14	4	10	10	10	18	17	4	15	4	2	6
05	5	9	5	5	15	5	16	16	16	17	4	5	9	5	3	2
06	6	10	6	6	17	6	8	8	8	4	5	6	10	6	11	3
07	7	11	7	7	18	7	11	11	11	5	6	7	0	2	12	12
08	8	12	8	8	19	9	12	12	12	6	7	19	16	3	13	13
09	9	13	9	9	0	10	13	13	13	7	19	1	18	11	8	8
10	10	14	10	10	1	16	18	18	18	19	1	2	17	12	7	7
11	11	15	11	11	2	8	19	1	17	1	2	3	4	13	19	19
12	12	16	12	12	3	11	1	2	4	2	3	14	5	8	14	14
13	13	18	13	13	4	12	2	3	5	3	14	15	6	7	15	15
14	14	19	14	14	5	13	3	17	6	14	15	9	2	19	0	0
15	15	0	15	15	6	18	17	4	7	15	9	10	3	14	16	16
16	16	1	16	16	7	19	4	5	19	9	10	11	11	15	17	17
17	17	2	18	17	9	1	5	6	1	10	12	12	12	0	1	1
18	18	3	19	18	10	2	6	7	2	12	13	13	13	16	9	9
19	19	17	17	19	16	3	7	19	3	13	8	8	8	17	10	10

# Durbin's Algorithm Example

X	00	01	02	03	04	05	06	07	08	09	10	11	12	13	14
00	1	0	0	1	0	0	0	0	0	0	0	1	1	0	1
01	1	0	0	1	1	0	0	1	0	0	0	0	0	1	1
02	1	0	0	1	1	0	0	1	0	0	0	1	0	0	1
03	1	0	0	1	1	0	0	1	0	0	0	1	0	0	1
04	0	1	0	1	0	1	0	0	0	0	0	1	0	0	1
05	0	1	0	1	0	1	0	0	0	0	0	1	0	0	1
06	0	1	0	1	0	1	0	0	0	0	0	1	0	0	1
07	0	1	0	1	0	1	0	0	0	0	0	0	1	0	1
08	0	1	0	0	1	0	0	0	0	1	1	1	0	0	1
09	0	1	0	1	0	0	0	0	1	0	0	0	0	1	1
10	0	1	0	1	0	0	0	0	1	0	0	0	0	1	1
11	0	1	0	0	1	0	0	0	0	0	1	1	0	0	0
12	0	1	0	0	1	0	0	0	1	0	1	1	0	0	1
13	0	1	0	0	1	0	0	0	1	0	1	1	0	0	1
14	0	1	0	0	0	0	0	0	1	0	0	0	1	0	1
15	0	1	0	0	0	0	0	0	1	0	0	0	1	0	1
16	0	1	0	1	0	0	0	0	0	0	0	1	1	0	1
17	1	1	0	0	0	1	0	0	0	0	0	1	1	0	1
18	0	1	1	0	1	0	0	0	0	0	0	1	0	0	1
19	0	1	1	0	1	0	1	0	0	0	0	0	1	0	1
z	0	1	0	0	1	0	1	0	0	0	1	1	1	0	1

$pre_e = 6$   
at 7

$pre_e = 8$   $pre_e = 13$   
at 11 at 13



X	00	01	02	03	04	05	06	07	08	09	10	11	12	13	14
00	1	0	0	1	0	0	0	0	0	0	0	1	1	0	1
01	1	0	0	1	1	0	0	1	0	0	0	0	0	1	1
02	1	0	0	1	1	0	0	1	0	0	0	1	0	0	1
03	1	0	0	1	1	0	0	1	0	0	0	1	0	0	1
04	0	1	0	1	0	1	0	0	0	0	0	1	0	0	1
05	0	1	0	1	0	1	0	0	0	0	0	1	0	0	1
06	0	1	0	1	0	1	0	0	0	0	0	1	0	0	1
07	0	1	0	1	0	1	0	0	0	0	0	0	1	0	1
08	0	1	0	0	1	0	0	0	0	1	1	1	0	0	1
09	0	1	0	1	0	0	0	0	1	0	0	0	0	1	1
10	0	1	0	1	0	0	0	0	1	0	0	0	0	1	1
11	0	1	0	0	1	0	0	0	0	0	1	1	0	0	0
12	0	1	0	0	1	0	0	0	1	0	1	1	0	0	1
13	0	1	0	0	1	0	0	0	1	0	1	1	0	0	1
14	0	1	0	0	0	0	0	0	1	0	0	0	1	0	1
15	0	1	0	0	0	0	0	0	1	0	0	0	1	0	1
16	0	1	0	1	0	0	0	0	0	0	0	1	1	0	1
17	1	1	0	0	0	1	0	0	0	0	0	1	1	0	1
18	0	1	1	0	1	0	0	0	0	0	0	1	0	0	1
19	0	1	1	0	1	0	1	0	0	0	0	0	1	0	1
z	0	1	0	0	1	0	1	0	0	0	1	1	1	0	1

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- 2 The New Idea
- 3 BitVector Version

# 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 sometimes 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.

# Two Pass on RLPBWT

In order to not save the *divergence/LCP array* we could make two *RLPBWT*, one for the normal order and one for the reverse order.

Instead of use the *divergence/LCP array* to retrieve matches that are overlapped in the panel we look for non overlapped matches only. To do this when we find a match we continue the search updating  $f$  and  $g$  based only on  $c$  and  $h$ , the total number of haplotypes in the panel:

$$(f, g) = \begin{cases} (0, c) & \text{if } \sigma = 0 \\ (c, h) & \text{if } \sigma = 1 \end{cases}$$

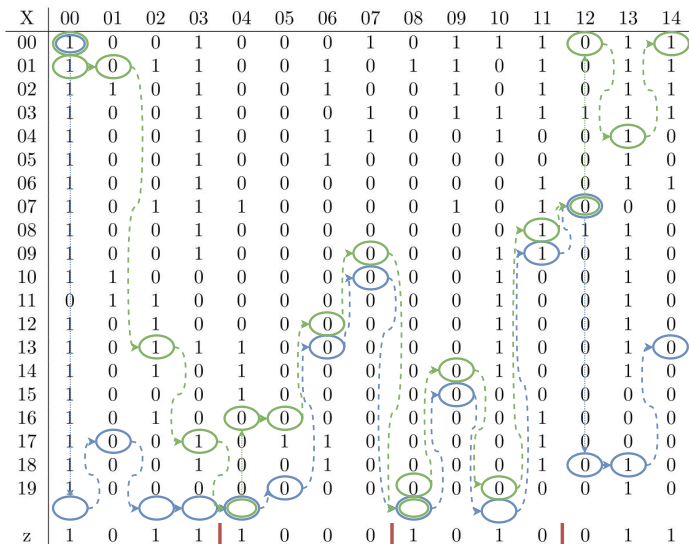
We query in this way the first one with the haplotype and the second one with the reverse of the haplotype. Than we intersect the results.



## Two Pass on RLPBWT

X	00	01	02	03	04	05	06	07	08	09	10	11	12	13	14
00	1	1	0	1	1	0	0	0	1	0	0	1	1	1	1
01	1	1	0	1	1	0	0	0	1	0	0	1	1	1	1
02	1	1	0	1	1	1	0	0	0	1	1	1	0	1	1
03	1	1	0	1	1	0	0	0	1	0	0	1	1	0	1
04	0	1	0	1	0	1	0	0	1	0	0	1	1	0	1
05	0	1	0	1	0	1	0	0	0	0	0	1	0	0	1
06	0	1	0	1	0	1	0	0	0	0	0	1	0	0	0
07	0	1	0	1	1	1	0	0	0	0	0	0	1	0	1
08	0	1	0	0	1	0	0	0	1	0	0	0	1	0	1
09	0	1	0	1	0	0	0	0	1	0	0	0	0	0	1
10	0	1	0	1	1	0	0	0	0	0	0	1	1	0	1
11	0	1	0	0	1	0	1	1	0	0	0	1	0	0	1
12	0	1	0	0	1	0	0	1	0	0	0	0	0	0	1
13	0	1	0	0	0	0	0	1	0	0	0	0	0	0	1
14	0	1	0	0	0	0	0	0	0	0	0	0	0	0	1
15	0	0	0	0	0	0	0	0	0	0	0	0	0	0	1
16	0	0	0	1	0	0	0	0	0	0	0	0	0	0	1
17	1	0	1	0	0	0	0	0	0	0	0	1	1	0	1
18	0	0	1	0	0	0	0	0	0	0	1	1	0	0	1
19	0	1	0	0	0	0	0	0	0	0	1	1	0	0	1
z	0	1	0	0	1	0	1	0	0	0	1	1	1	0	1

## Two Pass on RLPBWT



## Two Pass on RLPBWT

X	00	01	02	03	04	05	06	07	08	09	10	11	12	13	14
00	1	0	0	1	0	0	0	0	0	0	0	1	1	0	1
01	1	0	0	1	1	0	0	1	0	0	0	0	0	1	1
02	1	0	0	1	1	0	0	1	0	0	0	1	0	0	1
03	1	0	0	1	1	0	0	1	0	0	0	1	0	0	1
04	0	1	0	1	0	1	0	0	0	0	0	1	0	0	1
05	0	1	0	1	0	1	0	0	0	0	0	1	0	0	1
06	0	1	0	1	0	1	0	0	0	0	0	1	0	0	1
07	0	1	0	1	0	1	0	0	0	0	0	0	1	0	1
08	0	1	0	0	1	0	0	0	0	1	1	1	0	0	1
09	0	1	0	1	0	0	0	0	1	0	0	0	0	1	1
10	0	1	0	1	0	0	0	0	1	0	0	0	0	1	1
11	0	1	0	0	1	0	0	0	0	0	1	1	0	0	0
12	0	1	0	0	1	0	0	0	1	0	1	1	0	0	1
13	0	1	0	0	1	0	0	0	1	0	1	1	0	0	1
14	0	1	0	0	0	0	0	0	1	0	0	0	1	0	1
15	0	1	0	0	0	0	0	0	1	0	0	0	1	0	1
16	0	1	0	1	0	0	0	0	0	0	0	1	1	0	1
17	1	1	0	0	0	1	0	0	0	0	0	1	1	0	1
18	0	1	1	0	1	0	0	0	0	0	0	1	0	0	1
19	0	1	1	0	1	0	1	0	0	0	0	0	1	0	1
z	0	1	0	0	1	0	1	0	0	0	1	1	1	0	1

Match

Forward

Backward

# Testing

## Benchmark

At the moment I'm testing the implementation using the sample data (VCF files) at:

[https://github.com/ZhiGroup/Syllable-PBWT/tree/master/sample\\_data](https://github.com/ZhiGroup/Syllable-PBWT/tree/master/sample_data)  
(the panel is  $900 \times 500$  with 100 queries).

## Next Work

Trying to implement also non maximal matches with a query, as, for example, in Naseri's paper.

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# The column data I

We save, for every column:

- a bit vector of the same length of the dense PBWT column, with 1 in every position before a head of a run
- 2 bit vector that can be queried to obtain  $u$  and  $v$  value. So we have a bitvector for zeros that contains 1 in position  $i$  iff we have  $i$  zeros at point in the column when we change value anche similar for ones(**to be explained better**)
- the  $c$  value and a bool to indicate how a column start

# The column data II

## example

If we have a column:

$$c = 00001110001111110001111111$$

We save:

$$h = 000100100100000100010000000(1)$$

$$u = 00010010001 \text{ (to represent 4,3 and 4 zeros)}$$

$$v = 0010000010000001 \text{ (to represent 3,6 and 7 ones)}$$

# The column data III

## example

If we want to obtain, for example, the number of zeros before and index  $i$ , for example  $i = 18$ :

- we *rank*  $h$  to obtain the run that contain  $i$ ,  $rank_h(18) = 4$
- we know that the column start with 0 so we are in a run of zeros and we have  $\lfloor \frac{4}{2} \rfloor = 2$  run's of zeros before
- we know that the number of zeros in the previous complete runs is  $select_u(2) + 1 = 7$
- remain zeros in the run to compute are given by  $i - (select_h(4) + 1)$

## TODO

Re-implement the algorithm using bitvectors, *rank* and *select* instead of run head indices and *uv* values.