

# 1 Overview

In the last lecture we discussed **Suffix Arrays** and **Burrows-Wheeler Transform (BWT)**.

## Goal

The goal of this lecture is to be able to **store the human genome** efficiently.

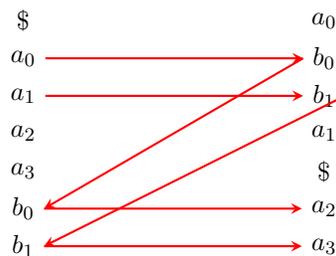
- Human genome: 3 billion base pairs
- Alphabet size:  $|\Sigma| = 4$  (DNA bases)
- Memory requirements:
  - Suffix tree: 47 GB
  - Suffix array: 12 GB
  - FM-index: 1.5 GB

# 2 BWT Pattern Search Review

Consider the string  $T = \text{abaaba\$}$  and the pattern  $P = \text{aba}$ .

The BWT produces first and last columns  $F$  and  $L$  (with subscripts to distinguish repeated letters):

$F$	\$	$a_0$	$a_1$	$a_2$	$a_3$	$b_0$	$b_1$
$L$	$a_0$	$b_0$	$b_1$	$a_1$	\$	$a_2$	$a_3$



**Explanation:** The red arrows show the **LF-mapping steps** as we search for the pattern **aba**. This corresponds to tracking the suffixes that match the pattern in reverse order via the BWT.

### 3 FM-Index

Ferragina and Manzini [2000]

The FM-index is an index that **combines the BWT with a few small auxiliary data structures**.

- Core of the index: **First (F) + Last (L) columns** from BWT
- $F$  can be represented very simply (1 integer per character)
- $L$  is highly compressible

#### 3.1 Issues with BWT Pattern Search

1. Scanning characters in the last column  $L$  can be very slow.
2. Need a way to find where matches occur in  $T$ .

#### 3.2 Solution 1: Tally Table

**Idea:** Precompute number of occurrences of each character up to each row (cumulative counts).

Without the tally table, we need to check all of  $a_0, a_1, a_2, a_3$  to see which has a corresponding  $b$  in the right column. With the tally table, we can narrow the search window to between  $a_1$  and  $a_2$ , significantly reducing the number of checks.

$F$		\$	$a_0$	$a_1$	$a_2$	$a_3$	$b_0$	$b_1$
$L$		$a_0$	$b_0$	$b_1$	$a_1$	\$	$a_2$	$a_3$

		1	2	3	4	5	6	7
Tally: $a$		1	1	1	2	2	3	4
$b$		0	1	2	2	2	2	2

**Space trade-off:** Storing every entry in the tally table is costly. Instead, we store every  $x$ -th tally and perform a few extra scans between stored entries (sparsifying), balancing space and speed.

#### 3.3 Solution 2: Sampled Suffix Array

**Idea:** Tally tables solve the problem of speeding up the search, but we still need a way to locate the matching substrings. The suffix array directly corresponds to the ranking of the permuted strings in the BWT and lets us retrieve the offsets of the matches.

- We sparsify and store only some entries of the suffix array.

## 4 FM-Index Memory Footprint

- **First Column (F):**  $\sim |\Sigma|$  integers
- **Last Column (L):**  $T$  characters
- **Sampled Suffix Array:**  $T \cdot a$  integers, where  $a$  is the fraction of rows sampled
- **Sampled Tally:**  $T \cdot |\Sigma| \cdot b$  integers, where  $b$  is the fraction of rows sampled

### 4.1 Example: Human Genome

- DNA alphabet:  $|\Sigma| = 4$
- Human genome: 3 billion base pairs
- F:  $4 \times 4 = 16$  bytes
- L: 2 bits per base  $\times 3\text{B}$  bases  $\approx 750$  MB
- Sampled SA:  $3\text{B} \times 4 \times 1/32 \approx 400$  MB
- Sampled Tally:  $3\text{B} \times 4 \times 4 \times 1/128 \approx 100$  MB
- **Total:**  $\lesssim 1.5$  GB

## References

P. Ferragina and G. Manzini. Opportunistic data structures with applications. In *Proceedings of the 41st Annual Symposium on Foundations of Computer Science, FOCS '00*, page 390, USA, 2000. IEEE Computer Society. ISBN 0769508502.