Ultrafast and memory-efficient alignment of short DNA sequences to the human genome

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We are in 2009

Outline of Bowtie

- Burrows-Wheeler Transform(Indexing)
- Exact and inexact alignment
- ► Excessive backtracking

Burrows- Wheeler Transform: Forward Transform

Let $T = \mathtt{BANANA}$. $\mathsf{BWT}(T)$ will be:

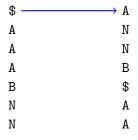
Burrows- Wheeler Transform: Forward Transform

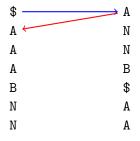
Let T = BANANA. BWT(T) will be:

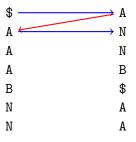
$$\begin{bmatrix} \$ & \texttt{B} & \texttt{A} & \texttt{N} & \texttt{A} & \texttt{N} & \texttt{A} \\ \texttt{A} & \$ & \texttt{B} & \texttt{A} & \texttt{N} & \texttt{A} & \texttt{N} \\ \texttt{N} & \texttt{A} & \$ & \texttt{B} & \texttt{A} & \texttt{N} & \texttt{A} \\ \texttt{A} & \texttt{N} & \texttt{A} & \$ & \texttt{B} & \texttt{A} & \texttt{N} \\ \texttt{A} & \texttt{N} & \texttt{A} & \$ & \texttt{B} & \texttt{A} & \texttt{N} \\ \texttt{N} & \texttt{A} & \texttt{N} & \texttt{A} & \$ & \texttt{B} & \texttt{A} & \texttt{N} \\ \texttt{A} & \texttt{N} & \texttt{A} & \texttt{N} & \texttt{A} & \$ & \texttt{B} & \texttt{A} & \texttt{N} \\ \texttt{A} & \texttt{N} & \texttt{A} & \texttt{N} & \texttt{A} & \$ & \texttt{B} & \texttt{A} & \texttt{N} \\ \texttt{A} & \texttt{N} & \texttt{A} & \texttt{N} & \texttt{A} & \$ & \texttt{B} & \texttt{A} & \texttt{N} \\ \texttt{B} & \texttt{A} & \texttt{N} & \texttt{A} & \texttt{N} & \texttt{A} & \$ & \texttt{B} \\ \texttt{B} & \texttt{A} & \texttt{N} & \texttt{A} & \texttt{N} & \texttt{A} & \$ & \texttt{B} \\ \texttt{B} & \texttt{A} & \texttt{N} & \texttt{A} & \texttt{N} & \texttt{A} & \$ & \texttt{B} \\ \texttt{N} & \texttt{A} & \$ & \texttt{B} & \texttt{A} & \texttt{N} & \texttt{A} \\ \texttt{N} & \texttt{A} & \$ & \texttt{B} & \texttt{A} & \texttt{N} & \texttt{A} \\ \texttt{N} & \texttt{A} & \texttt{N} & \texttt{A} & \$ & \texttt{B} & \texttt{A} \\ \end{bmatrix}$$

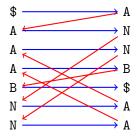
$$BWT(T) \to \mathtt{ANNB\$AA}$$

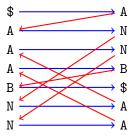
\$	A
Α	N
Α	N
Α	В
В	\$
N	A
N	Α











follow the tip of blue arrows to get back the initial message: BANANA\$

Pattern to find is $P={\tt NAN}.$ $\Sigma=B_1A_1N_1A_2N_2A_3$

\$	\mathbf{A}_1
\mathtt{A}_1	N_1
\mathtt{A}_2	N_2
A_3	B_1
\mathtt{B}_1	\$
N_1	\mathtt{A}_2
N_2	\mathtt{A}_3

Pattern to find is P = NAN. $\Sigma = B_1 A_1 N_1 A_2 N_2 A_3$

0	\$	\mathtt{A}_1	1
1	\mathtt{A}_1	N_1	5
2	\mathtt{A}_2	N_2	6
3	\mathtt{A}_3	B_1	4
4	\mathtt{B}_1	\$	0
5	N_1	\mathtt{A}_2	2
6	\mathtt{N}_2	\mathtt{A}_3	3

Pattern to find is $P={\tt NAN}.$ $\Sigma=B_1A_1N_1A_2N_2A_3$

top→0	\$	\mathtt{A}_1	1
1	\mathtt{A}_1	N_1	5
2	\mathtt{A}_2	N_2	6
3	\mathtt{A}_3	B_1	4
4	\mathtt{B}_1	\$	0
5	N_1	\mathtt{A}_2	2
$bot \rightarrow 6$	N_2	\mathtt{A}_3	3

Pattern to find is P = NAN. $\Sigma = B_1 A_1 N_1 A_2 N_2 A_3$

	0	\$	\mathtt{A}_1	1
	1	\mathtt{A}_1	\mathtt{N}_1	5
	2	\mathtt{A}_2	\mathtt{N}_2	6
	3	A_3	\mathtt{B}_1	4
	4	\mathtt{B}_1	\$	0
$\mathtt{N}_1{ ightarrow}$	5	N_1	\mathtt{A}_2	2
$ exttt{N}_2 ightarrow$	6	N_2	\mathtt{A}_3	3

Pattern to find is P = NAN. $\Sigma = B_1 A_1 N_1 A_2 N_2 A_3$

	0	\$	\mathtt{A}_1	1
	1	\mathtt{A}_1	N_1	5
$\mathtt{A}_{2}\mathtt{N}_{1}{\rightarrow}$	2	\mathtt{A}_2	N_2	6
${\tt A}_3{\tt N}_2{\to}$	3	\mathtt{A}_3	\mathtt{B}_1	4
	4	B_1	\$	0
	5	N_1	\mathtt{A}_2	2
	6	\mathtt{N}_2	\mathtt{A}_3	3

Pattern to find is $P={\tt NAN}.$ $\Sigma=B_1A_1N_1A_2N_2A_3$

	0	\$	\mathtt{A}_1	1
	1	\mathtt{A}_1	\mathtt{N}_1	5
	2	\mathtt{A}_2	\mathtt{N}_2	6
	3	\mathtt{A}_3	B_1	4
$\mathtt{B}_{1}\mathtt{A}_{3}\mathtt{N}_{2}{\rightarrow}$	4	\mathtt{B}_1	\$	0
	5	N_1	\mathtt{A}_2	2
$\mathtt{N}_{3}\mathtt{A}_{2}\mathtt{N}_{1}{ ightarrow}$	6	N_2	\mathtt{A}_3	3