Dissertation Title

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Abstract

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Part I APPROXIMATE STRING MATCHING

Introduction

- 1.1 Bioinformatics applications
- 1.2 Problem definition
- 1.3 Overview of existing methods
- 1.3.1 Online methods
- 1.3.2 Indexed methods
- 1.3.3 Filtering methods
- 1.4 Related problems
- 1.4.1 Local similarity search
- 1.4.2 Dictionary search
- 1.4.3 Overlaps computation

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Online Methods

- 2.1 Myers' bit-vector algorithm
- 2.2 Banded Myers' bit-vector algorithm
- 2.3 Increased bit-parallelism using SIMD instructions

Indexed Methods

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- 3.1.3 Suffix array
- 3.1.4 q-Gram index

3.2 Compressed Full-Text Indices

- 3.2.1 Burrows-Wheeler transform
- 3.2.2 FM-index
- 3.2.3 Rank dictionaries
- 3.3 Backtracking
- 3.3.1 Pruning methods
- 3.3.2 Multiple backtracking

Filtering Methods

- 4.1 Counting filters
- 4.1.1 Exact seeds
- 4.1.2 Gapped seeds
- 4.2 Factor filters
- 4.2.1 Exact seeds
- 4.2.2 Approximate seeds
- 4.3 Suffix filters

Part II APPLICATIONS

Read Mapping

5.1	Related work
5.1.1	Best mappers
5.1.2	All mappers
5.2	The Masai mapper
5.2.1	Single-end mapping
5.2.2	Paired-end mapping
5.3	The Masai 2 mapper
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5.3.3	Parallelization
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5.4	Experimental results
5.4.1	Comparison of filtration strategies
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5.4.3	Variant detection results
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5.5 Discussion

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String Similarity Search / Join

- 6.1 The competition
- 6.2 Our method
- 6.2.1 Implementation
- 6.2.2 Parallelization
- 6.3 Related methods
- 6.4 Experimental results
- **6.4.1** Search
- 6.4.2 Join
- 6.5 Discussion

APPENDIX



Declaration

I declare that this thesis is my own work and has not been submitted in any form for another degree or diploma at any university or other institute of tertiary education. Information derived from the published and unpublished work of others has been acknowledged in the text and a list of references is given.

Enrico Siragusa October 5, 2013

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LIST OF ALGORITHMS

LIST OF NOTATIONS

bp	base pair, character of the alphabet {A,C,G,T}	?
Mb	megabase, 1 million base pairs or characters	??
MB	megabyte, 1 MB = $1024 \text{ kB} = 1,048,576 \text{ byte } \dots \dots \dots \dots$??
GB	gigabyte, 1 GB = 1024 MB	??
Σ, Ψ, Φ	finite alphabets	??
Σ^*	set of all possible strings over the alphabet Σ	??
Σ^n	set of all possible strings over the alphabet Σ with length n	??
ϵ	empty string	??
s	length of string s	??
s[i]	character of s at position i (counting from 0)	??
s_i , suf(s , i)	suffix of s beginning at position i	??
[ij]	set of integers $i, i + 1,, j$??
[ij)	set of integers $i, i+1,, j-1$??
\mathbb{N}_0	set of non-negative integers	??
≺, ≤	strict and non-strict substring relation	??
〈 〉	definition of a string analogous to the set notation	??
$\operatorname{lcp} \mathcal{S}$	longest common prefix of a set S of strings	??
< _{lex}	lexicographical order	??
< _q	lexicographical prefix order, compares only prefixes of length $q \ \dots \ \dots$??
\$, \$ ^j	(virtual) sentinel characters to well-define the suffix tree	??
concat(v)	edge label concatenation on the path from root to suffix tree node $v \;\;$.	?
\overline{s}	suffix tree node whose edge label concatenation is $s \ldots \ldots$??
rank(a)	rank of character a in the underlying alphabet	?
R,D,I	edit operations that replace, delete, or insert a character	??
$ T _{\mathrm{E}}$	number of edit operations in transcript T	??
$ T _{\mathbb{R}}$	number of matches, replacements, and deletions in transcript T	??
${\mathcal R}$	sequenced reads, set of strings	??
G	reference sequence, string	??
${\mathcal D}$	database, set of strings	?
$freq(\phi, \mathcal{D})$	absolute number of strings in ${\mathcal D}$ that contain ϕ at least once $\dots \dots$?
$supp(\phi, \mathcal{D})$	relative number of strings in \mathcal{D} that contain ϕ at least once	??