

Practical significance of some full-text indices for read mapping

Bowtie2



Based on FM-index for seed finding

Novel strategy / heuristic for seed scoring and exploration

Makes use of SIMD-accelerated alignment DP

Capable of global (end-to-end) or local alignment

No spliced alignment (i.e. for DNA-seq or RNA-seq -> txome)

Bowtie2

■ Bowtie 2 ● BWA ▲ SOAP2 ◆ Bowtie

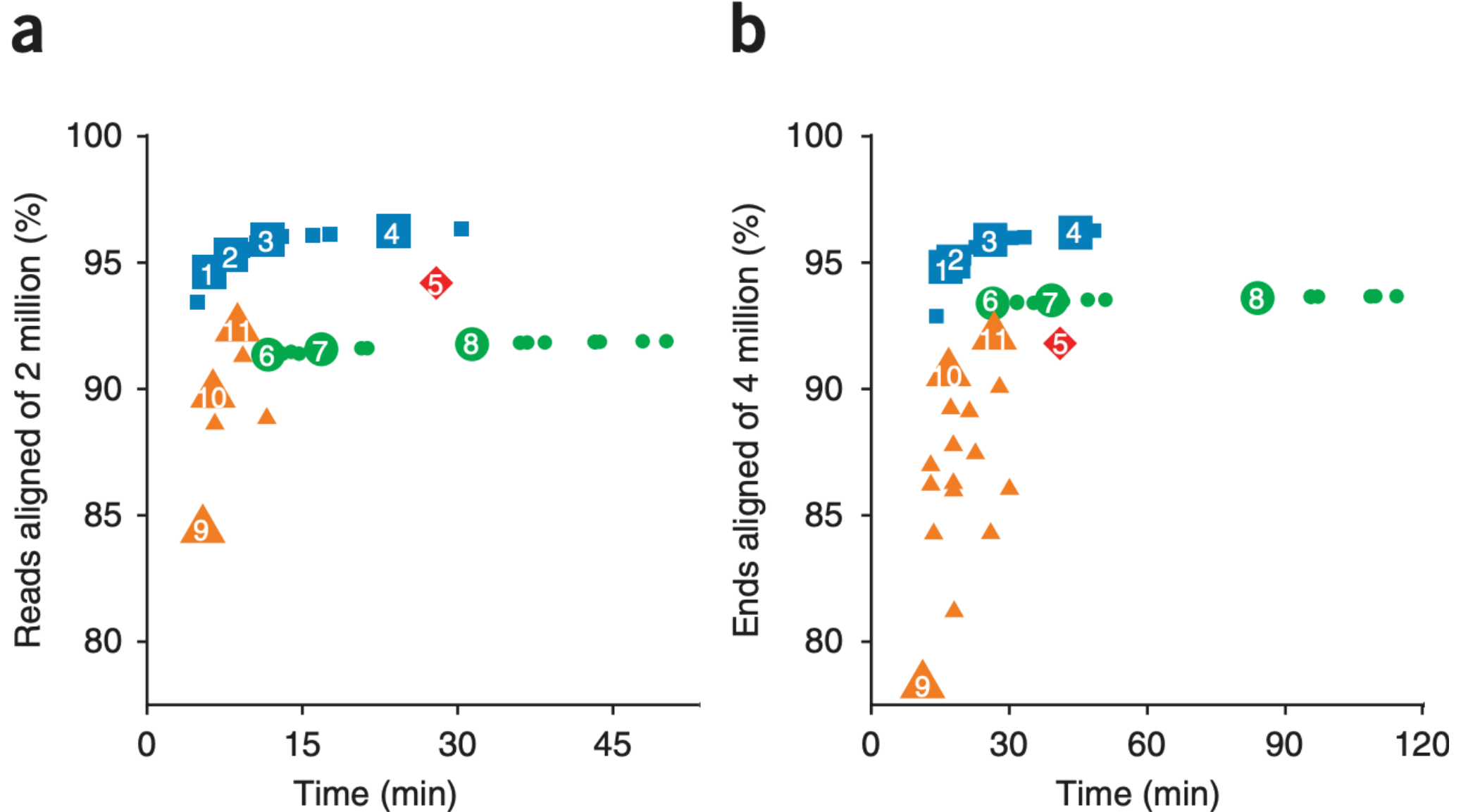


Figure 1 | Alignment comparison using HiSeq 2000, 454 and Ion Torrent reads. (a–d) Bowtie 2, BWA, SOAP2 and Bowtie were used to align two million 100 nt × 100 nt paired-end HiSeq 2000 reads from a resequencing study¹¹. Shown are results for unpaired alignment of end 1 (a), paired-end alignment (b), Bowtie 2 and BWA-SW alignment of 1 million 454 reads from the 1000 Genomes Project Pilot¹² (c), and Bowtie 2 and BWA-SW to align one million Ion Torrent reads from the G. Moore resequencing project¹³ (d). Plotted is the percentage of reads for which at least one alignment was found. Each numbered point is data obtained using command-line parameters shown in **Supplementary Table 1**.

Bowtie2

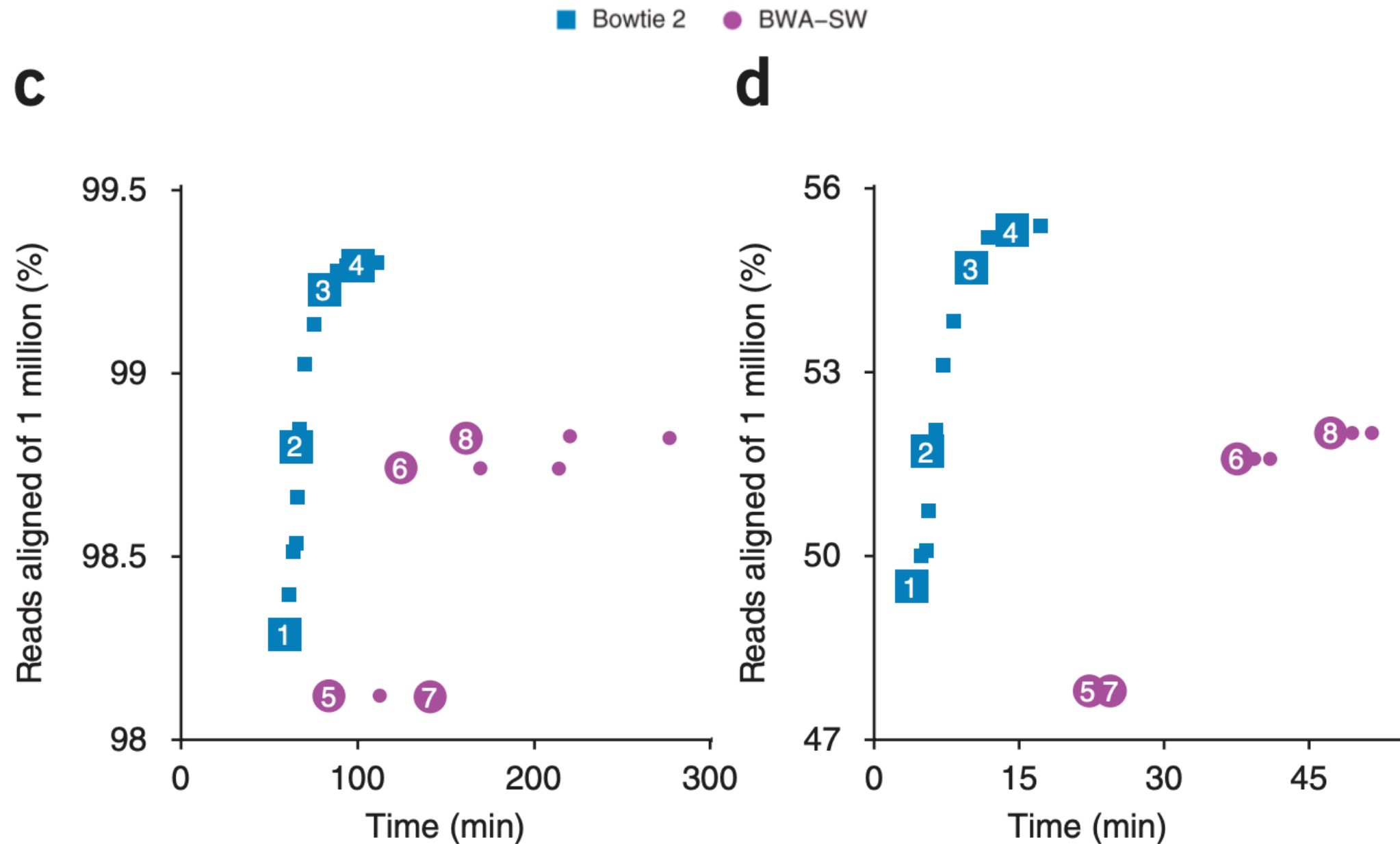
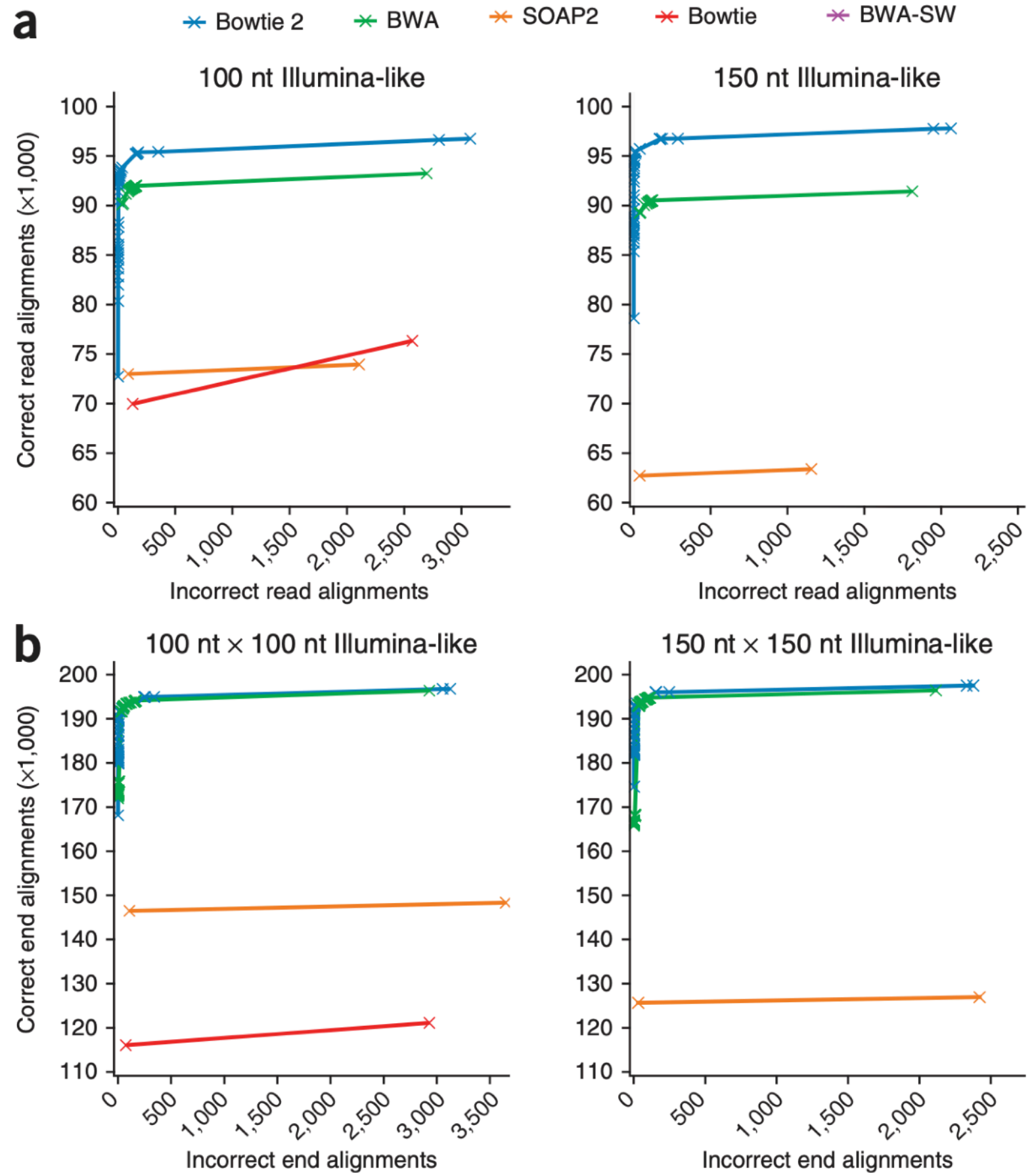


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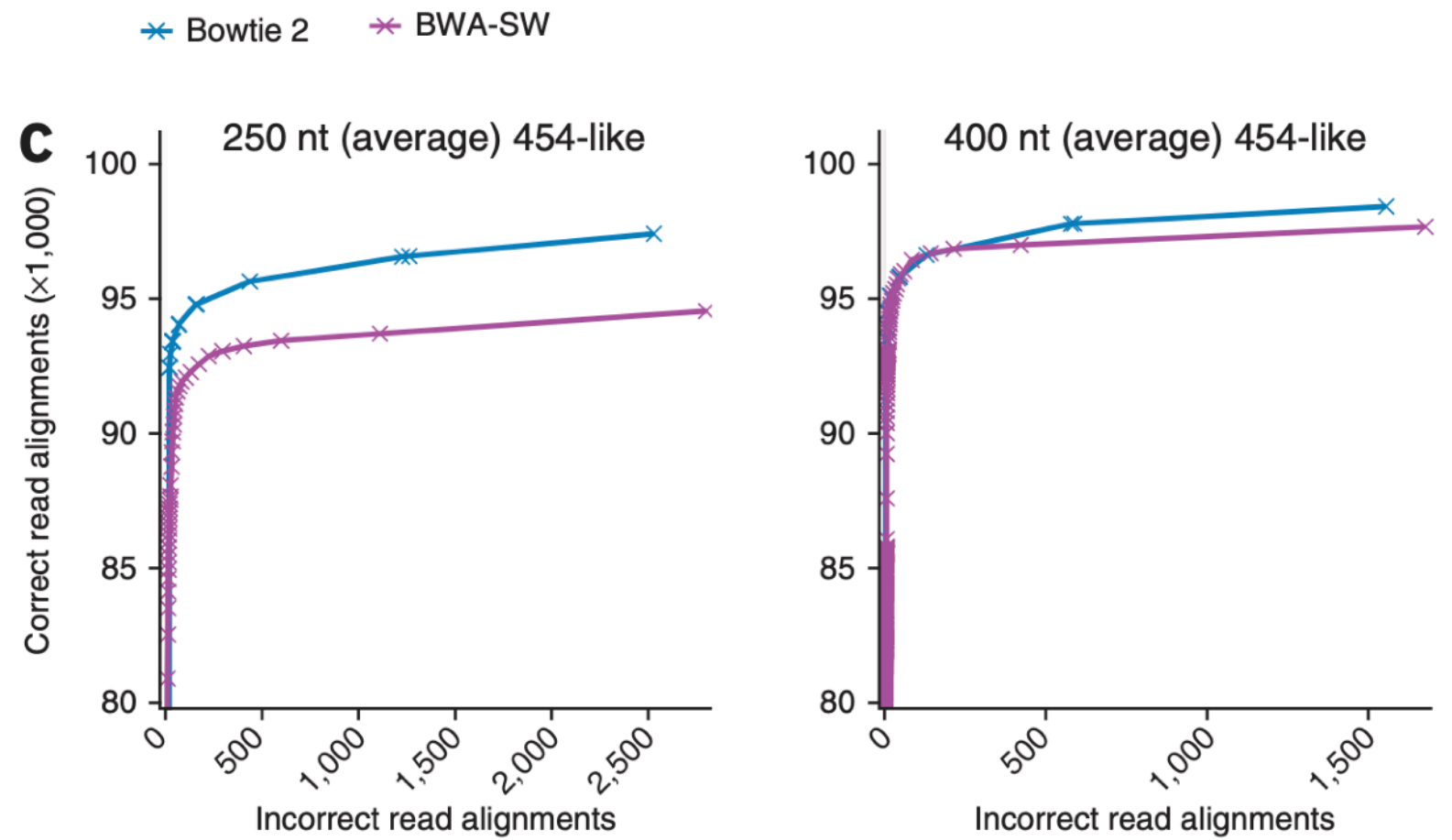
Bowtie2

Results on simulated data



Bowtie2

Results on simulated data



Bowtie2

Dataset	Bowtie 2 versus	Reads or ends aligned by neither	Reads or ends aligned by only Bowtie 2	Reads or ends aligned by only other tool	Reads or ends aligned by both
Unpaired HiSeq 2K	BWA	79,842 (3.99%)	84,136 (4.21%)	449 (0.09%)	1,834,243 (91.71%)
Paired HiSeq 2K	BWA	154,799 (3.87%)	99,852 (2.50%)	9,137 (0.23%)	3,736,212 (93.41%)
454	BWA-SW	7,458 (0.75%)	11,344 (1.13%)	266 (0.03%)	988,390 (98.84%)
Ion Torrent	BWA-SW	450,602 (45.06%)	71,423 (7.14%)	2,270 (0.23%)	475,705 (47.57%)

Bowtie2

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BWA-MEM

Quantitative Biology > Genomics

Aligning sequence reads, clone sequences and assembly contigs with BWA-MEM

Heng Li

(Submitted on 16 Mar 2013 ([v1](#)), last revised 26 May 2013 (this version, v2))

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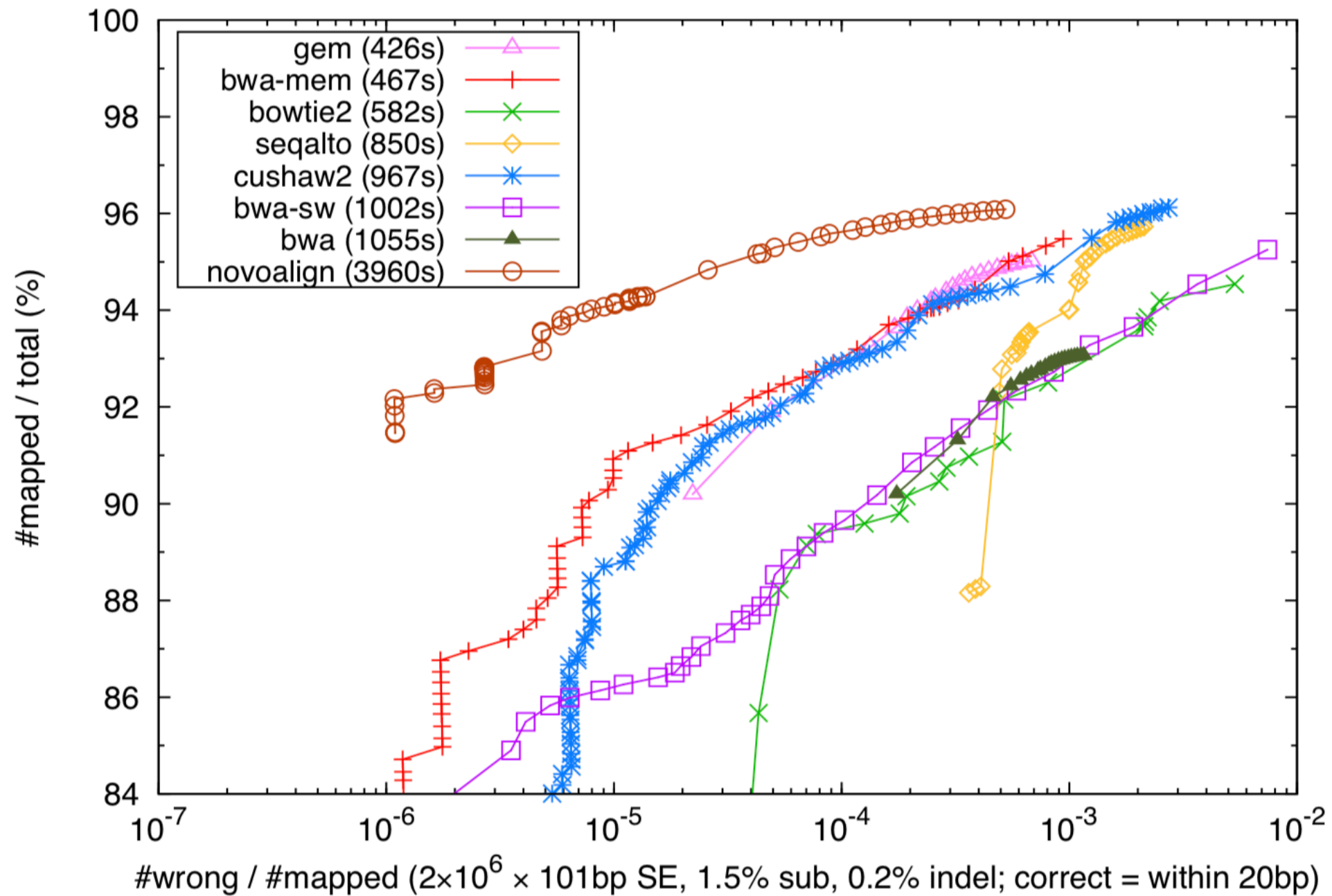
Based on FMD-index for seed finding

Novel “chaining” strategy to find potential alignment loci

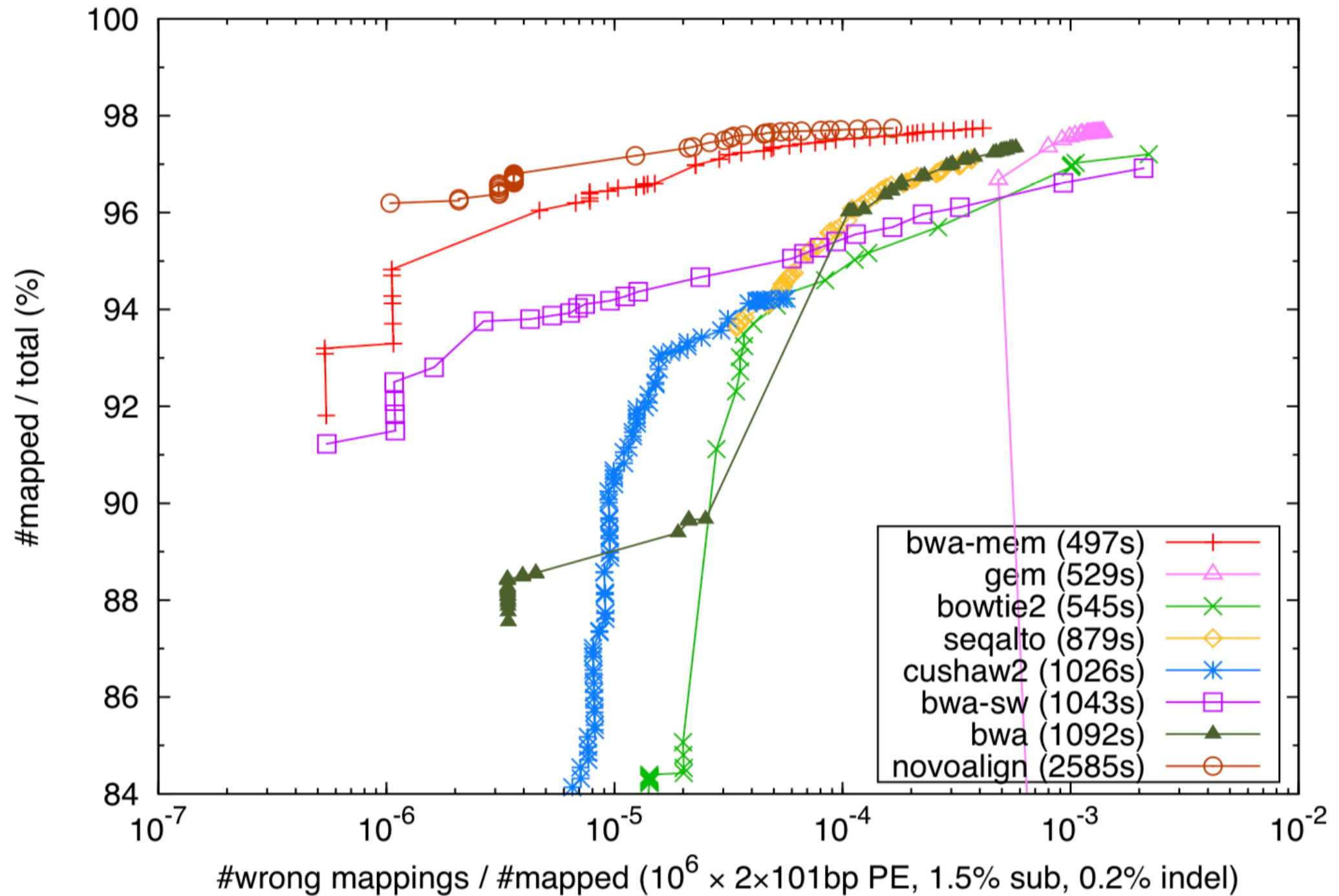
No spliced alignment (i.e. for DNA-seq or RNA-seq -> txome)

Note: The BWA-MEM “paper” is this arXiv pre-print. The manuscript itself was never “published” in a traditional journal. This is a great example of software with *huge* impact that was nonetheless never published.

BWA-MEM



BWA-MEM



STAR

BIOINFORMATICS

ORIGINAL PAPER

Vol. 29 no. 1 2013, pages 15–21
doi:10.1093/bioinformatics/bts635

Sequence analysis

Advance Access publication October 25, 2012

STAR: ultrafast universal RNA-seq aligner

Alexander Dobin^{1,*}, Carrie A. Davis¹, Felix Schlesinger¹, Jorg Drenkow¹, Chris Zaleski¹, Sonali Jha¹, Philippe Batut¹, Mark Chaisson² and Thomas R. Gingeras¹

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Associate Editor: Inanc Birol

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Based on suffix array + prefix-table for seed finding

Custom “chaining” & between match alignment strategy

Capable of both contiguous and spliced alignment,
behavior is *highly* configurable via parameters (DNA-seq or
RNA-seq alignment directly to the genome)

STAR

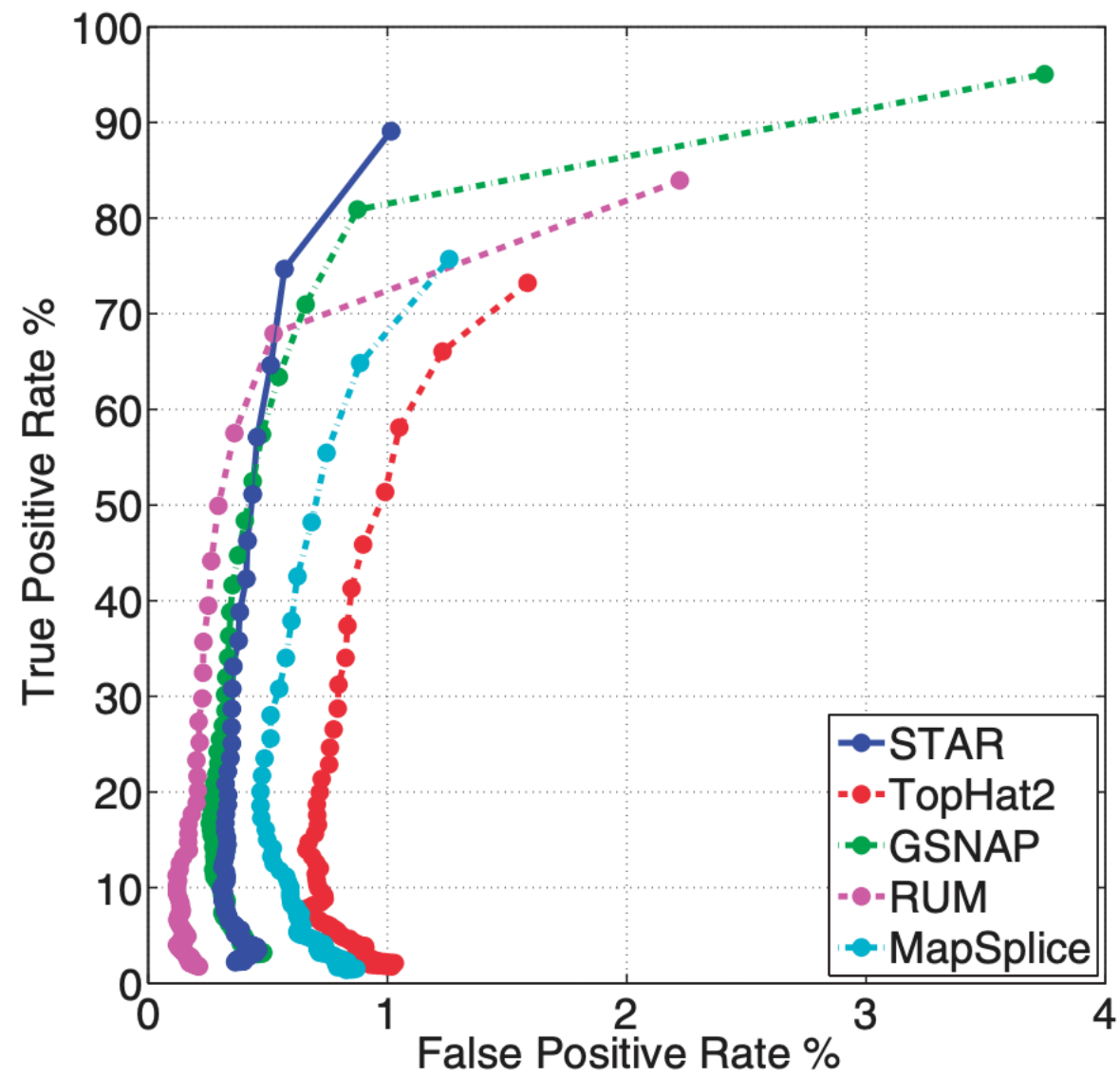
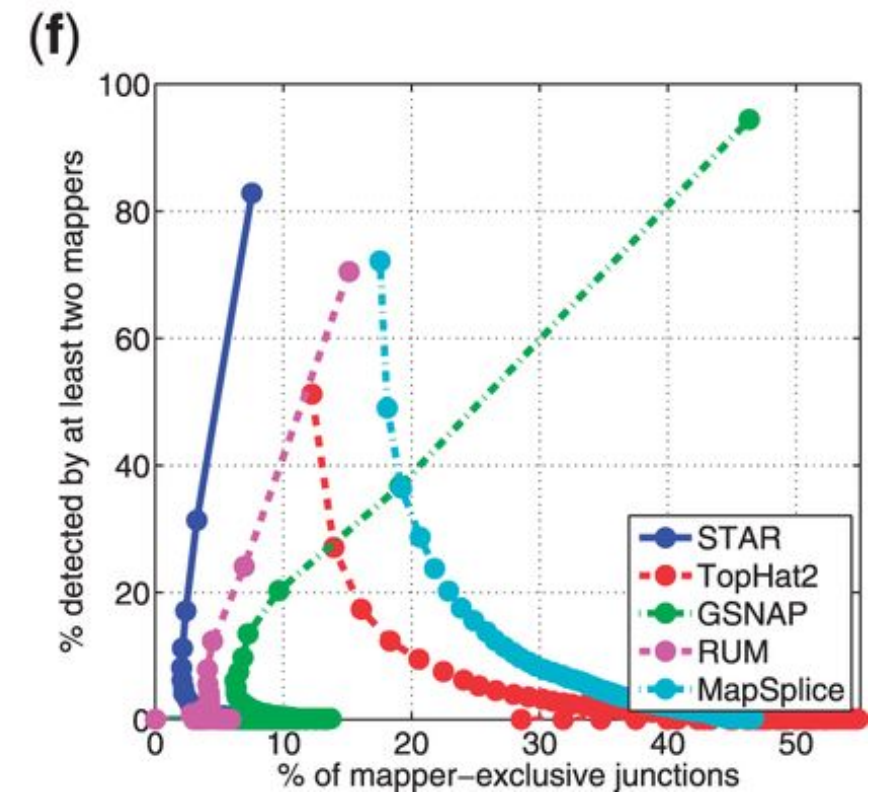
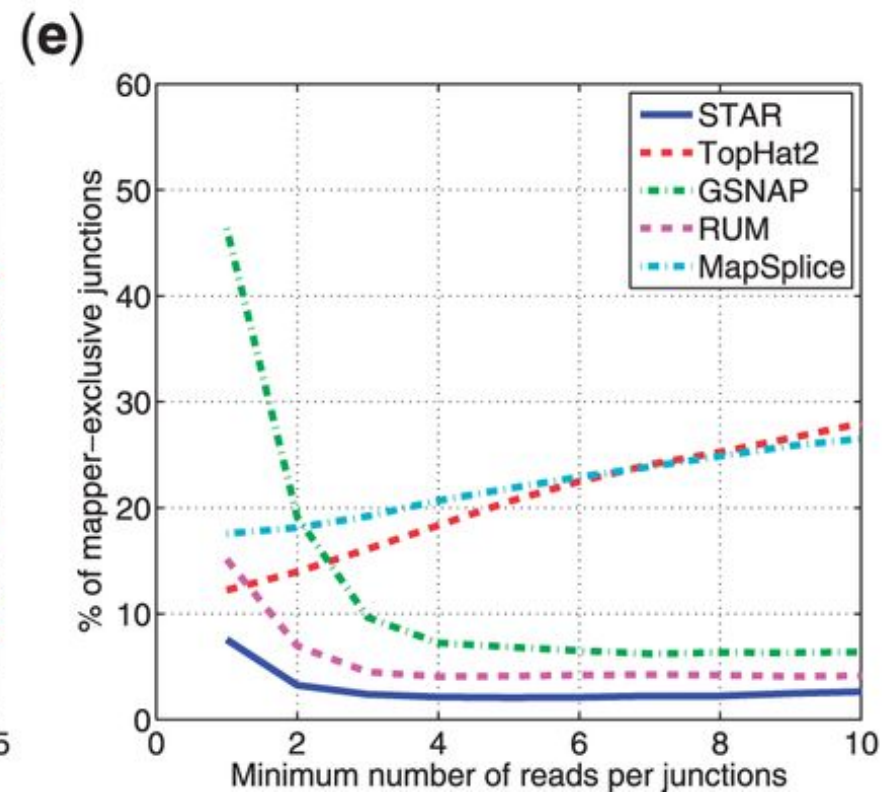
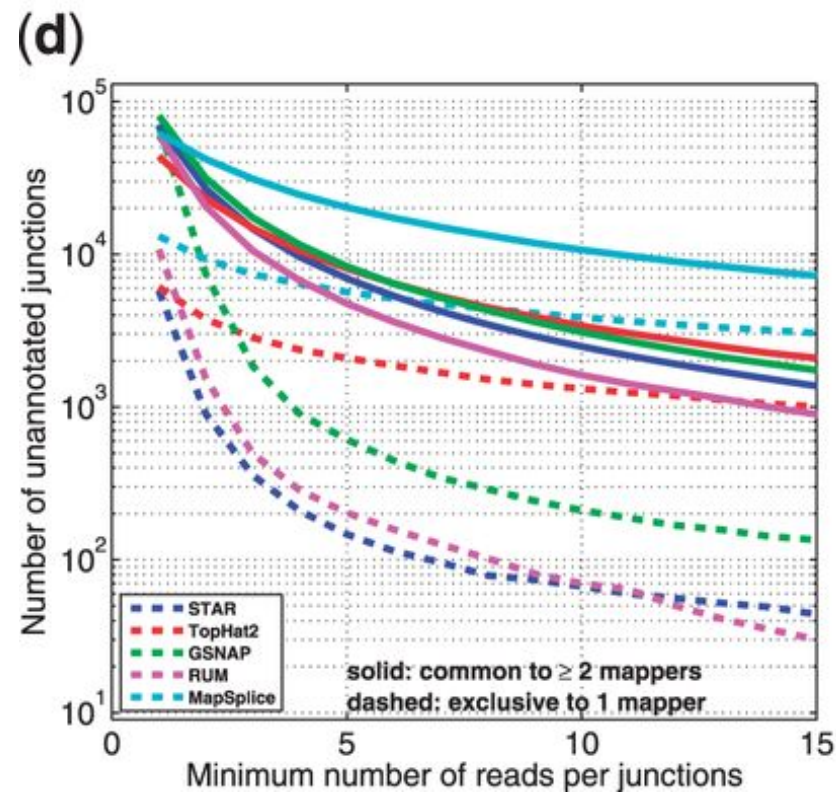
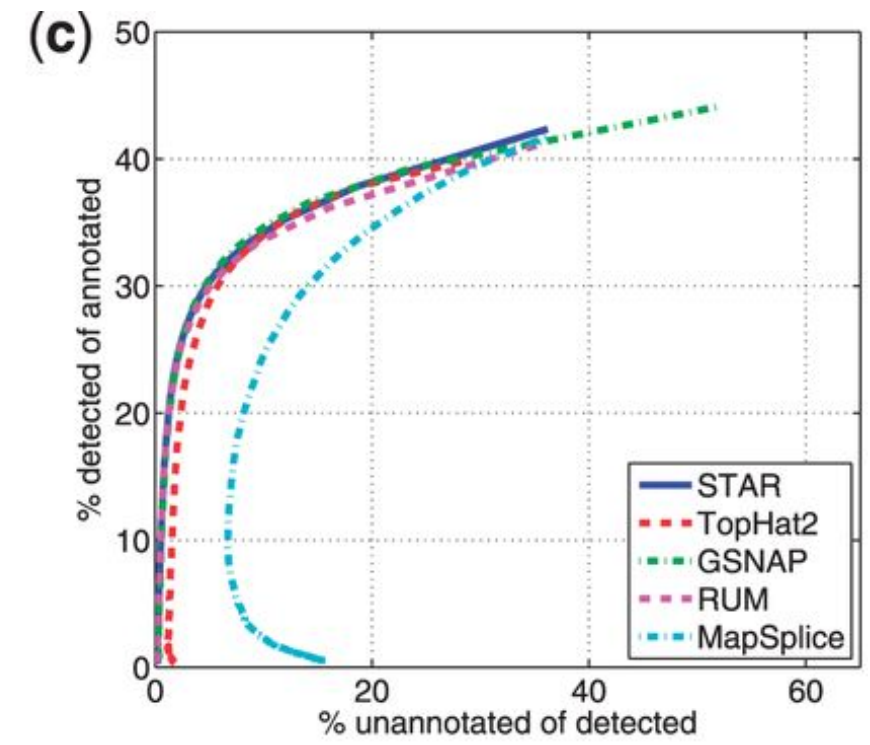
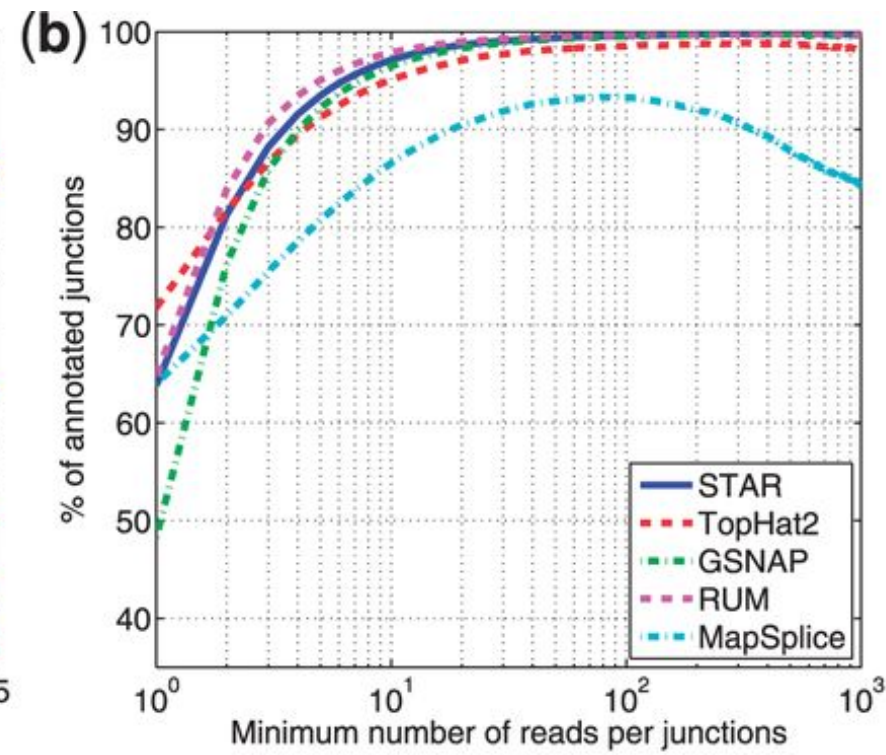
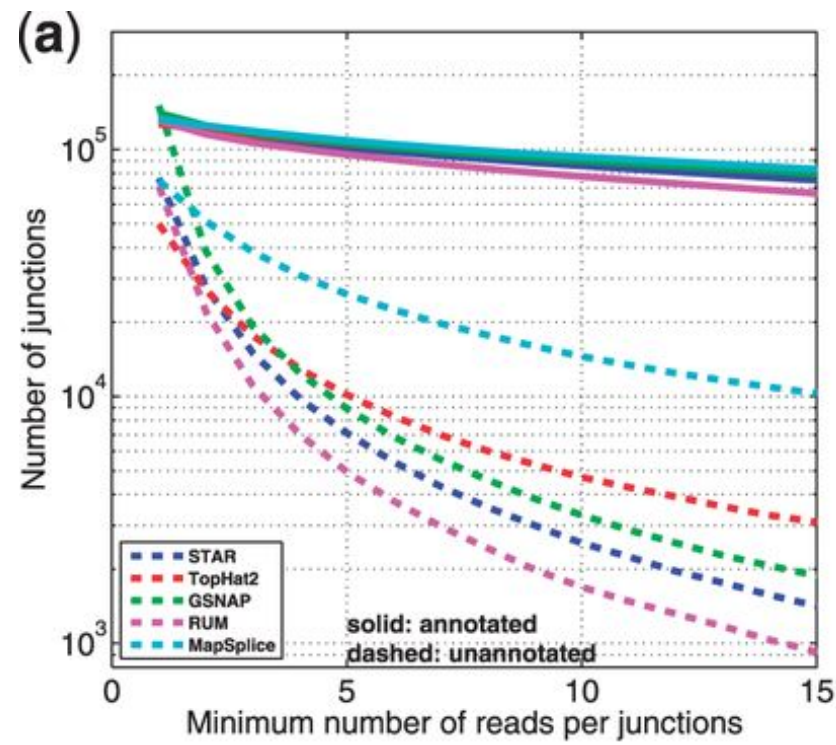


Fig. 2. True-positive rate versus false-positive rate (ROC-curve) for simulated RNA-seq data for STAR, TopHat2, GSNAP, RUM and MapSplice

STAR



STAR

Table 1. Mapping speed and RAM benchmarks on the experimental RNA-seq dataset

Aligner	Mapping speed: million read pairs/hour		Peak physical RAM, GB	
	6 threads	12 threads	6 threads	12 threads
STAR	309.2	549.9	27.0	28.4
STAR sparse	227.6	423.1	15.6	16.0
TopHat2	8.0	10.1	4.1	11.3
RUM	5.1	7.6	26.9	53.8
MapSplice	3.0	3.1	3.3	3.3
GSNAP	1.8	2.8	25.9	27.0

HISAT2

Article | Published: 02 August 2019

Graph-based genome alignment and genotyping with HISAT2 and HISAT-genotype

Daehwan Kim , Joseph M. Paggi, Chanhee Park, Christopher Bennett & Steven L. Salzberg

Based on hierarchical graph FM-index for alignment

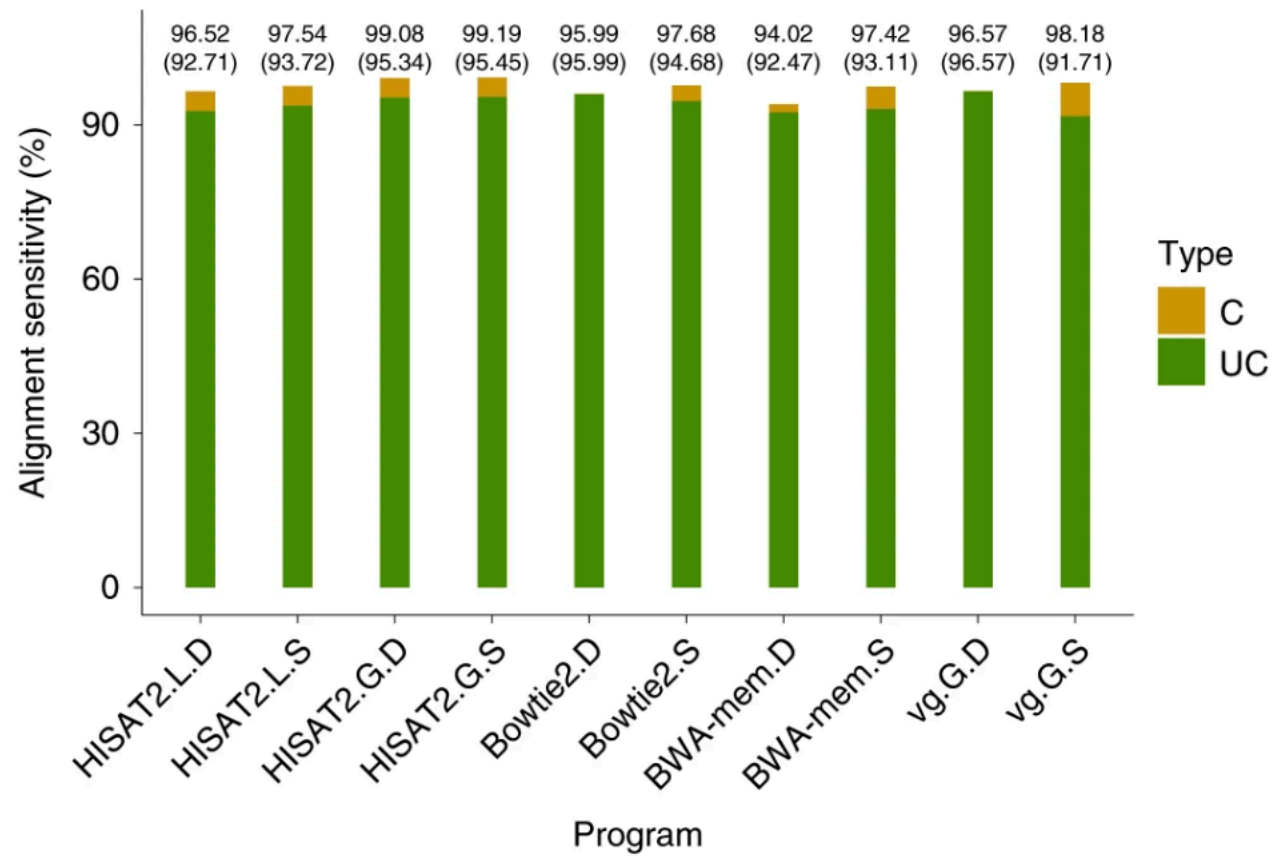
Custom strategy to deal with highly-repetitive regions

Capable of both contiguous and spliced alignment,
behavior is *highly* configurable via parameters (built for both
DNA-seq and RNA-seq alignment directly to the genome)

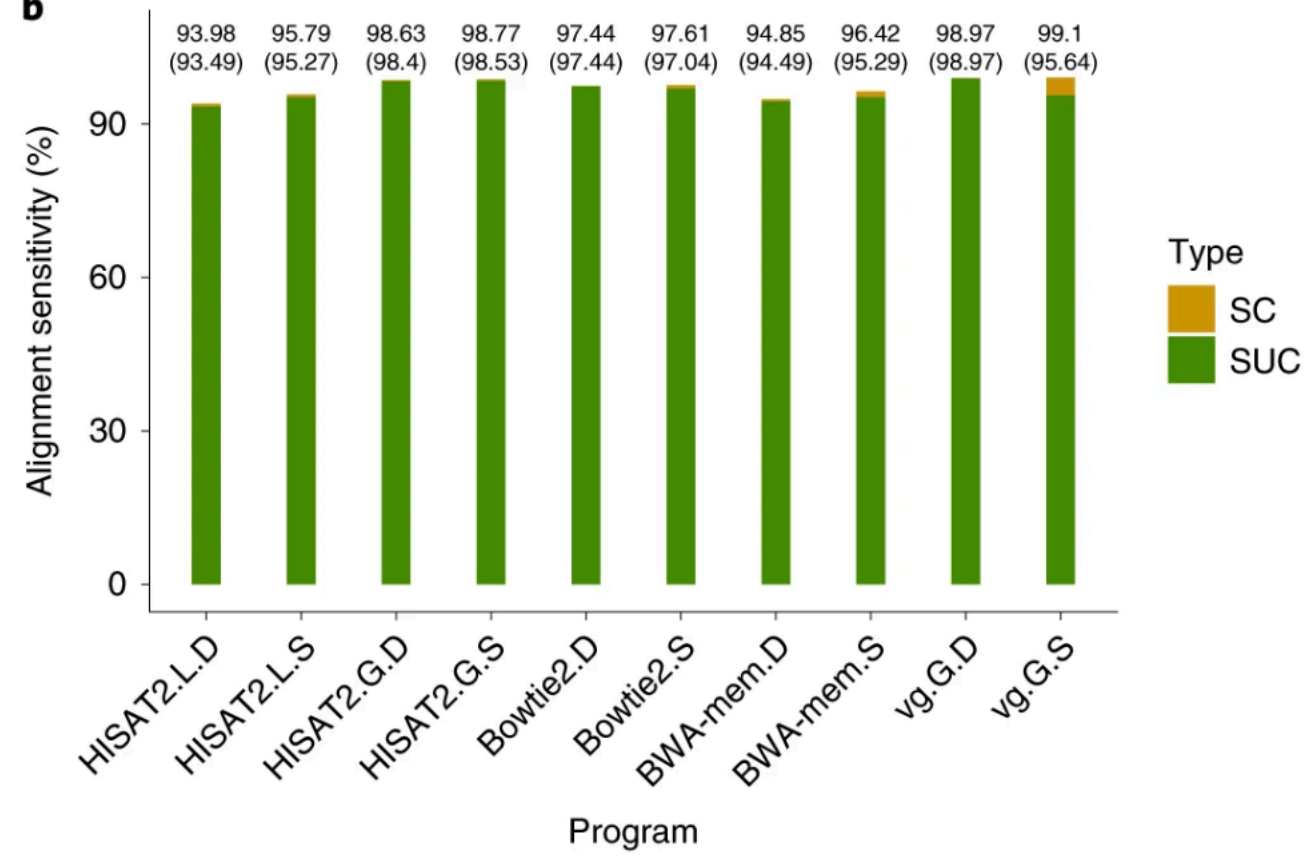
Built-in algorithm to do HLA-typing over aligned reads

HISAT2

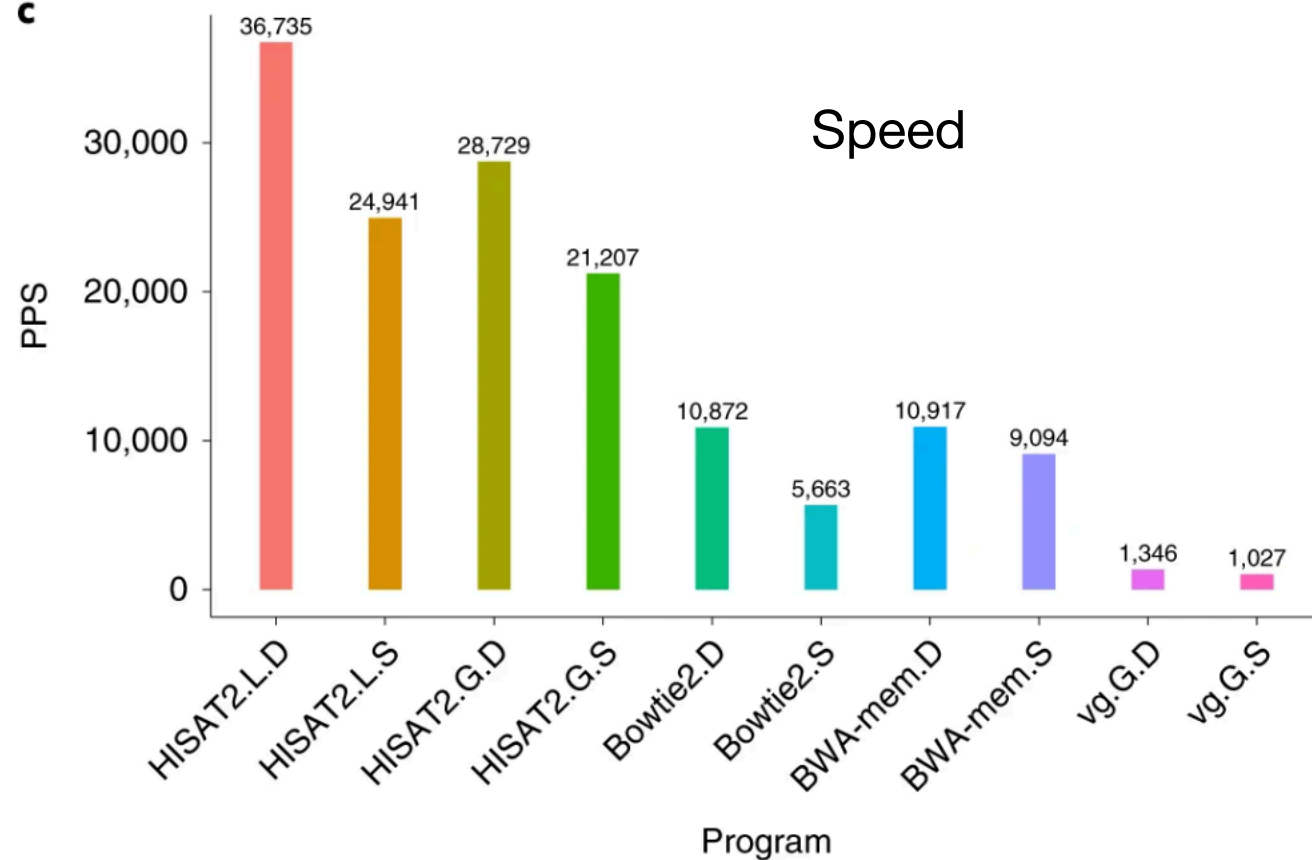
a



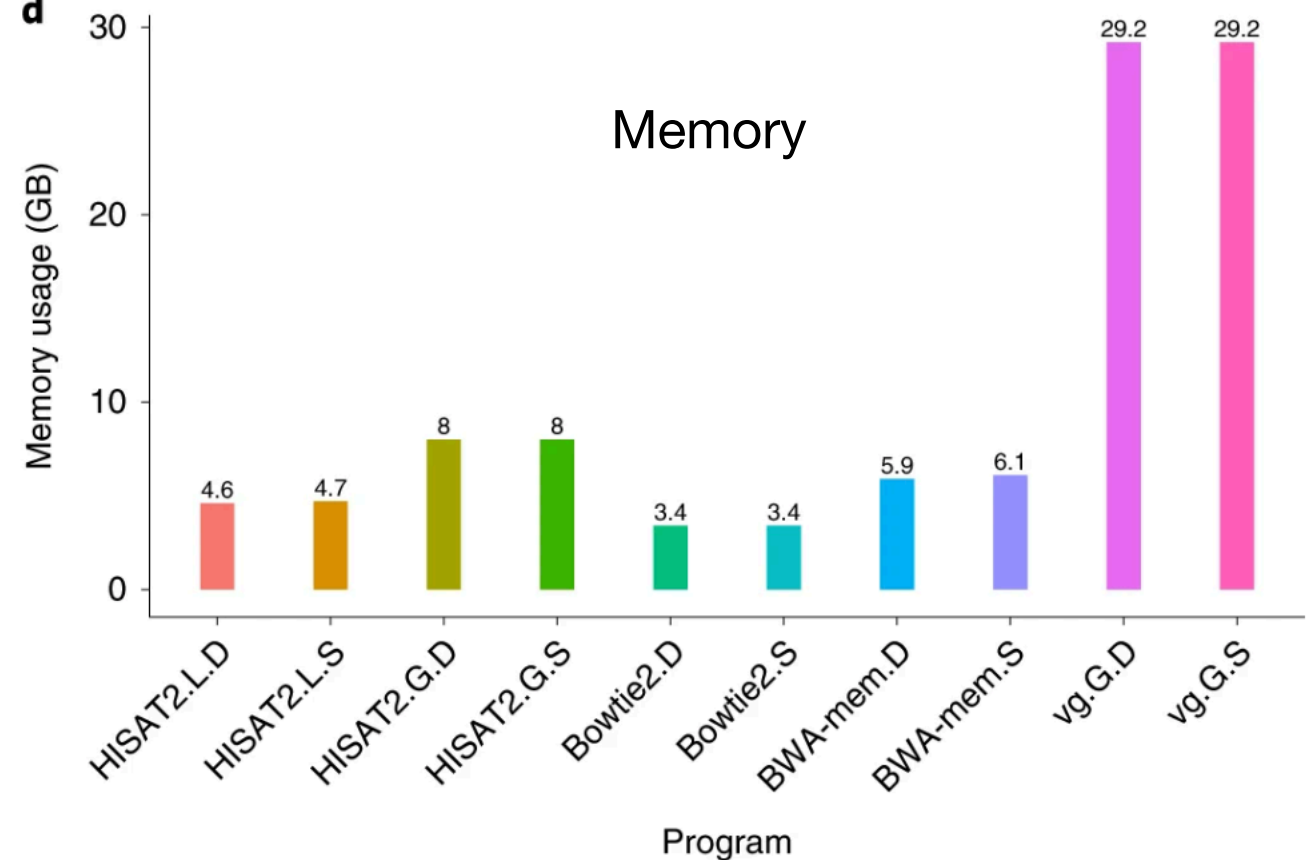
b



c



d



Graph alignment improves sensitivity

	10 million read pairs with SNPs and 0.2% per base sequencing error					10 million read pairs with SNPs and no sequencing error				
	C	UC	SC	SUC	PPS	C	UC	SC	SUC	PPS
HISAT2.Linear (default)	96.52%	92.71%	93.98%	93.49%	36,735	97.05%	93.15%	94.65%	94.15%	37,934
HISAT2.Linear (sensitive)	97.54%	93.72%	95.79%	95.27%	24,941	97.83%	93.92%	96.07%	95.55%	27,331
HISAT2.Graph (default)	99.08%	95.34%	98.63%	98.40%	28,729	99.36%	95.54%	98.84%	98.62%	32,096
HISAT2.Graph (sensitive)	99.19%	95.45%	98.77%	98.53%	21,207	99.36%	95.54%	98.84%	98.61%	25,639
Bowtie2 (default)	95.99%	95.99%	97.44%	97.44%	10,872	96.05%	96.05%	97.50%	97.50%	10,575
Bowtie2 (sensitive)	97.68%	94.68%	97.61%	97.04%	5,663	97.85%	94.77%	97.63%	97.07%	5,597
BWA-mem (default)	94.02%	92.47%	94.85%	94.49%	10,917	94.03%	92.49%	94.83%	94.47%	12,110
BWA-mem (sensitive)	97.42%	93.11%	96.42%	95.29%	9,094	97.57%	93.15%	96.40%	95.28%	10,106
VG.Linear (default)	95.56%	95.56%	96.91%	96.91%	1,315	95.34%	95.34%	96.65%	96.65%	1,367
VG.Linear (sensitive)	97.31%	89.74%	97.27%	92.27%	1,012	97.18%	90.31%	97.14%	92.71%	1,028
VG.Graph (default)	96.57%	96.57%	98.97%	98.97%	1,346	96.64%	96.64%	99.02%	99.02%	1,413
VG.Graph (sensitive)	98.18%	91.71%	99.10%	95.64%	1,027	98.37%	91.51%	99.16%	95.40%	1,083