

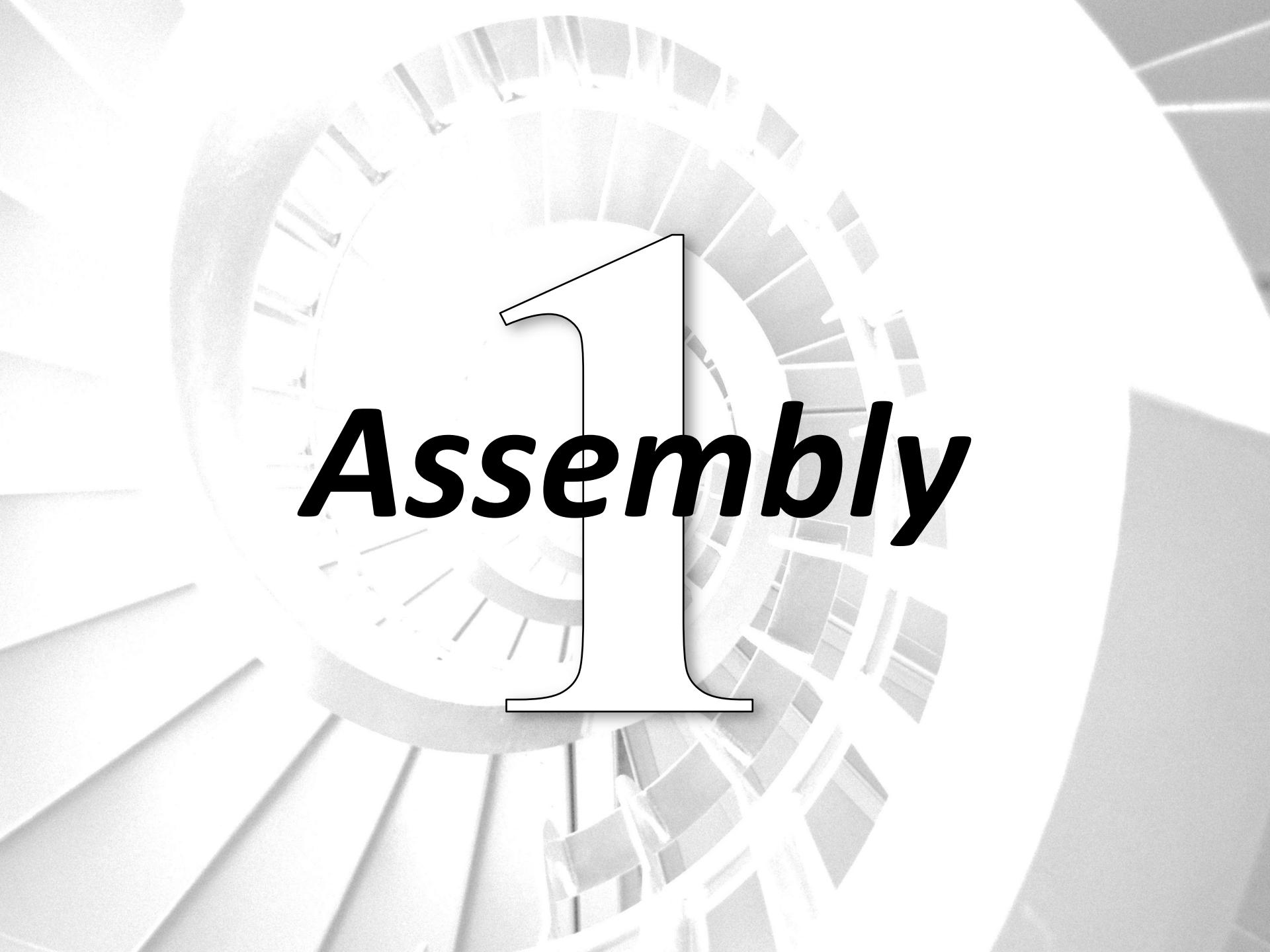
5-step, scalable solutions for *de novo* genome assembly

René L Warren 03/2019



CANADA'S MICHAEL SMITH
GENOME
SCIENCES
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b1



Assembly



2009 : Parallel DBG assembler

MPI to aggregate memory

Assembled 20Gb spruce genome

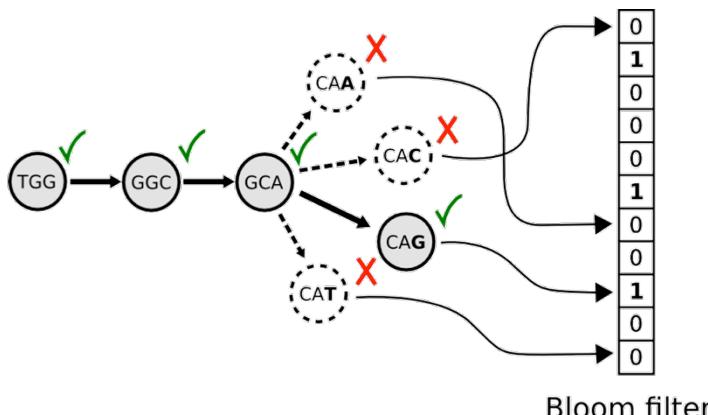
GENOME
RESEARCH
Resource

ABySS: A parallel assembler for short read sequence data

Jared T. Simpson,¹ Kim Wong, Shaun D. Jackman, Jacqueline E. Schein, Steven J.M. Jones, and İnanç Birol²

2017 : Bloom filter representation

1/10th RAM, single computer, scalable to 20 Gbp spruce



GENOME
RESEARCH
Method

ABySS 2.0: resource-efficient assembly of large genomes using a Bloom filter

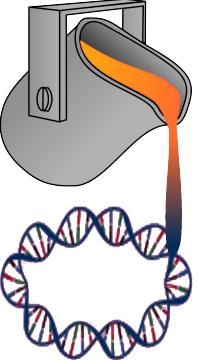
Shaun D. Jackman,¹ Benjamin P. Vandervalk,¹ Hamid Mohamadi, Justin Chu, Sarah Yeo, S. Austin Hammond, Golnaz Jahesh, Hamza Khan, Lauren Coombe, Rene L. Warren, and Inanc Birol

<https://github.com/bcgsc/abyss>



Correction

Tigmint



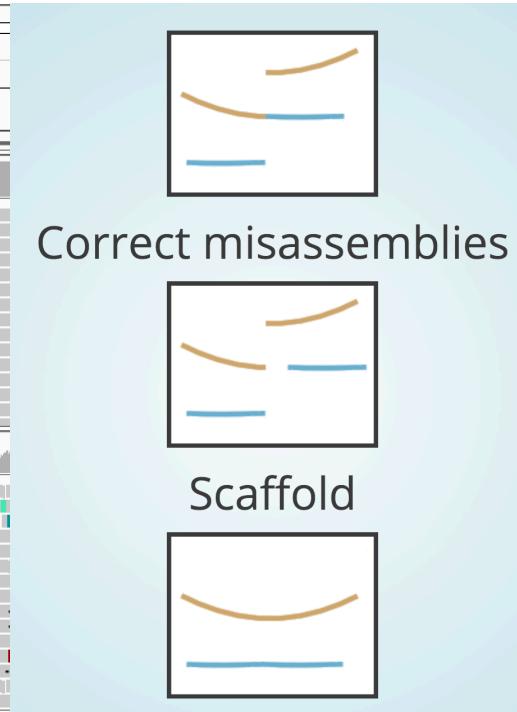
linked reads misassembly correction



SOFTWARE

Tigmint: correcting assembly errors using linked reads from large molecules

Shaun D. Jackman^{1*} , Lauren Coombe¹, Justin Chu¹, Rene L. Warren¹, Benjamin P. Vandervalk¹, Sarah Yeo¹, Zhuyi Xue¹, Hamid Mohamadi¹, Joerg Bohlmann², Steven J.M. Jones¹ and Inanc Birol¹

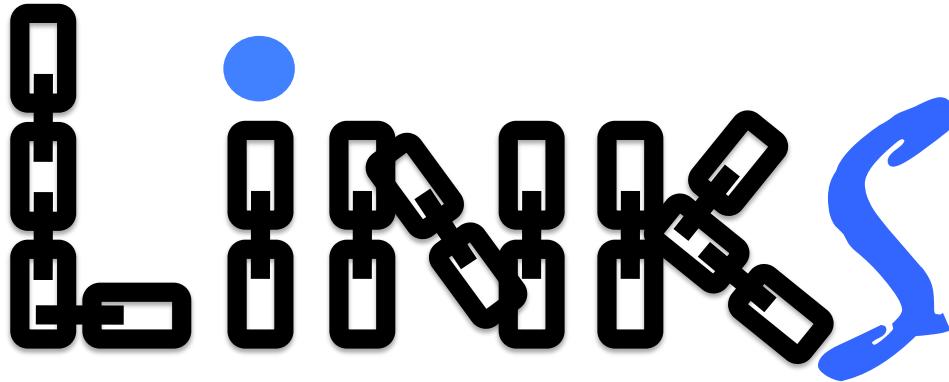


IGV screenshot: Tigmint breakpoint in human genome NA24143

<https://github.com/bcgsc/tigmint>



Scaffolding



Warren et al. *GigaScience* (2015) 4:35
DOI 10.1186/s13742-015-0076-3

(GIGA)ⁿ
SCIENCE

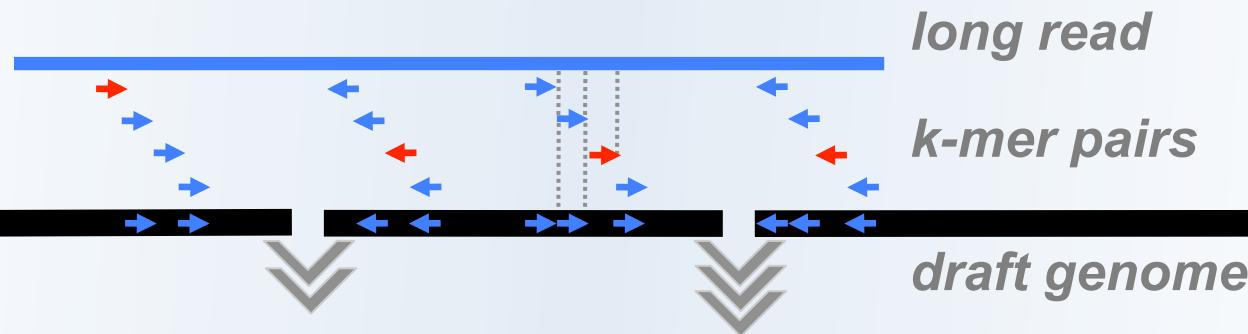
RESEARCH Open Access



LINKS: Scalable, alignment-free scaffolding of draft genomes with long reads

René L. Warren*, Chen Yang, Benjamin P. Vandervalk, Bahar Behsaz, Albert Lagman, Steven J. M. Jones and Inanç Birol

Long read kmer scaffolding



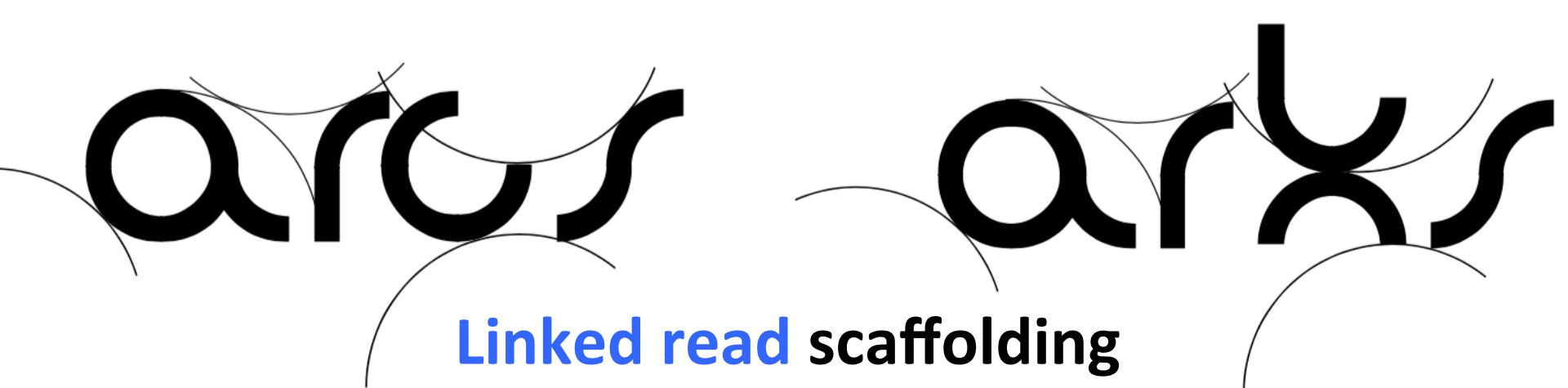
- **Scaffolder** : order & orient sequences
- ***k*-mer based** : no alignments
- **Vast *k*-mer space** : no fragment length limitation
- **Versatile** : long-reads, draft sequences, MPET

length

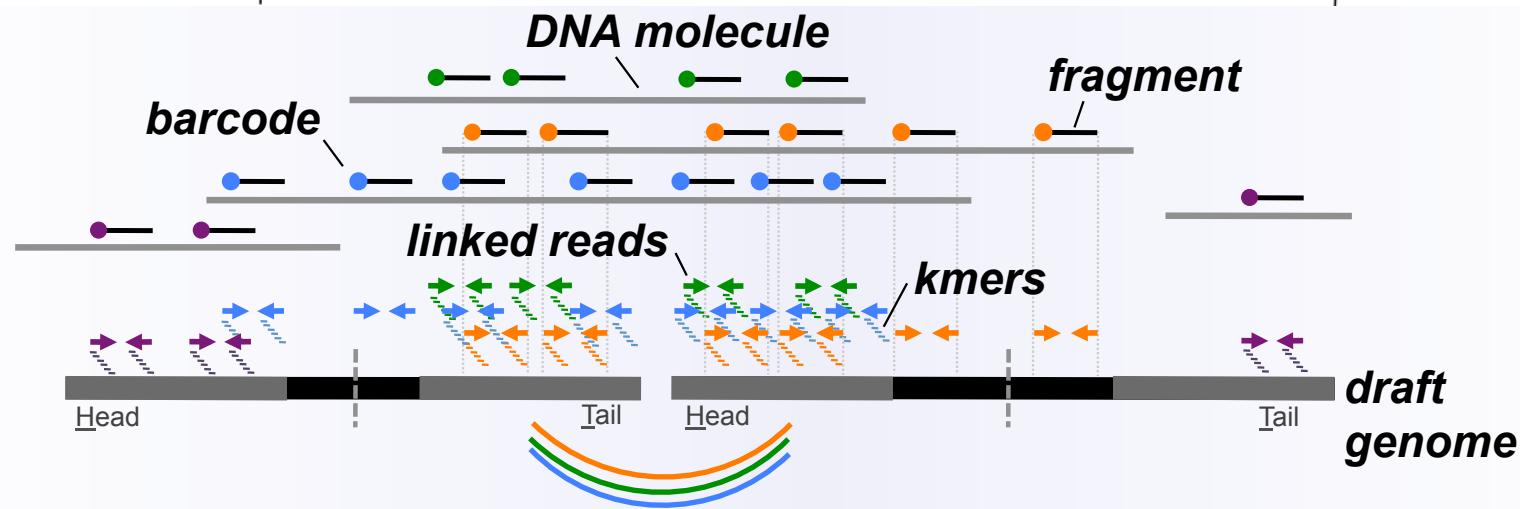
errors

∅ base correction

<https://github.com/bcgsc/links>



Linked read scaffolding



ARCS: scaffolding genome drafts with linked reads



Sarah Yeo, Lauren Coombe, René L Warren ✉, Justin Chu, Inanç Birol Author Notes

Bioinformatics, Volume 34, Issue 5, 1 March 2018, Pages 725–731,

<https://doi.org/10.1093/bioinformatics/btx675>

Coombe et al. BMC Bioinformatics (2018) 19:234
<https://doi.org/10.1186/s12859-018-2243-x>

BMC Bioinformatics

SOFTWARE

Open Access

ARKS: chromosome-scale scaffolding of human genome drafts with linked read kmers

Lauren Coombe[†], Jessica Zhang[†], Benjamin P. Vandervalk, Justin Chu, Shaun D. Jackman, Inanc Birol and René L. Warren^{*}



<https://github.com/bcgsc/arcs>

<https://github.com/bcgsc/arks>

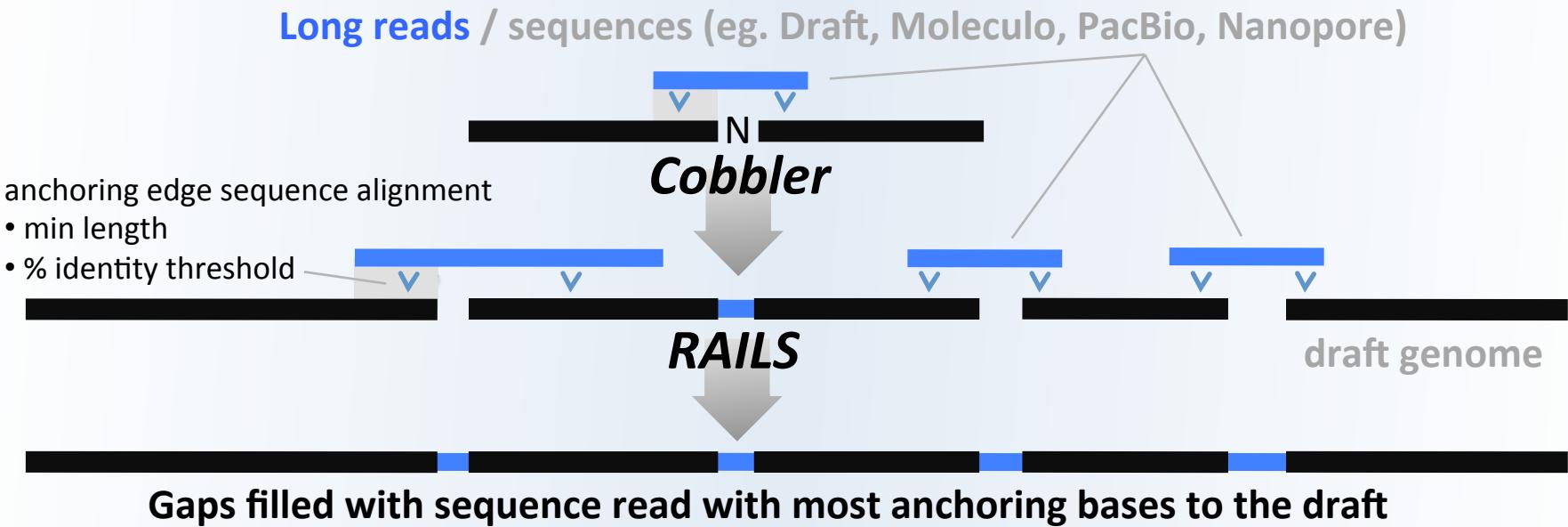
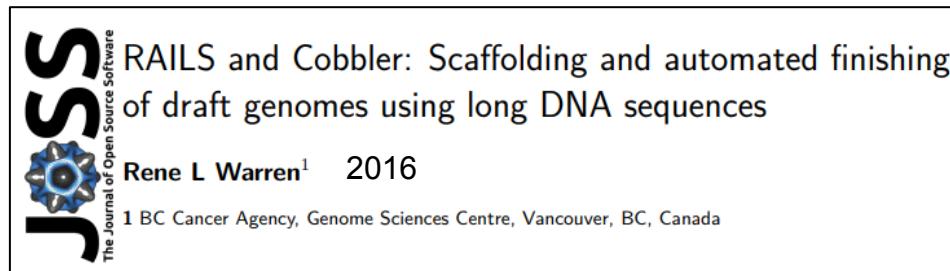


Gap-filling



Scaffolding and gap-filling

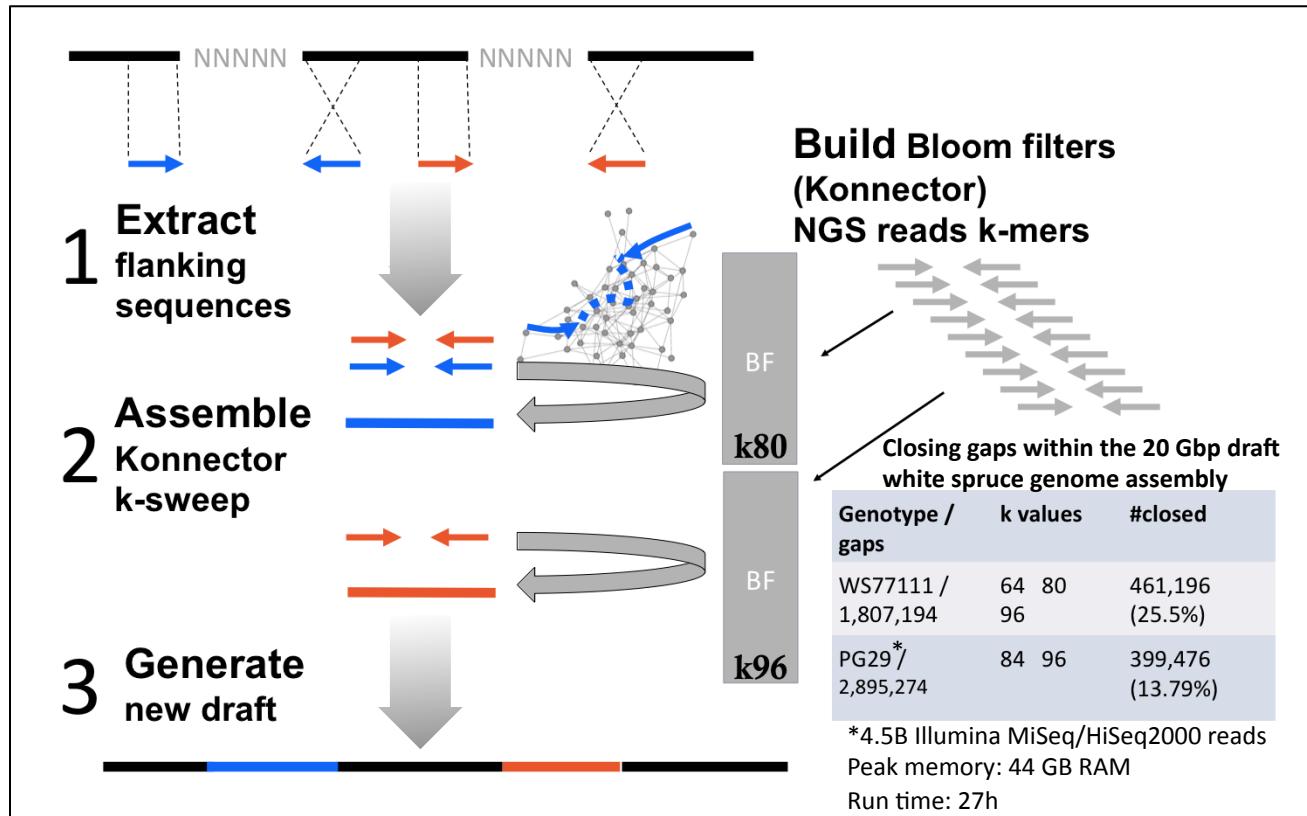
Uses LINKS scaffolding algorithm



Sealer

Automated genome finishing

- Gap-filler (resolve Ns)
- Implements Bloom filter de Bruijn graph



Vandervalk et al. BMC Medical Genomics 2015, 8(Suppl 3):S1
http://www.biomedcentral.com/1755-8794/8/S3/S1

BMC
Medical Genomics

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Konnector v2.0: pseudo-long reads from paired-end sequencing data

Paulino et al. BMC Bioinformatics (2015) 16:230
DOI 10.1186/s12859-015-0663-4

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Sealer: a scalable gap-closing application for finishing draft genomes

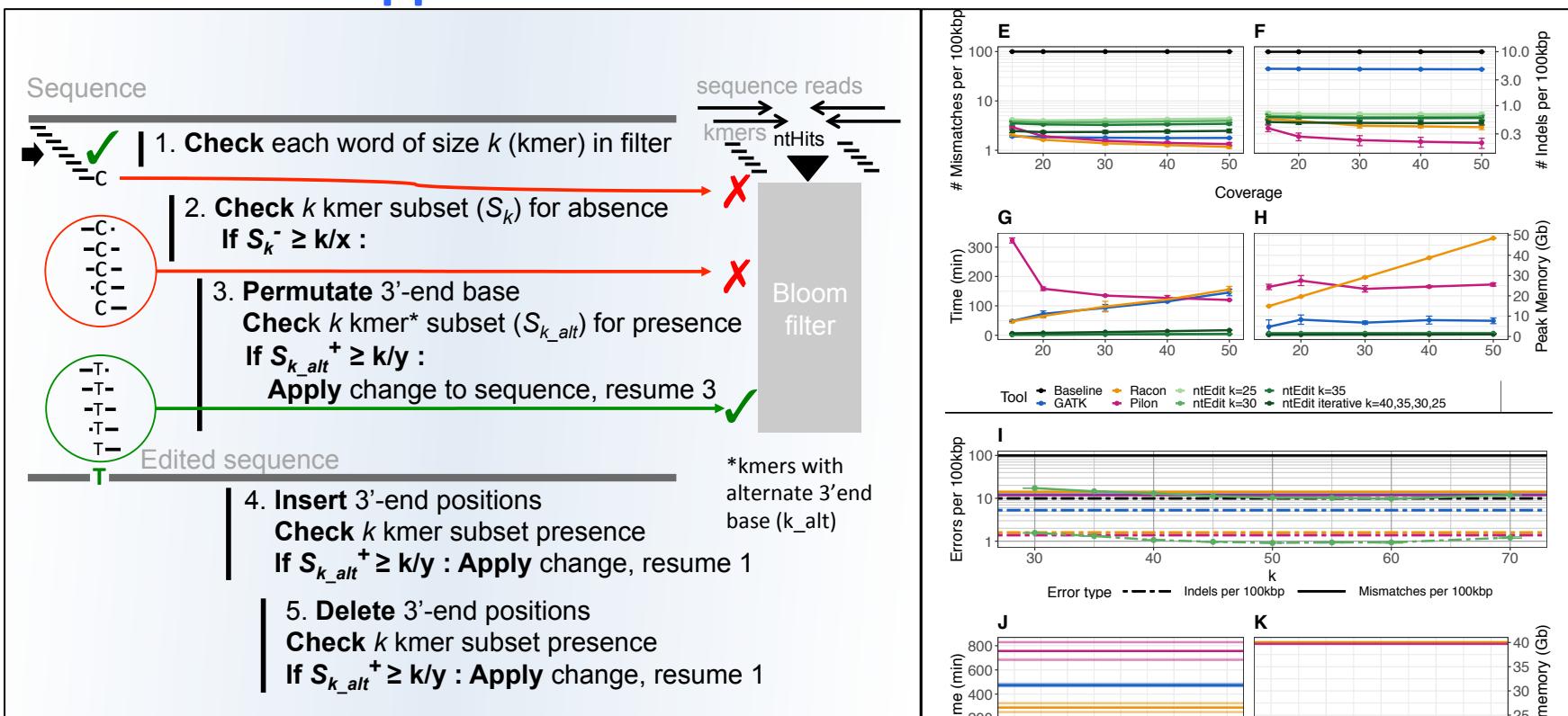




Polishing

ntEdit

Fast homozygous error correction / genome “haploidization” Approach Results



ntEdit: scalable genome sequence polishing
2019



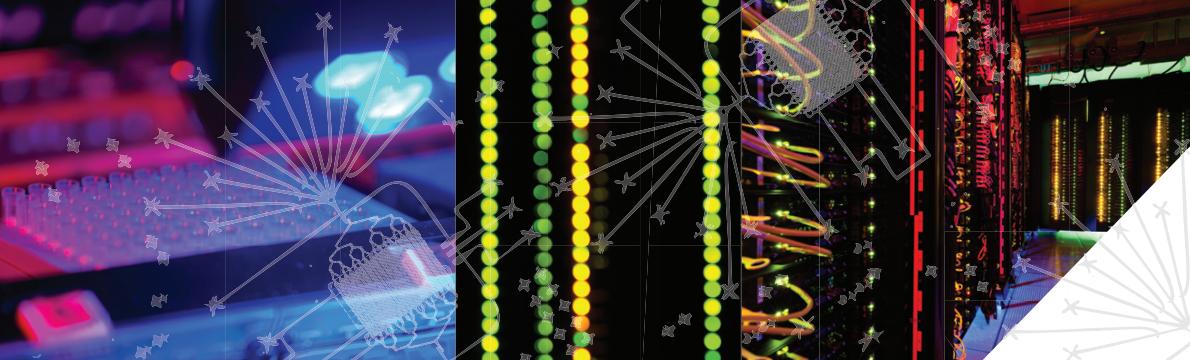
bioRxiv
THE PREPRINT SERVER FOR BIOLOGY

René L Warren, Lauren Coombe, Hamid Mohamadi, Jessica Zhang, Barry Jaquish, Nathalie Isabel, Steven JM Jones, Jean Bousquet, Joerg Bohlmann, Inanç Birol

doi: <https://doi.org/10.1101/565374>

<https://github.com/bcgsc/ntedit>

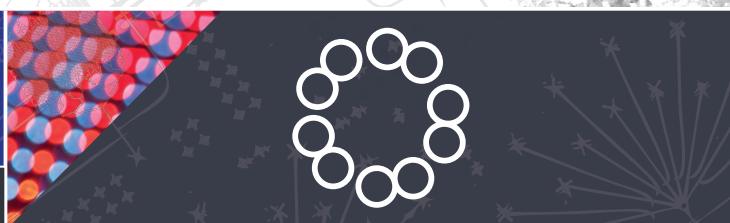
C. elegans
H. Sapiens chr21



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AFFILIATIONS BC Cancer Research Center • BC Cancer Agency • BC Cancer Foundation • Genome BC • Simon Fraser University • University of British Columbia • Genome Sciences Institute



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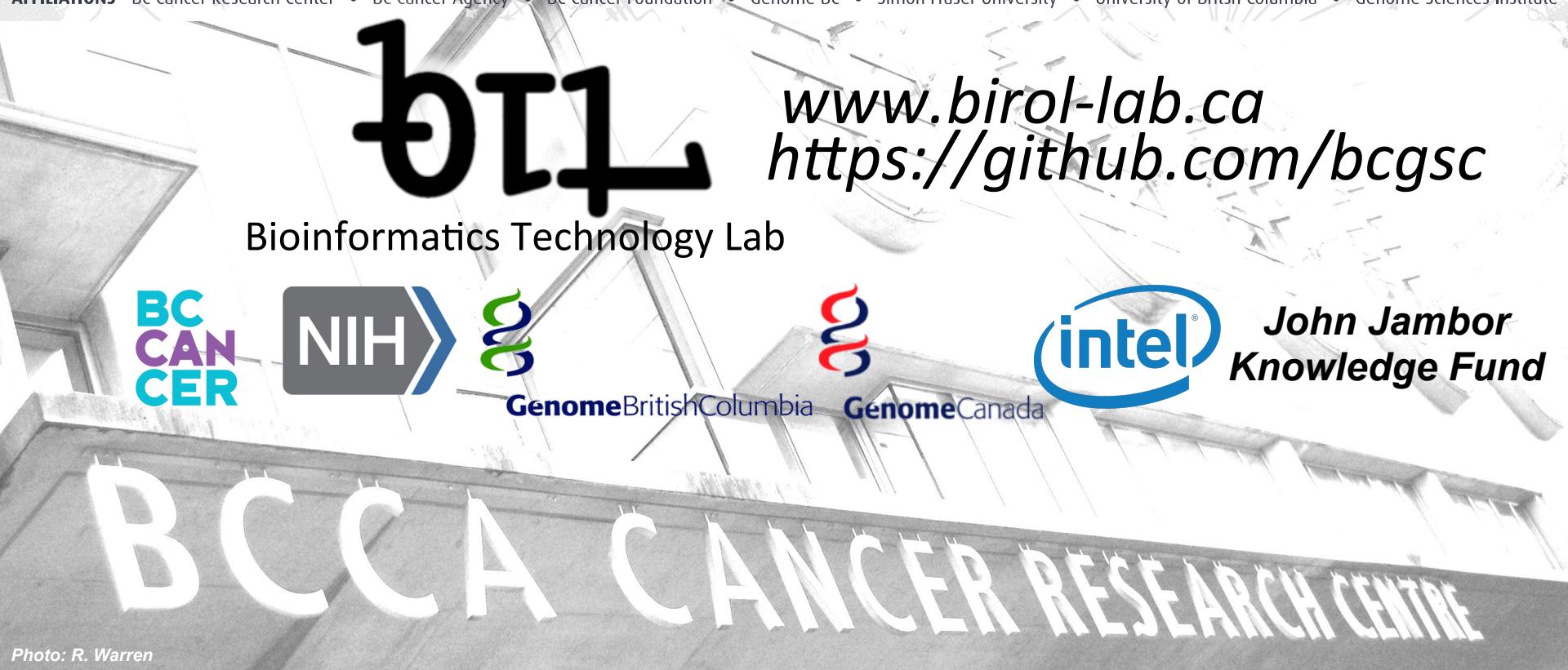
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