GOETHE UNIVERSITÄT FRANKFURT AM MAIN

Assembly of long, error-prone reads using repeat graphs

Mikhail Kolmogorov, Jeffrey Yuan, Yu Lin, and Pavel A. Pevzner

July 1, 2021

Johannes Hausmann, Luis Kress

Background

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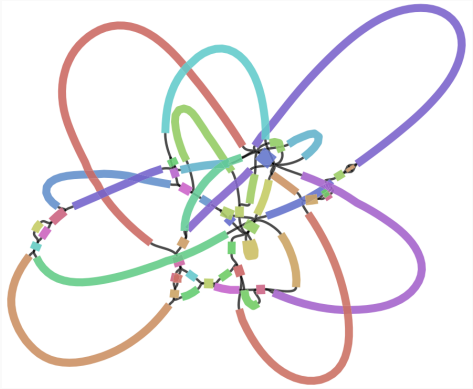

Figure 1: Tangled assembly graph[1]

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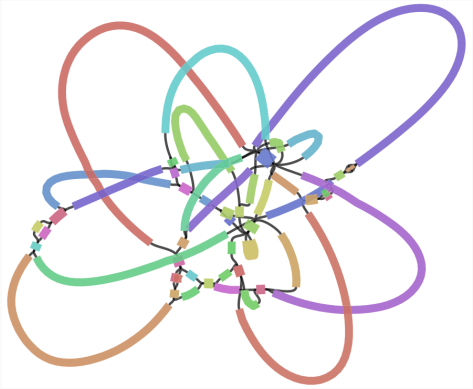

Figure 1: Tangled assembly graph[1]

- Small differences between repeat copies $\rightarrow$ hard to resolve with error-prone reads

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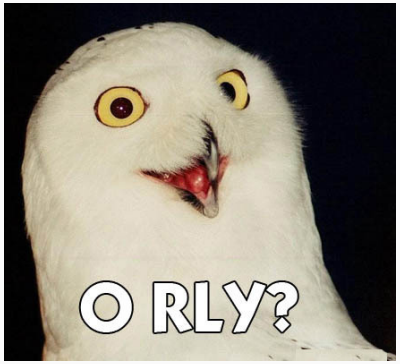
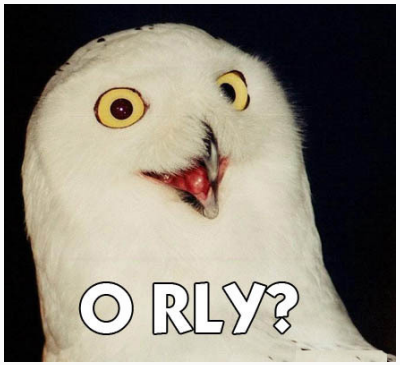


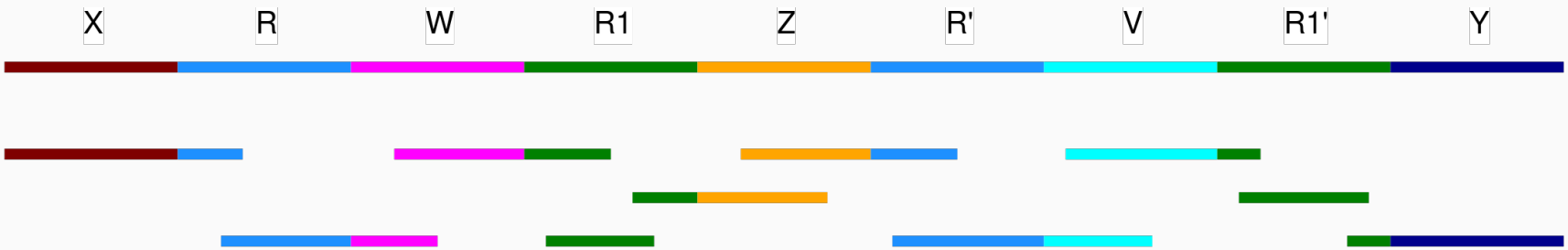
Disjointigs

- Most assemblers spend much time on correct contig assembly
- Flye uses a different approach [1]:
  - we don't care (at least at the initial stage)



- Generate paths from overlapping reads without checking for correct repeat resolution $\rightarrow$ Disjointigs

Repeat Graph Creation



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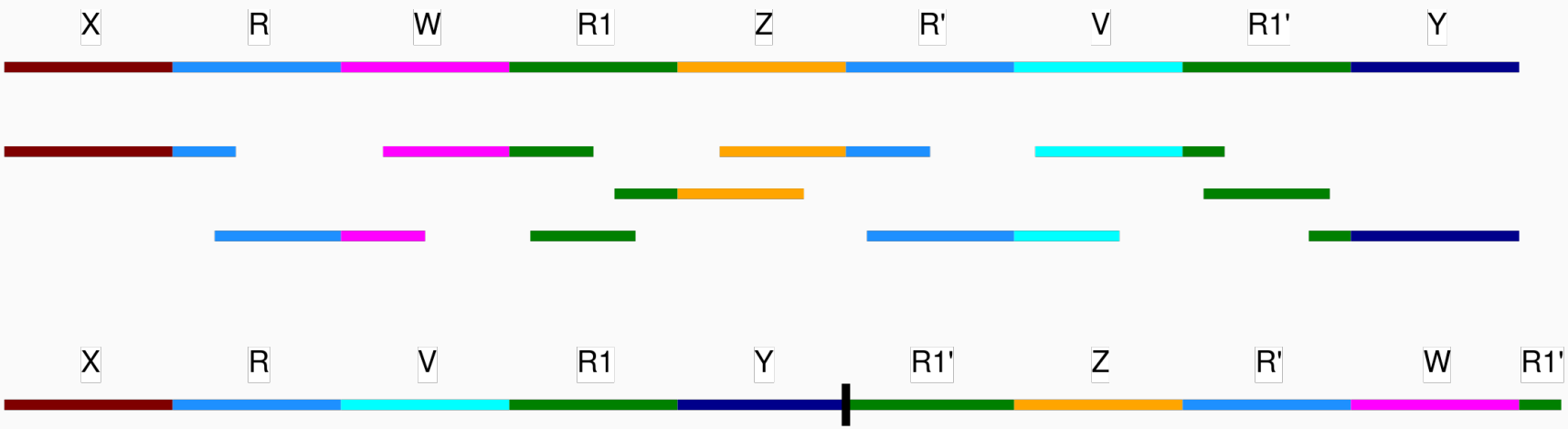

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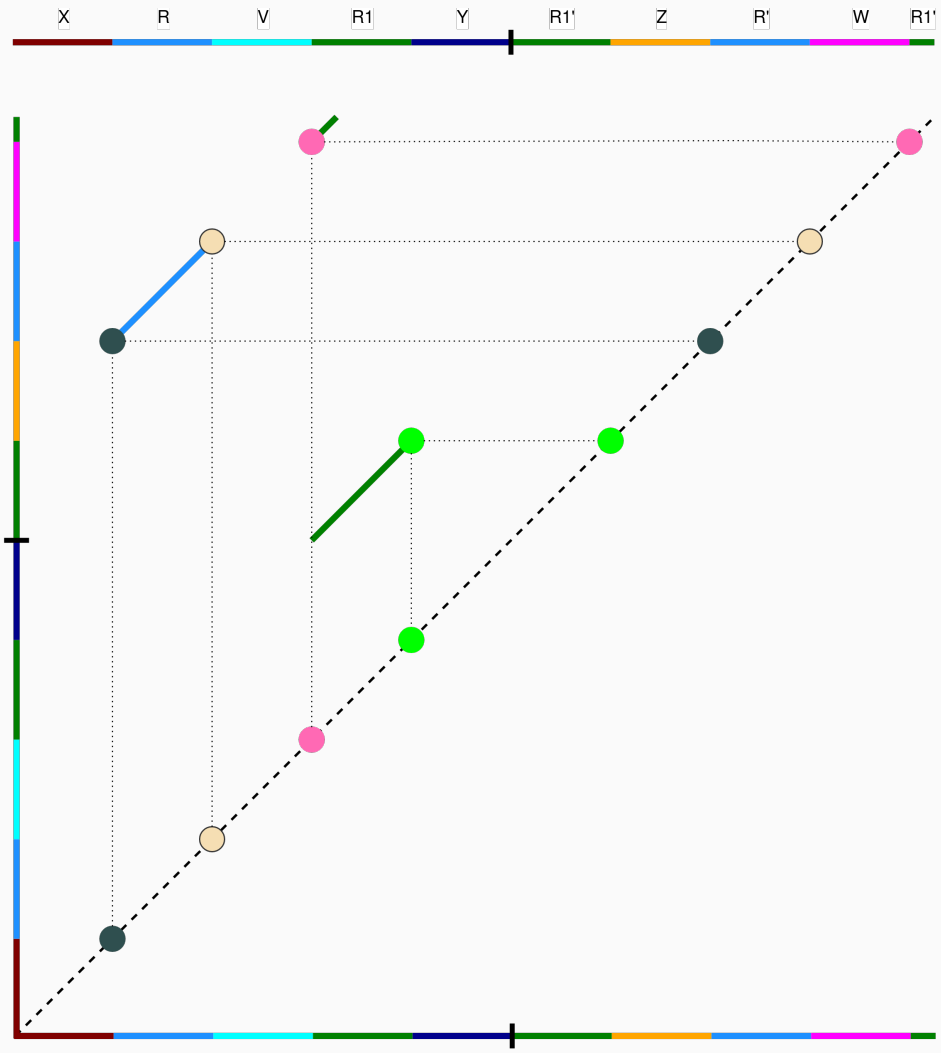

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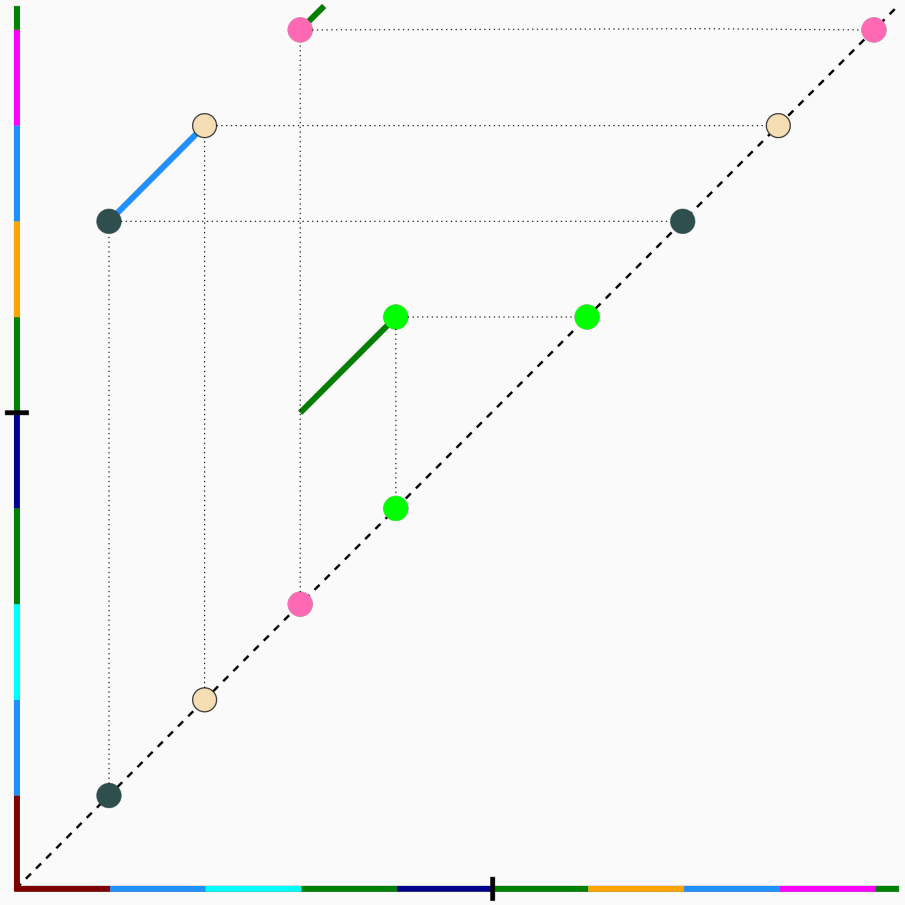

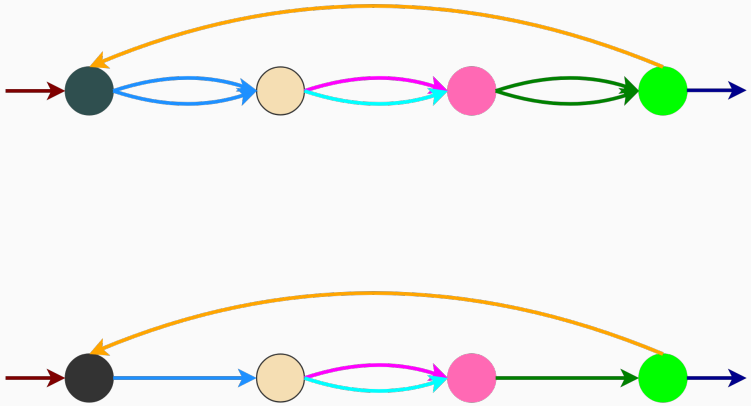

Results

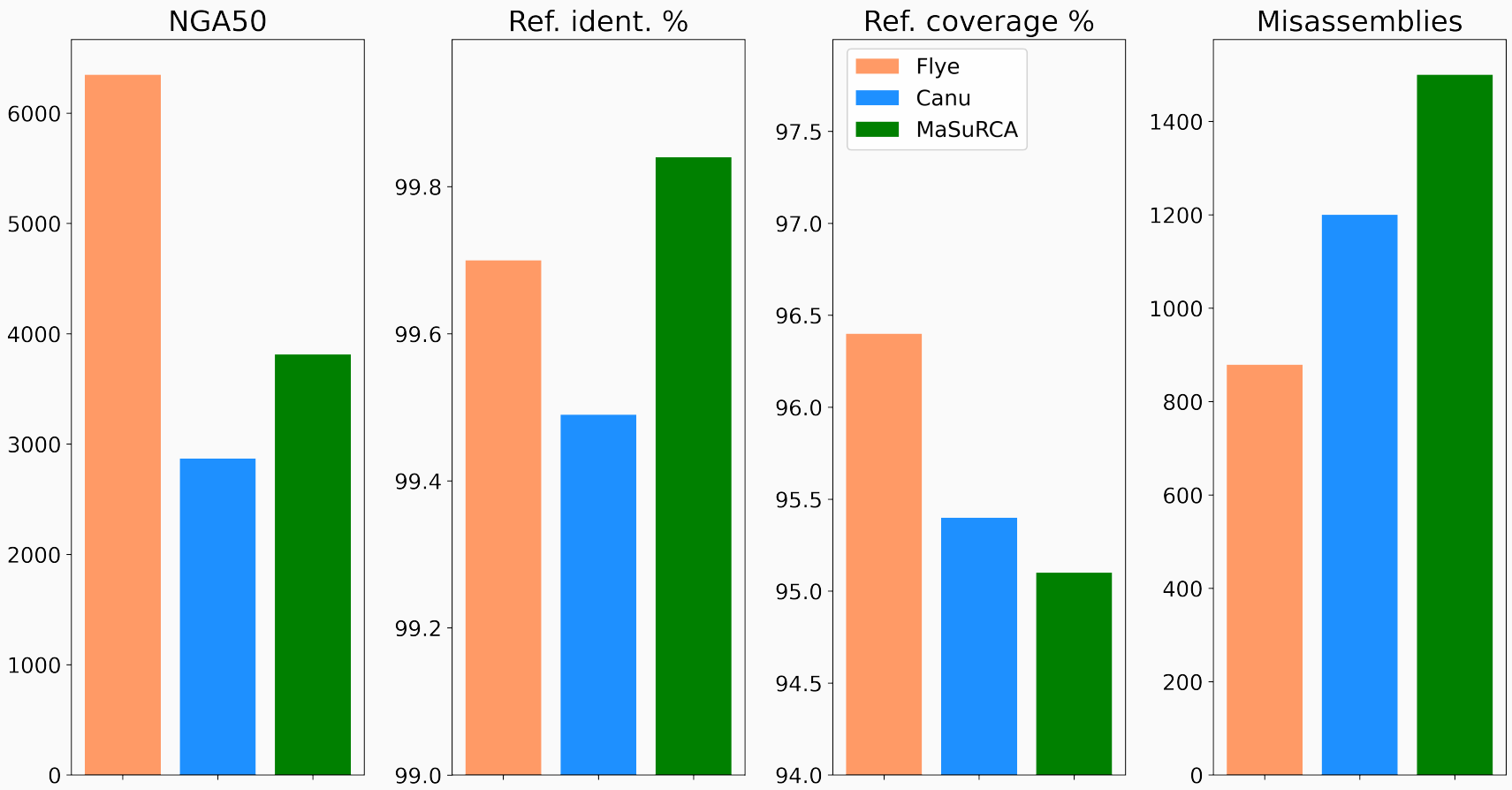

Figure 2: Results for HUMAN testset

References i

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Assembly of long, error-prone reads using repeat graphs.
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What is the difference between the breakpoint graph and the de Bruijn graph?
BMC genomics, 15 Suppl 6:S6, 2014.

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Genome Research, 14(9):1786–1796, Sept. 2004.

References ii

B. J. Walker, T. Abeel, T. Shea, M. Priest, A. Abouelliel, S. Sakthikumar, C. A. Cuomo, Q. Zeng, J. Wortman, S. K. Young, and A. M. Earl.
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PloS One, 9(11):e112963, 2014.

Git (presentation and poster)

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Appendix

Dot plot creation

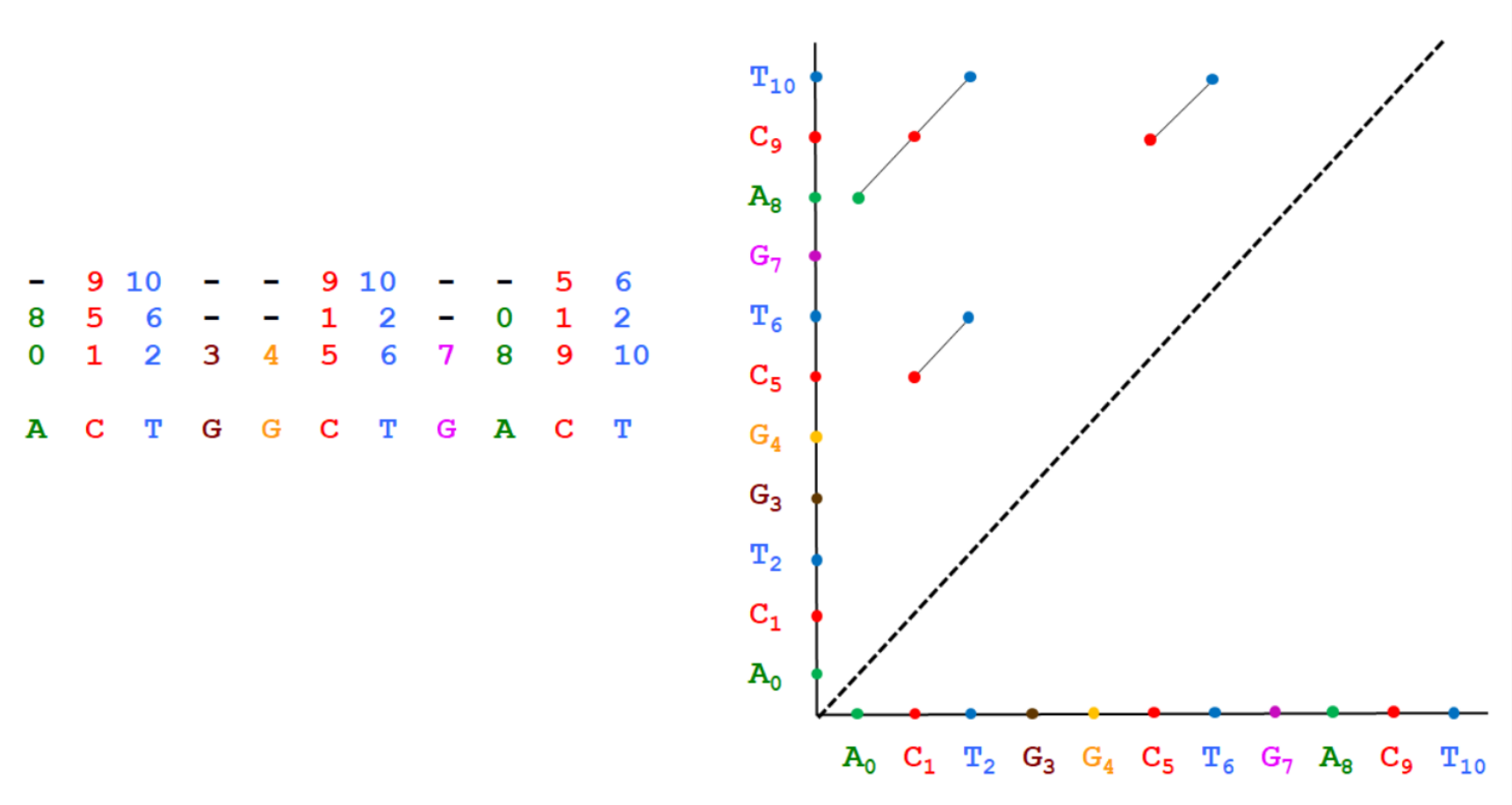

Figure 3: Dot plot creation

Repeat graphs

- generalization of de bruijn graphs
- structure
- creation
  - from disjointigs = random walk of reads on the repeat graph
  - means the repeat graph hasn't to be known

Difference repeat graph de Bruijn graph

- A-Bruijn graph (alignments) generalizes the de Bruijn graph
- "We thus argue that the time has come to explain that the breakpoint graphs and the de Bruijn graphs are two identical data structures (if one ignores a cosmetic difference between them) as they both represent specific instances of a general notion of the A-Bruijn graph introduced in [2]. The A-Bruijn graphs are based on representing genomes as sets of labeled paths and further gluing identically labeled edges (breakpoint graphs) or vertices (de Bruijn graphs) in the resulting paths." [3]
- de Bruijn graphs need correct bases
- otherwise tangled graph

Segmental duplications

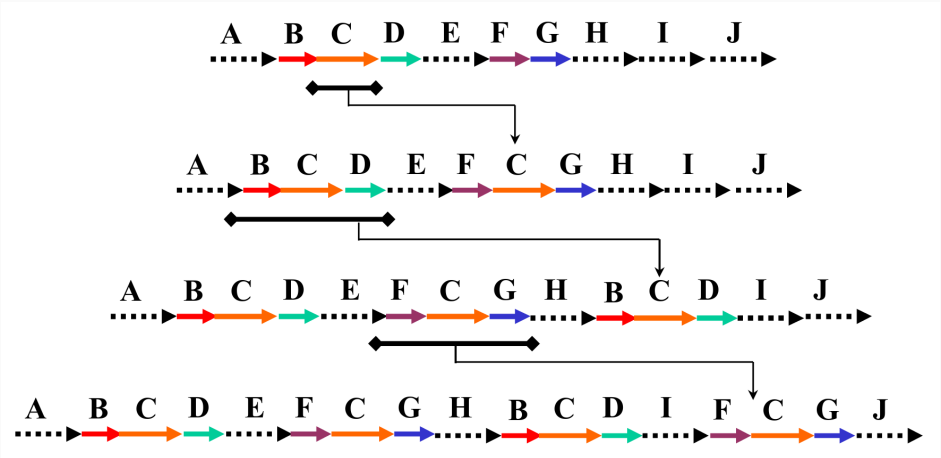

Figure 4: Segmental Duplications

- Segmental duplications are duplicated blocks of genomic DNA typically ranging in size from 1-200 kb (IHGSC 2001)
- They often contain sequence features such as high-copy repeats and gene sequences with intron-exon structure.

Contigity improvement

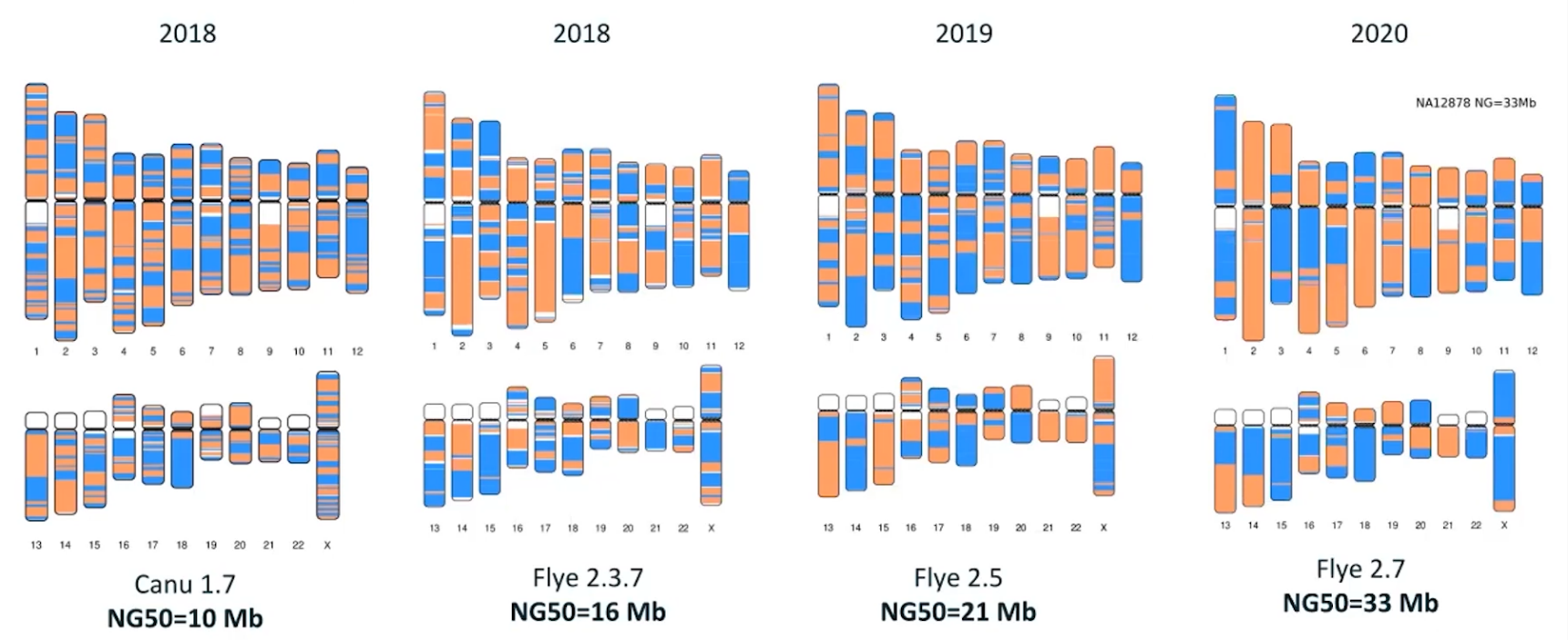

Figure 5: Contigity improvements

- colors are contigs $\rightarrow$ change in color means fragmentation