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Assembly of long, error-prone reads using repeat graphs

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

June 30, 2021

Johannes Hausmann, Luis Kress

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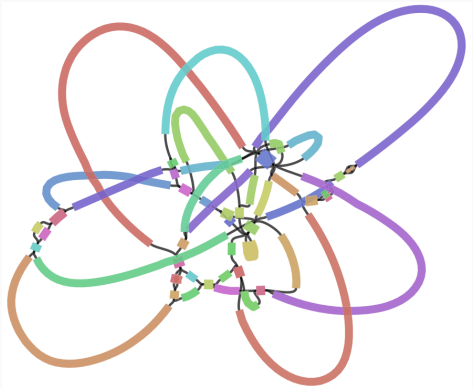

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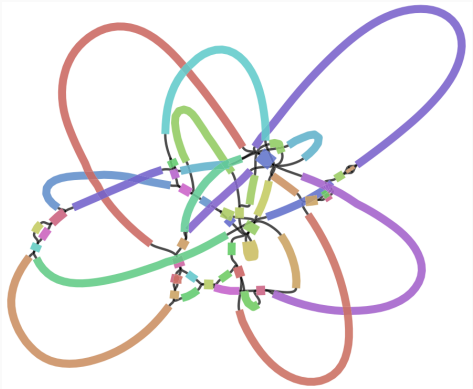

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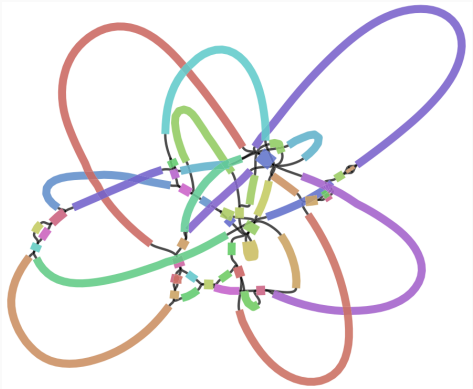

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- Error rate long read $\leftrightarrow$ short read
- Flye $\rightarrow$ resolve these repeats correctly, create contiguous assemblies

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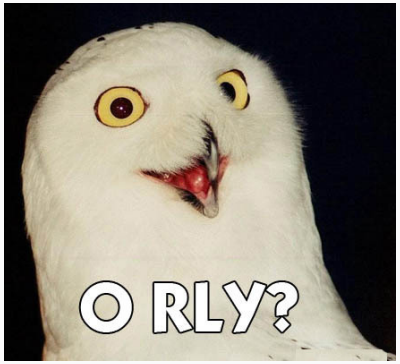
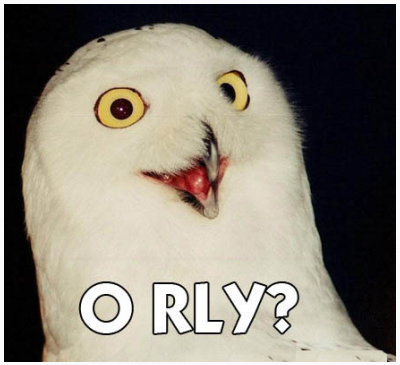


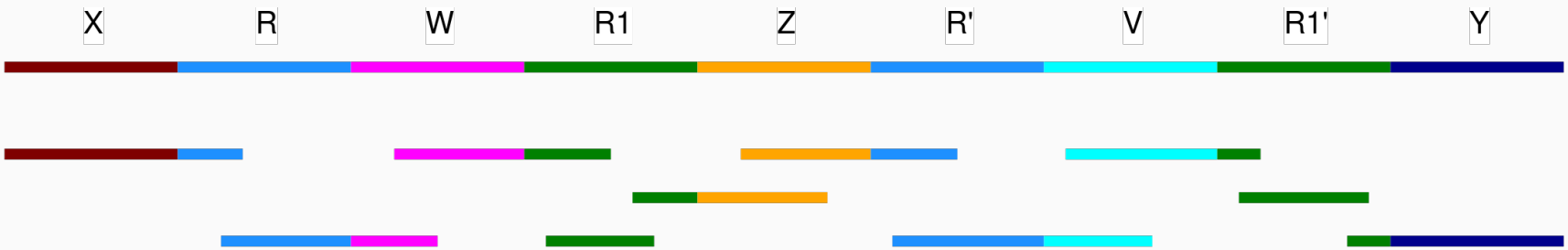
Disjointigs

- 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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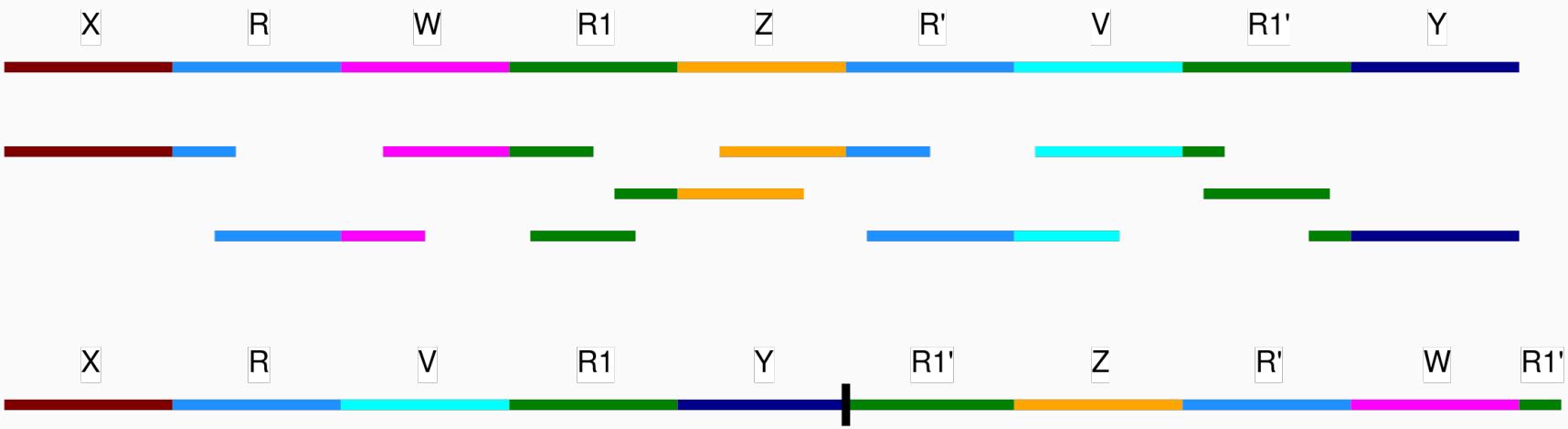
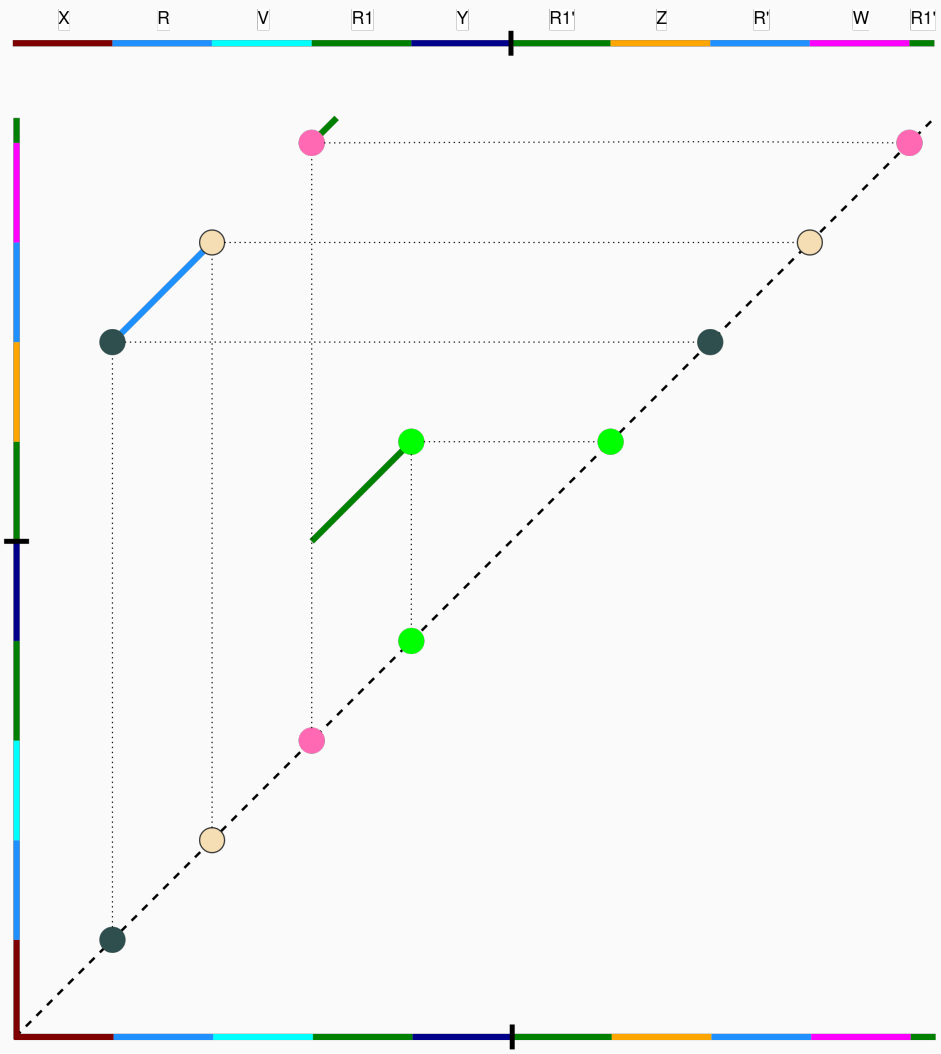


Repeat Graph Creation



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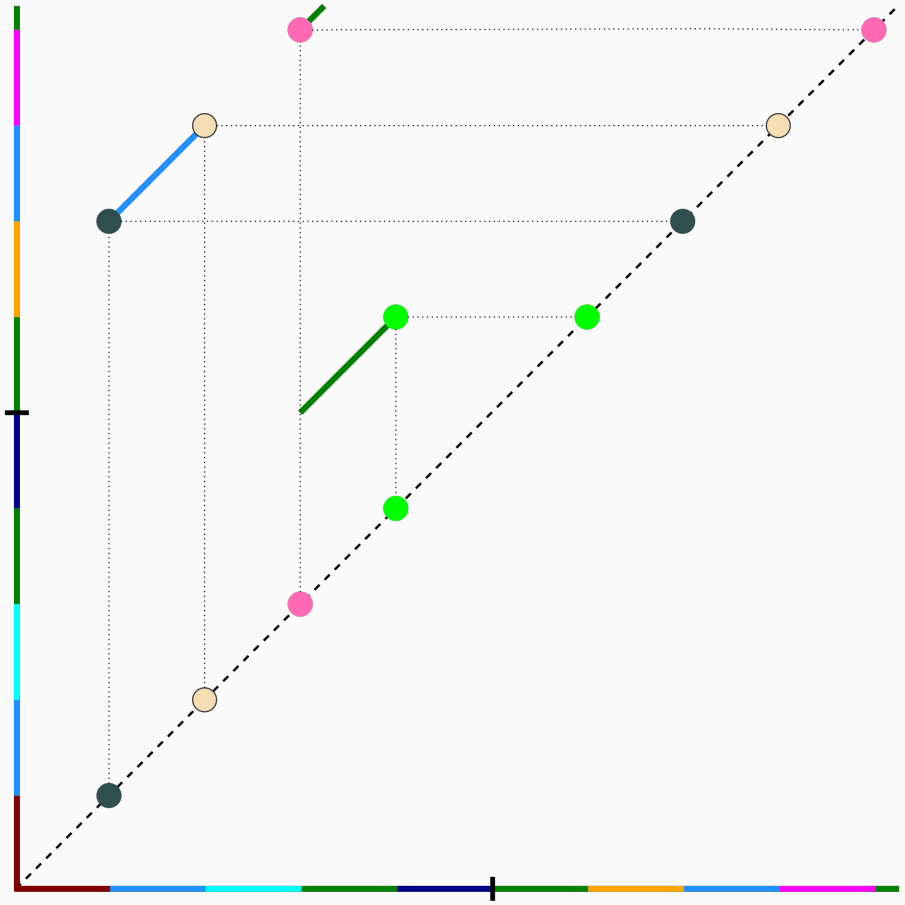

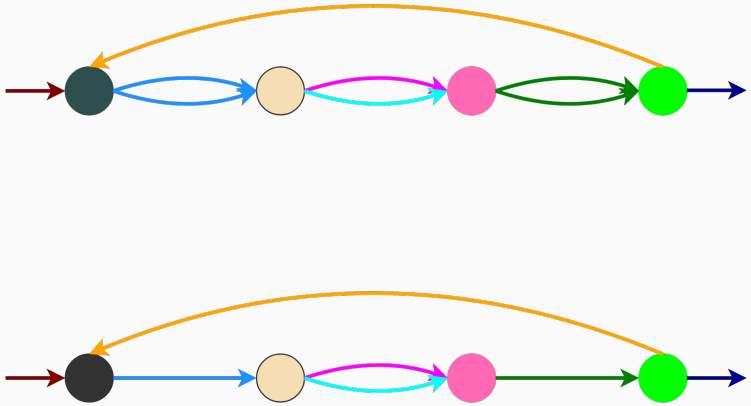

Results

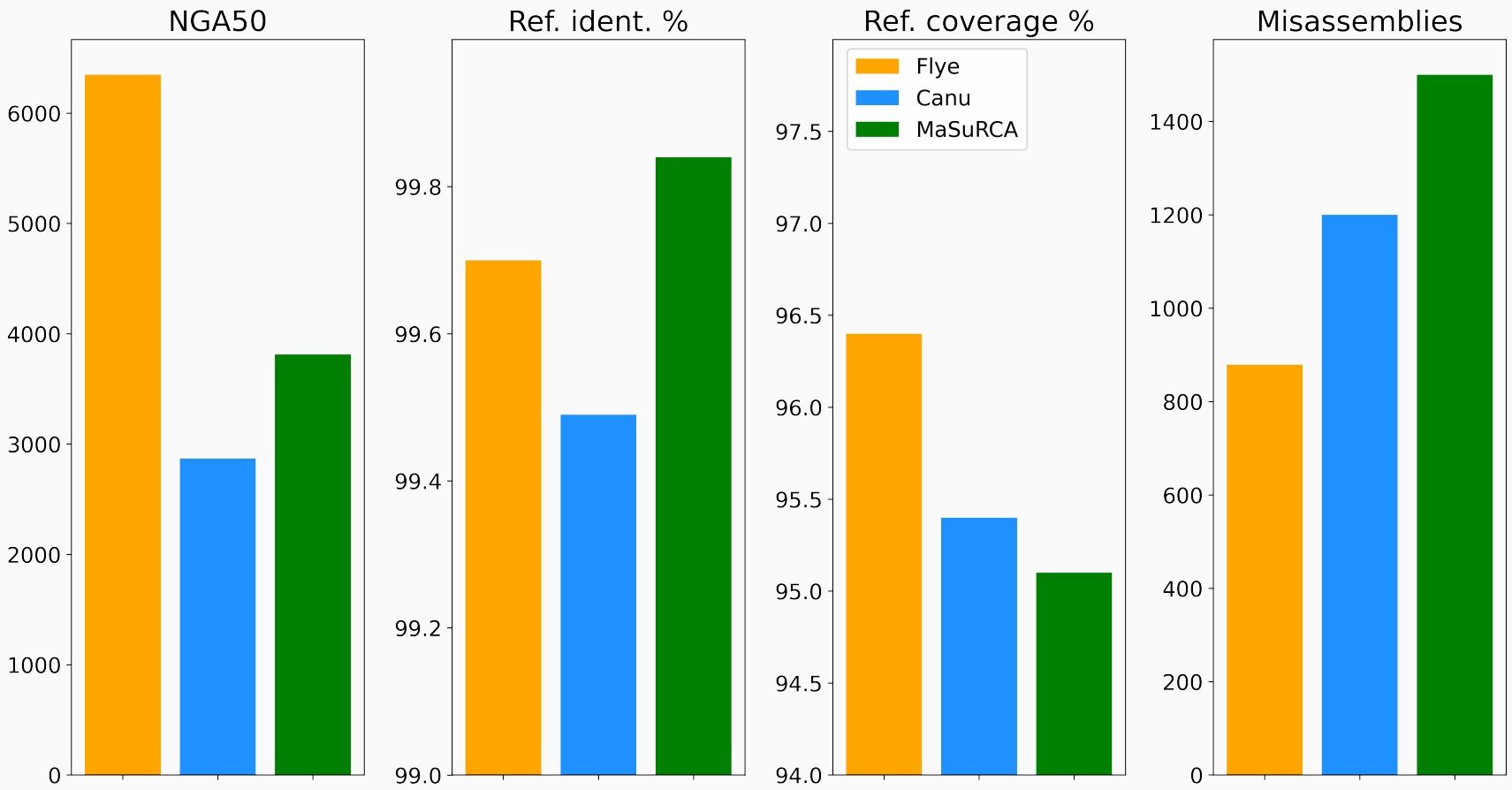

Figure 2: Results for HUMAN testset

References i


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Assembly of long, error-prone reads using repeat graphs.
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References ii

Git (presentation and poster)

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Appendix

Dot plot creation

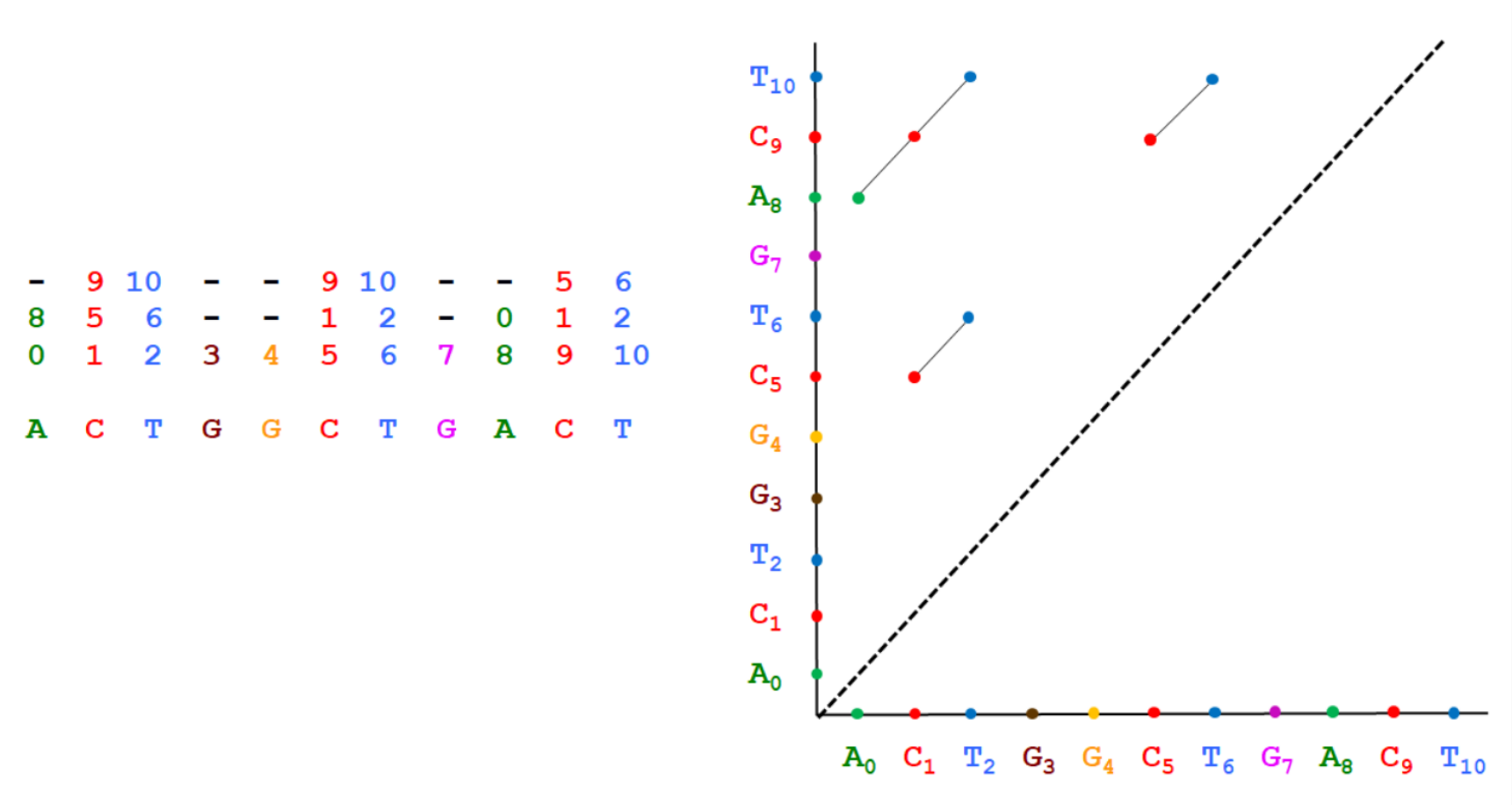

Figure 3: Dot plot creation

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 [13]. 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." [2]
- de Bruijn graphs need correct bases
- otherwise tangled graph

Segmental duplications

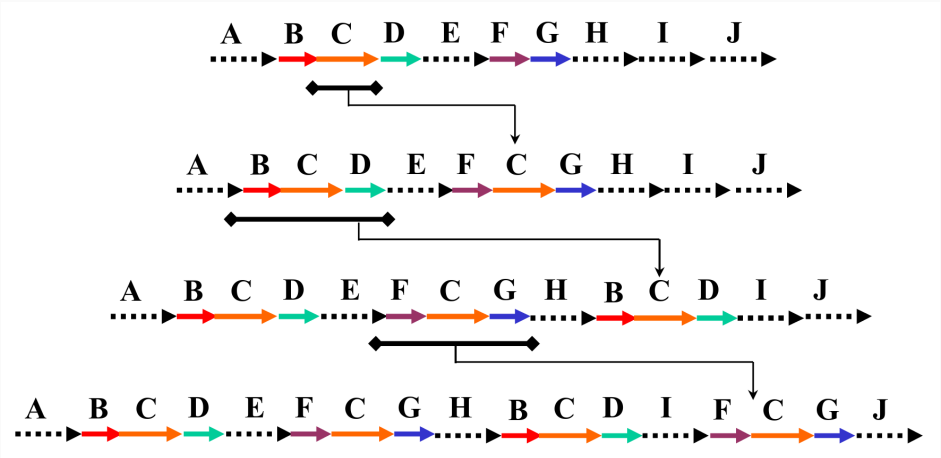

Figure 4: Segmental Duplications

- 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

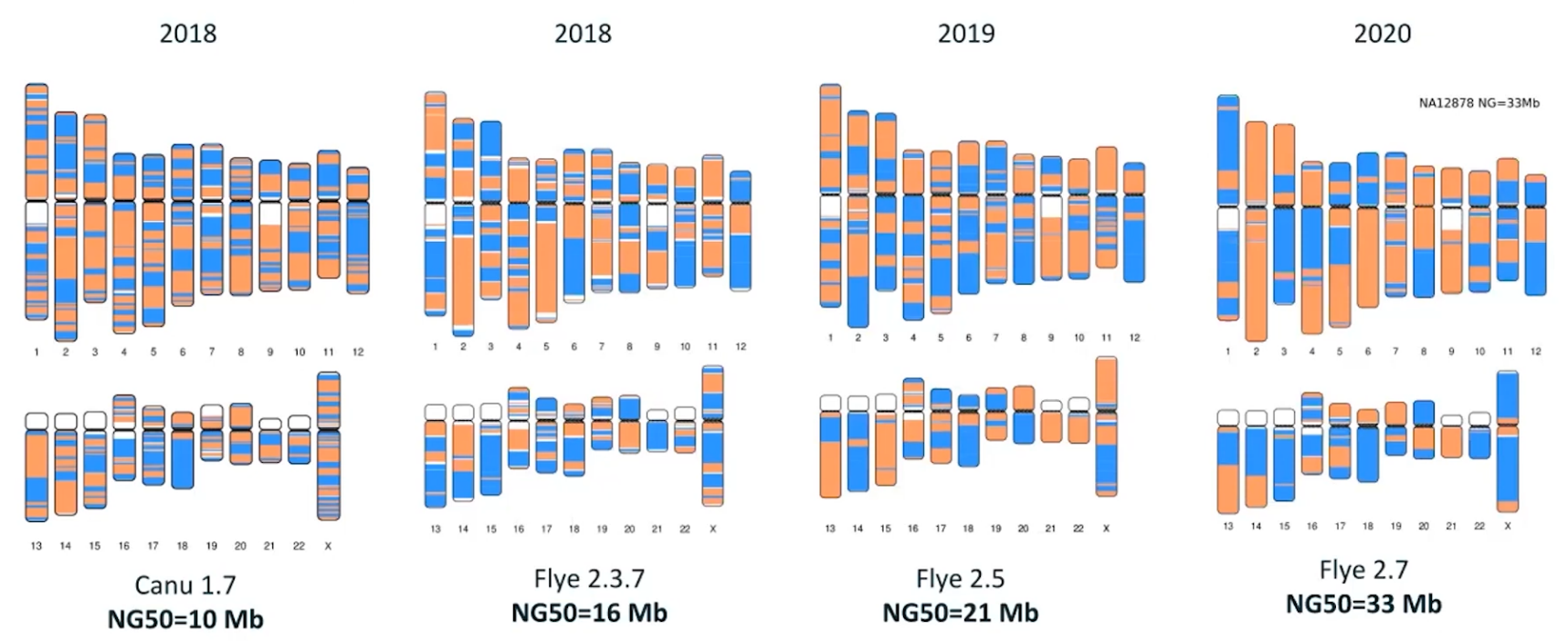

Figure 5: Contigity improvements

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