GOETHE UNIVERSITÄT FRANKFURT AM MAIN

Assembly of long, error-prone reads using repeat graphs

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

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Johannes Hausmann, Luis Kress

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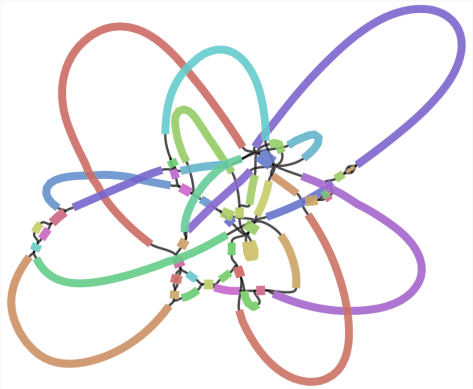

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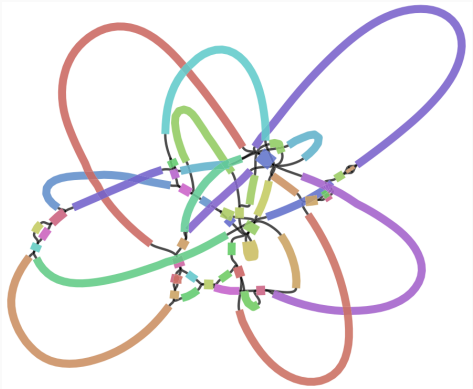

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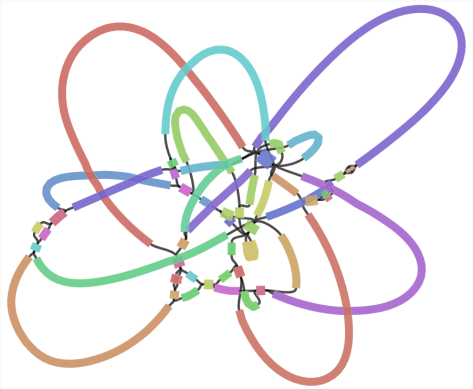

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- Error rate long read $\leftrightarrow$ short read
- Flye $\rightarrow$ resolve these repeats correctly, create contiguous assemblies

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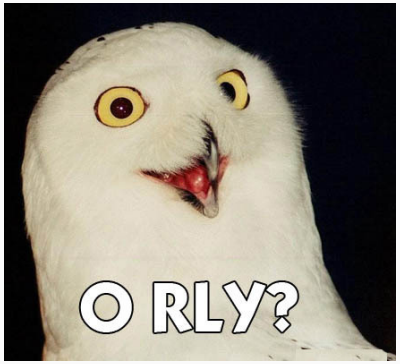
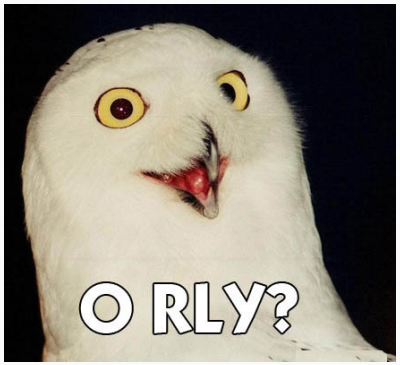


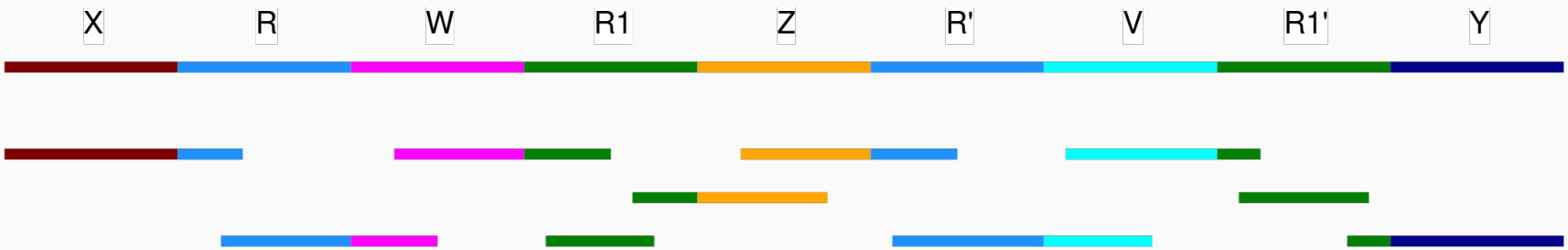
Disjointigs

- 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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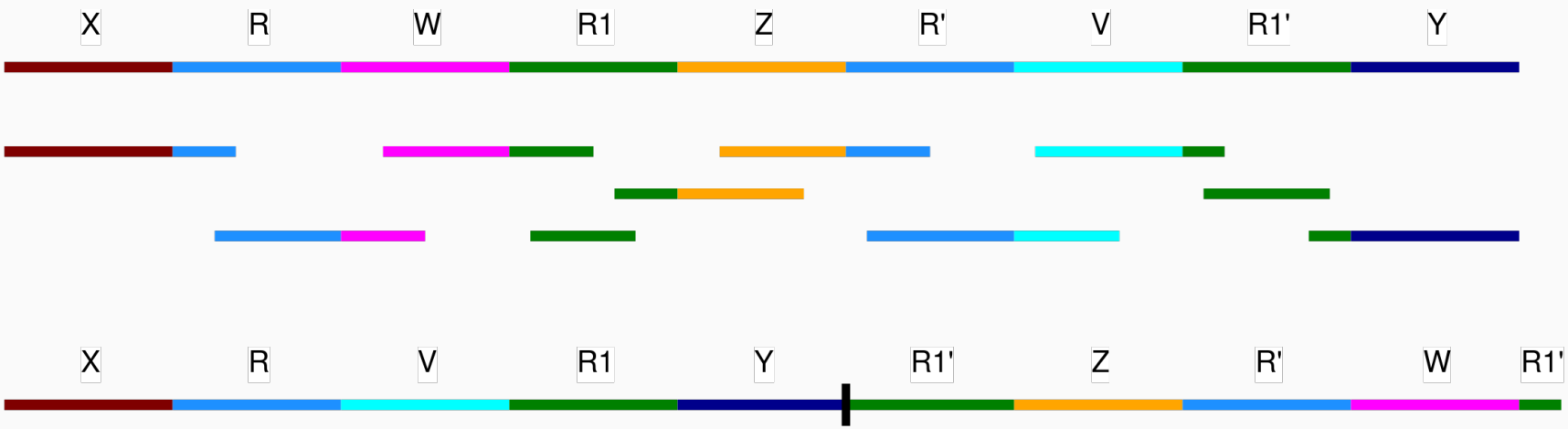

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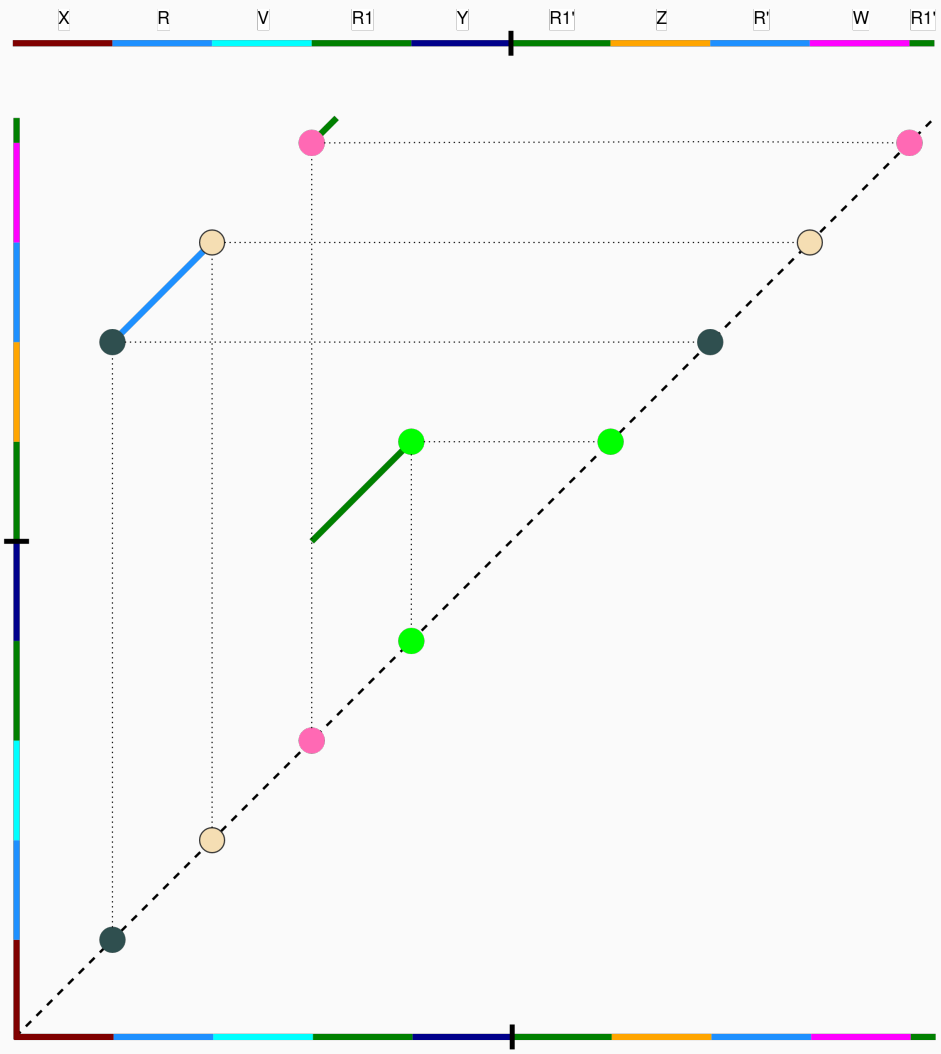

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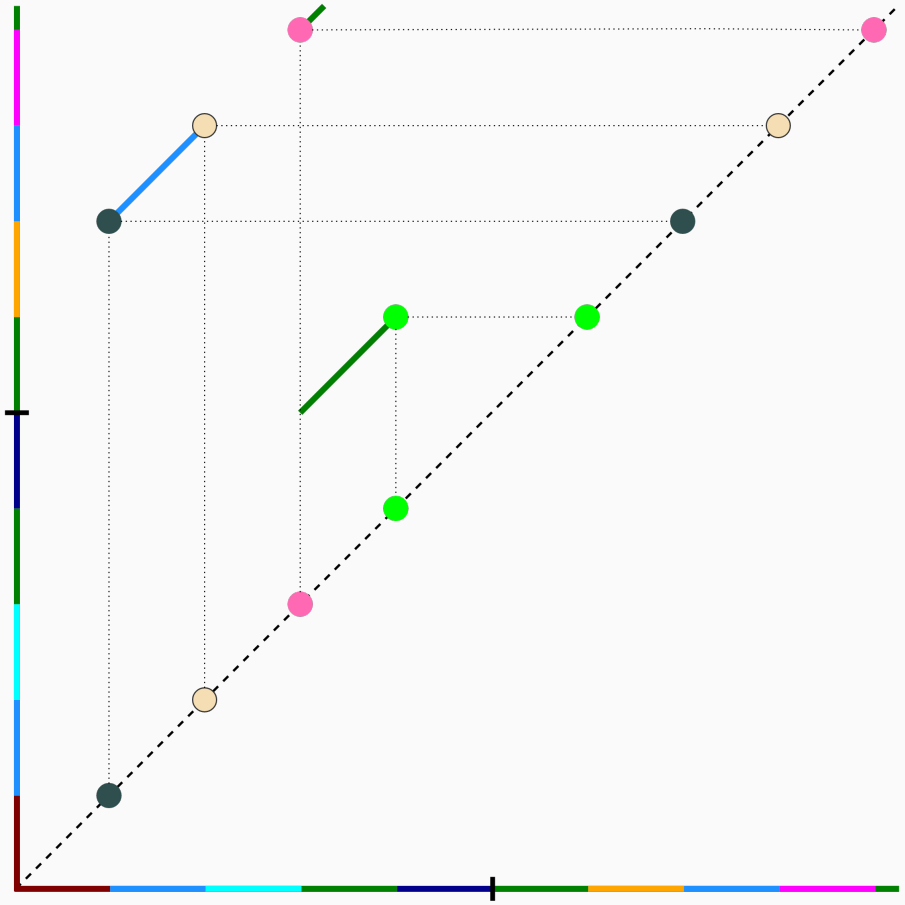
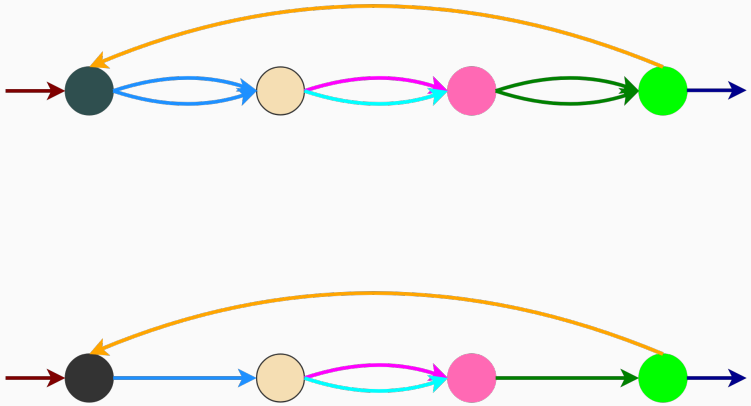
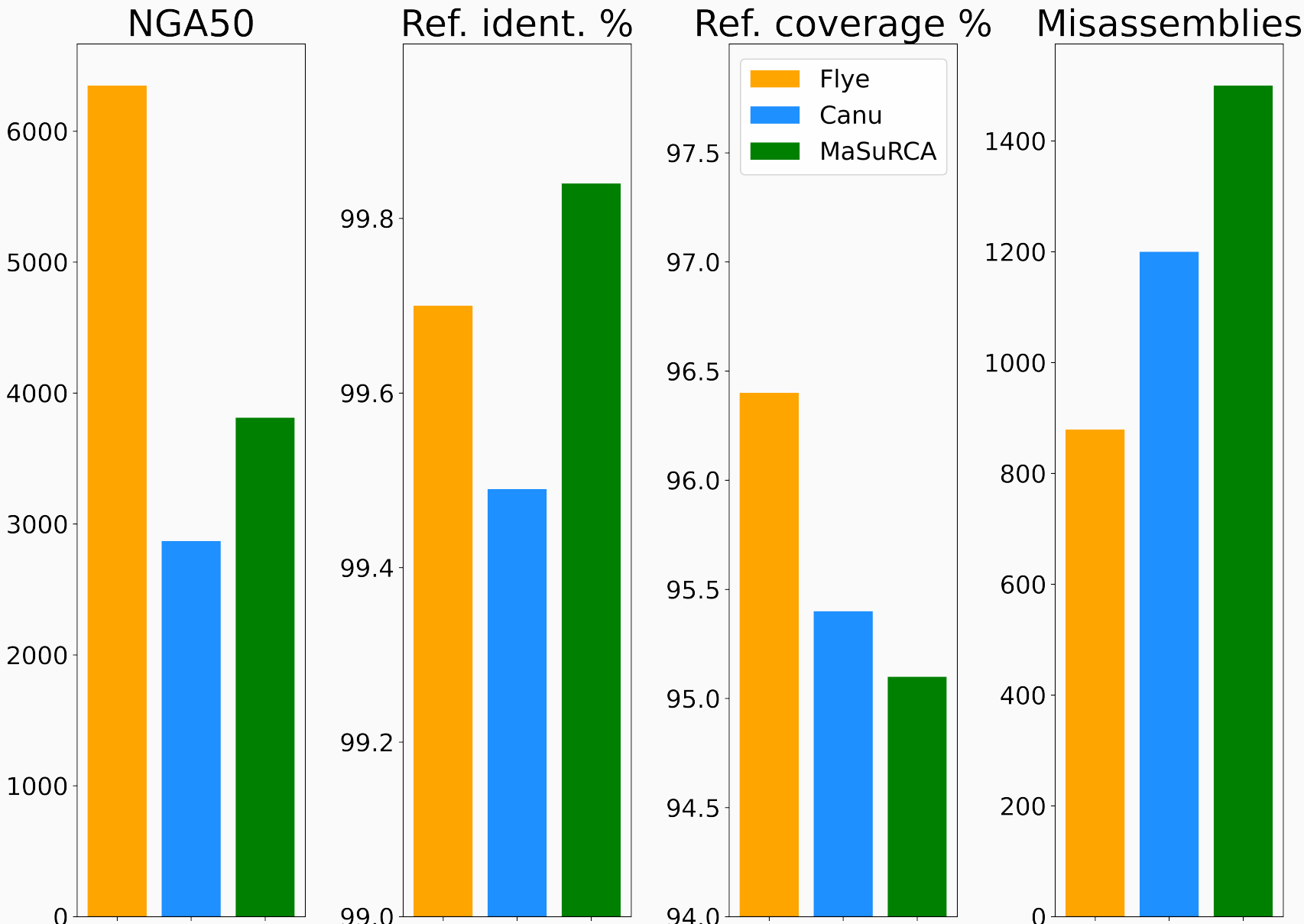




Results


NGA50
Ref. ident. %
Ref. coverage %
Misassemblies
Flye
Canu
MaSuRCA
6000
5000
4000
3000
2000
1000
0
99.8
99.6
99.4
99.2
99.0
97.5
97.0
96.5
96.0
95.5
95.0
94.5
94.0
1400
1200
1000
800
600
400
200
0

References i

M. Kolmogorov, J. Yuan, Y. Lin, and P. A. Pevzner.
Assembly of long, error-prone reads using repeat graphs.
Nature Biotechnology, 37(5):540–546, May 2019.

Git (presentation and poster)

Appendix

Dot plot creation

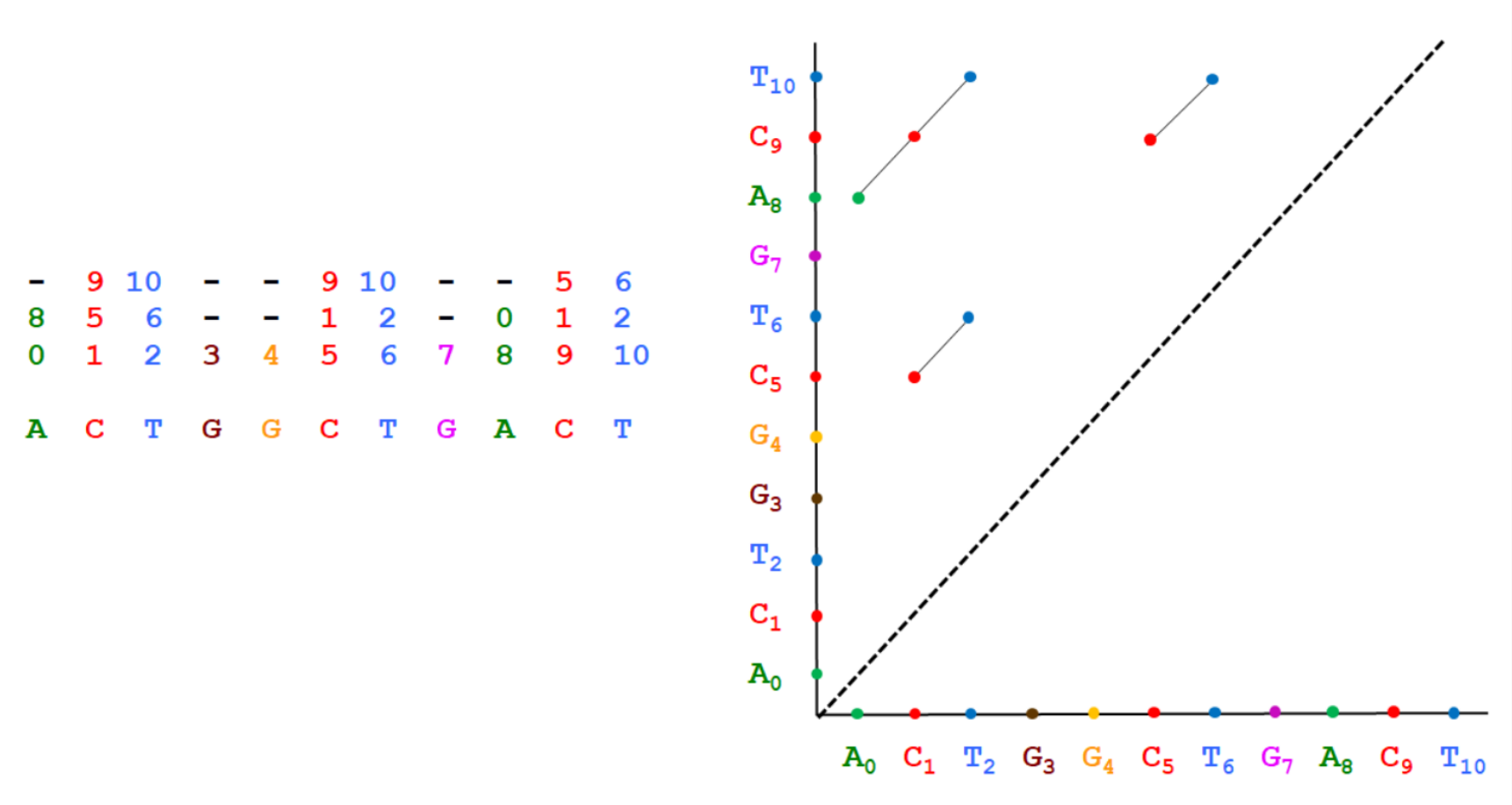


Figure 3: Dot plot creation

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.
- de Bruijn graphs need correct bases
- otherwise tangled graph

Segmental duplications

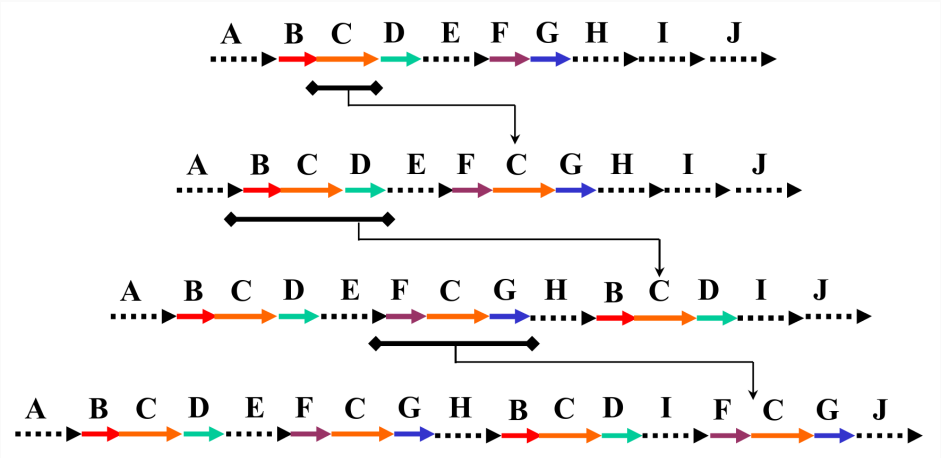


Figure 4: Segmental Duplications

- 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

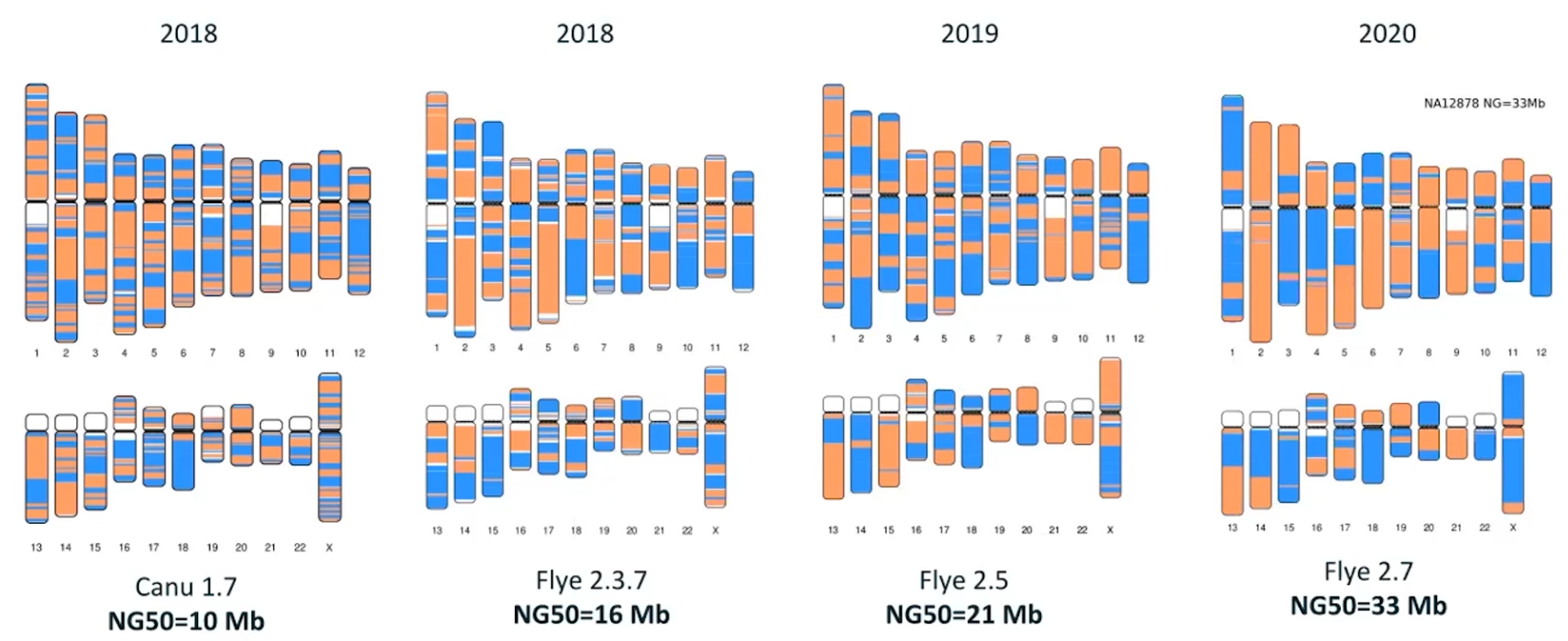


Figure 5: Contigity improvements

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