# Assembly of long, error-pront reads using repeat graphs

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- error rate long read ↔ short read
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  - we don't care (at least at the initial stage)
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- generate paths from overlapping reads without checking for correct assembly → disjointigs

# Repeat Graph Creation

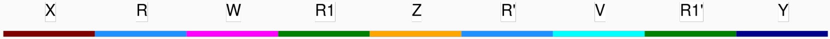

**Figure 1:** Example Genome

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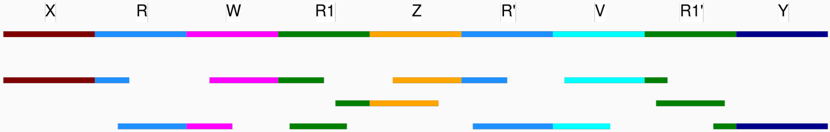

**Figure 2:** Example Genome and Reads

# Repeat Graph Creation

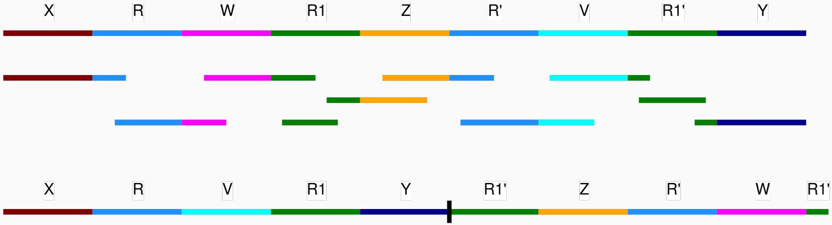

**Figure 3:** Example Genome, Reads and Disjointigs

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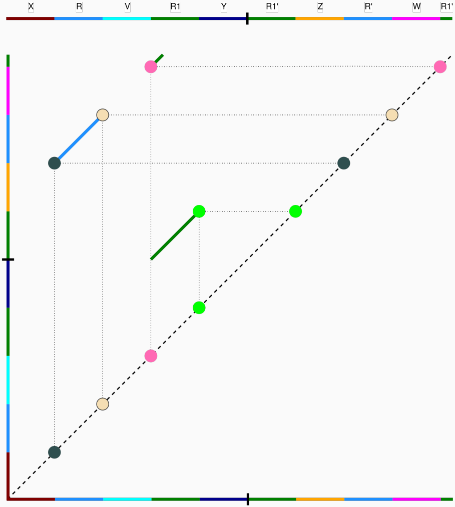

**Figure 4:** Breakpoint Graph

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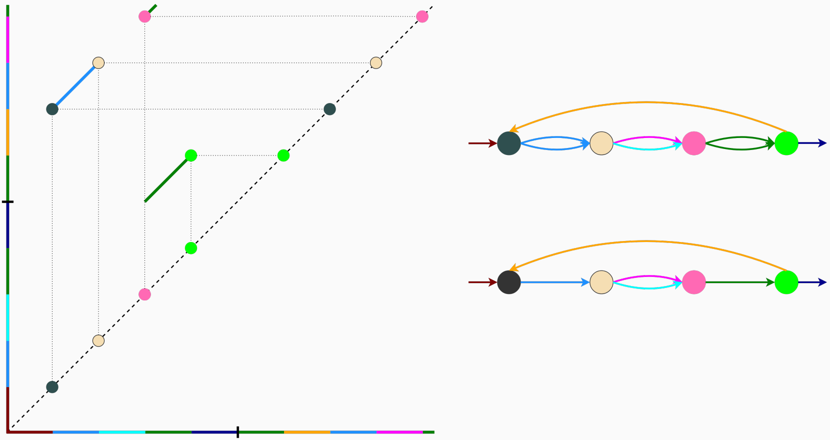


**Figure 5:** Repeat Graph

# Results

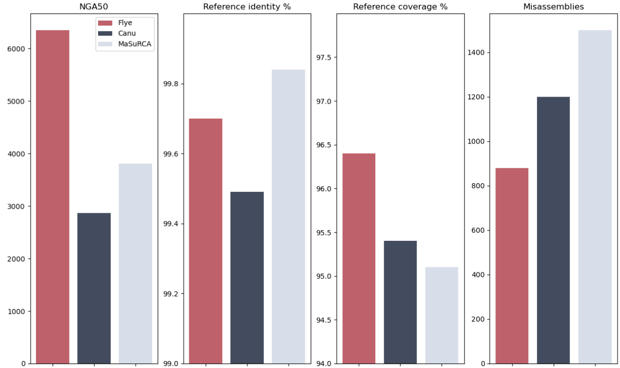


**Figure 6:** Results for HUMAN testset

# References i

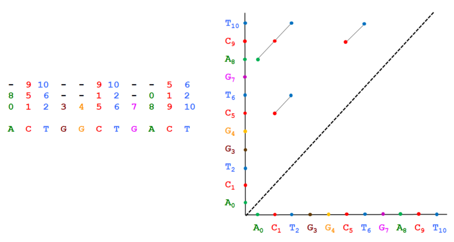
M. Kolmogorov, J. Yuan, Y. Lin, and P. A. Pevzner.
**Assembly of long, error-prone reads using repeat graphs.**
*Nature Biotechnology*, 37(5):540–546, May 2019.

# Git (presentation)

**Figure 7:** Link to our git repo

# Appendix

# Dot plot creation



| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| - | 9 | 10 | - | - | 9 | 10 | - | - | 5 | 6 |
| 8 | 5 | 6 | - | - | 1 | 2 | - | 0 | 1 | 2 |
| 0 | 1 | 2 | 3 | 4 | 5 | 6 | 7 | 8 | 9 | 10 |
| A | C | T | G | G | C | T | G | A | C | T |

$T_{10}$
$C_9$
$A_8$
$G_7$
$T_6$
$C_5$
$G_4$
$G_3$
$T_2$
$C_1$
$A_0$
$A_0$ $C_1$ $T_2$ $G_3$ $G_4$ $C_5$ $T_6$ $G_7$ $A_8$ $C_9$ $T_{10}$

**Figure 8:** Dot plot creation

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  - from disjointigs = random walk of reads on the repeat graph
  - means the repeat graph hasn't to be known

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- de Bruijn graphs need correct bases
- otherwise tangled graph

# Segmental duplications

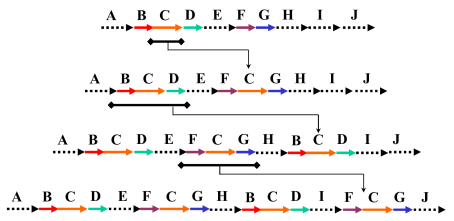
A B C D E F G H I J
A B C D E F C G H I J
A B C D E F C G H B C D I J
A B C D E F C G H B C D I F C G J

**Figure 9:** Segmental Duplications

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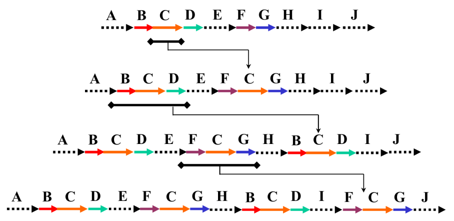
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**Figure 9:** Segmental Duplications

- 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

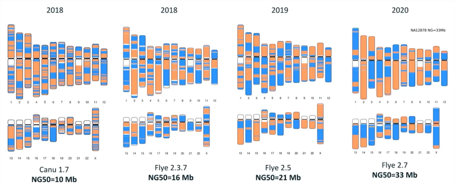

**Figure 10:** Contigity improvements

- colors are contigs

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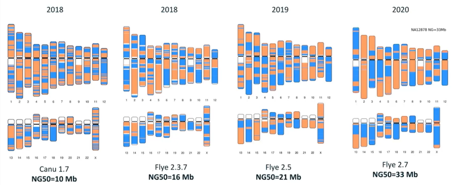

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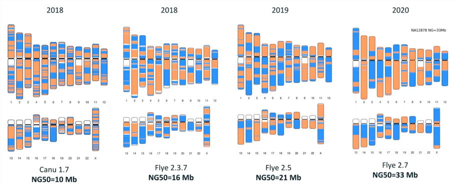

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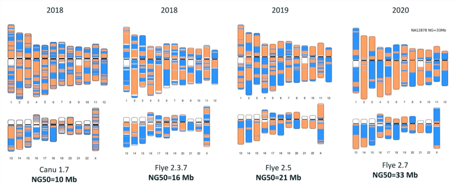

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