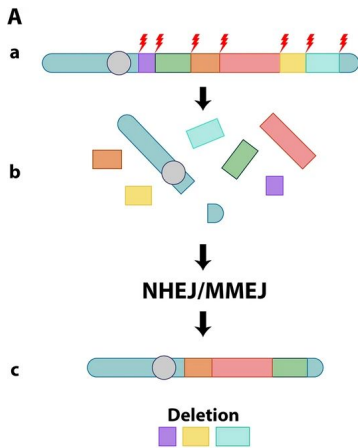
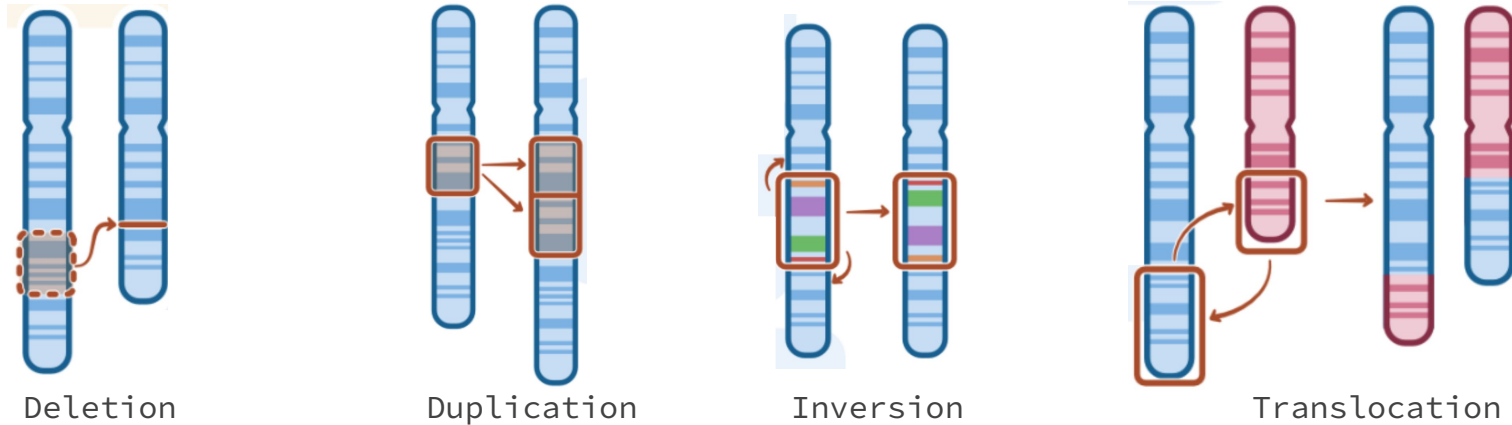


Barbara Poszewiecka

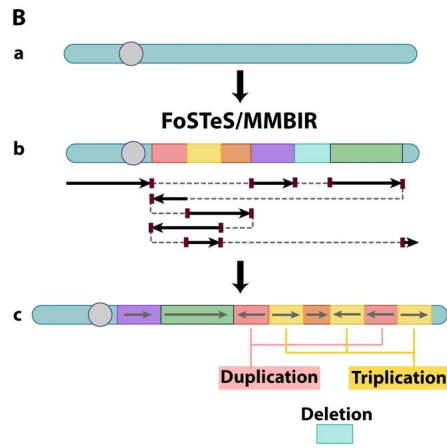


Bioinformatics

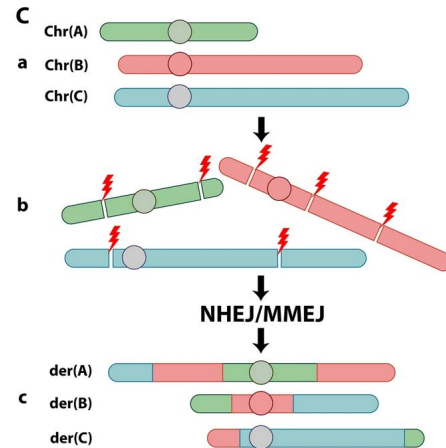
Chromosomal rearrangements



Chromoanagenesis

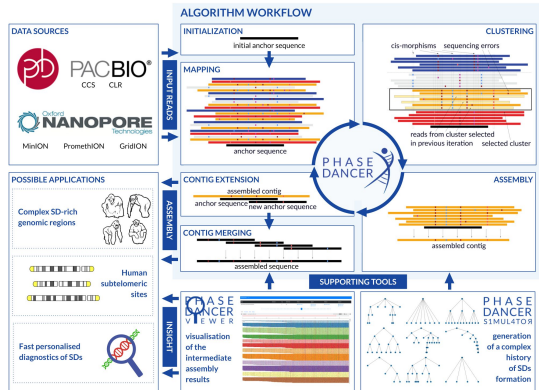


Chromoanasythesis

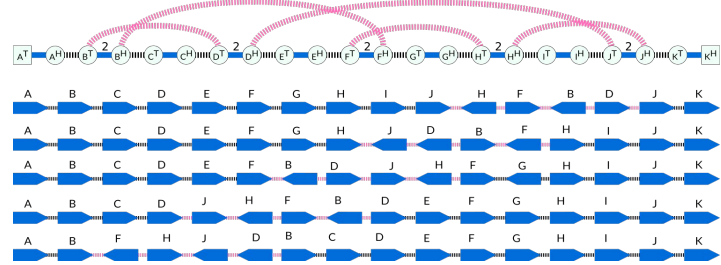


Chromoplexy

Assembling regions enriched in segmental duplications
PhaseDancer Assembler; Genome Biology 2023



An efficient enumeration algorithm for minimal linear Eulerian decompositions of Karyote graph

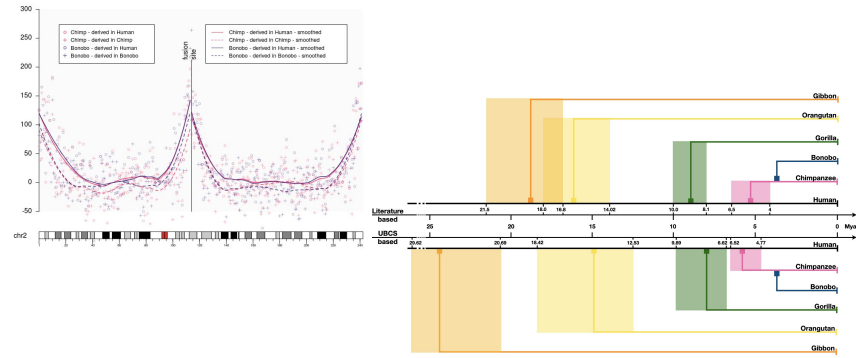


A server supporting the clinical diagnosis of structural variants that disrupt chromatin structure; Nucleic Acids Research 2022

The screenshot shows the TADeUS2 web server interface, which is a Django-based application. It includes sections for:

- Visualization:**
 - Syntentic mode:** Displays a heatmap of syntentic relationships between genomic regions.
 - Breakpoint mode:** Displays a heatmap of structural variant breakpoints.
- Datasets:** Lists various genomic datasets and annotations, including Ensembl, OMIM, 4D Nucleome, ClinGen, LUSC Genome Browser, ENCODE, and Genotek/ClassifyCNV.
- Evaluation:**
 - Chromosome conformation changes:** Shows a diagram of chromatin interactions.
 - Gene ranking:** Lists genes and their scores, such as HDAC1, TSC2, and PML.
 - Pathogenicity:** Shows the TADA-CNV annotation tool.

Methods for dating large evolutionary events; BMC Genomics 2



ALGORITHM WORKFLOW

INITIALIZATION

initial anchor sequence

PHASE
DANCER



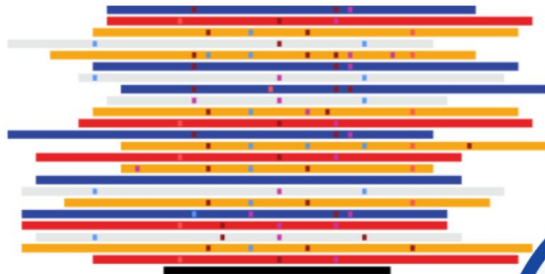
ALGORITHM WORKFLOW

INITIALIZATION

initial anchor sequence



MAPPING



anchor sequence



PHASE
DANCER

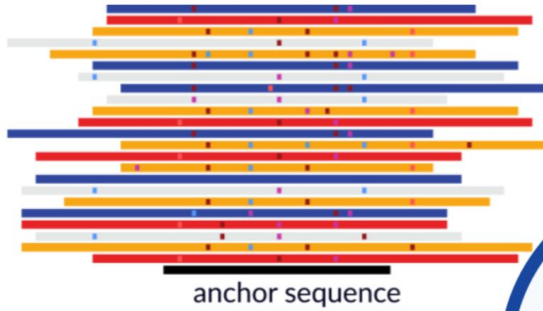


ALGORITHM WORKFLOW

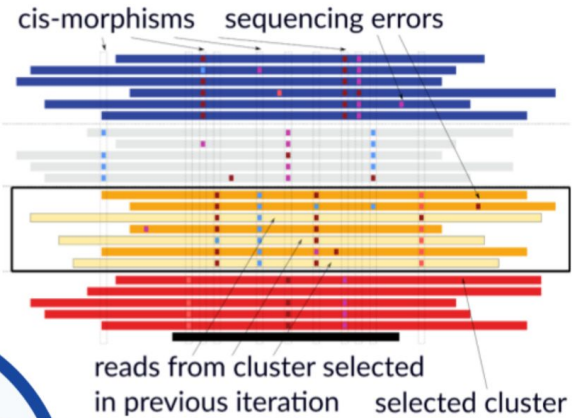
INITIALIZATION

initial anchor sequence

MAPPING



CLUSTERING



PHASE
DANCER

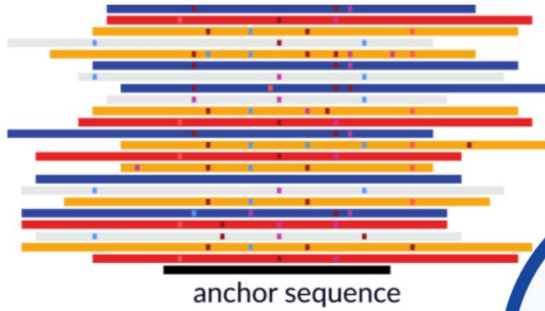


ALGORITHM WORKFLOW

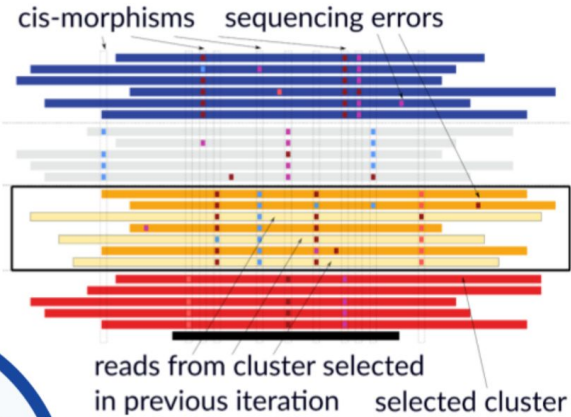
INITIALIZATION

initial anchor sequence

MAPPING

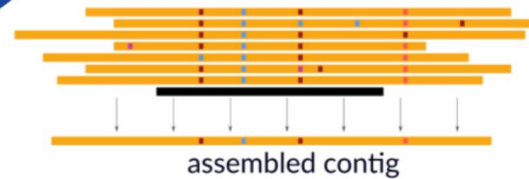


CLUSTERING



PHASE
DANCER

ASSEMBLY

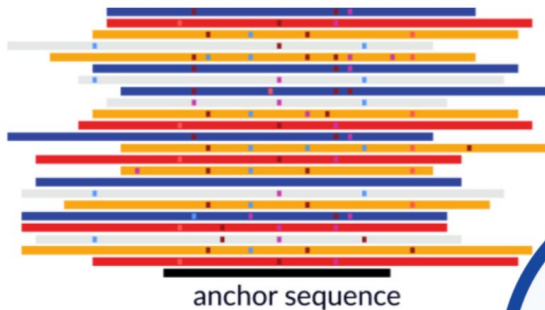


ALGORITHM WORKFLOW

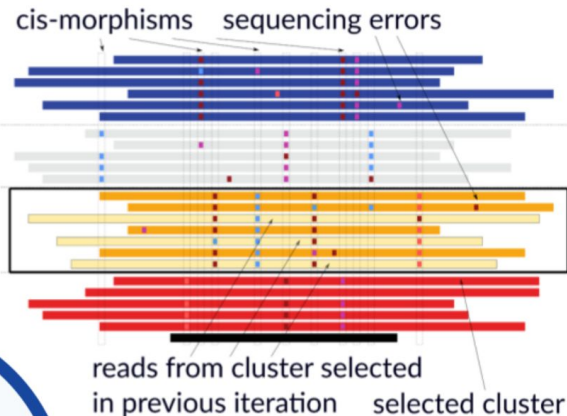
INITIALIZATION

initial anchor sequence

MAPPING



CLUSTERING

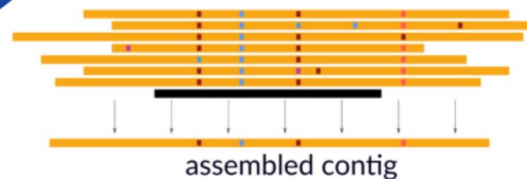


CONTIG EXTENSION



PHASE
DANCER

ASSEMBLY

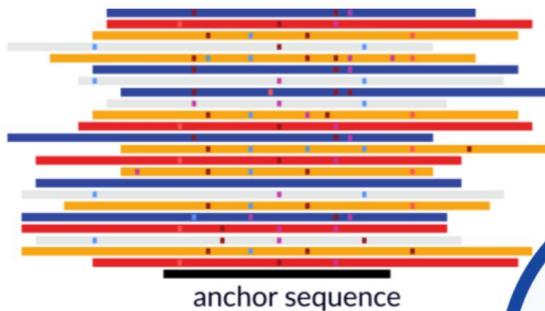


ALGORITHM WORKFLOW

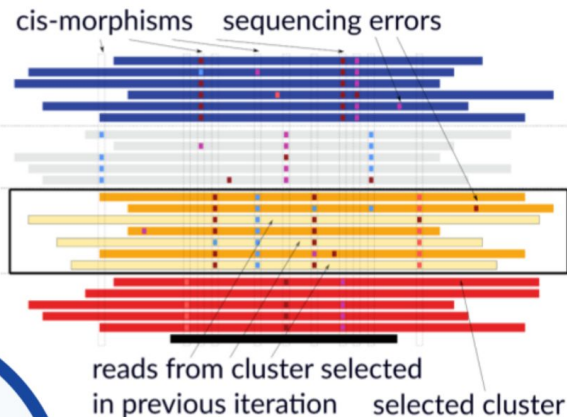
INITIALIZATION

initial anchor sequence

MAPPING



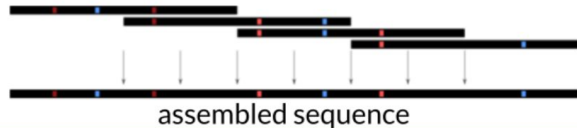
CLUSTERING



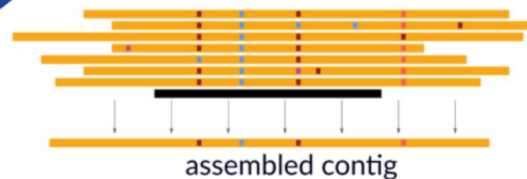
CONTIG EXTENSION



CONTIG MERGING

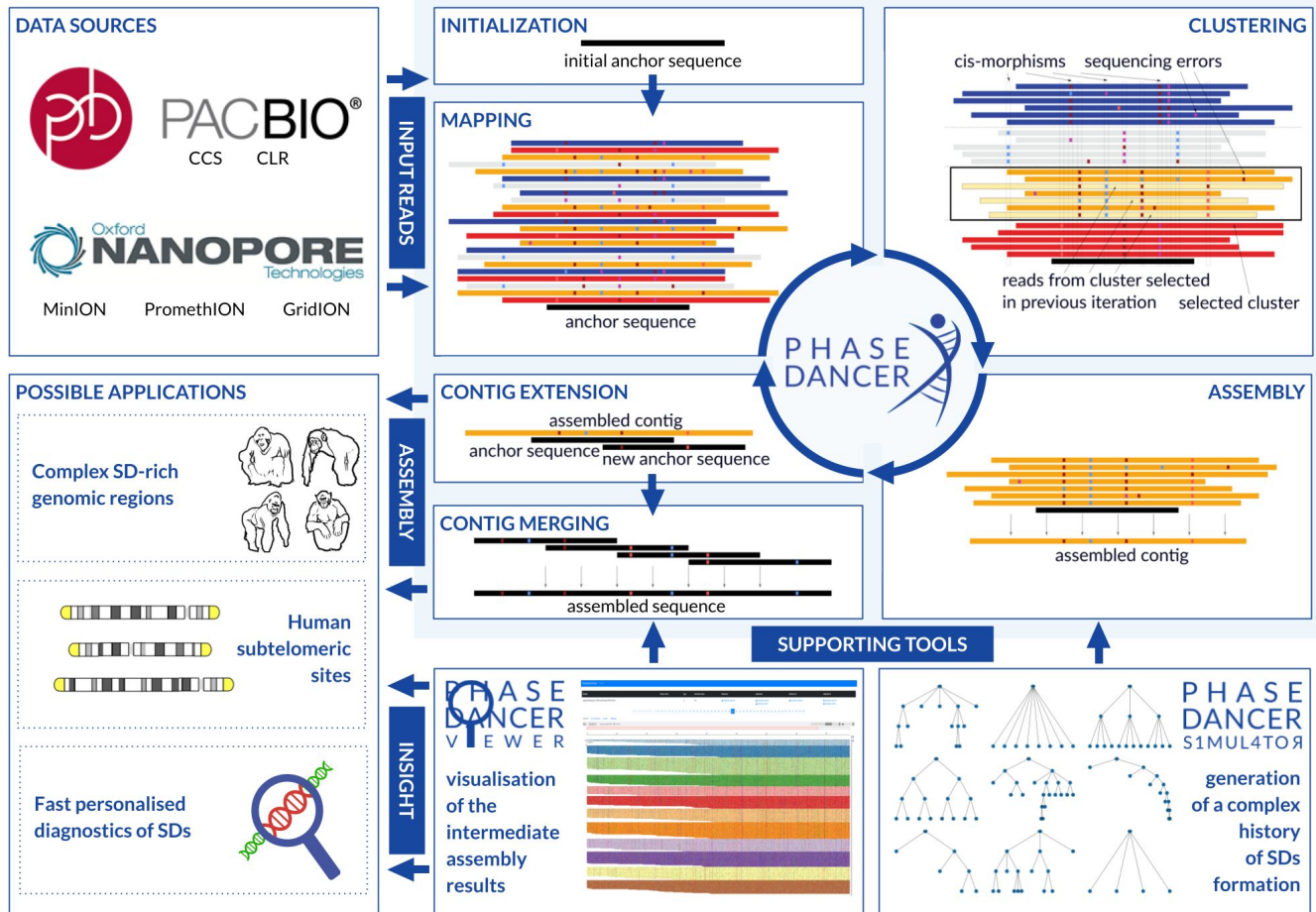


ASSEMBLY

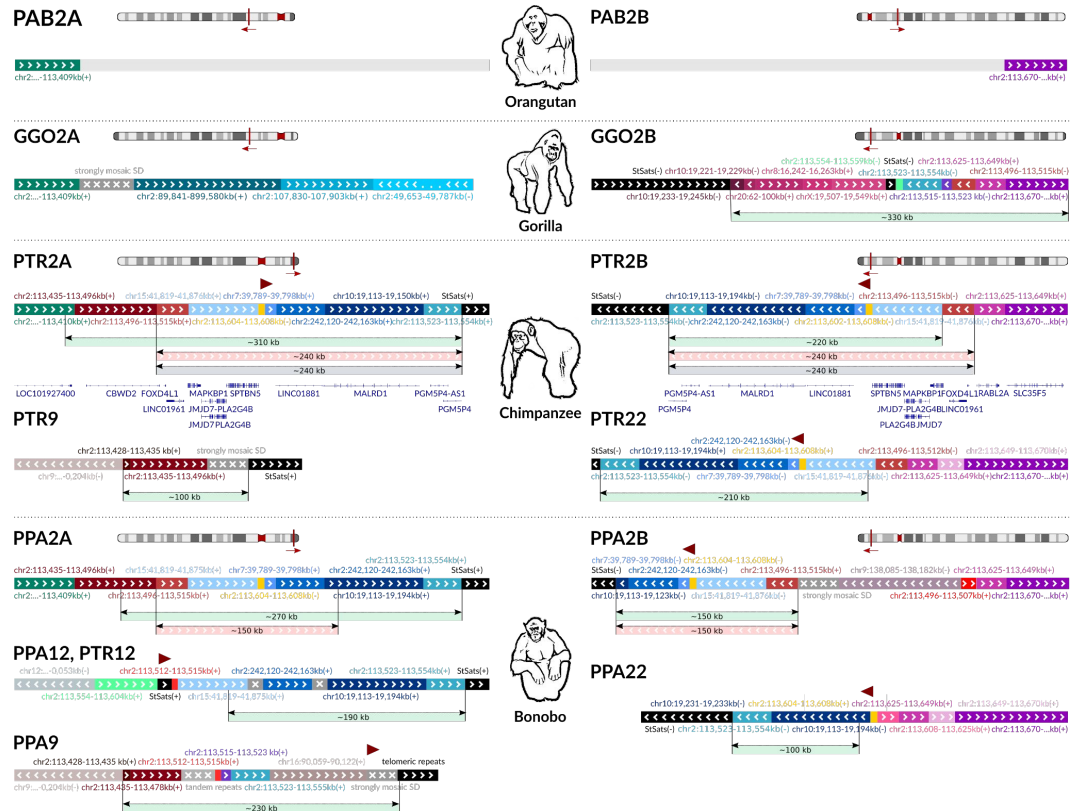


PHASE
DANCER

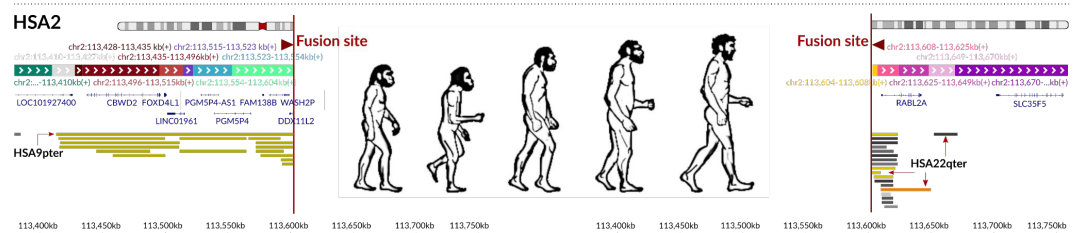


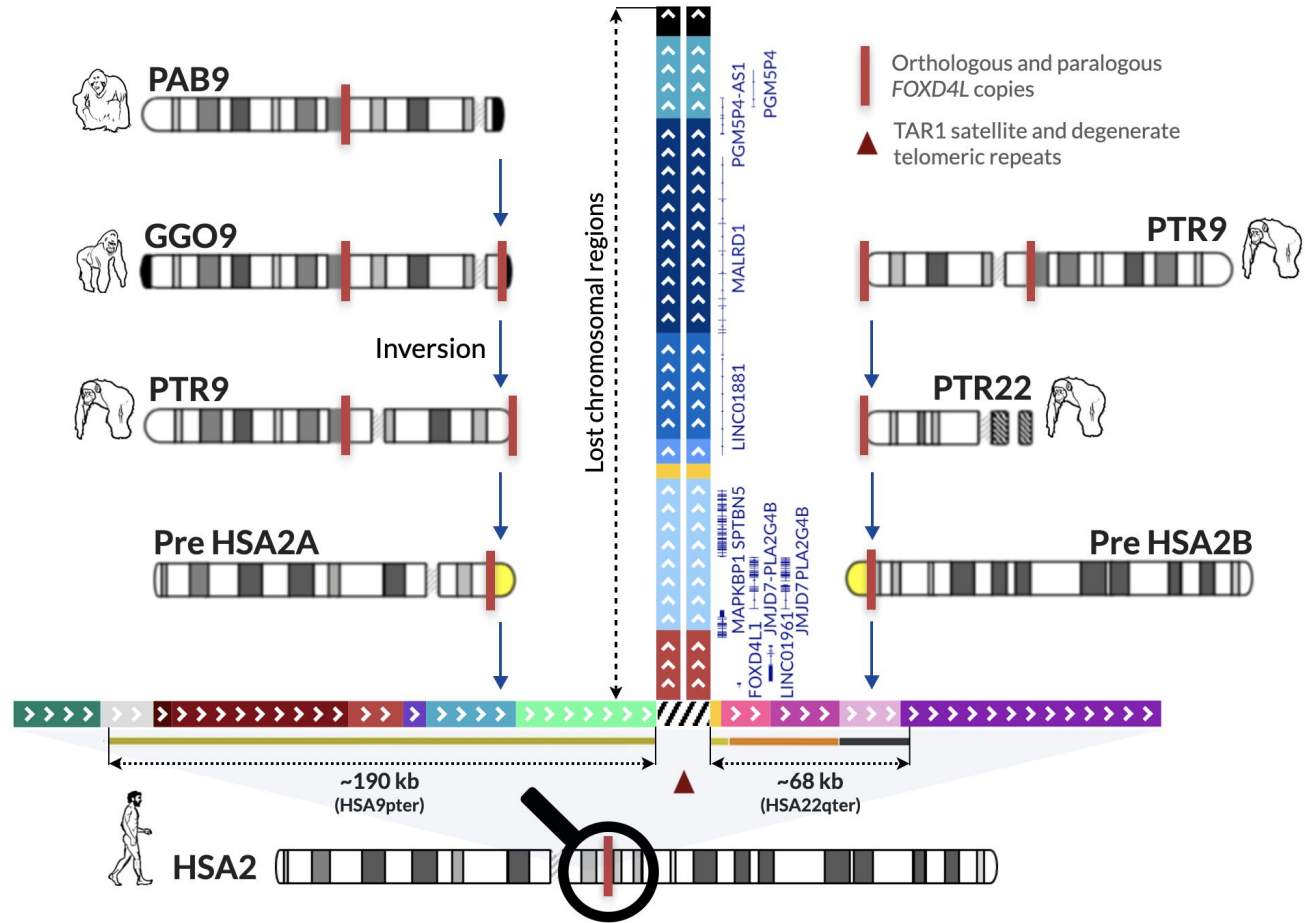


Poszewiecka, Barbara, et al. "PhaseDancer: a novel targeted assembler of segmental duplications unravels the complexity of the human chromosome 2 fusion going from 48 to 46 chromosomes in hominin evolution." *Genome Biology* 24.1 (2023):205.



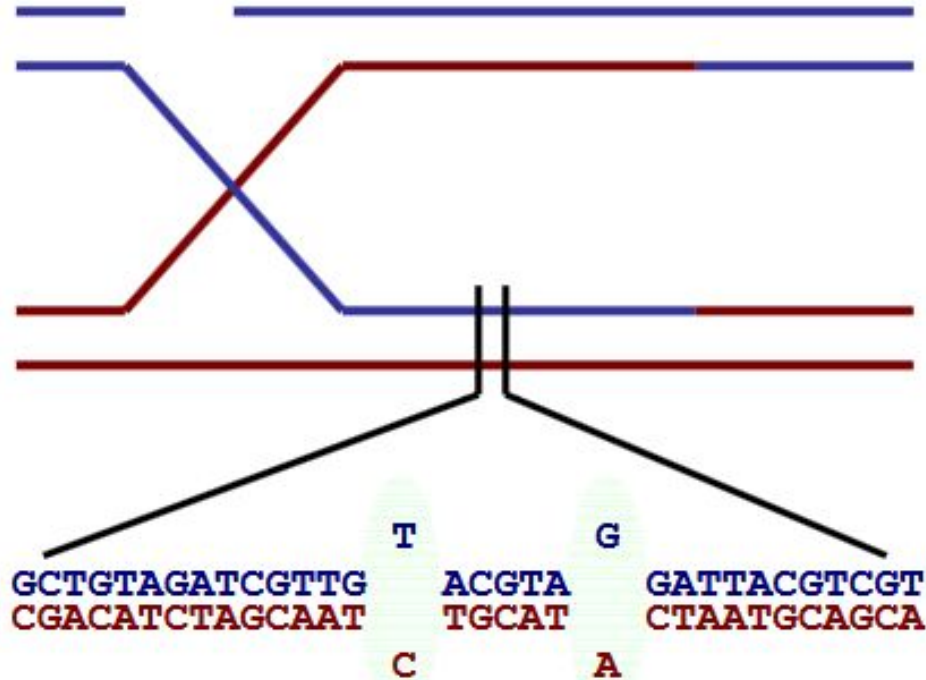
Poszewiecka, Barbara, et al.
 "PhaseDancer: a novel targeted assembler of segmental duplications unravels the complexity of the human chromosome 2 fusion going from 48 to 46 chromosomes in hominin evolution."
 Genome Biology 24.1 (2023): 205.





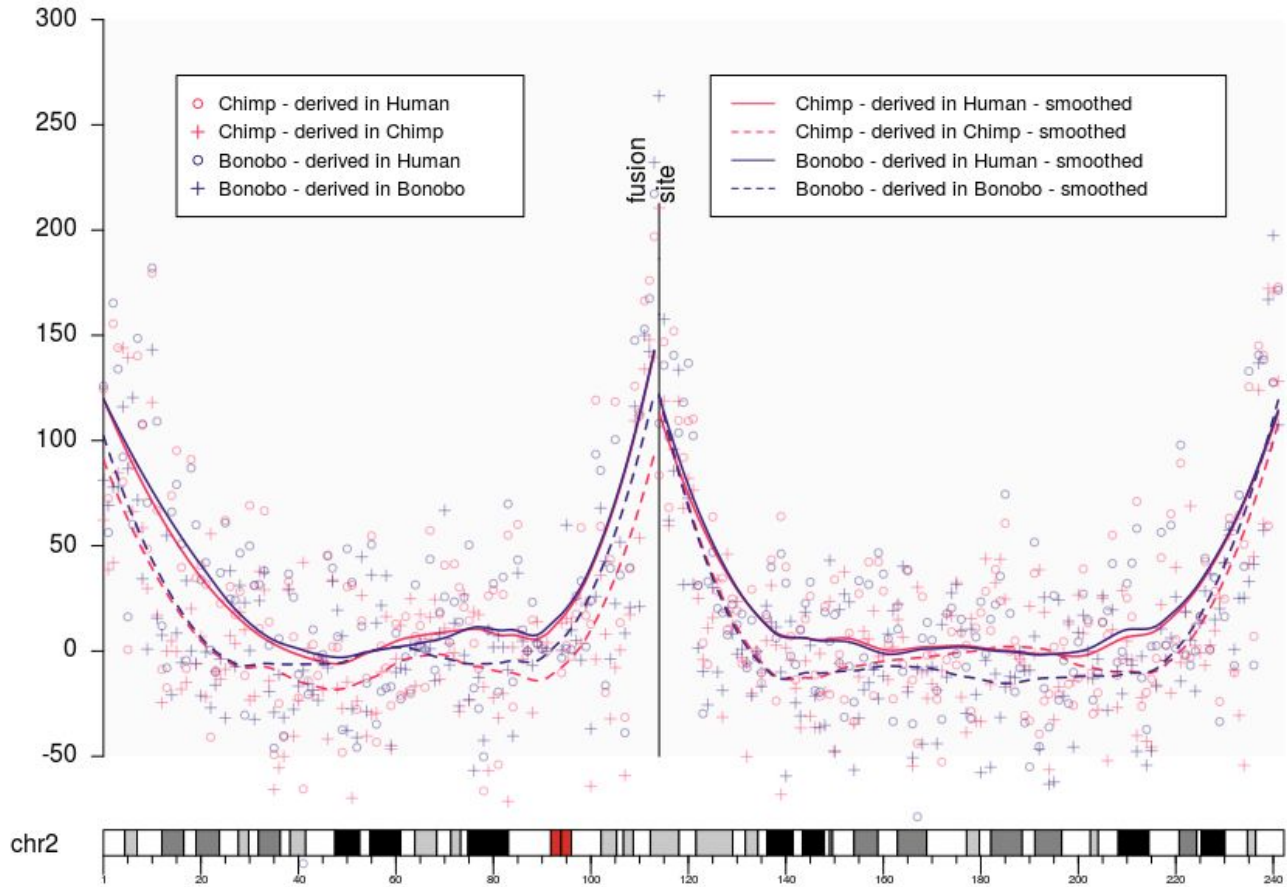
Poszewiecka, Barbara, et al.
 "PhaseDancer: a novel targeted assembler of segmental duplications unravels the complexity of the human chromosome 2 fusion going from 48 to 46 chromosomes in hominin evolution."
 Genome Biology 24.1 (2023): 205.

Biased Gene Conversion: Mismatched SNP Repair During Recombination



Both mismatches are converted to "strong" G-C pairs, replacing "weak" SNPs.

This phenomenon occurs during recombination (Strathern et al., 1995) and is a consequence of the more frequent use of strongly stable nucleotide pairs (G, C) than weakly stable ones (A, T) in heterozygous sites in heteroduplex DNA in the process of DNA repair.



Poszewiecka, Barbara, et al.
 "Revised time estimation
 of the ancestral
 human chromosome 2 fusion."
 BMC genomics 23.6 (2022): 1-16.

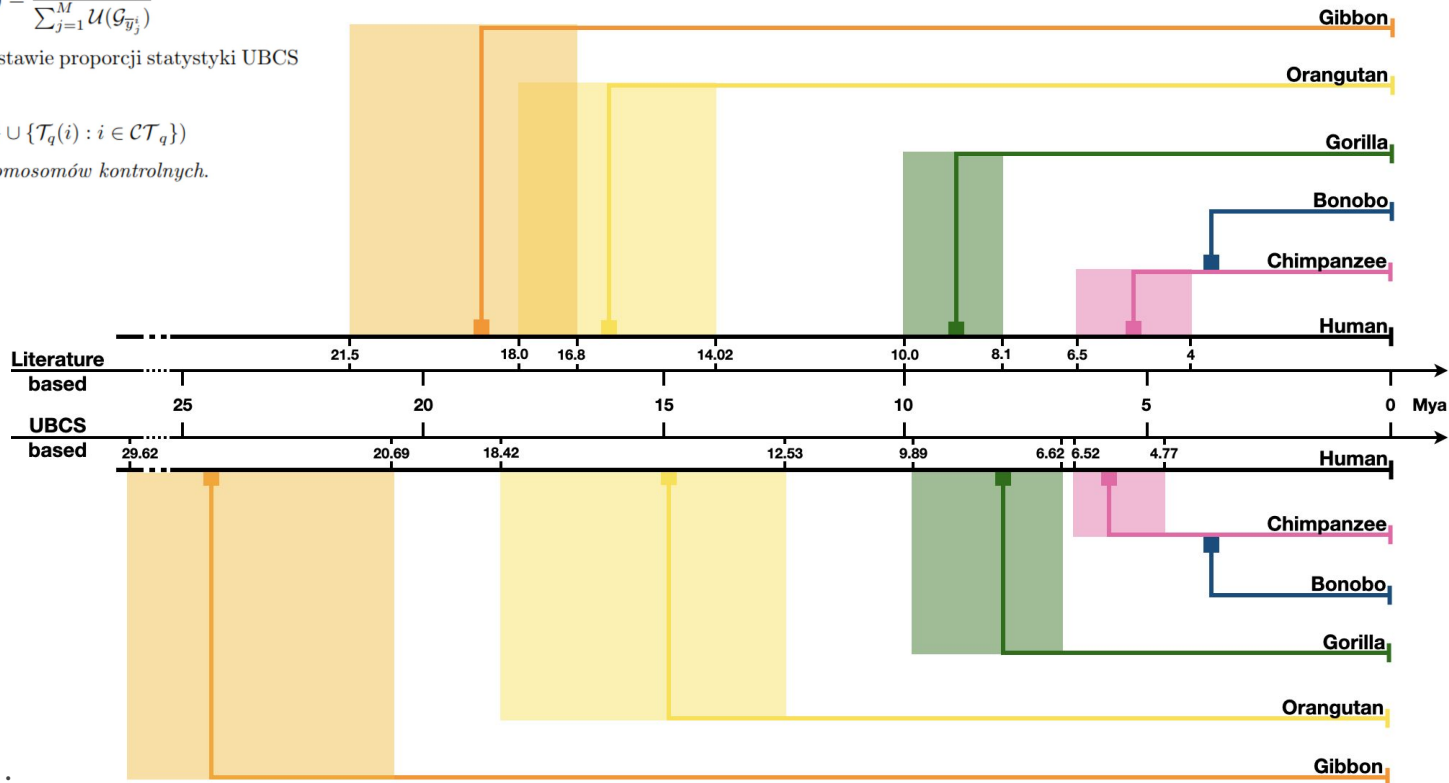
Niech \mathcal{G}_x i \mathcal{G}_y oznaczają dwa genomy o sekwencjach odpowiednio x i y , N liczbę chromosomów i M liczbę okien o wielkości 1 Mb na regionach subteleromowych chromosomów. Określamy $\mathcal{G}_{x_j^i}$ jako j -te okno na i -tym chromosomie w genomie \mathcal{G}_x , wartość statystyki UBCS jako $\mathcal{U}(\mathcal{G}_{x_j^i})$, a \bar{x} jako odwróconą sekwencję x . Obliczamy średnią proporcję statystyki UBCS pomiędzy telomerami na ramionach p i q i -ego chromosomu pomiędzy genomami \mathcal{G}_x i \mathcal{G}_y jako:

$$\mathcal{T}_p(i) = \frac{\sum_{j=1}^M \mathcal{U}(\mathcal{G}_{x_j^i})}{\sum_{j=1}^M \mathcal{U}(\mathcal{G}_{y_j^i})} \quad \mathcal{T}_q(i) = \frac{\sum_{j=1}^M \mathcal{U}(\mathcal{G}_{\bar{x}_j^i})}{\sum_{j=1}^M \mathcal{U}(\mathcal{G}_{\bar{y}_j^i})}$$

a odległość ewolucyjna jest określana na podstawie proporcji statystyki UBCS pomiędzy genomami \mathcal{G}_x i \mathcal{G}_y jako:

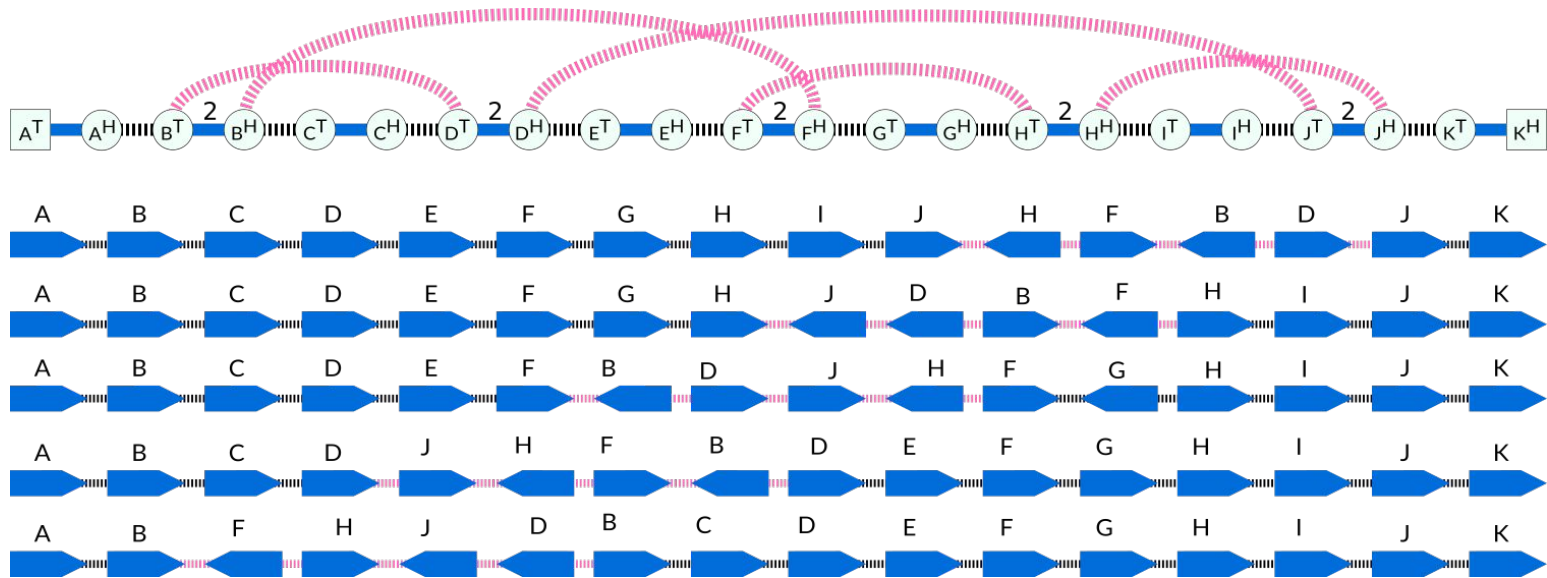
$$\mathcal{G}_x || \mathcal{G}_y = \text{median}(\{\mathcal{T}_p(i) : i \in \mathcal{CT}_p\} \cup \{\mathcal{T}_q(i) : i \in \mathcal{CT}_q\})$$

gdzie \mathcal{CT}_p i \mathcal{CT}_q to zbiór tak zwanych *chromosomów kontrolnych*.

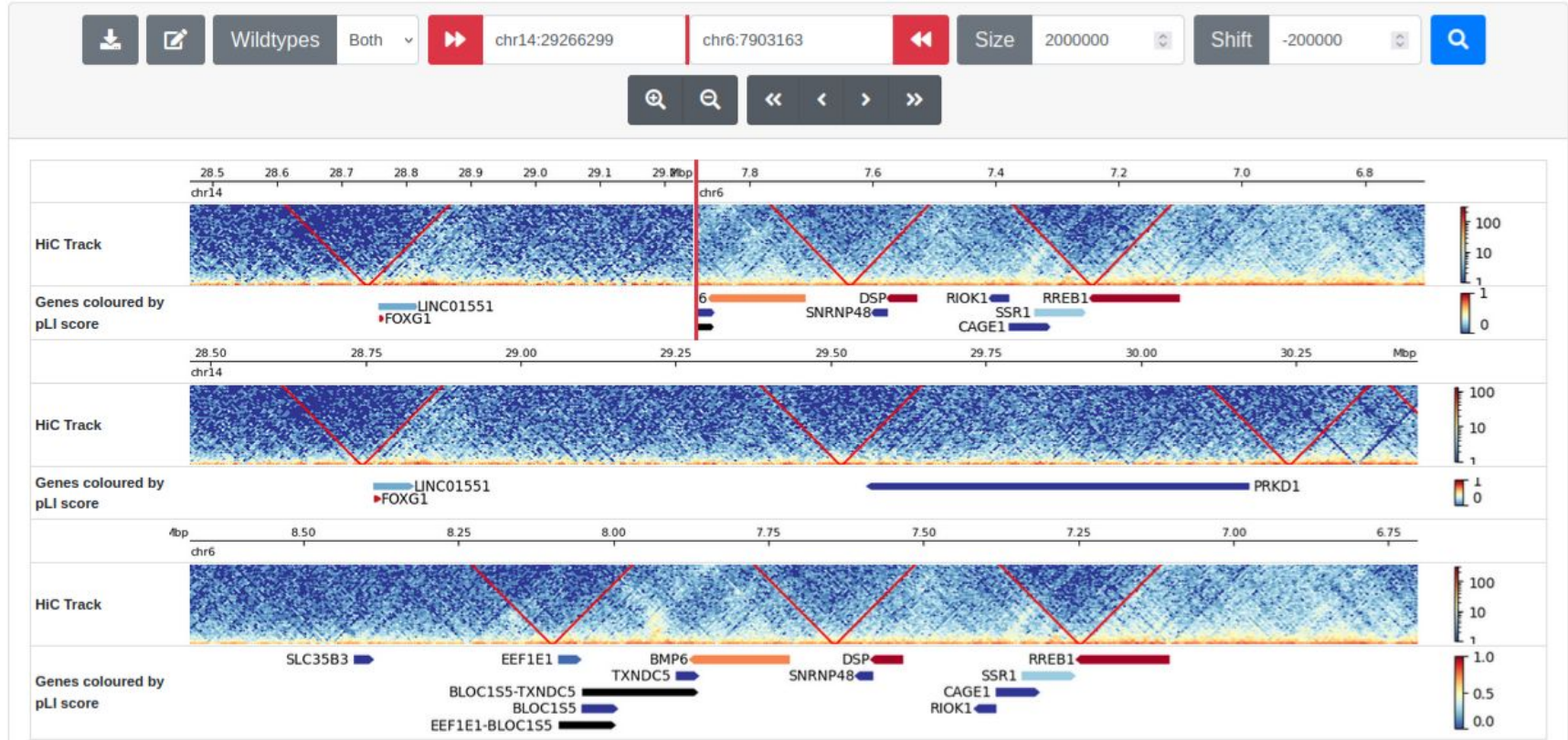


Poszewiecka, Barbara, et al.
 "Revised time estimation
 of the ancestral
 human chromosome 2 fusion."
 BMC genomics 23.6 (2022): 1-16.

Scenarios of complex chromosomal rearrangement in a patient from the study by Nazaryan-Petersen et al. (2018)



TADeus2 - genome browser in breakpoint view mode



"TADeus2: a web server facilitating the clinical diagnosis by pathogenicity assessment of structural variations disarranging 3D chromatin structure." *Nucleic Acids Research* 50.W1 (2022): W744-W752.

Main functionality offered by application website TADeus2

"TADeus2: a web server facilitating the clinical diagnosis by pathogenicity assessment of structural variations disarranging 3D chromatin structure."
 Nucleic Acids Research 50.W1 (2022): W744–W752.

TADeus2

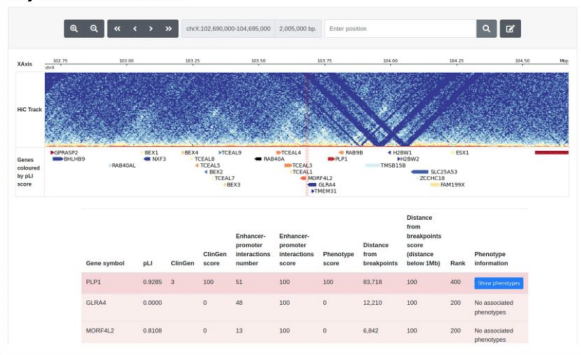
<https://tadeus2.mimuw.edu.pl>

a web server facilitating the clinical diagnosis by pathogenicity assessment of structural variations disarranging 3D chromatin structure

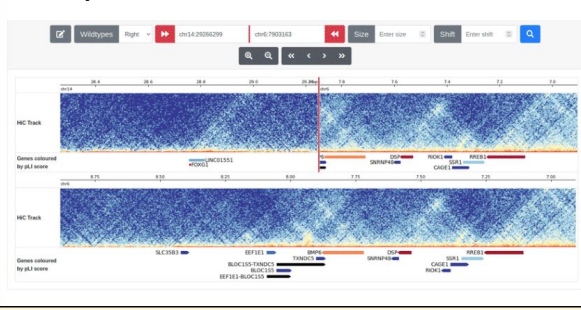


Visualization

Syntenic mode



Breakpoint mode



Datasets

Ontologies

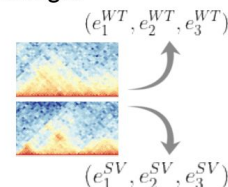


Genomic data and annotations



Evaluation

Chromosome conformation changes

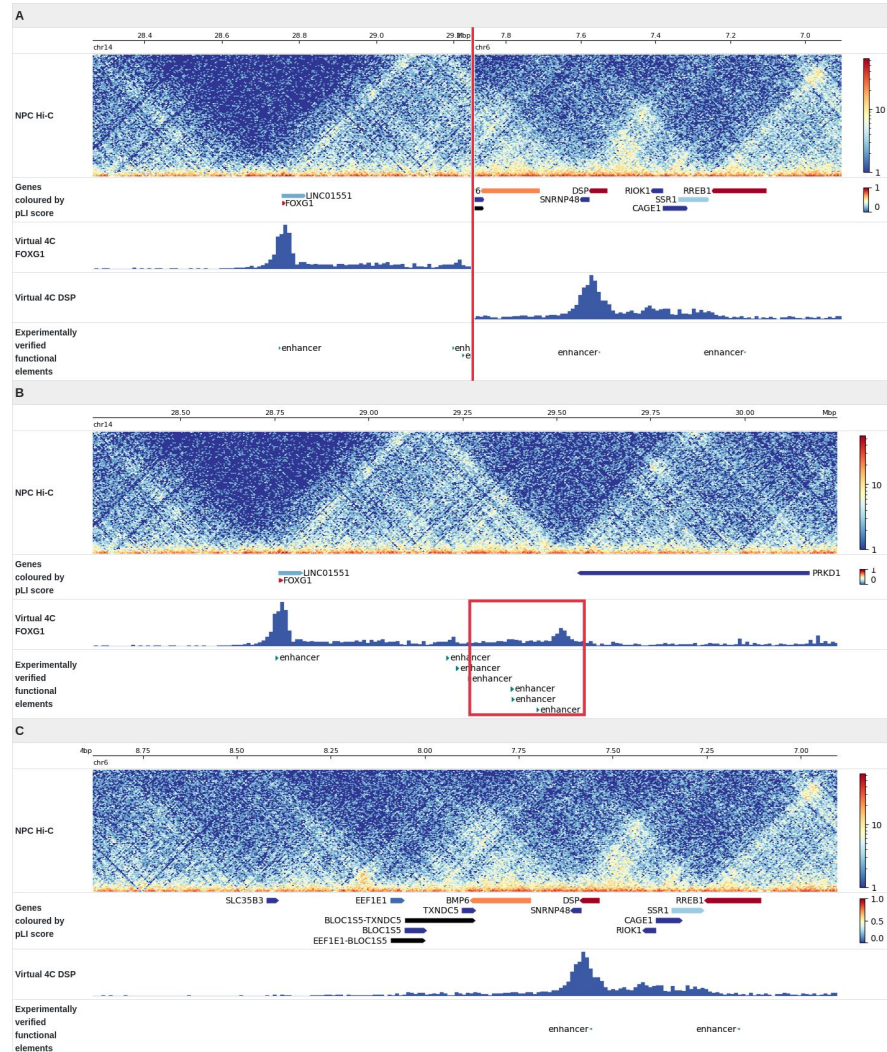


Gene ranking

HDAC1	score1	↑↑↑
TXLNA	score2	↑↑↑
...		
HCRTR1	score3	↑↑
PUM1	score4	↑↑
...		
SDC3	score5	↑
YARS1	score6	↑
...		

Pathogenicity





"TADeus2: a web server facilitating the clinical diagnosis by pathogenicity assessment of structural variations disarranging 3D chromatin structure."
 Nucleic Acids Research 50.W1 (2022): W744-W752.