

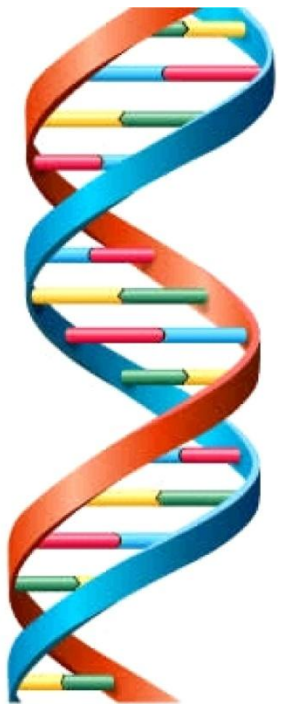
Heterozygous Genome Display

A decorative graphic consisting of a solid teal horizontal bar that spans the width of the slide. Below this bar, on the right side, there are three thin, parallel white horizontal lines that extend to the right edge of the slide.

Outline

- Background: Biology
- Alignment Approach
- Synteny Approach

Background: DNA

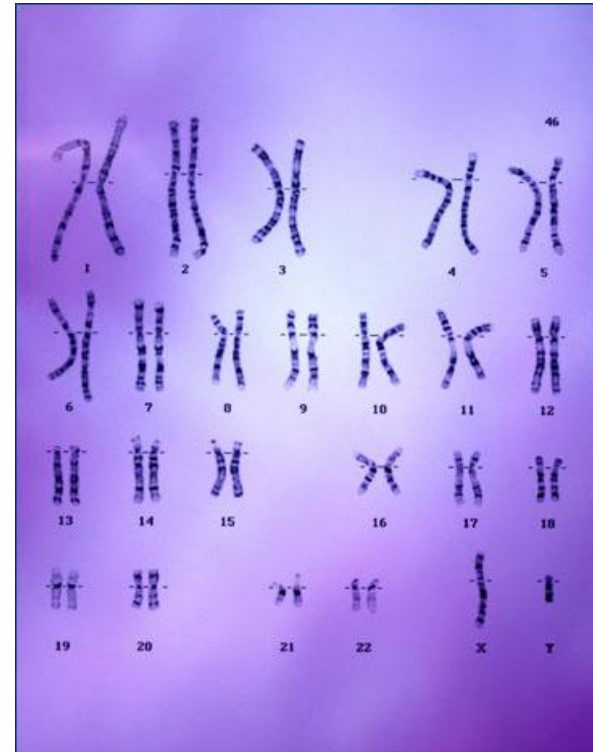


Yellow < A
Green > T
Blue < C
Red > G



Background: Definitions

- **Diploid:** having two copies of DNA
- **Homozygous:** both copies are the same
- **Heterozygous:** the two copies are different
- **Polyploidy:** having multiple copies of DNA (commonly 2, 4, or 6).
- **Variant:** A change in the DNA
- **Haplotype:** A set of variants that are in the same copy of DNA.



Background: Variants

- SNPS: Single nucleotide change (ACT -> ATT)
- Insertions/Deletions: Changes that add or remove bases (ATT -> AT)
 - Variable Tandem Repeats: change in the number of repeats (ACTACT -> ACTACTACT)
- Rearrangements: Changes that move sections of DNA to a new location (TAAT -> TAA)
- Inversions: Changes that switch the direction of the DNA (ACTT -> TTCA)

Background: Assembly

ACTGG**C**AACAAGCTGCCGCGCGT**T**TACAGT
ACTGGGAACAAGCTGCCGCGCGT**T**TACAGT



Sequencing

CGCGT**T**T

GAACAAGCTGCCGC

ACTGG**C**

TGGGAAC

GTTTACAGT



Assembly

ACTGG**C**

GTTTACAGT

TGGGAAC

CGCGT**T**T

GAACAAGCTGCCGC



ACTGGGAACAAGCTGCCGCGCGT**T**TACAGT

C

Heterozygous Genome Display

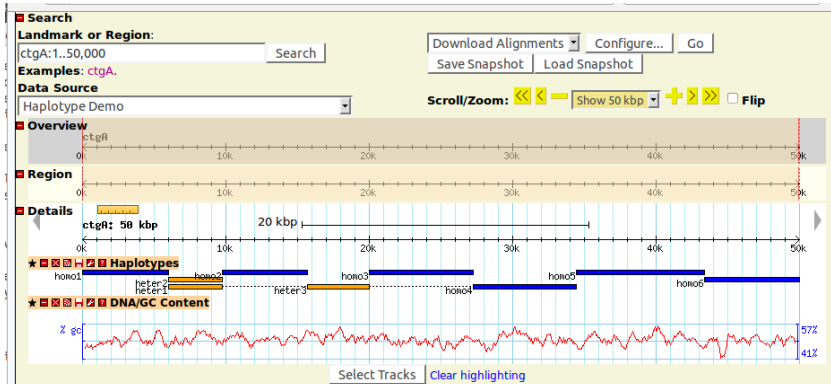
- What if our sequence is very heterozygous. As an extreme, what if it is so heterozygous that we can't form a consensus sequence? (Raspberry)

We need a way to display heterozygous genomes.

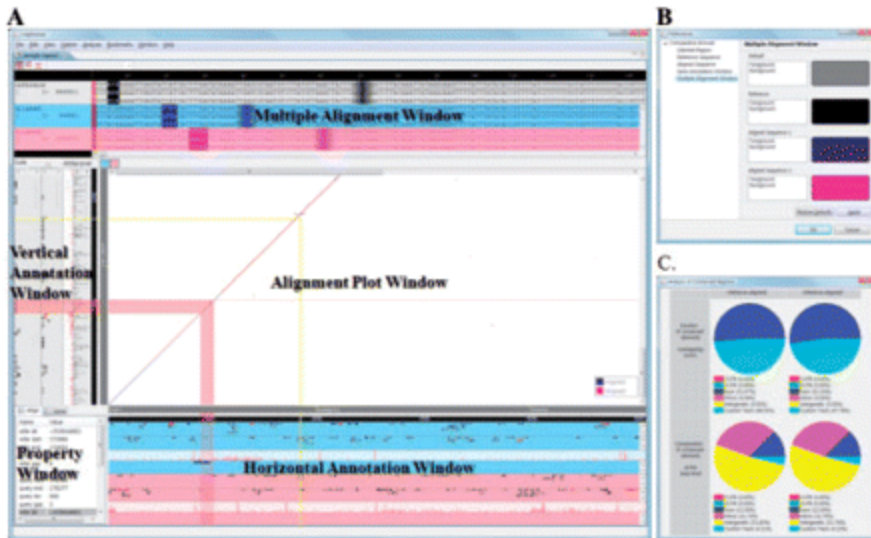
Current Options

Alignment Approach: Highlights Differences

Gbrowse

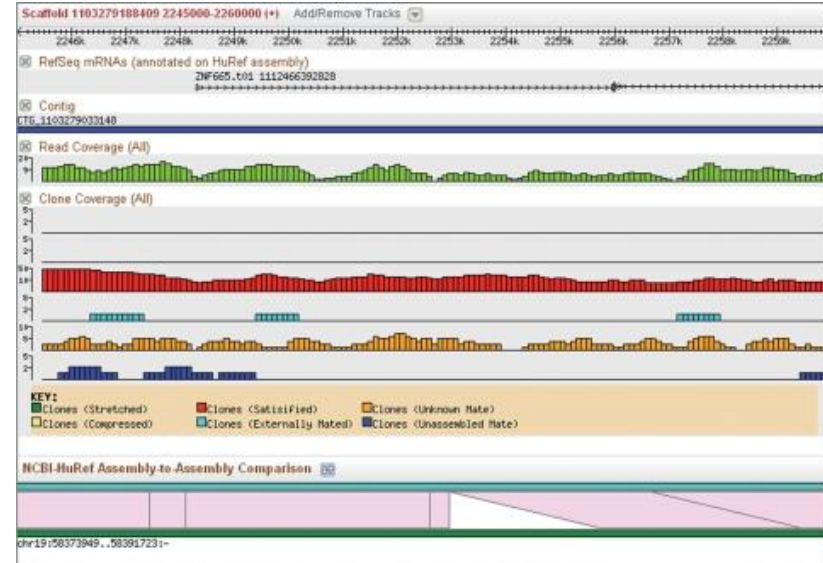


Haplowser

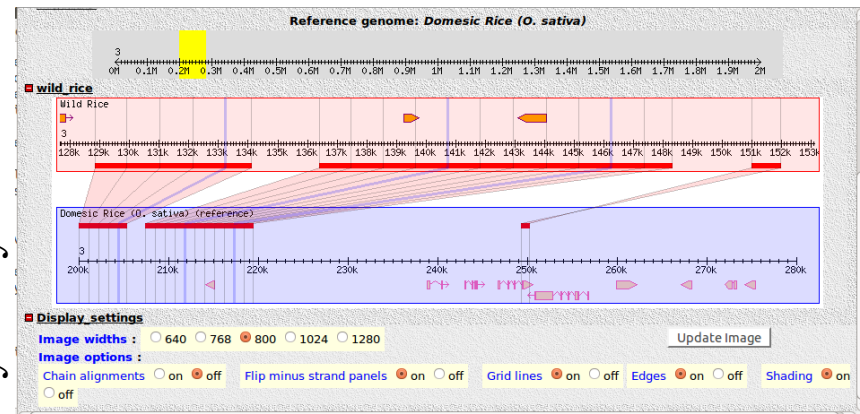


Synteny Approach: Highlights Similarities

HuRef Browser

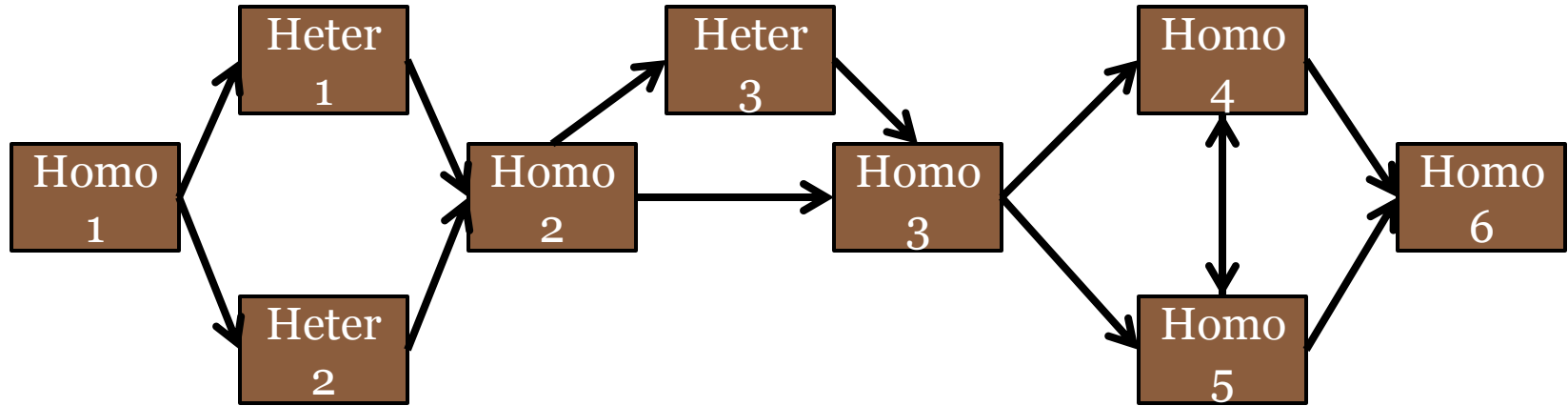


Synteny Browser



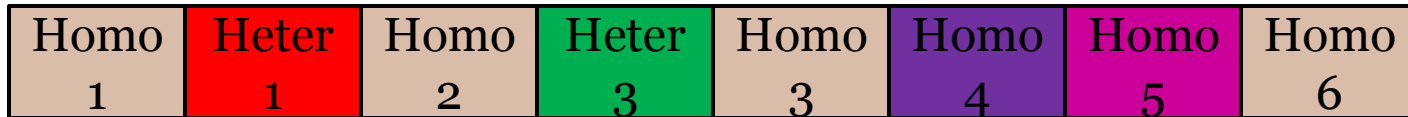
Example

Assembly
Graph

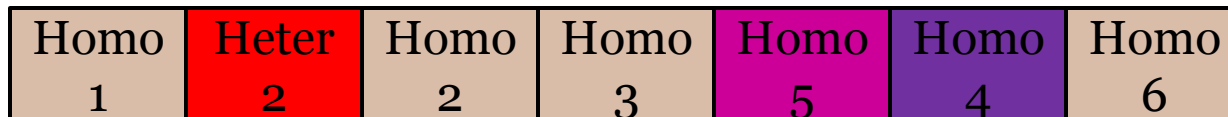


Haplotypes

Insertion



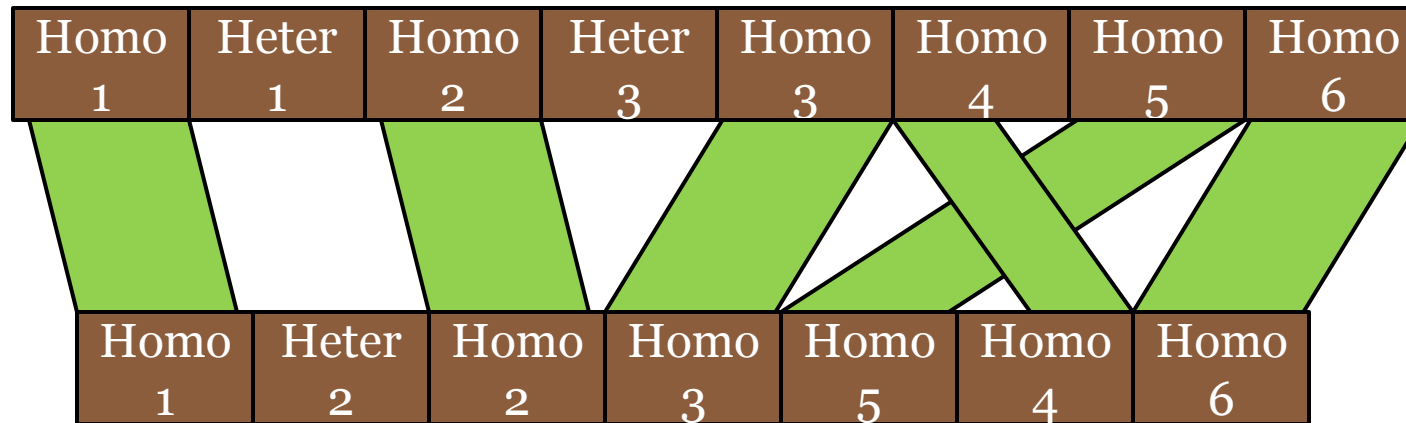
Alternative Allele



Rearrangement

Deletion

Synteny Browser



Pros:

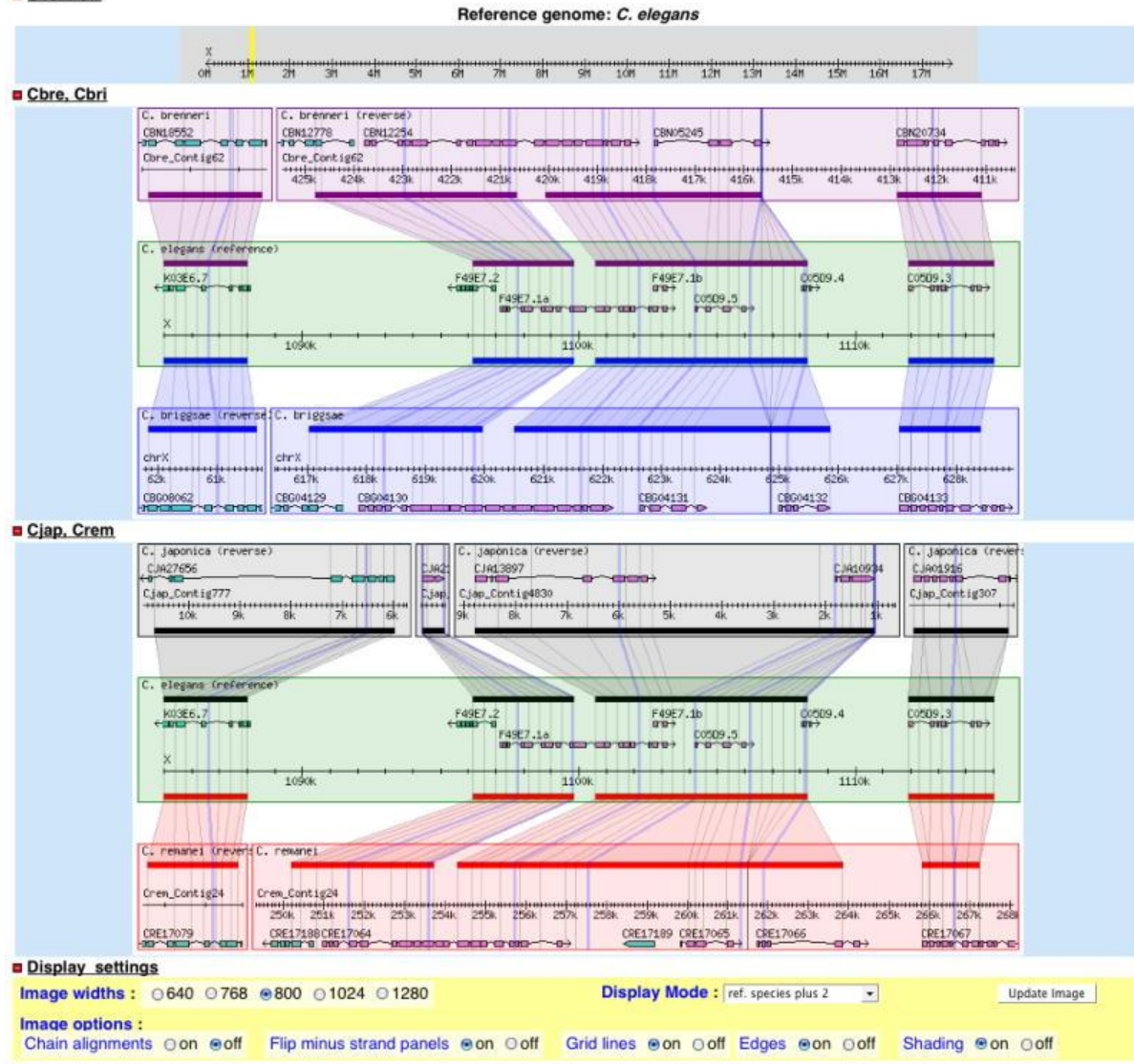
- Displays rearrangements.
- Can display haplotype specific genes.
- Better overview

Cons:

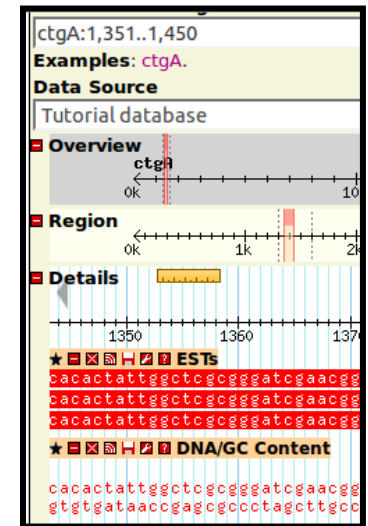
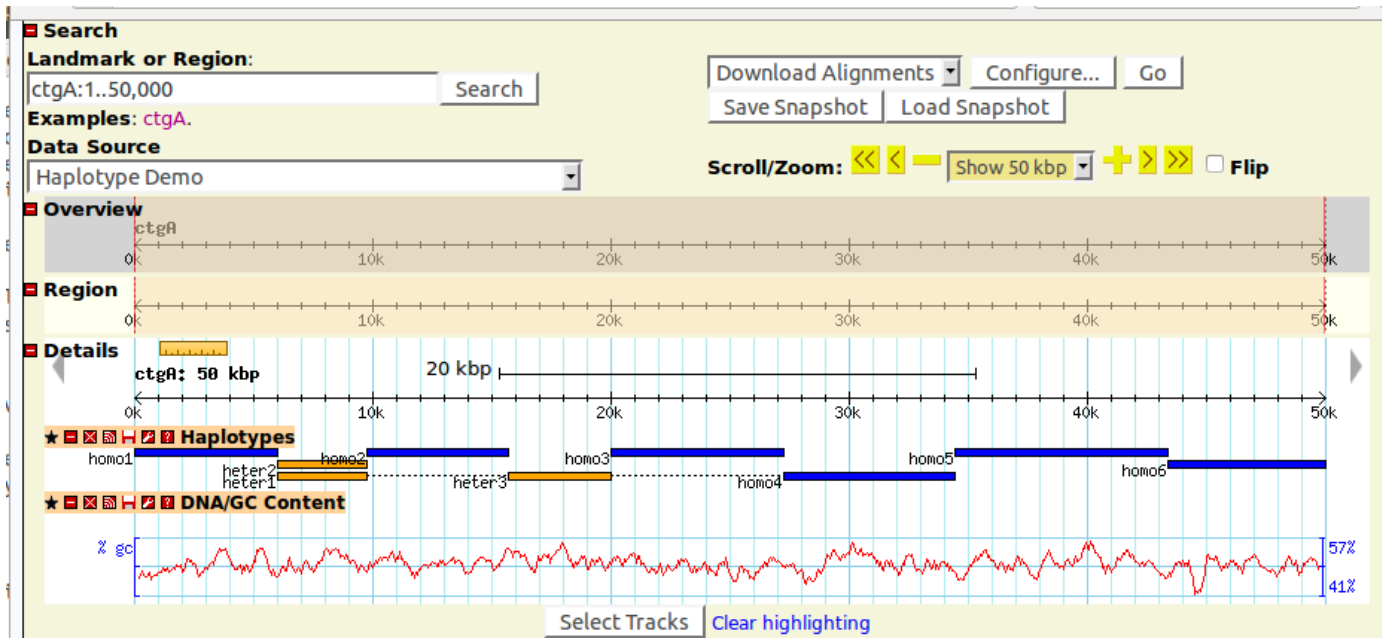
- Requires perfect haplotypes.
- Large homozygosity results in mostly duplicate tracks.
- Polyploidy quickly becomes overwhelming.

Screenshot

Overview



Alignment View



Pros:

- Limits duplicate tracks
- Better for polyploidy
- Handles imperfect assemblies.

Cons:

- No rearrangements or inversions could be shown.
- Haplotype specific genes would be hard to display
- Generates a fake consensus sequence



Questions?