Sequence Surveyor

Leveraging Overview for Scalable Genomic Alignment Visualization

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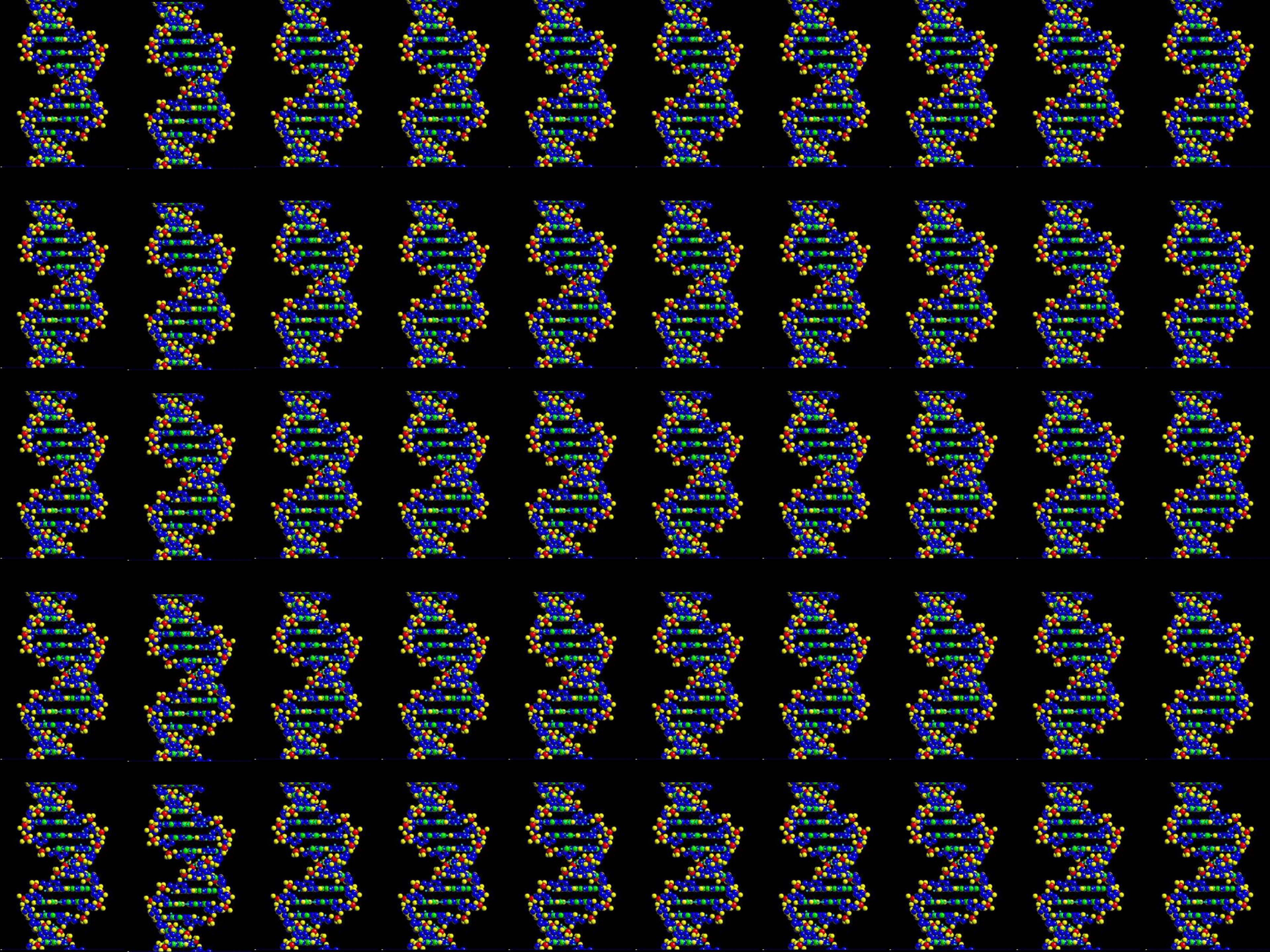
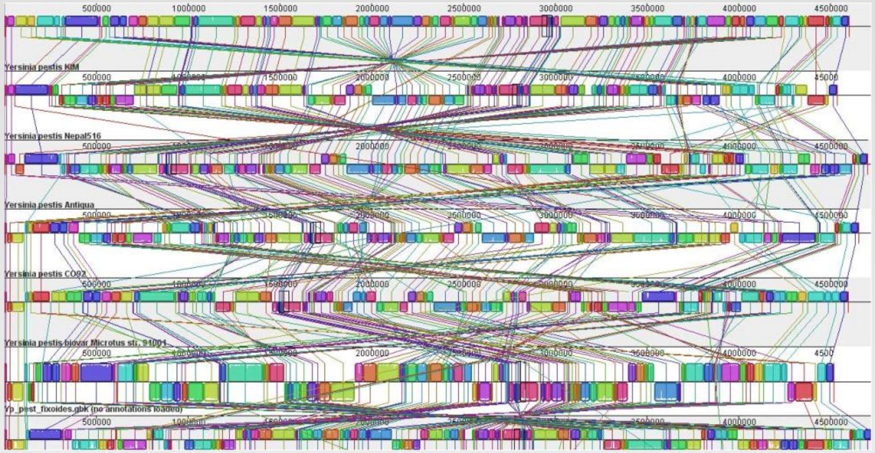
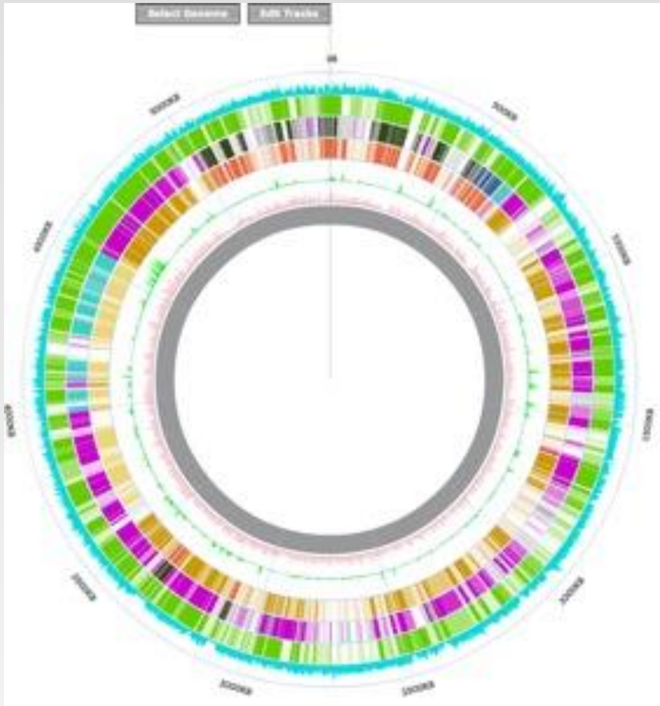
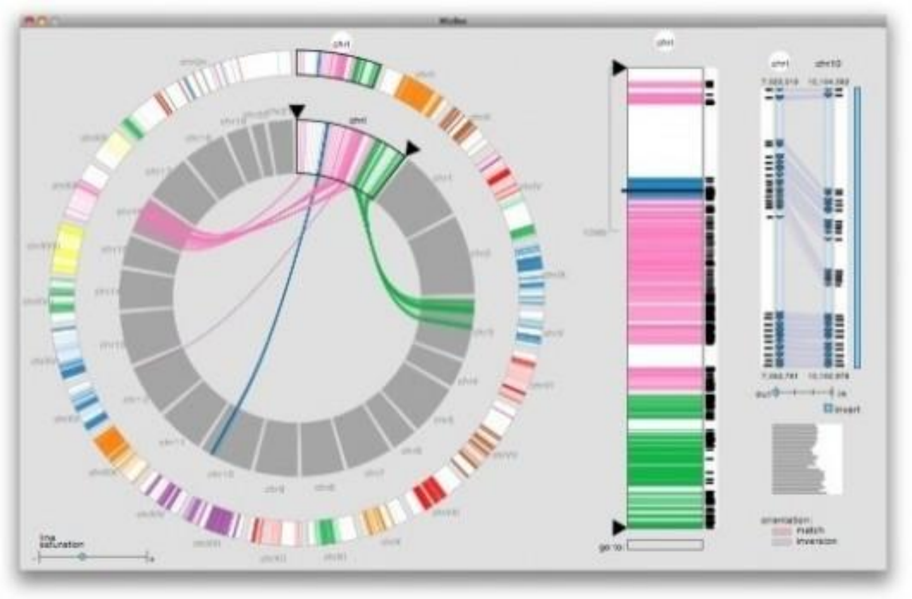
IEEE VisWeek 2011

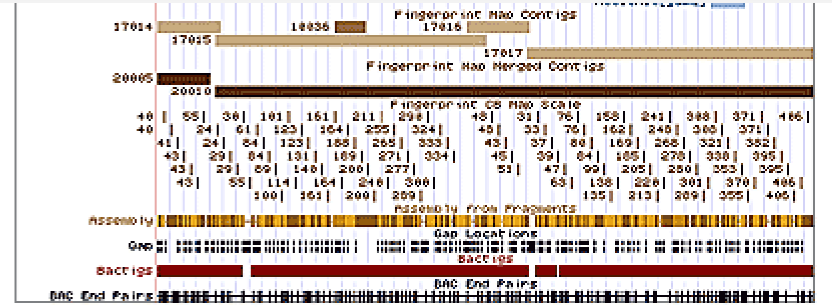


Viewing Genome Alignments



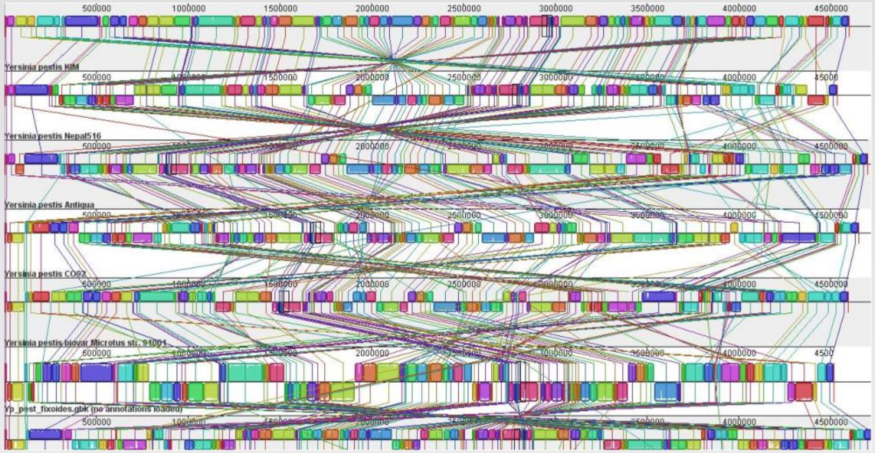


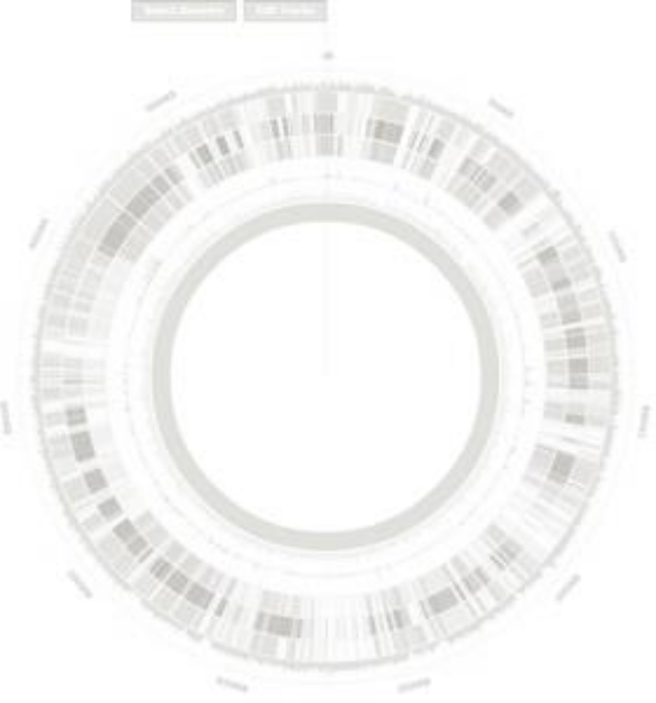


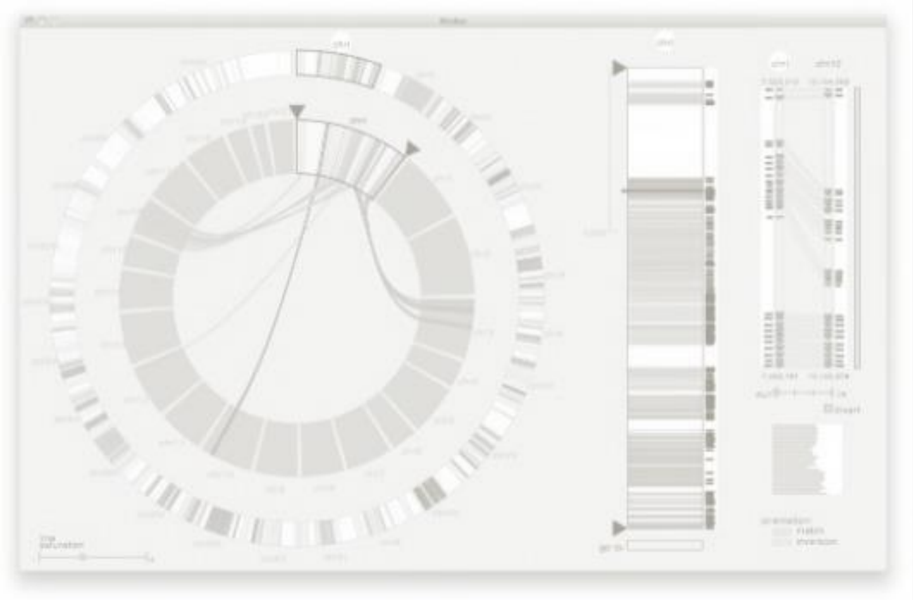


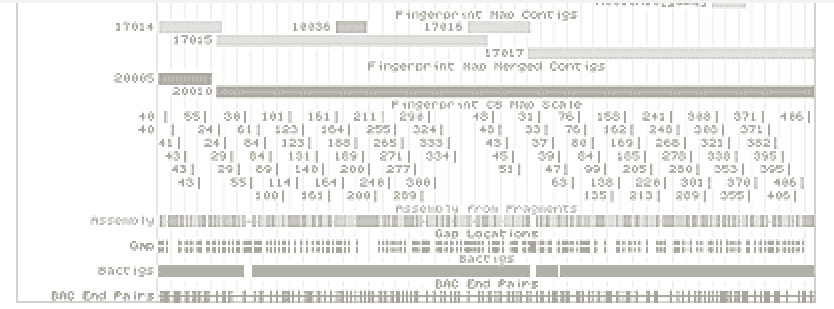
rat fingerprint map tracks | *UCSC Genome Browser*

Viewing Genome Alignments

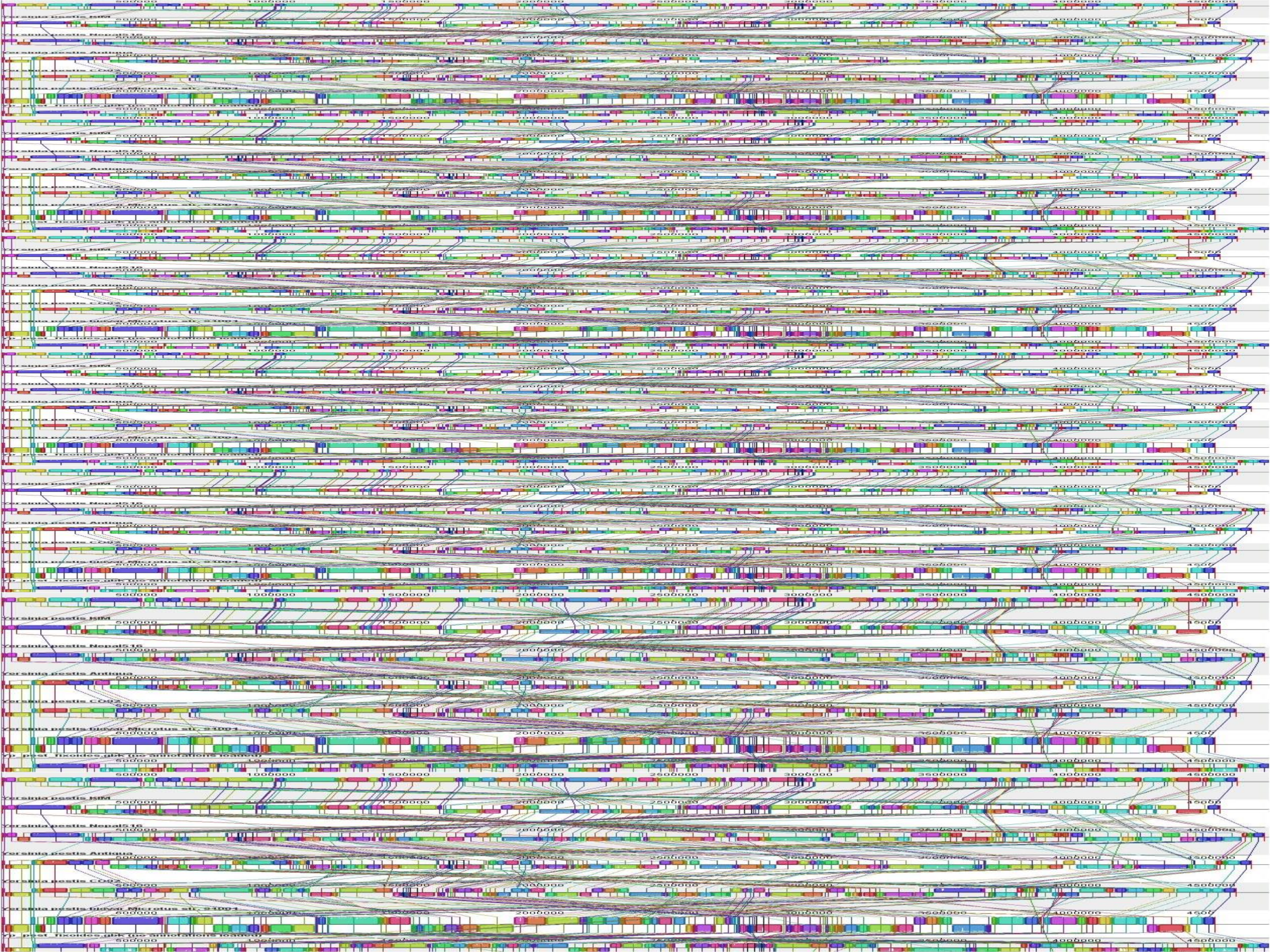








rat fingerprint map tracks | *UCSC Genome Browser*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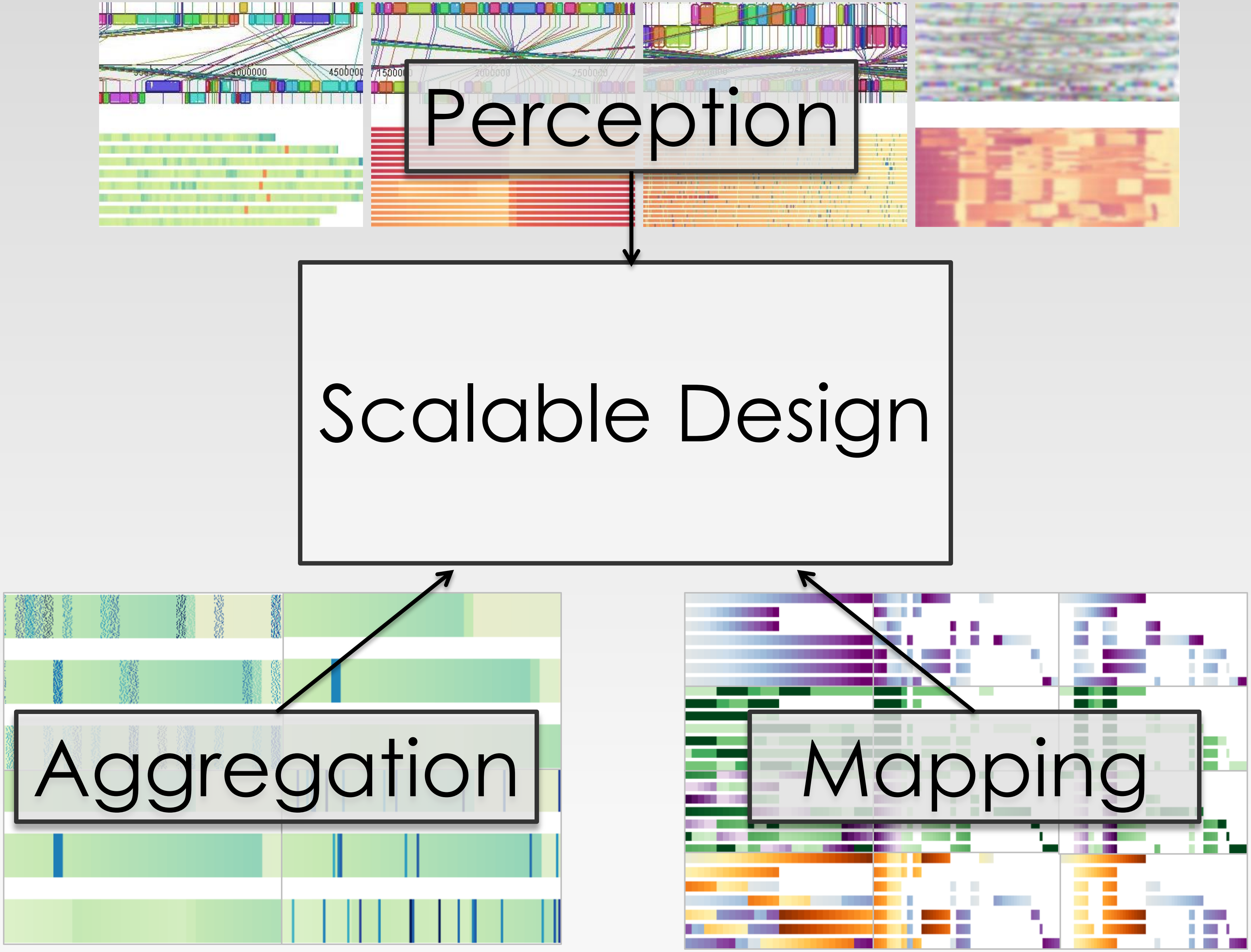
Yersinia pestis KIM
Yersinia pestis Nepal516
Yersinia pestis Antiqua
Yersinia pestis CO92
Yersinia pestis biovar Microtus str. 91001
Y. pest_fixoides.gbk (no annotations loaded)



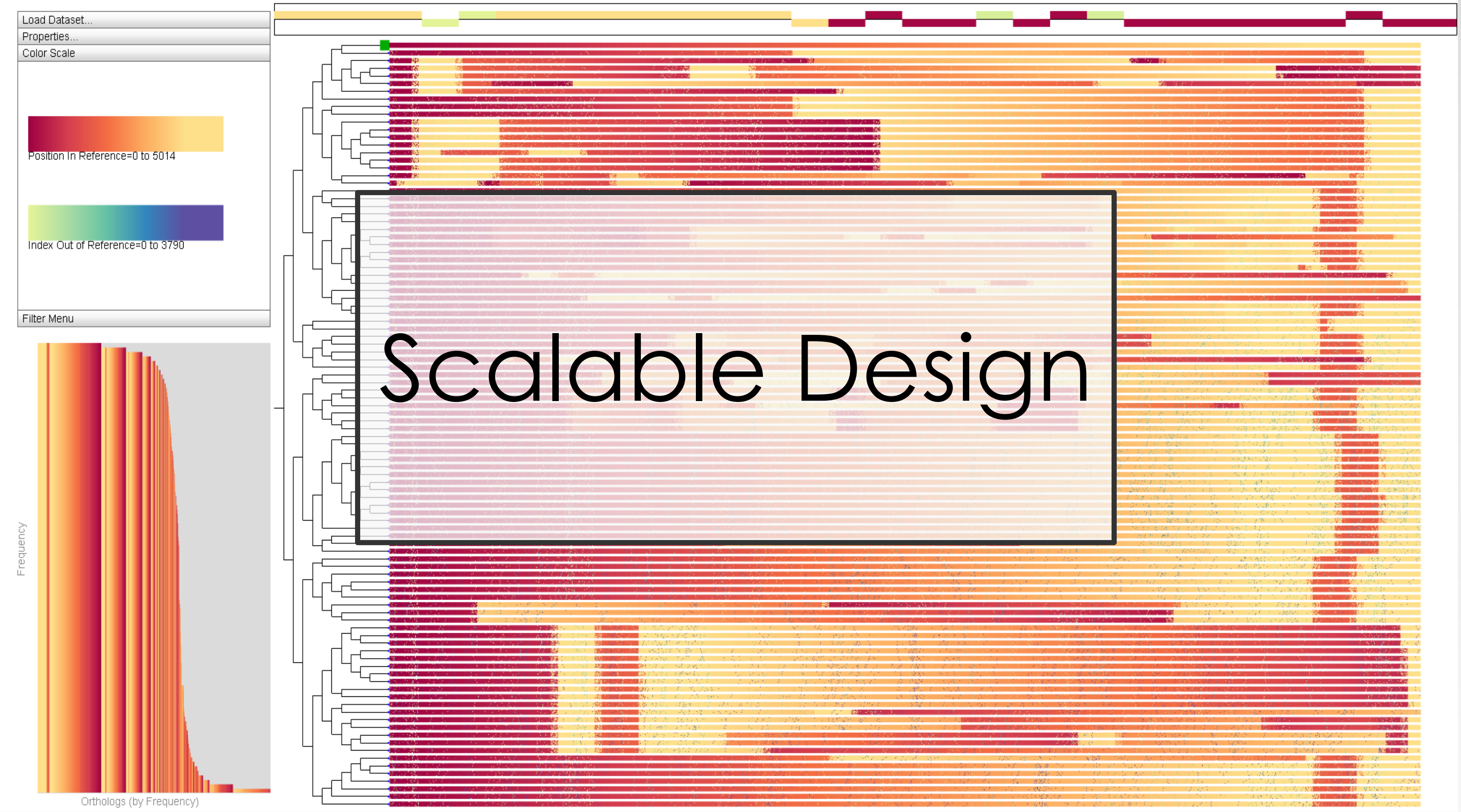
Perception

Scalable Design

Aggregation

Mapping

Scalable Design



Outline

The Data Domain

Sequence Surveyor

Design in Theory

- Perception
- Mapping
- Aggregation

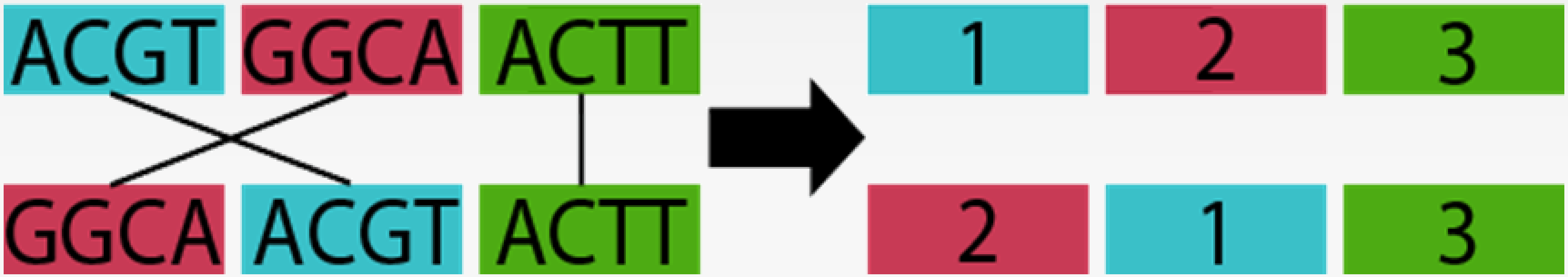
Design in Practice

Whole Genome Alignment

Identify related groups of genes appearing in a set of organisms

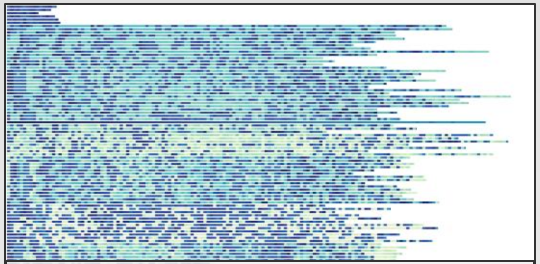
Organism One: ACGTGGCAACTT

Organism Two: GGCAACGTACTT



Defining Scale

Number of Genomes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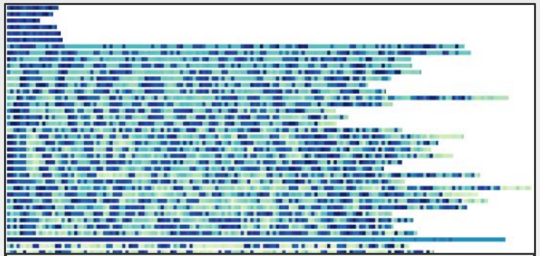
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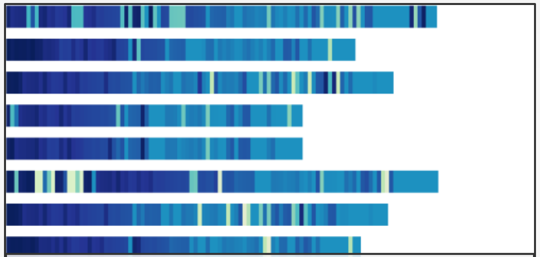
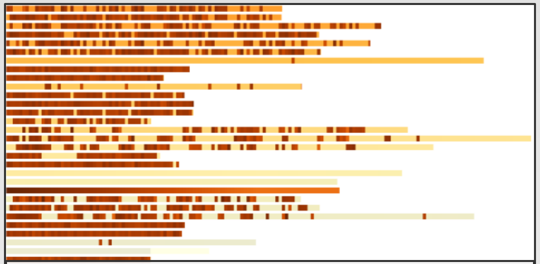
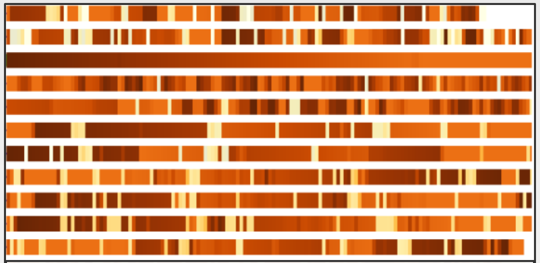
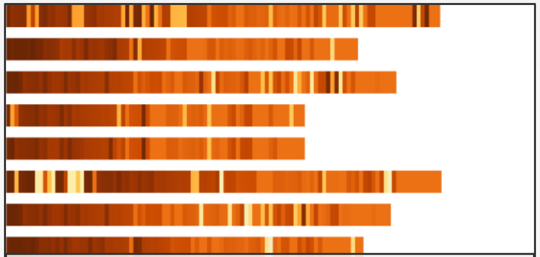

8

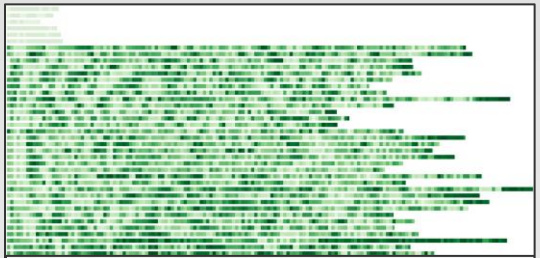
Length of Genomes

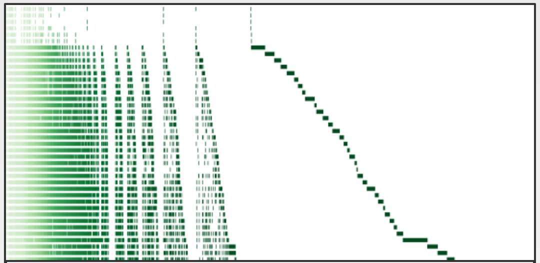

Fungi (17,000+ genes)

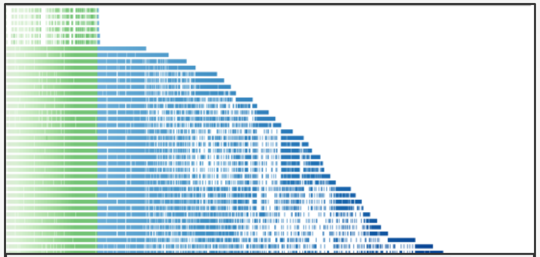

Bacteria (6,000+ genes)


Viral (300+ genes)

Types of Inquiry


Locality


Co-occurrence


Reference-Based

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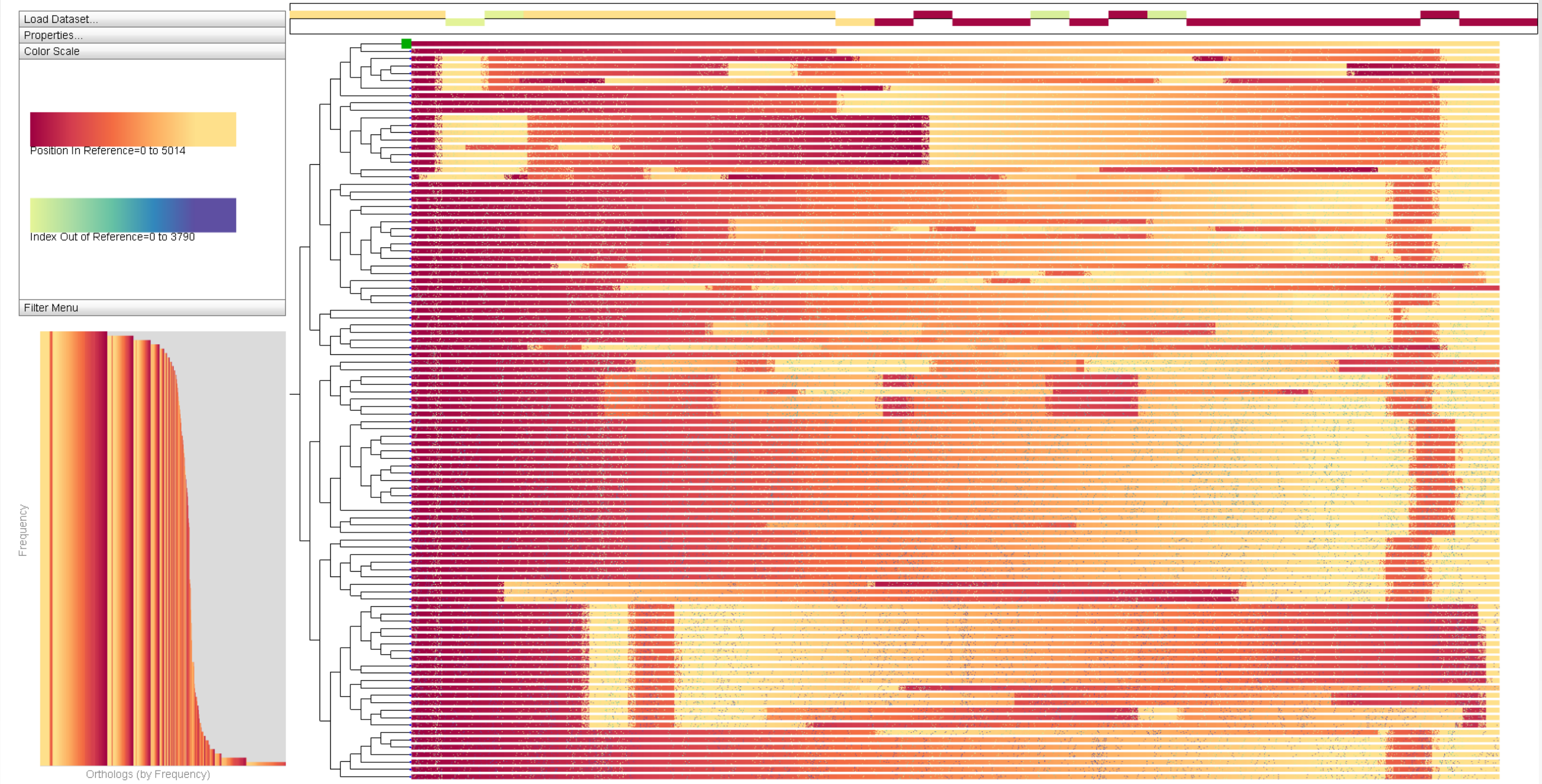
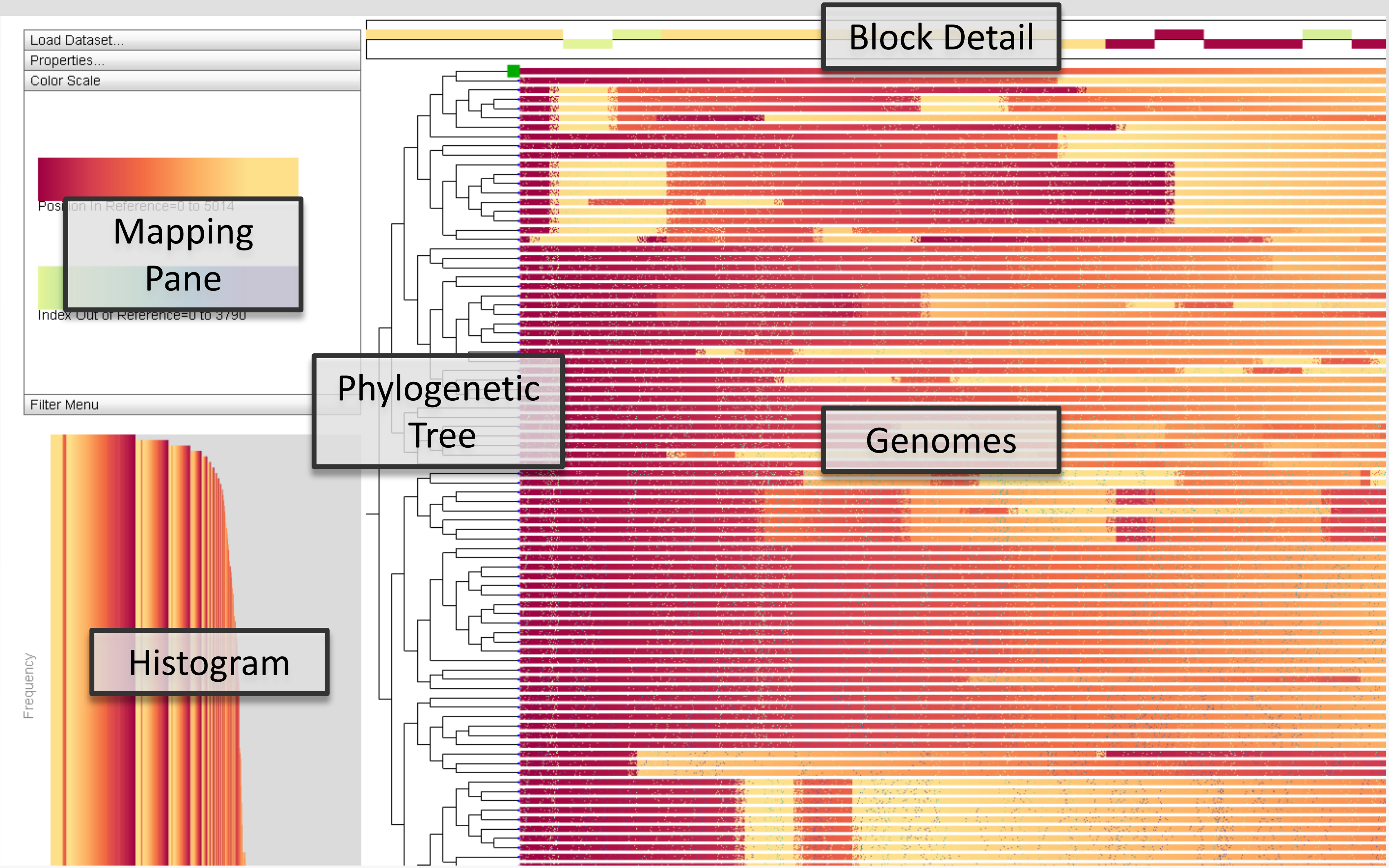
Design in Practice

Our Solution



Our Solution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Properties...
Color Scale
Position In Reference=0 to 5014
Index Out of Reference=0 to 3790
Filter Menu
Frequency

Block Detail

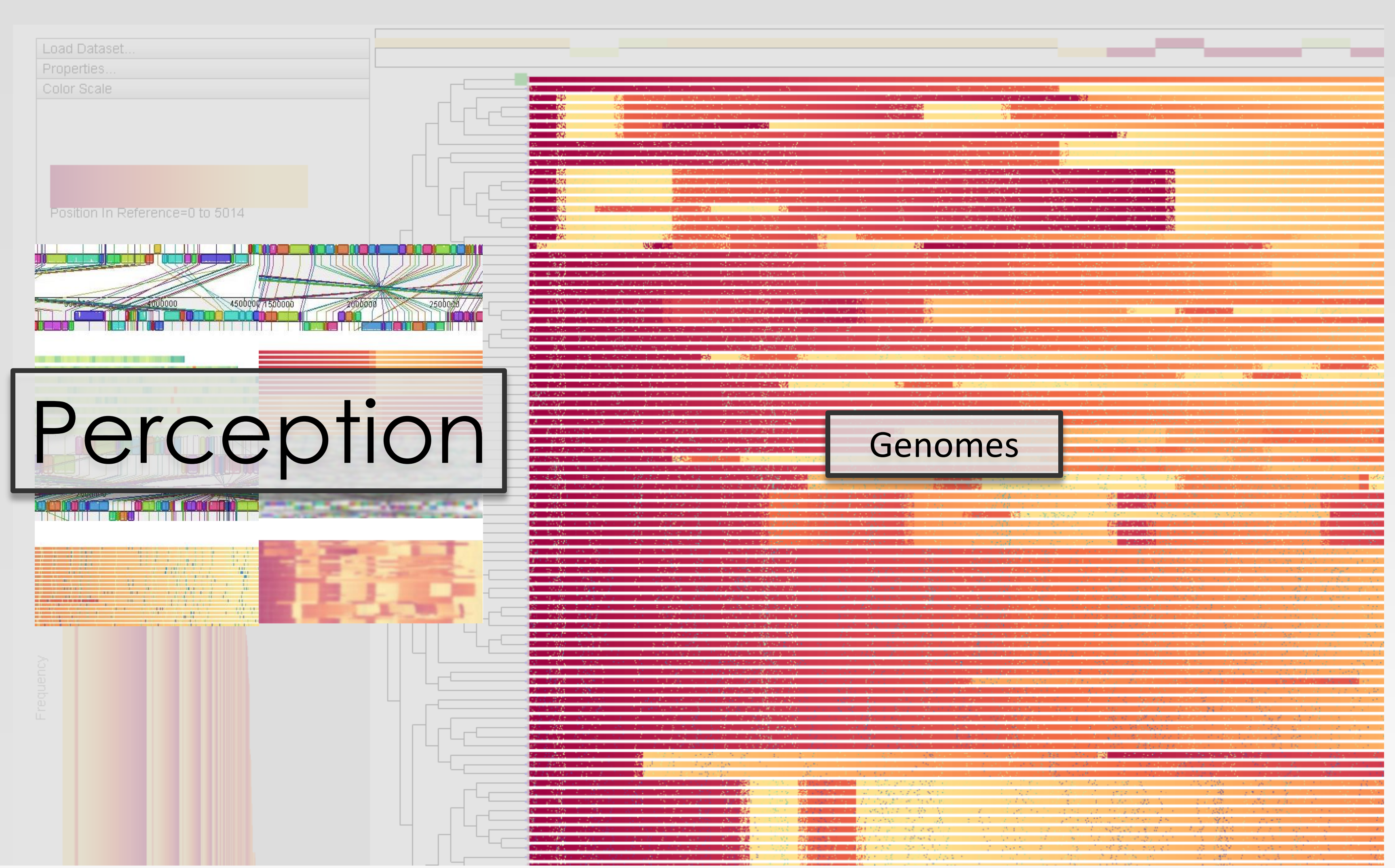
Mapping Pane

Phylogenetic Tree

Genomes

Histogram

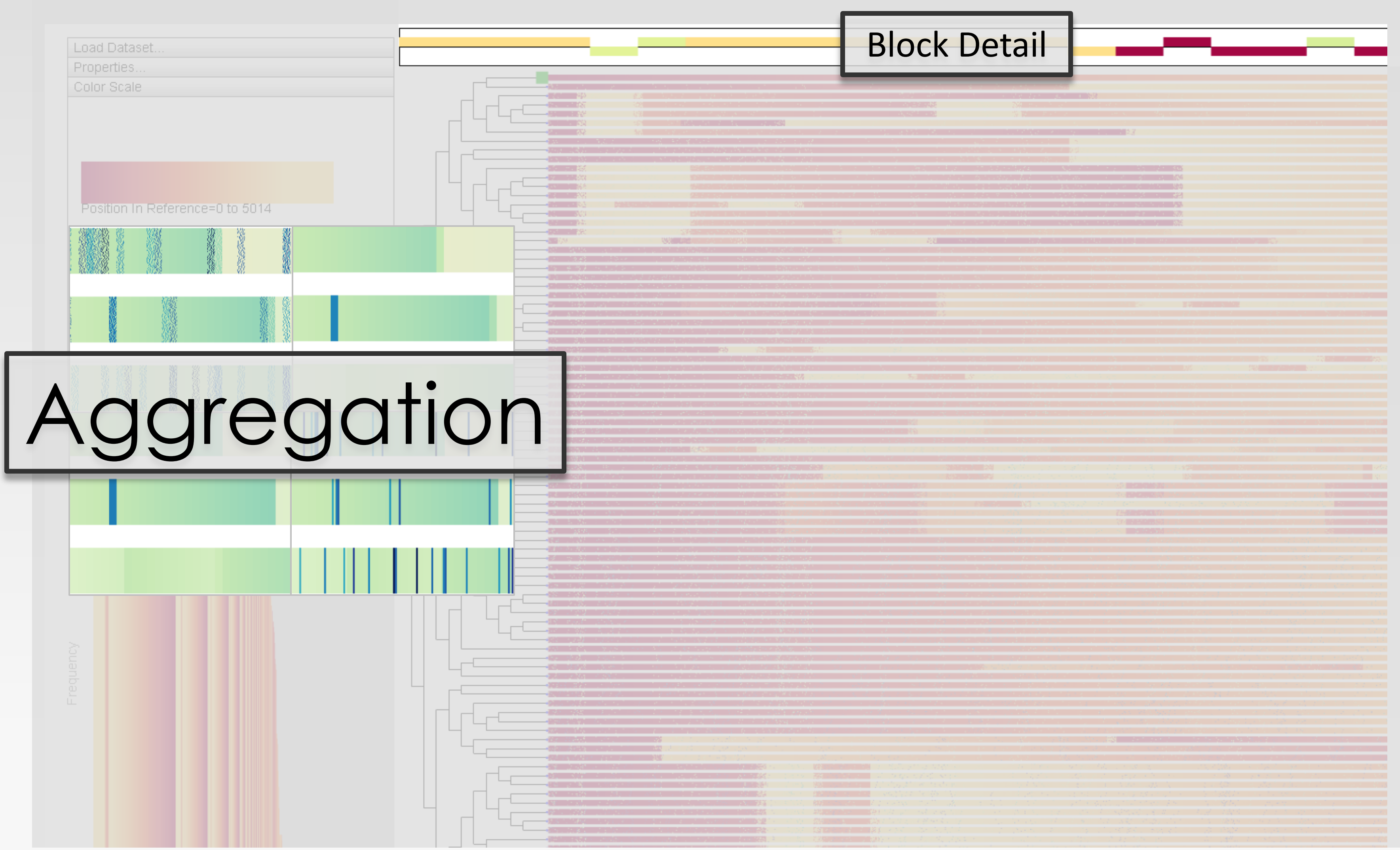
Our Solution



Perception

Genomes

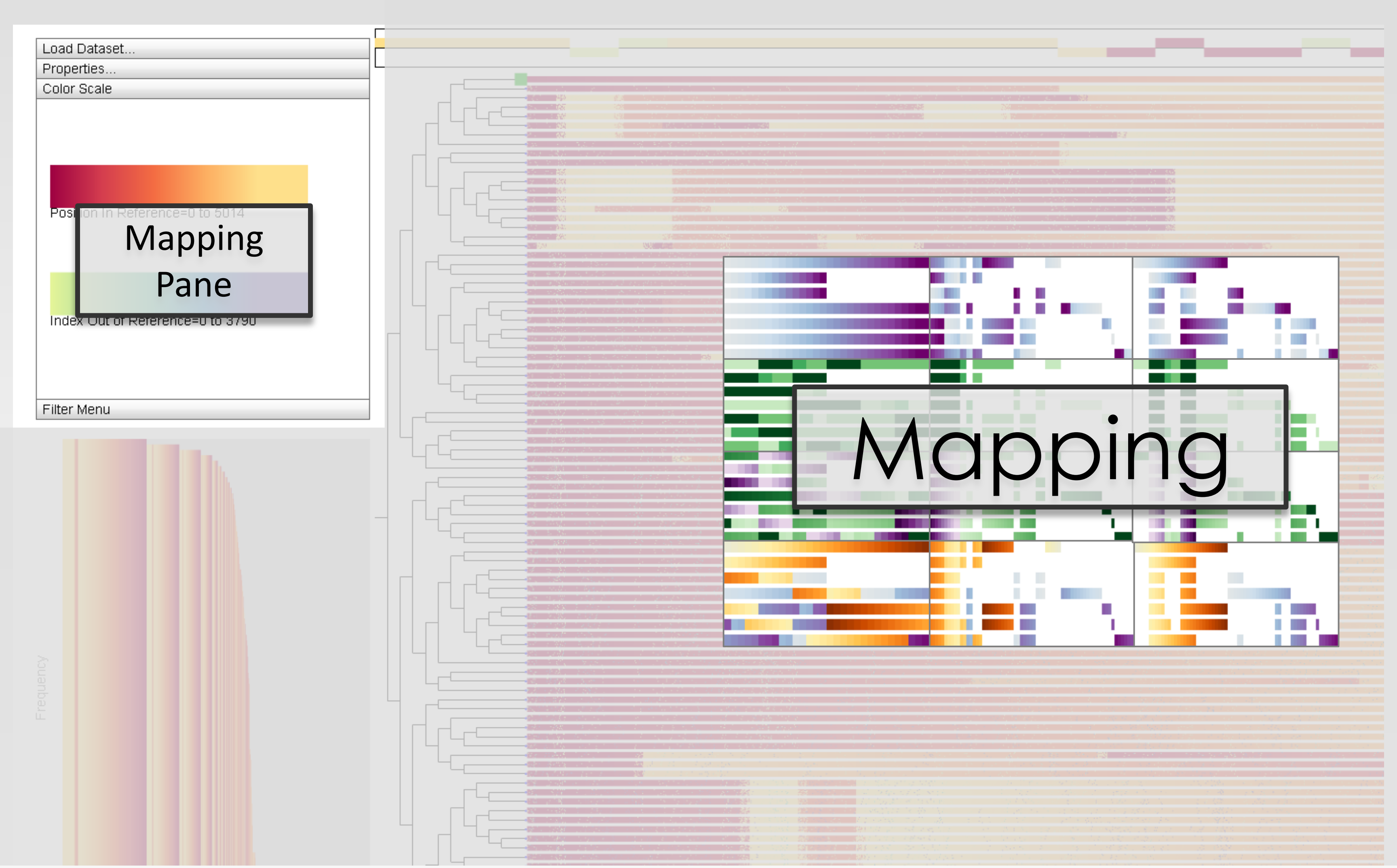
Our Solution



Block Detail

Aggregation

Our Solution


Load Dataset...
Properties...
Color Scale
Position In Reference=0 to 5014
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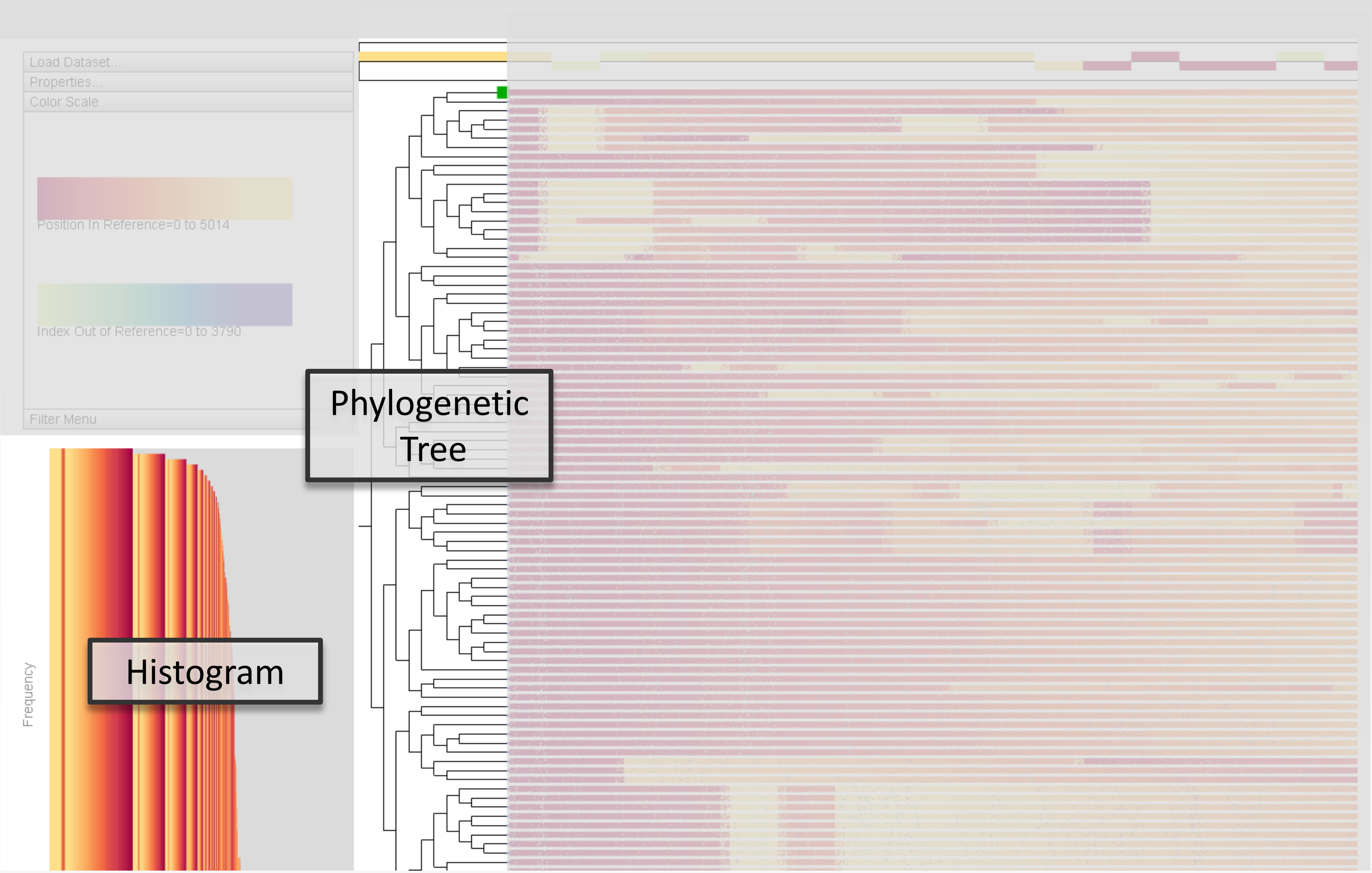
Filter Menu
Frequency

Mapping Pane

Mapping

Our Solution



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Phylogenetic Tree

Histogram

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Design in Practice

Perception

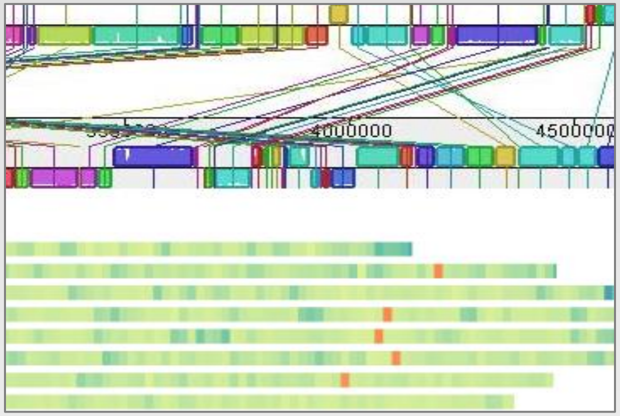
How the user processes dense data

Inform scalable design

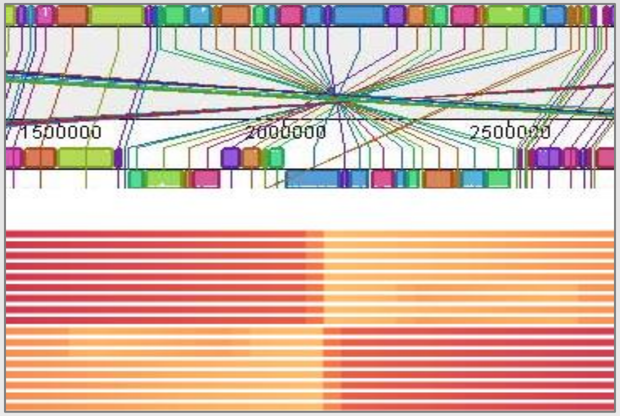
- Limitations of current designs
- Insight into future designs

Four principles

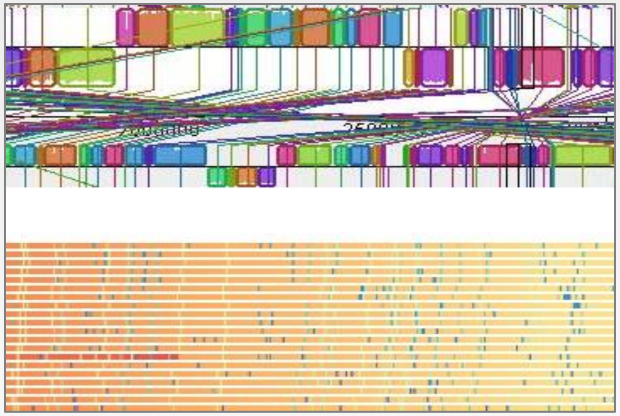
Perceptual Principles



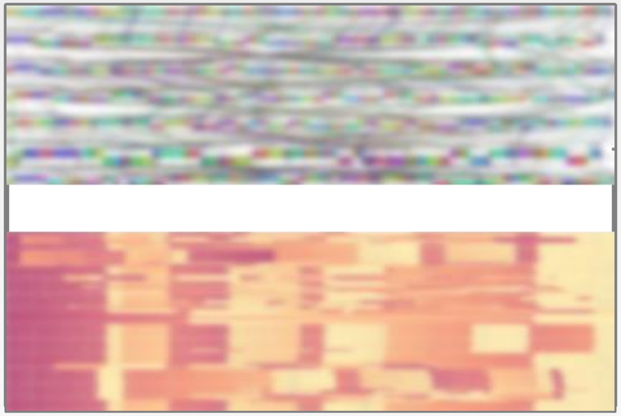
Pre-Attentive Phenomena



Visual Search

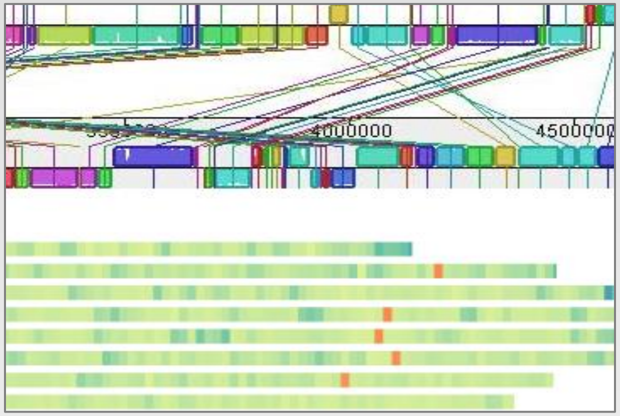


Visual Clutter

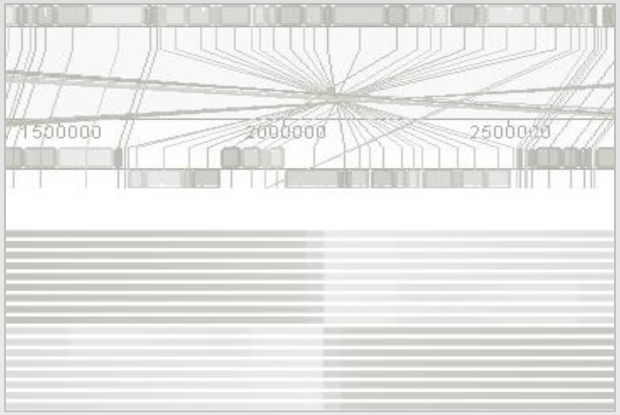


Summarization

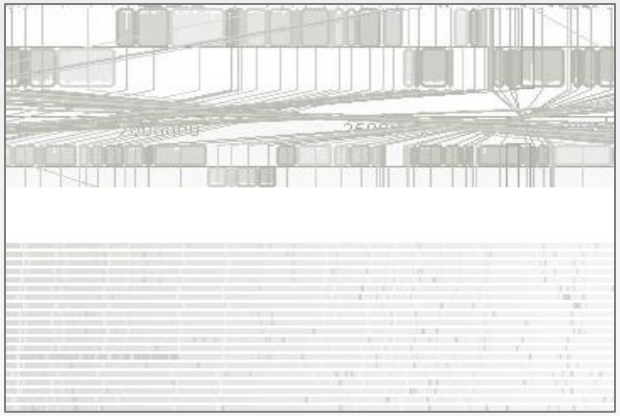
Perceptual Principles



Pre-Attentive Phenomena



Visual Search



Visual Clutter

Summarization

Perceptual Principles

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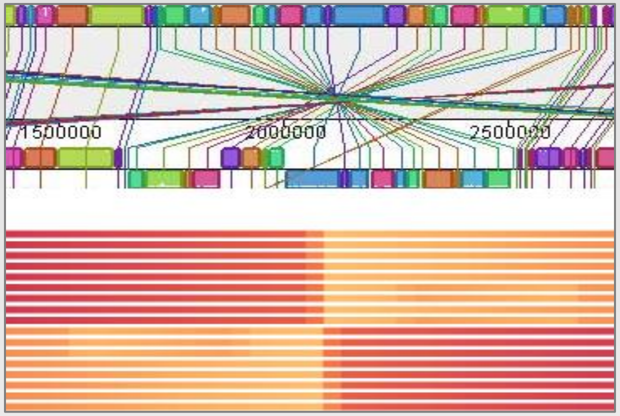
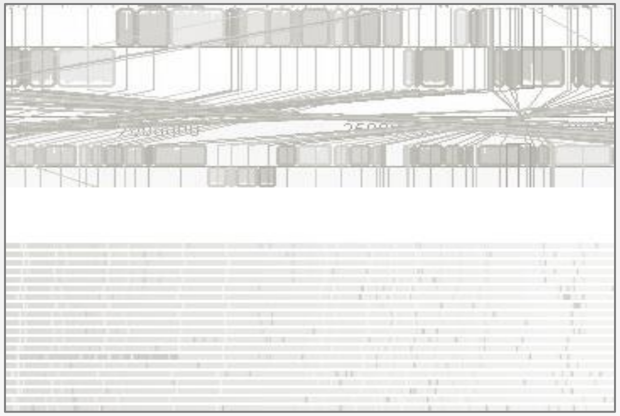


Visual Search



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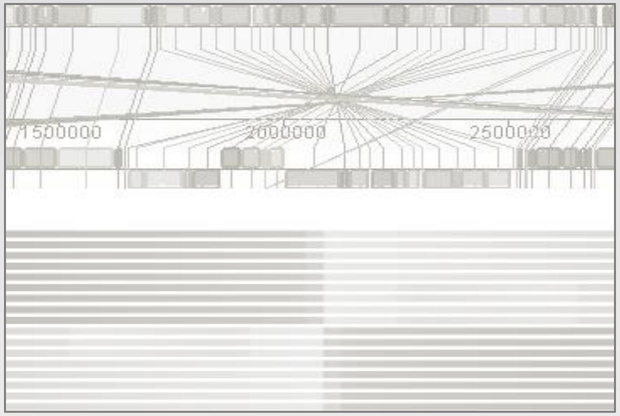

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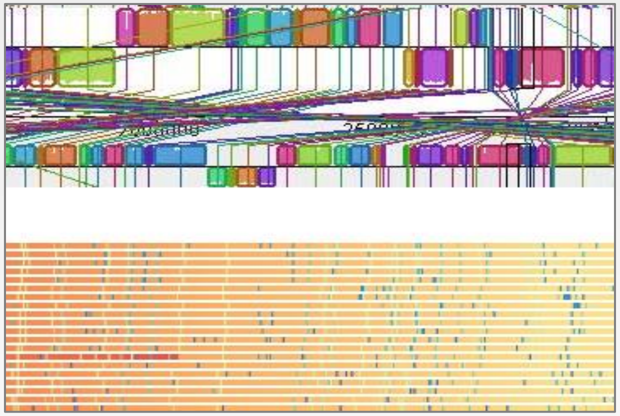

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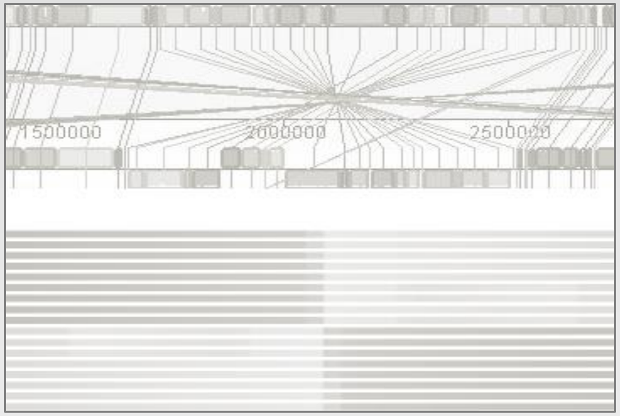
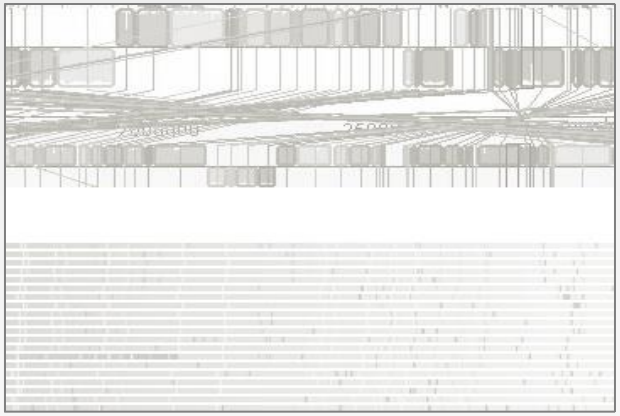
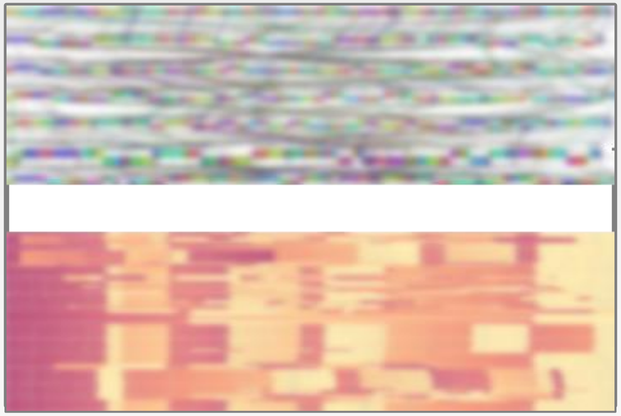


Visual Search



Visual Clutter



Summarization

Perception

Overview - Sacrifice detail for high-level comparison

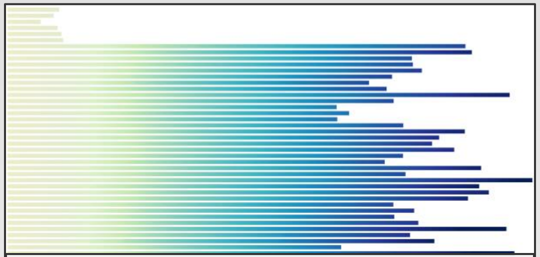
Colorfield - Emphasize visual structure

Mappings – Emphasize key details

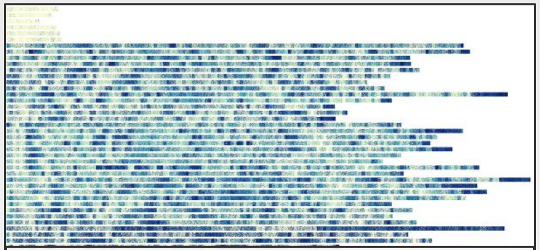
Aggregation – Do not overwhelm viewers

Mapping

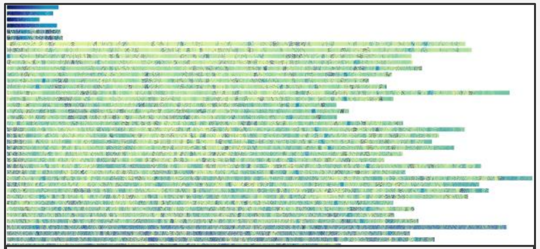
Color Mapping



Locality

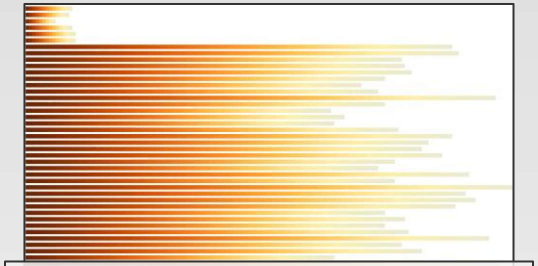


Frequency

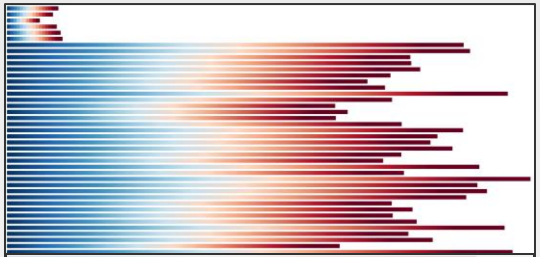


Reference

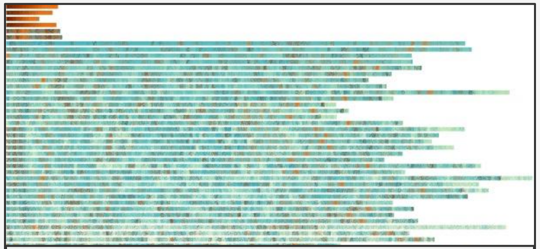
Color Schemes



Sequential

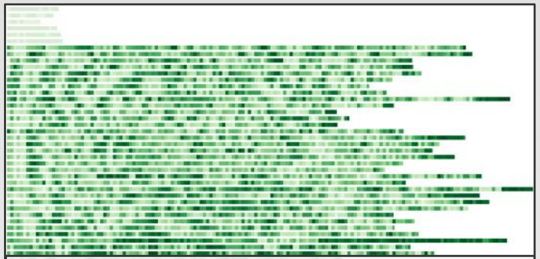


Diverging

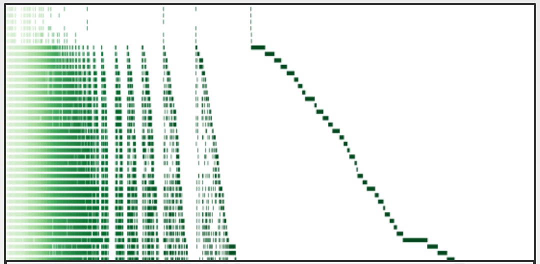


Split Schemes

Position Mapping



Locality



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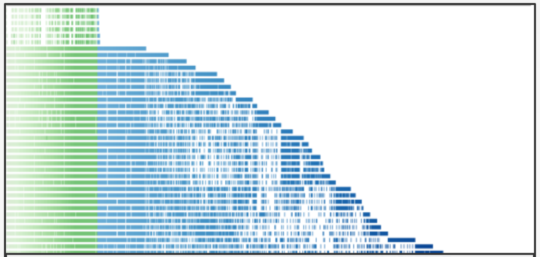
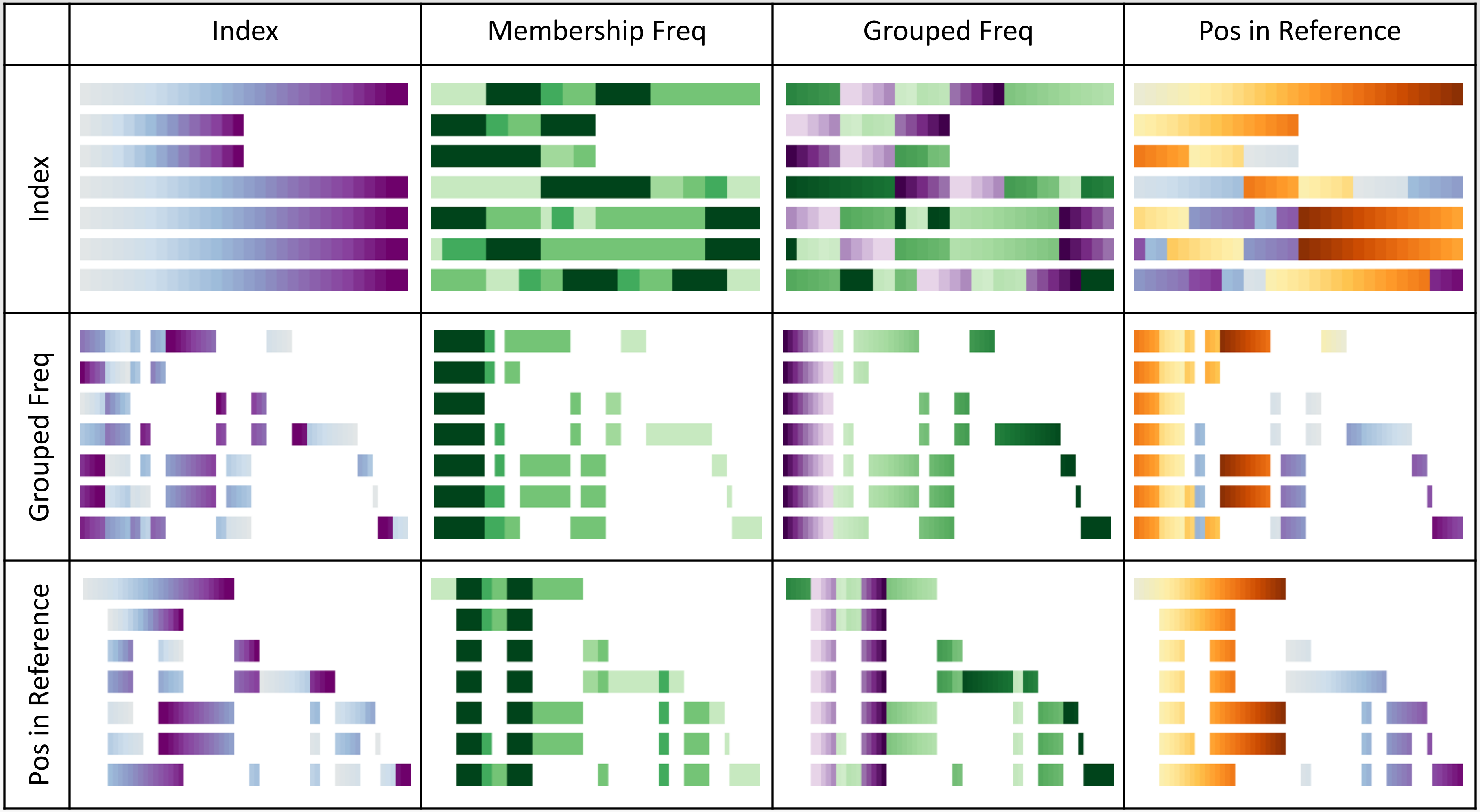


Reference

Combinations of different color and position mappings reveal interesting trends in the data



Aggregation

Cannot show all the data at once

- Limited screen real estate
- Clutter

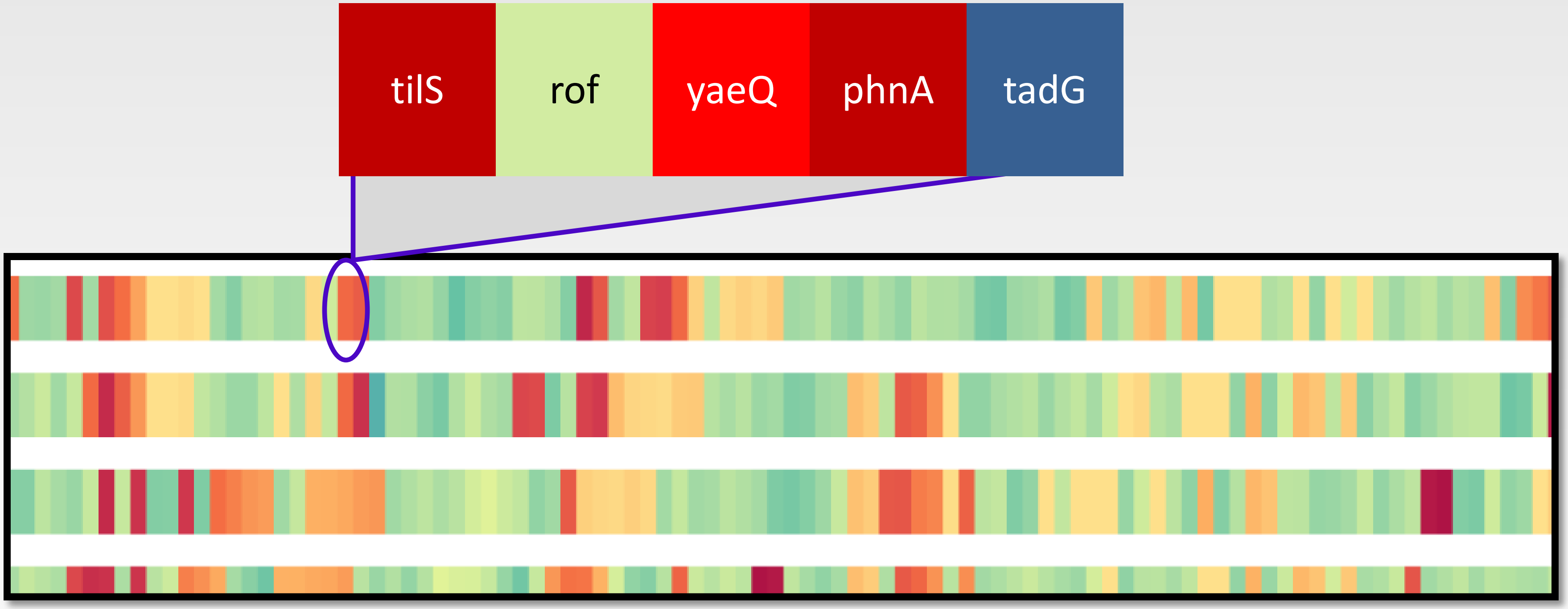
Blocking preserves local control

- Display gene neighborhoods as glyphs

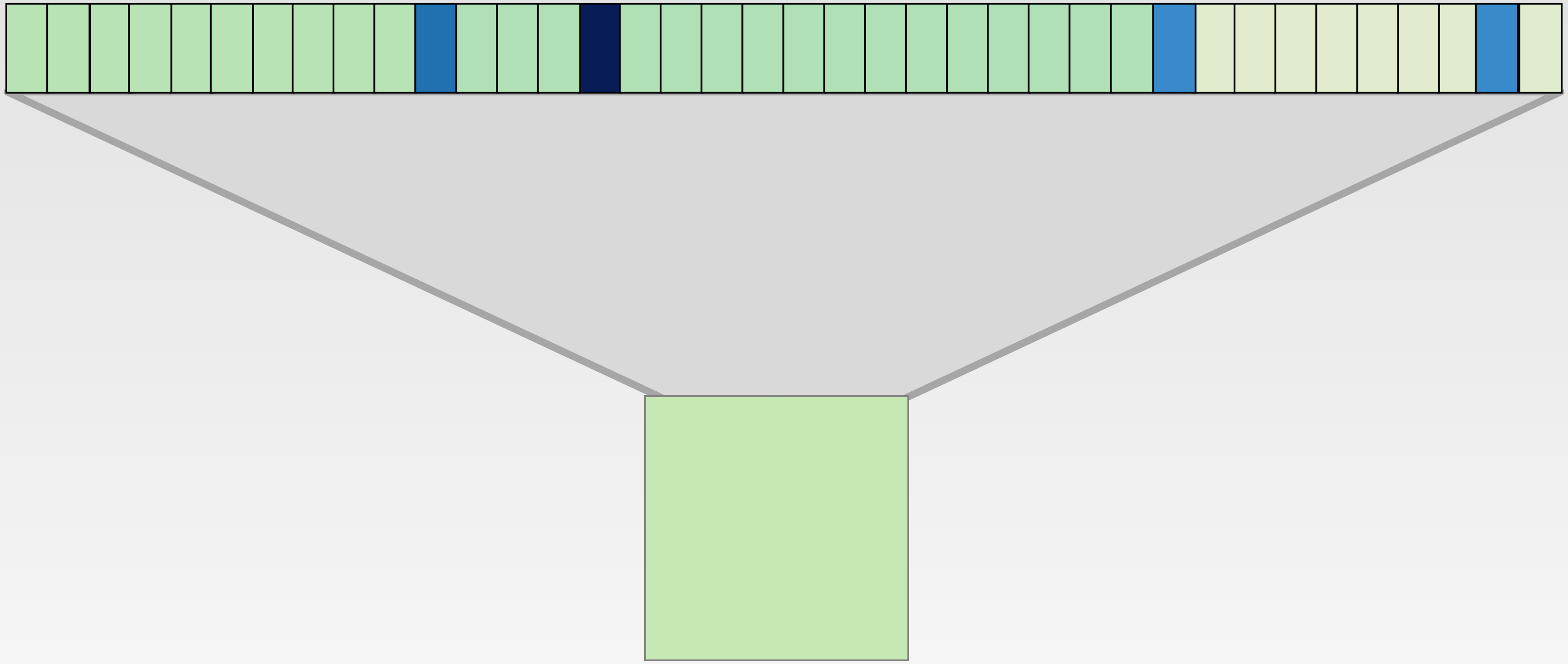
Four block encodings

Blocking

Group (relatively) continuous sets of neighboring genes into a single unit

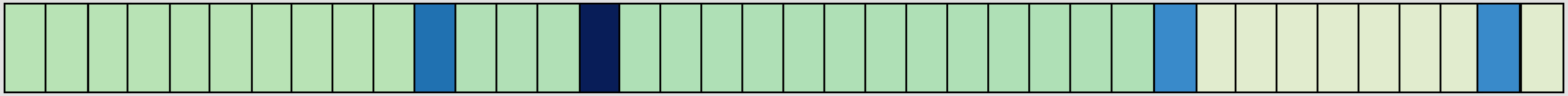


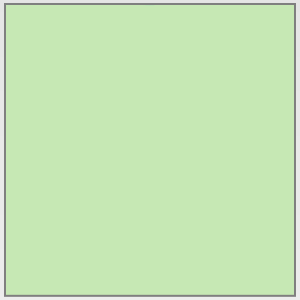
Aggregate Encodings



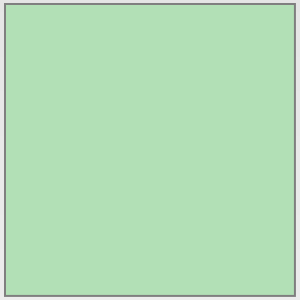
Average

Aggregate Encodings

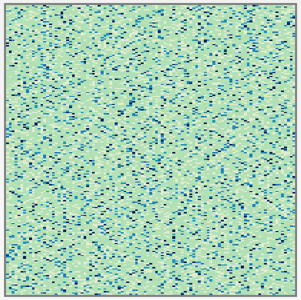




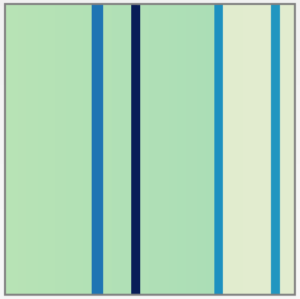
Average



Robust Average



Color Weaving



Event Striping

Interaction

Block Brushing: Highlight locations of block contents in overview, phylogeny, and histogram on mouse-over

Block Linking: Link locations of block contents in overview on click

Detail Notes: Details of genes in a block and matching genes of the set are presented in a separate window

Non-locality Zoom: Explore the contents of an aggregate block in the Block Detail Window on mouse-over

Zoom Lock: Fix the contents of a block in the zoom window to explore the distributions of specific genes

Zoomed Gene Brushing: Highlight locations of genes in overview, phylogeny, and histogram

Zoomed Gene Linking: Link locations of a set of matching genes in the overview

Manual Rearrangement: Drag-and-drop rearrangement of sequences and indicate branch crossings by opacity

Filtering: Highlight genes matching a set of names, id numbers, frequencies, genomes, or chromosomes

Load Filter: Load a filter set from a CSV

Save Filter: Save the current filter set to a CSV

Histogram Brushing: Highlight the locations of genes in a region of the frequency distribution in the overview and phylogenetic tree by mouse-over

Load Tree: Load different trees and arrangements from a tree file

Save Tree: Save the current tree structure and sequence arrangement to a tree file

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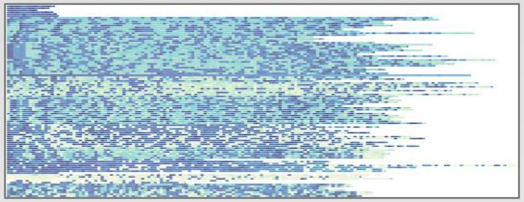
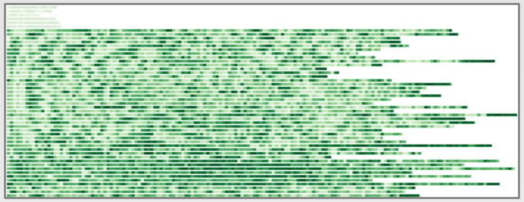
Design in Practice

Use Cases

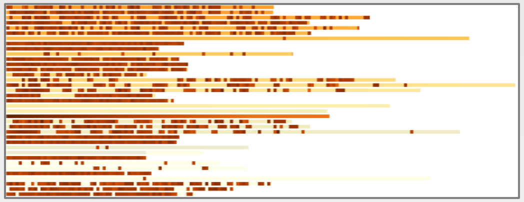
100 Bacteria
6,000 genes



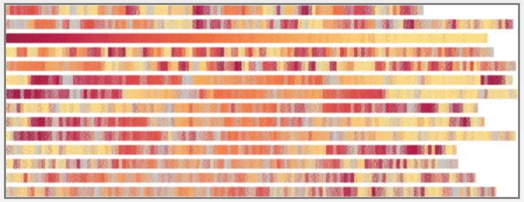
50 Bacteria
5,000 genes



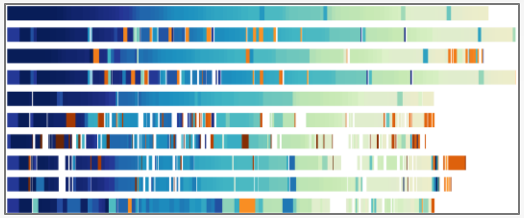
35 Fungi
17,000 genes



14 Pathogens
4,000 genes

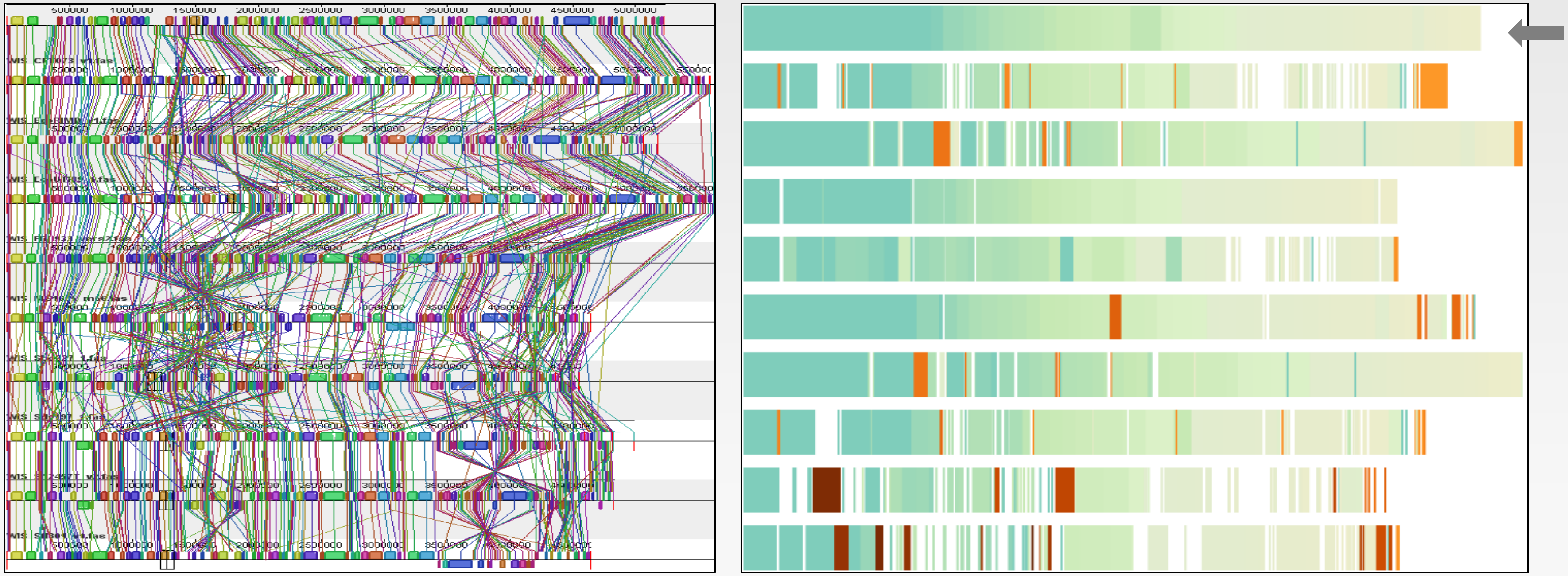


8 partial *E. coli* sequences
300 genes



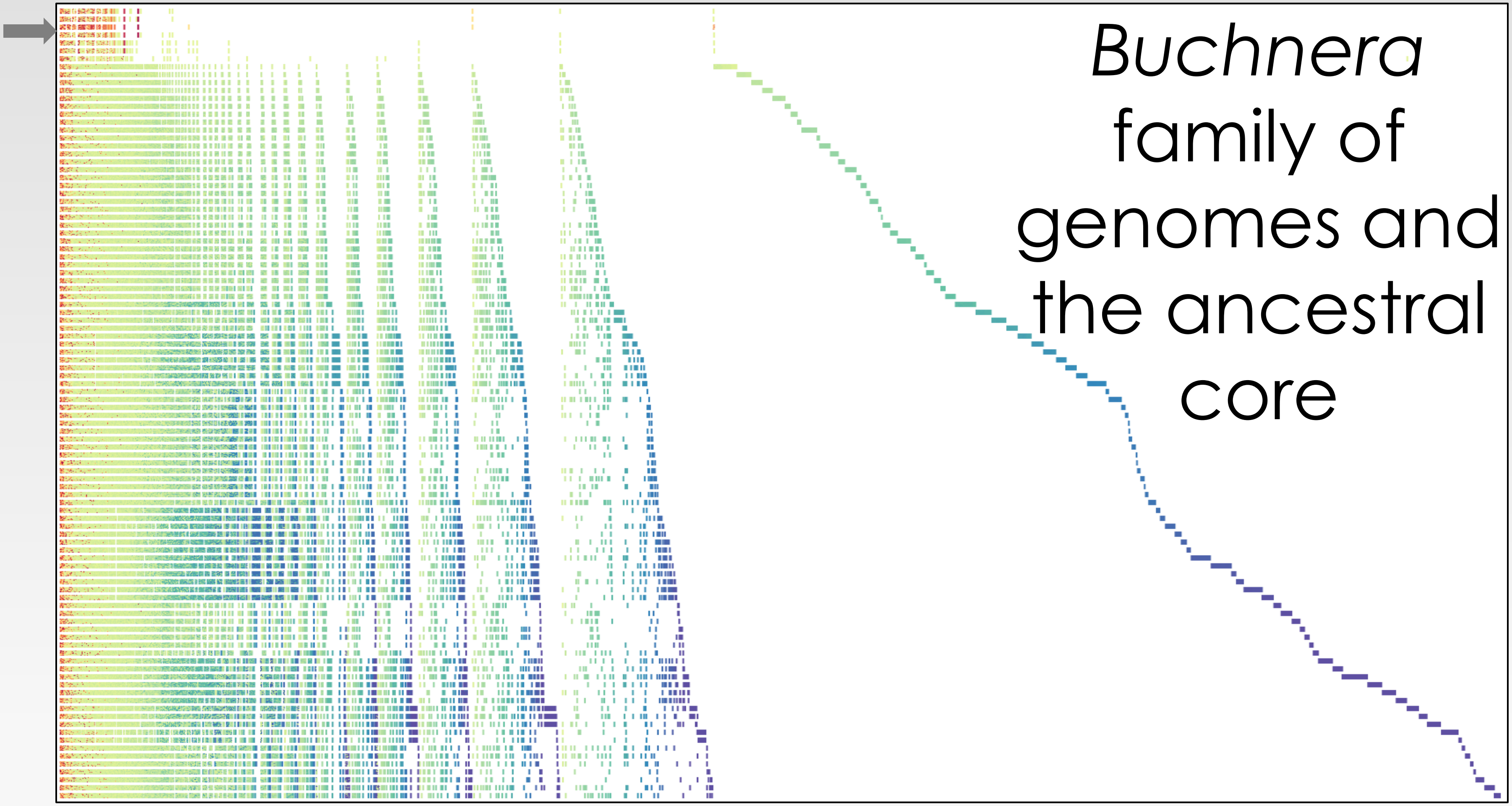
Parallels

Can use Sequence Surveyor to obtain information presented in existing tools at scale.



Mauve: Color by position in reference (arrow), order by start position

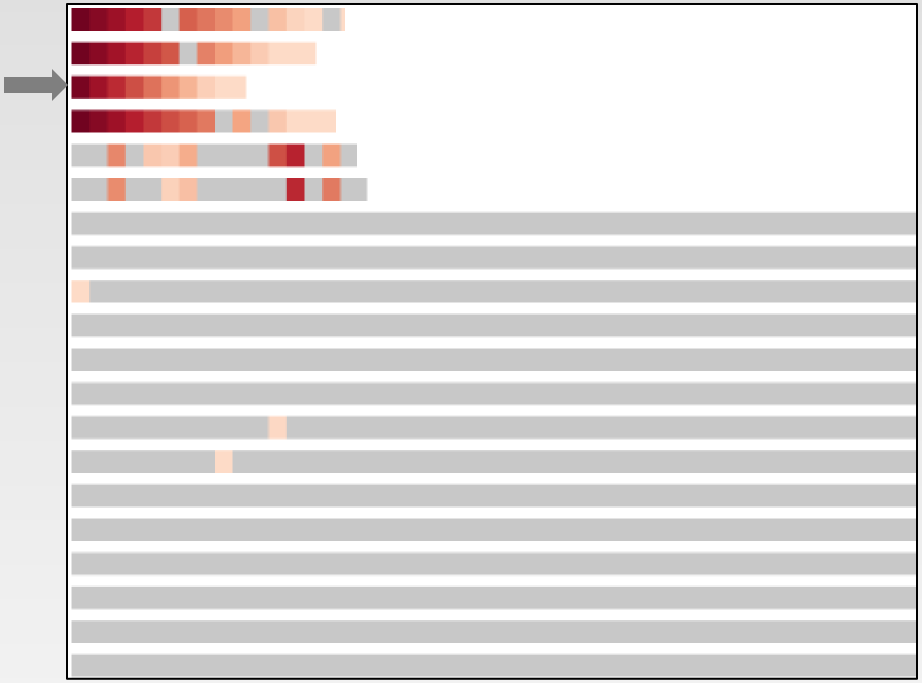
Anecdotes: *Buchnera*



Buchnera family of genomes and the ancestral core

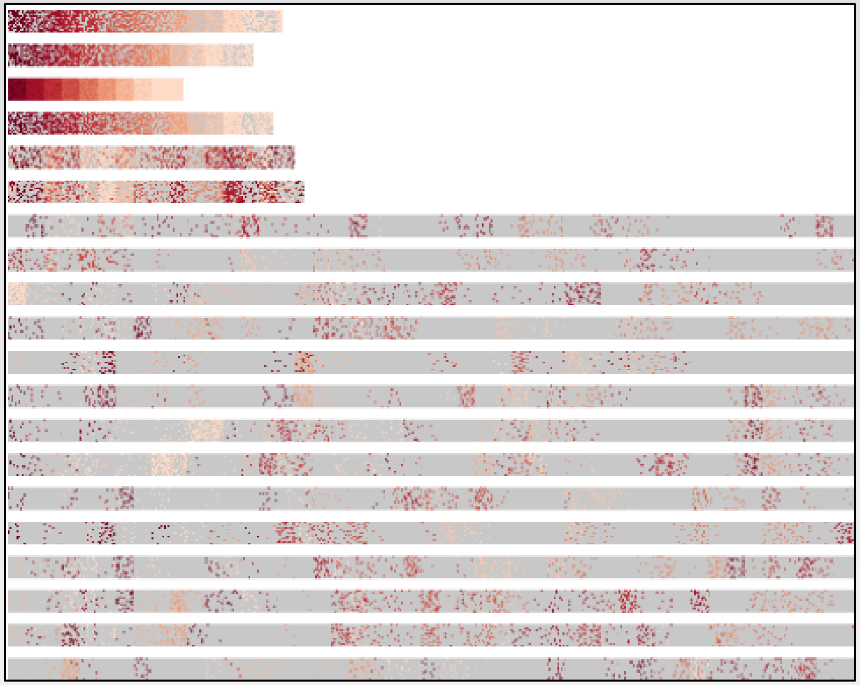
Color by position in reference (arrow), order by set of genomes containing each gene

Anecdotes: *Buchnera*



Averaging:

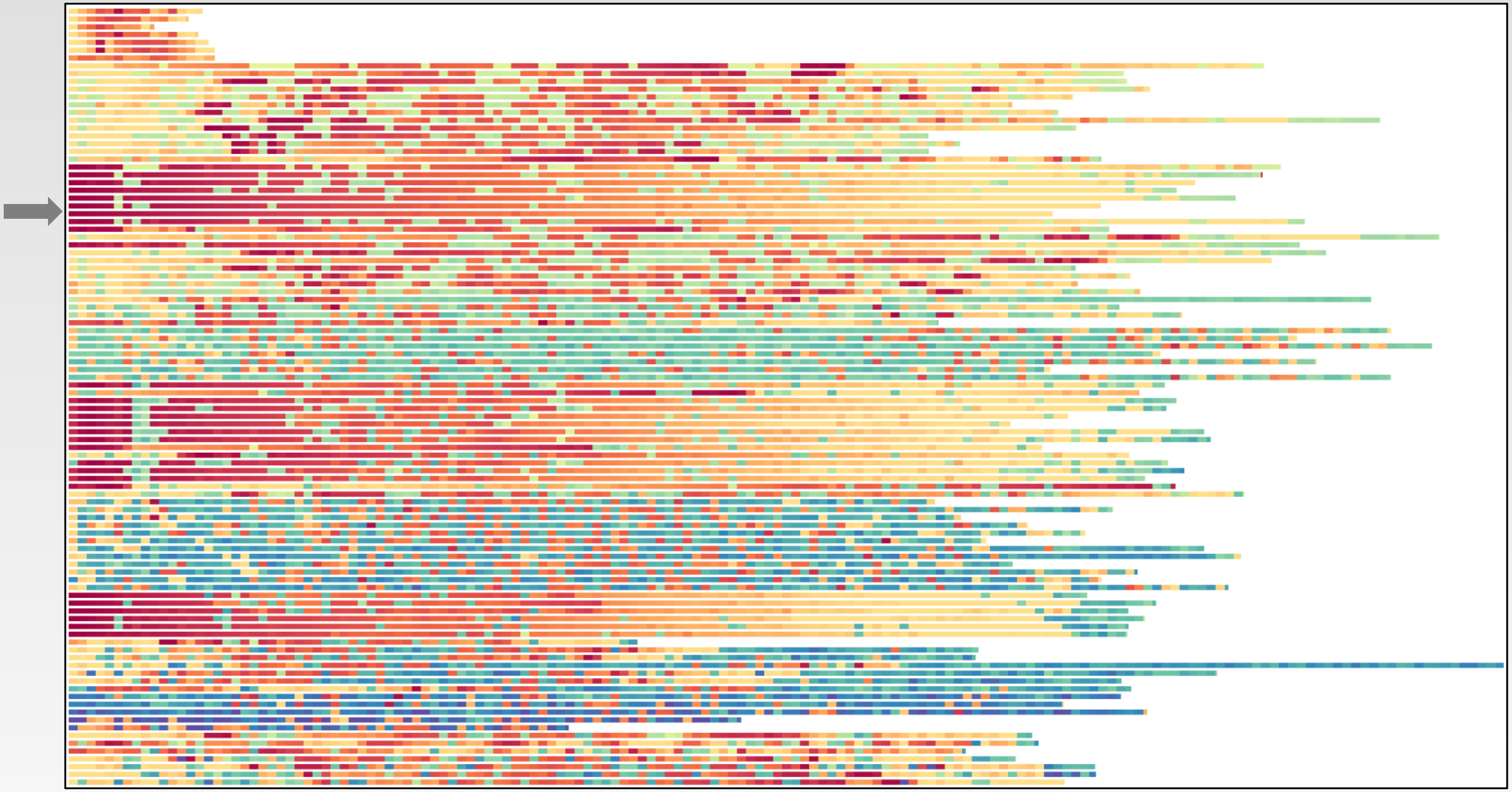
No significant trend



Color Weaving:

Overall distribution

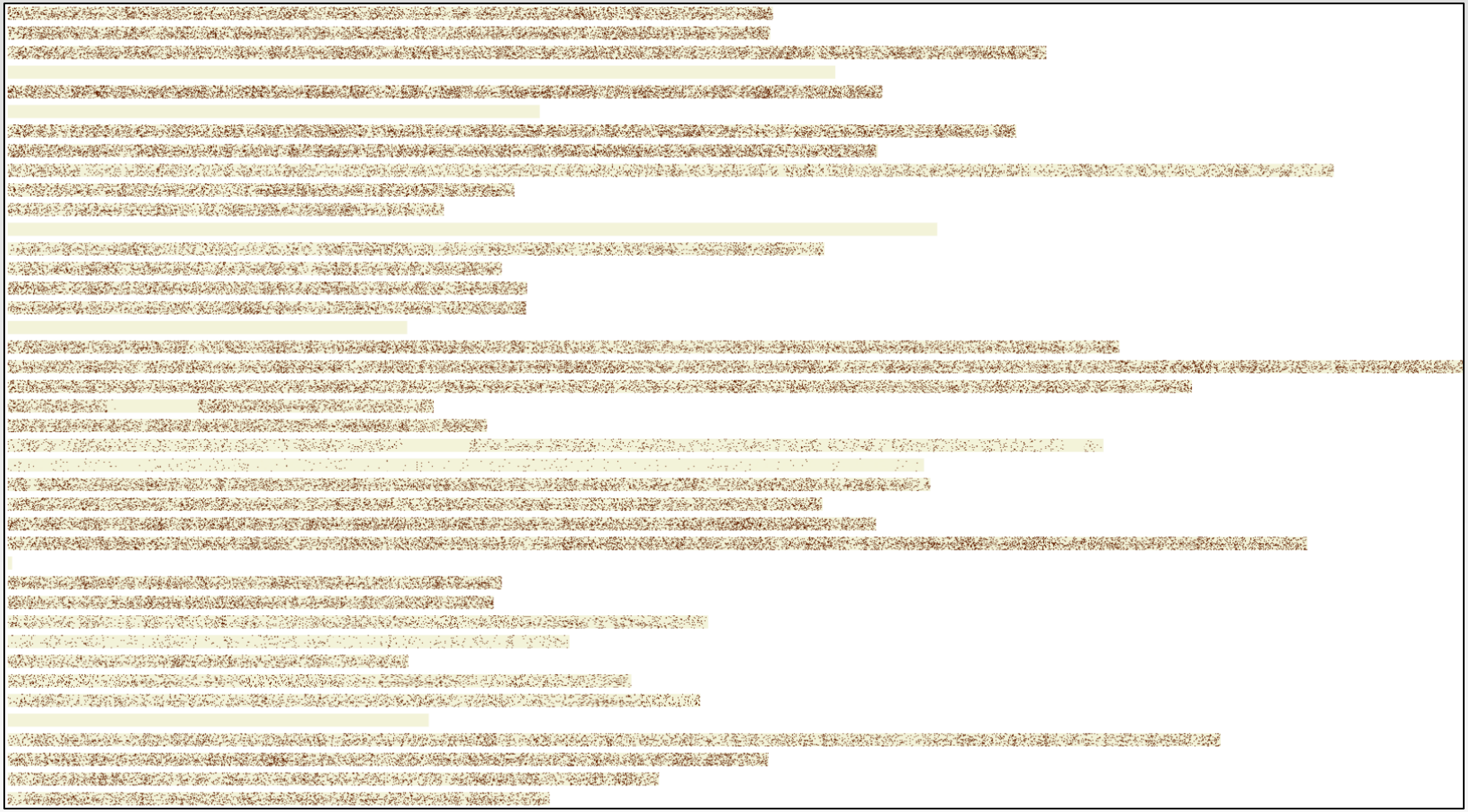
Anecdotes: *E. Coli*



Conservation relationships between different families of genomes

Color by position in reference (arrow), order by relative ordering

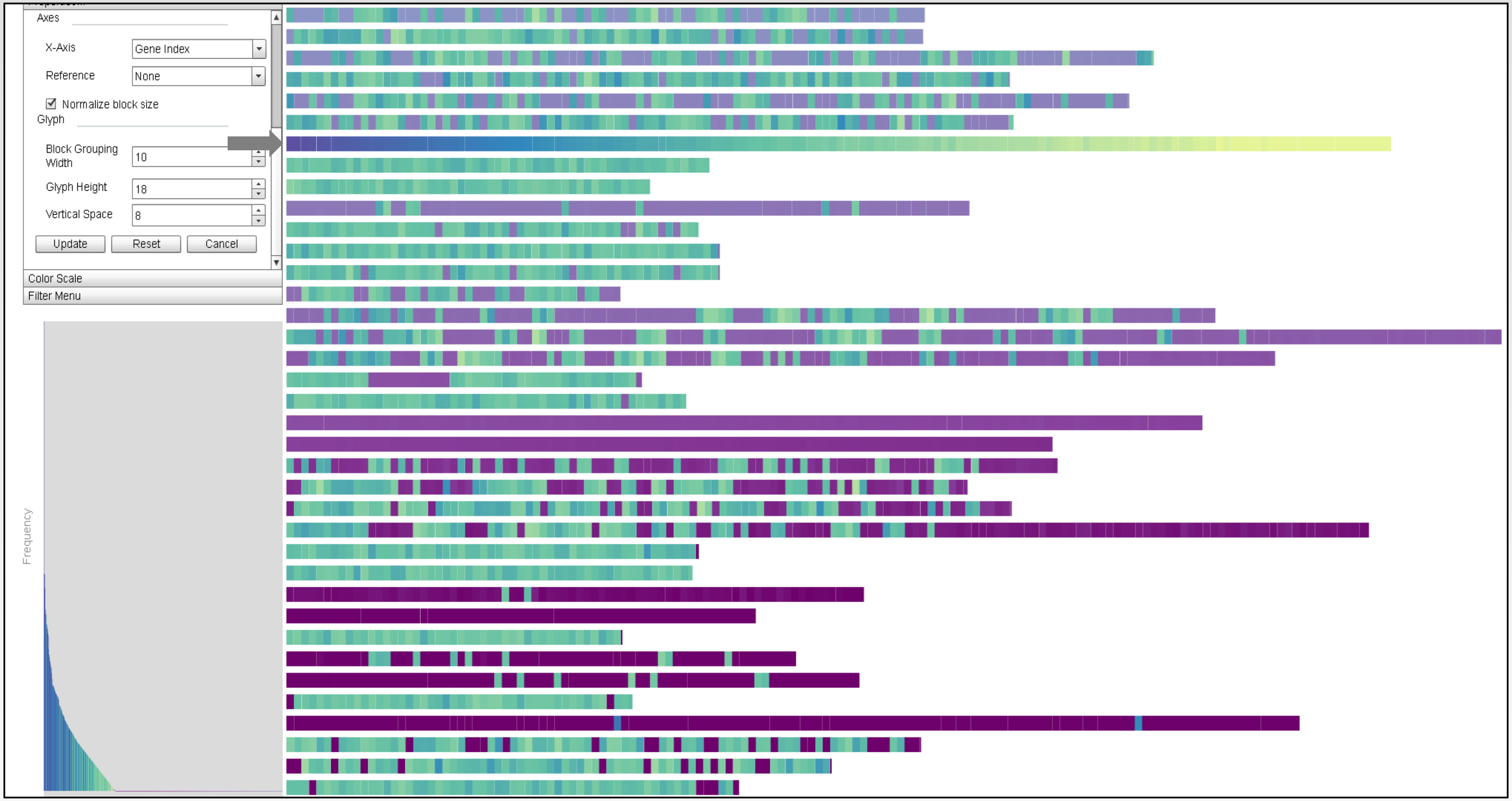
Anecdotes: Fungi



Bioinformatics applications allow users to test algorithms using visual checks

Color by overall frequency, order by relative ordering

Anecdotes: Fungi



Axes

X-Axis Gene Index

Reference None

☑ Normalize block size

Glyph

Block Grouping Width 10

Glyph Height 18

Vertical Space 8

Bioinformatics applications allow users to test algorithms using visual checks

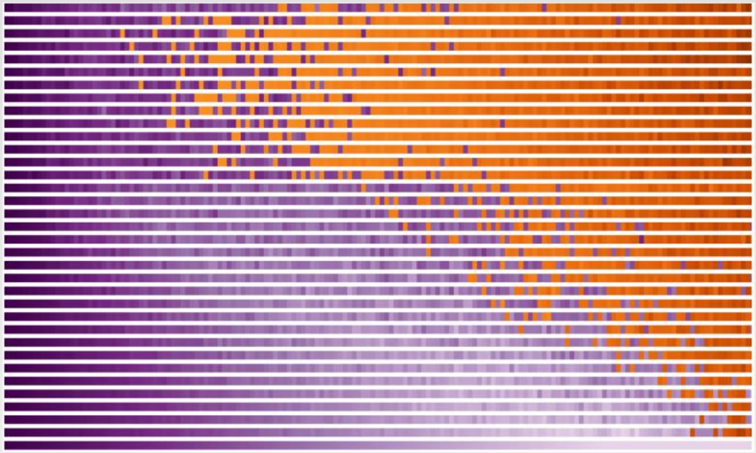
Color by position in a reference, order by relative ordering

Extensions

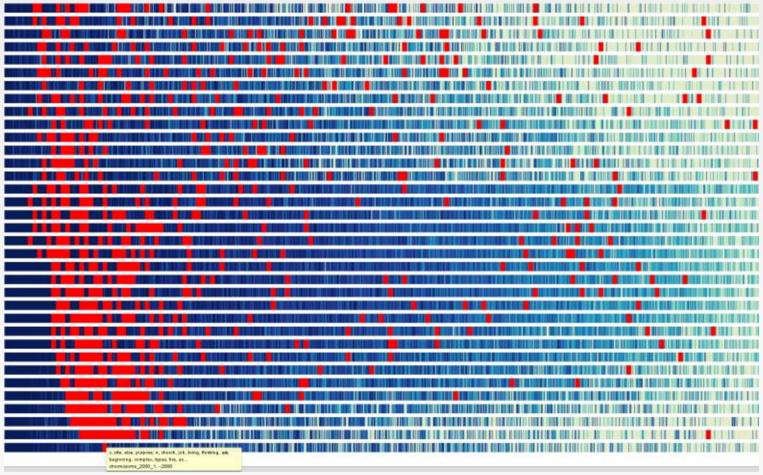
Proteins and nucleotide MSA

Any data with an orthology and ordered sets

Google N-Grams



Top 5,000 most popular words since 1660



Distribution of a word set in 2000 across time

Summary

Scalable whole genome alignment overview

Perception informs design

User-controlled mapping scales across queries

Aggregation filters data

Extends beyond the immediate biology

Acknowledgements

University of Wisconsin – Madison
Department of Computer
Sciences Graphics & Vision Lab

University of Wisconsin – Madison
BACTER Institute for Computational
Biology

University of Wisconsin – Madison
Genome Center Genome Evolution
Laboratory

Dr. David Baumler
Dr. Eric Neeno-Eckwall
Dr. Jeremy Glasner
Dr. Nicole Perna

Funding by NSF awards IIS-0946598, CMMI-0941013 and DEB-0936214 and DoE Genomics: GTL and SciDAC Programs (DE-FG02-04ER25627)

Availability

Prototype and sample data package (coming soon):

http://graphics.cs.wisc.edu/Vis/SequenceSurveyor/

dalbers@cs.wisc.edu