# Sequence Surveyor

#### Leveraging Overview for Scalable Genomic Alignment Visualization

Danielle Albers, Colin Dewey, and Michael Gleicher University of Wisconsin-Madison Department of Computer Sciences IEEE VisWeek 2011

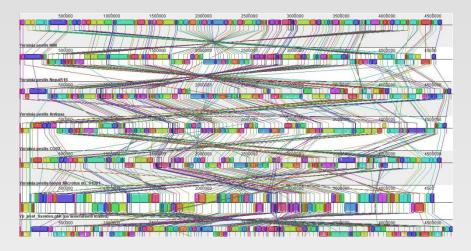


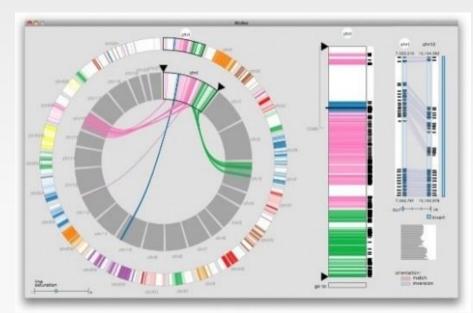


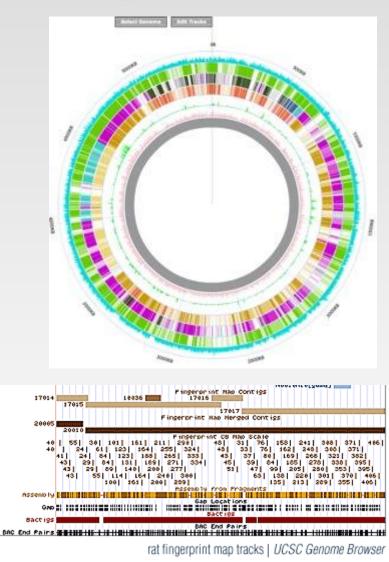
University of Wisconsin, Madison Computer Graphics



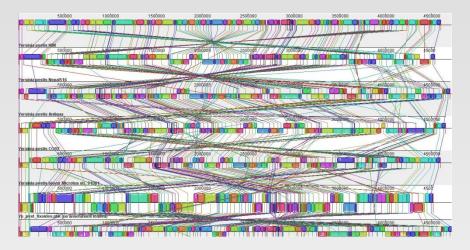
### Viewing Genome Alignments

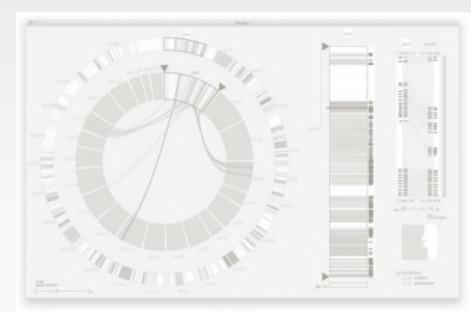


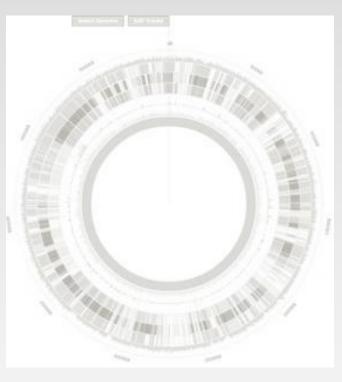




### Viewing Genome Alignments



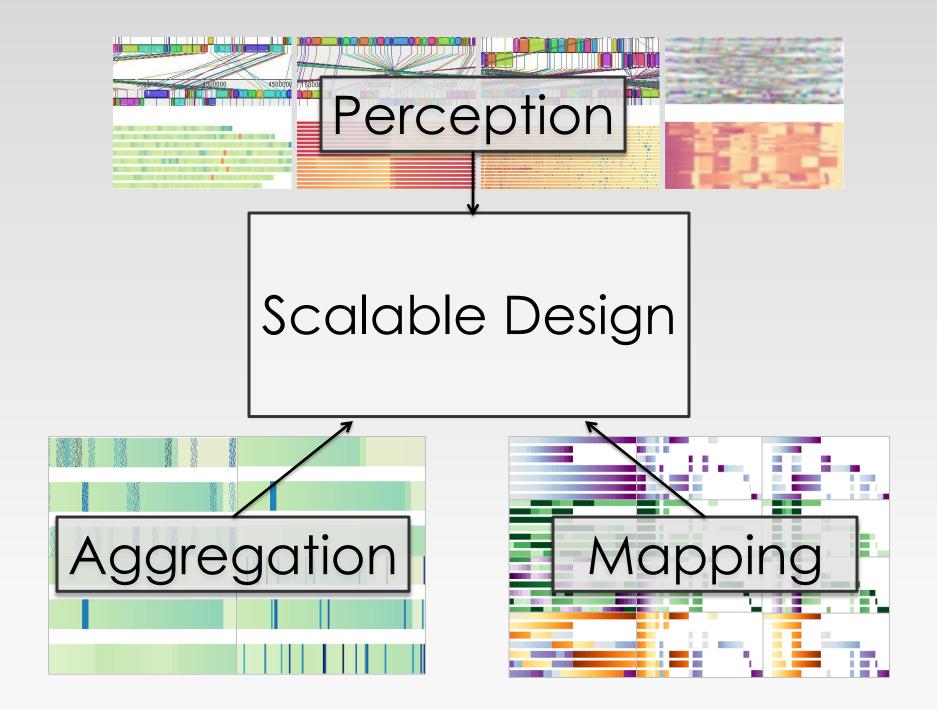


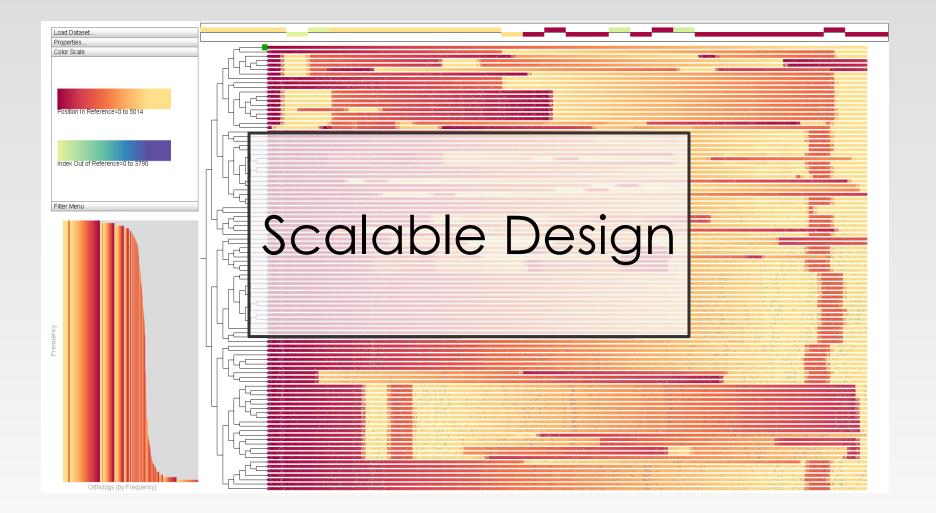




rat fingerprint map tracks | UCSC Genome Browser

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## Outline

The Data Domain Sequence Surveyor Design in Theory

- Perception
- Mapping
- Aggregation

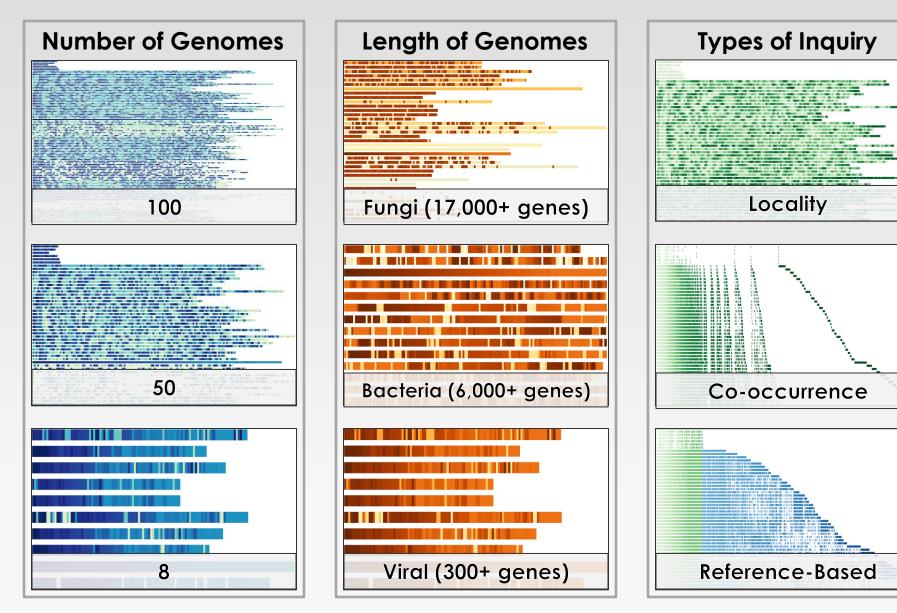
Design in Practice

### Whole Genome Alignment

Identify related groups of genes appearing in a set of organisms



## **Defining Scale**



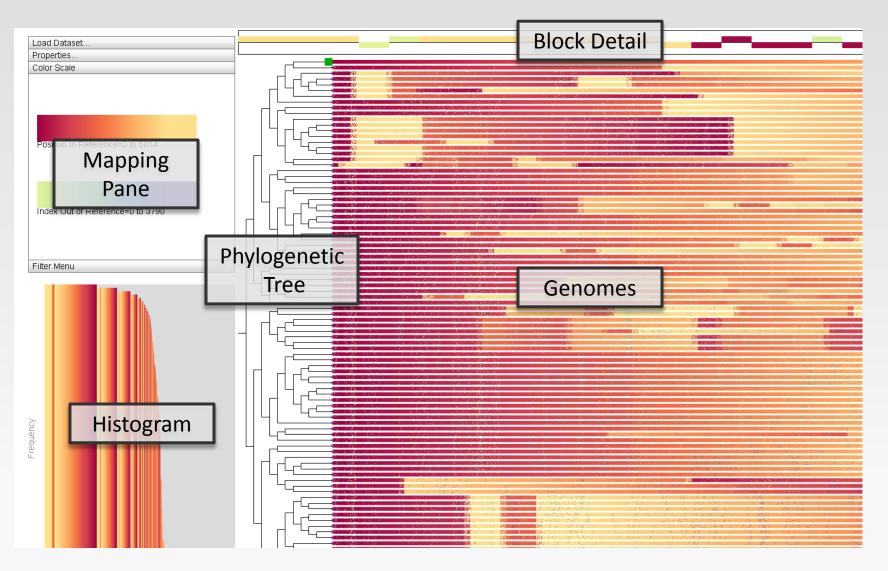
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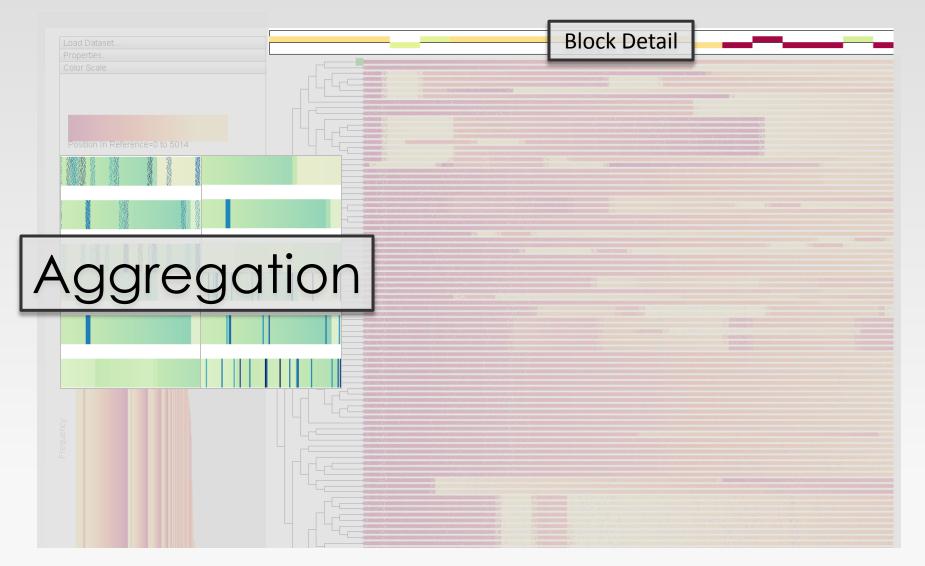
- Perception
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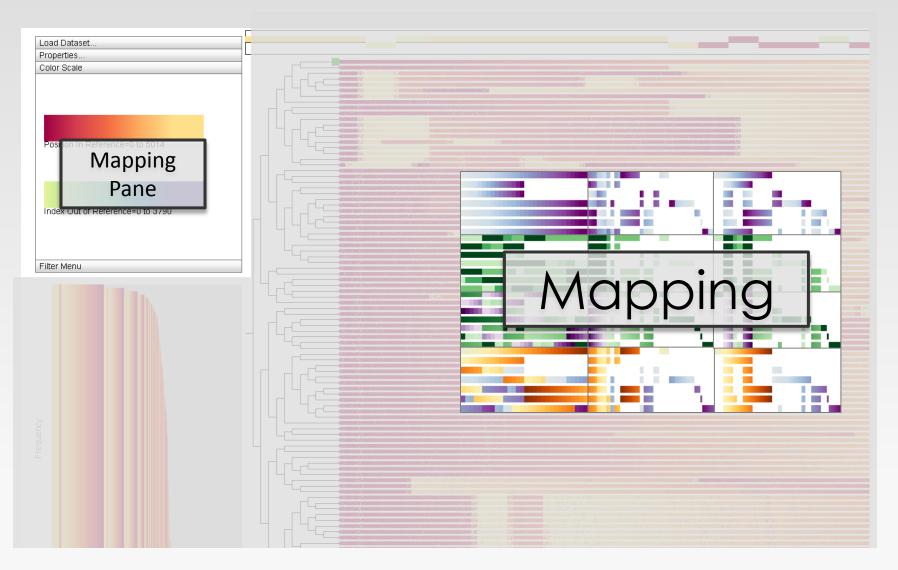
Design in Practice

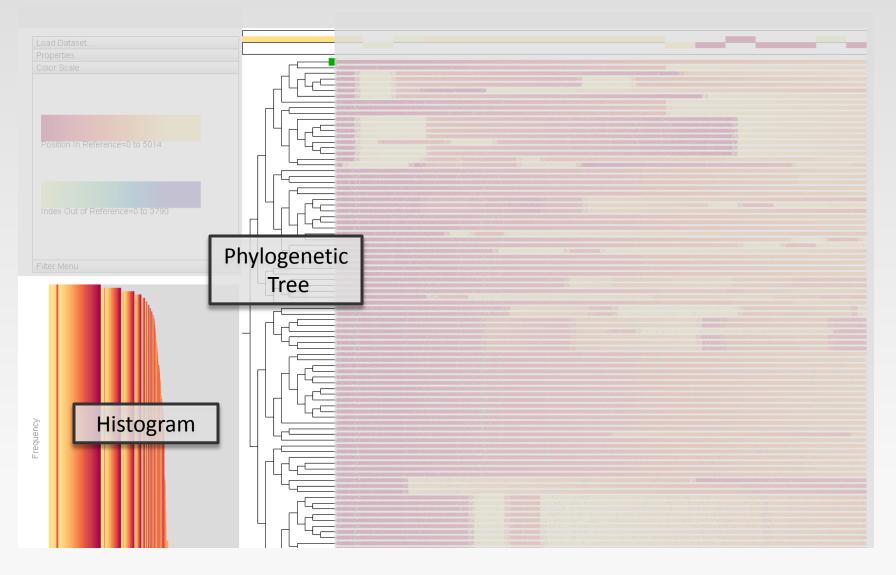












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Design in Practice

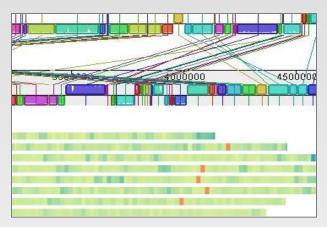
### Perception

How the user processes dense data

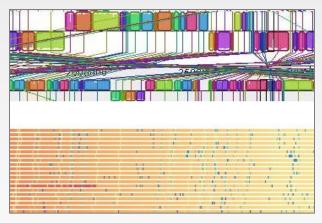
Inform scalable design

- Limitations of current designs
- Insight into future designs

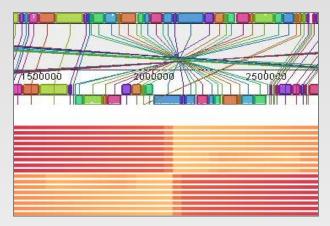
Four principles



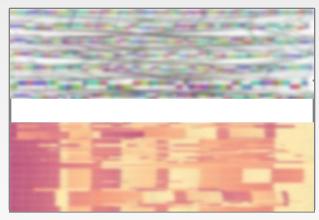
#### Pre-Attentive Phenomena

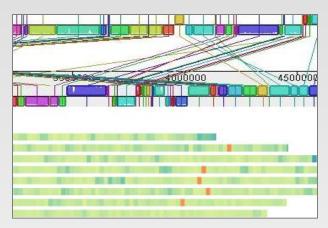


Visual Clutter

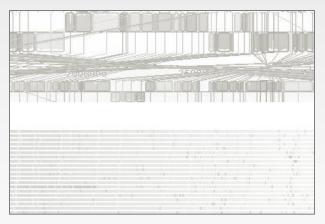


#### Visual Search

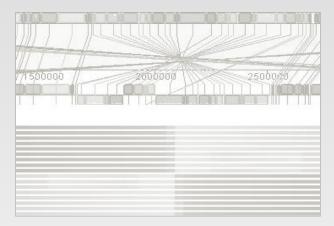




#### Pre-Attentive Phenomena



Visual Clutter

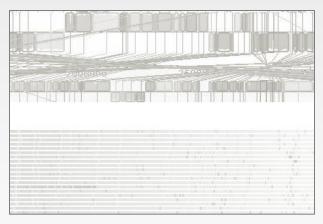


#### Visual Search

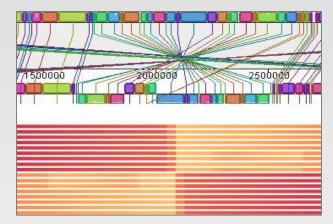




#### Pre-Attentive Phenomena



Visual Clutter

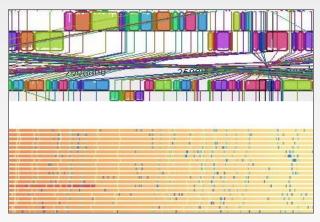


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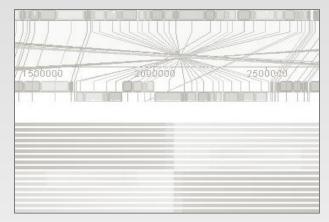




#### Pre-Attentive Phenomena



Visual Clutter

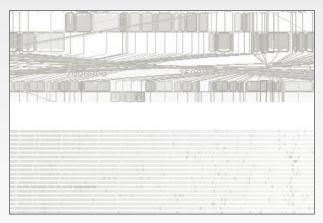


#### Visual Search

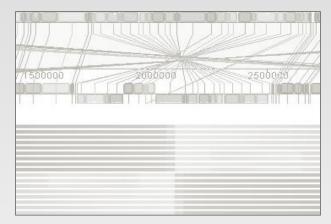




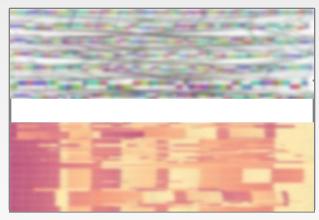
#### Pre-Attentive Phenomena



Visual Clutter



#### Visual Search



### Perception

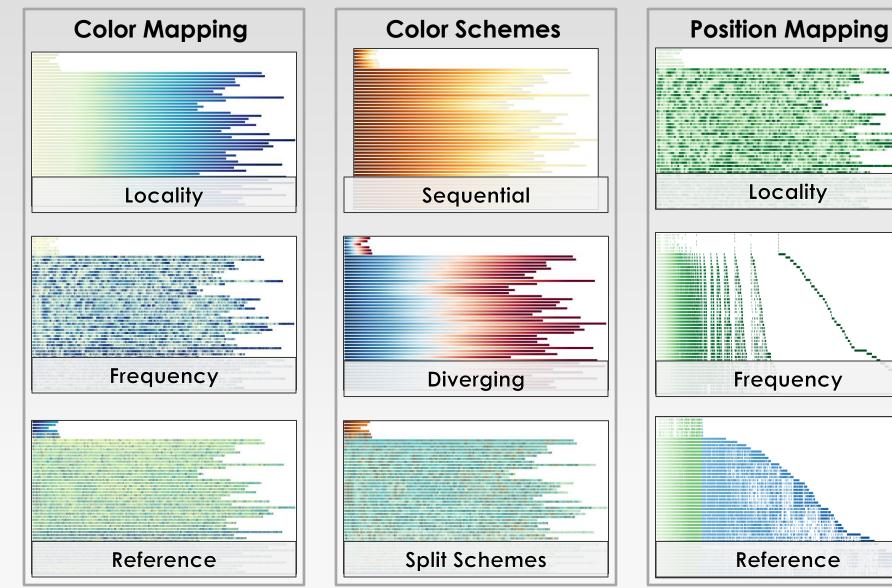
Overview - Sacrifice detail for high-level comparison

Colorfield - Emphasize visual structure

Mappings – Emphasize key details

Aggregation – Do not overwhelm viewers

# Mapping



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

	Index	Membership Freq	Grouped Freq	Pos in Reference	
Index					
Grouped Freq					
Pos in Reference					

## Aggregation

#### Cannot show all the data at once

- Limited screen real estate
- Clutter

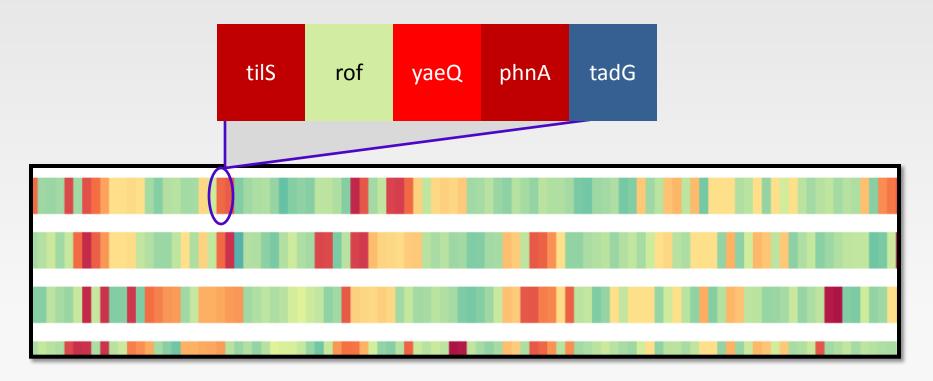
Blocking preserves local control

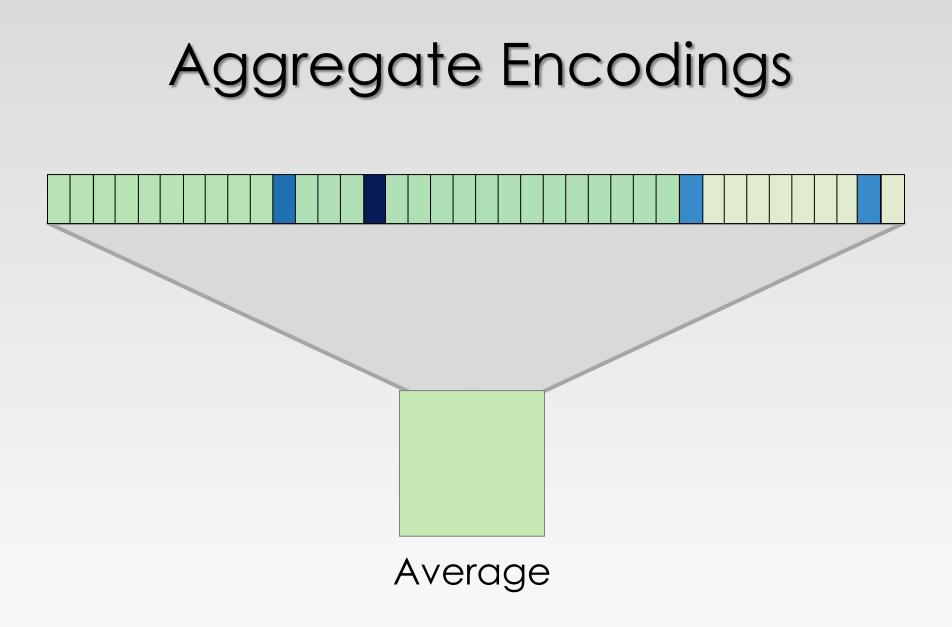
- Display gene neighborhoods as glyphs

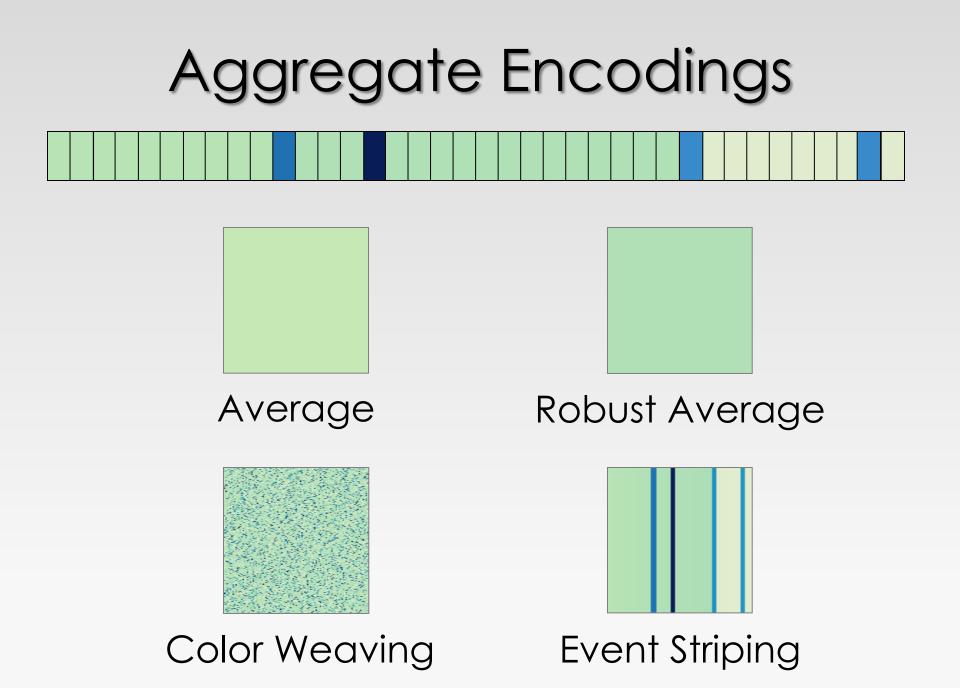
Four block encodings

### Blocking

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







### 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

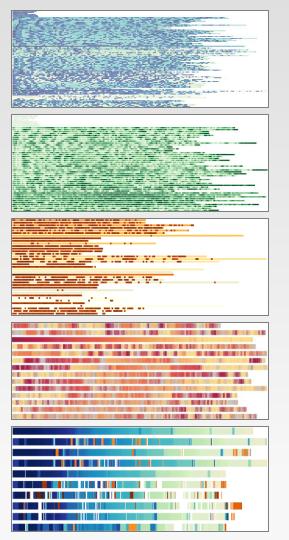
100 Bacteria 6,000 genes

50 Bacteria 5,000 genes

35 Fungi 17,000 genes

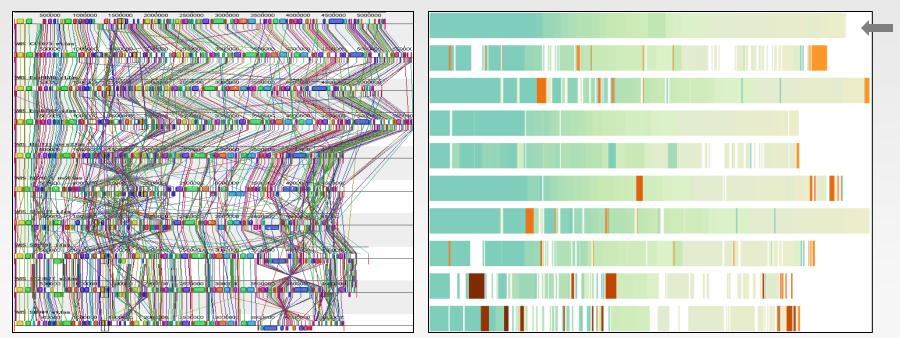
14 Pathogens 4,000 genes

8 partial E. coli sequences 300 genes



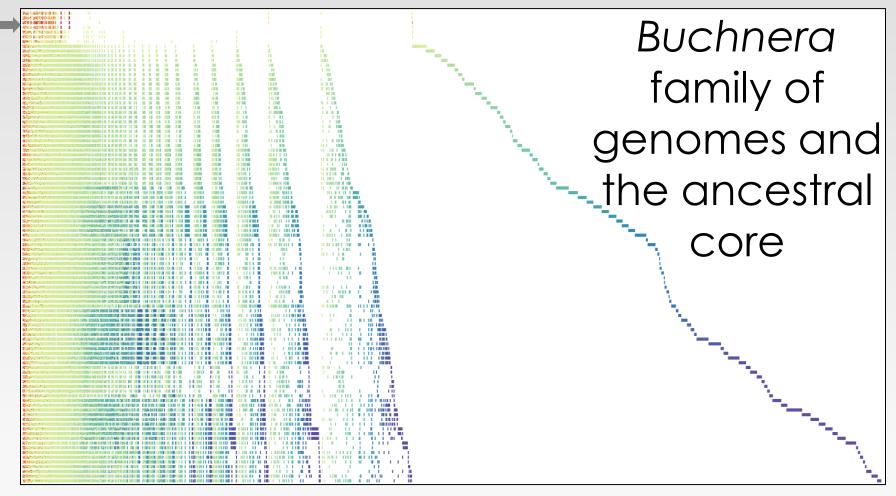
### Parallels

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



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

#### Anecdotes: Buchnera

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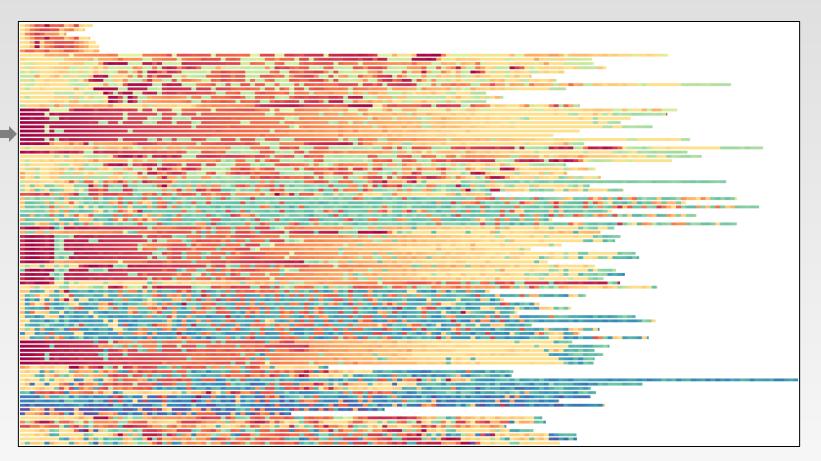
Averaging:

#### Color Weaving:

Overall distribution

No significant trend

#### Anecdotes: E. Coli



Conservation relationships between different families of genomes

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

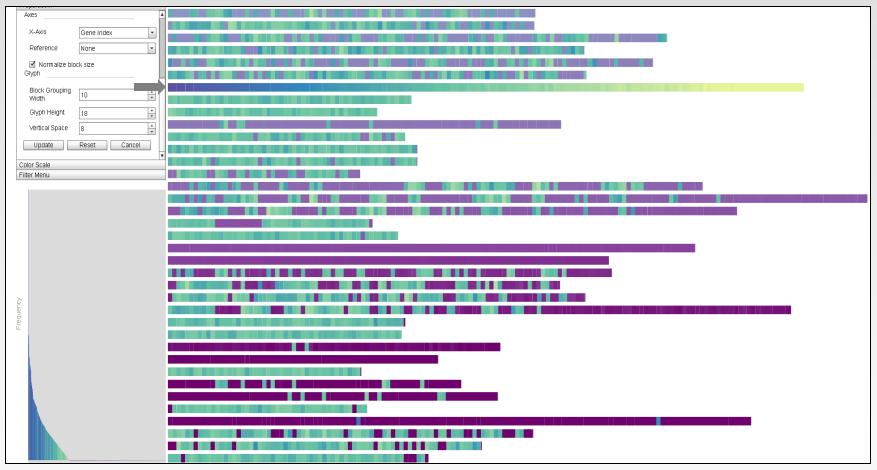
#### Anecdotes: Fungi

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Bioinformatics applications allow users to test algorithms using visual checks

Color by overall frequency, order by relative ordering

#### Anecdotes: Fungi



Bioinformatics applications allow users to test algorithms using visual checks

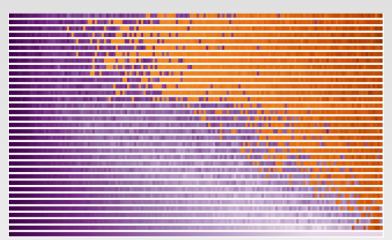
Color by position in a reference, order by relative ordering

#### Extensions

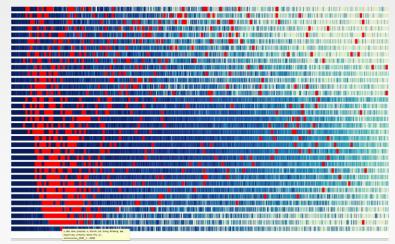
Proteins and nucleotide MSA

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

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University of Wisconsin – Madison Genome Center Genome Evolution Laboratory

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### Availability

Prototype and sample data package (coming soon): http://graphics.cs.wisc.edu/Vis/SequenceSurveyor/



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