

# Visualization Analysis & Design for Genomics

**Tamara Munzner**

Department of Computer Science  
University of British Columbia

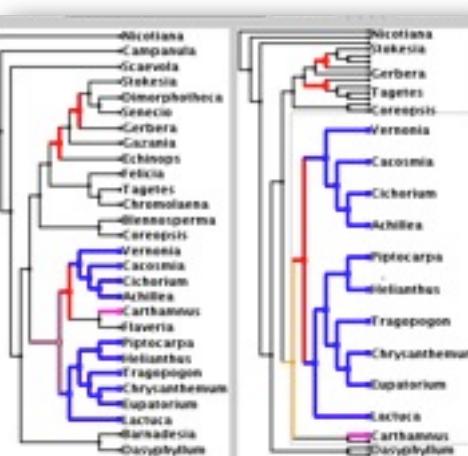
**Centre for Heart Lung Innovation, UBC and St. Paul's Hospital**  
30 July 2015, Vancouver BC

<http://www.cs.ubc.ca/~tmm/talks.html#daley15>

@tamaramunzner

# Outline

- introduction
- Cerebral
- MizBee
- TreeJuxtaposer
- wrapup



# Why have a human in the loop?

**Computer-based visualization systems provide visual representations of datasets designed to help people carry out tasks more effectively.**

# Why have a human in the loop?

**Computer-based visualization systems provide visual representations of datasets designed to help people carry out tasks more effectively.**

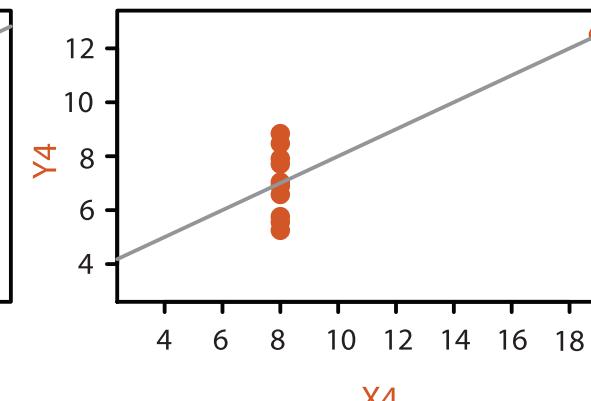
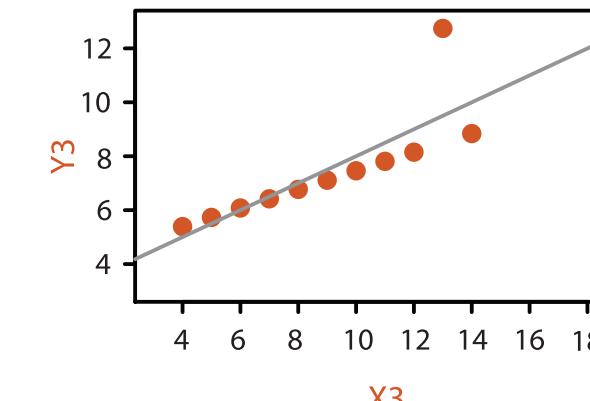
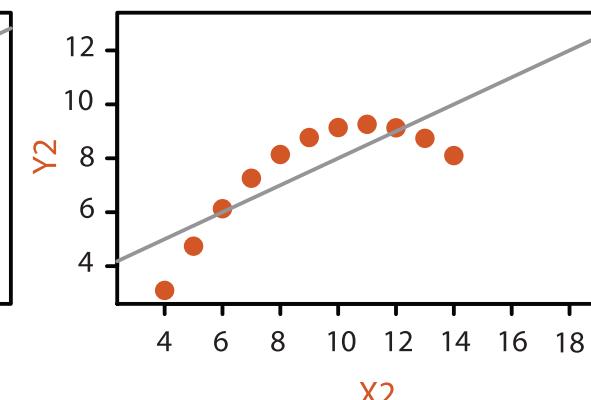
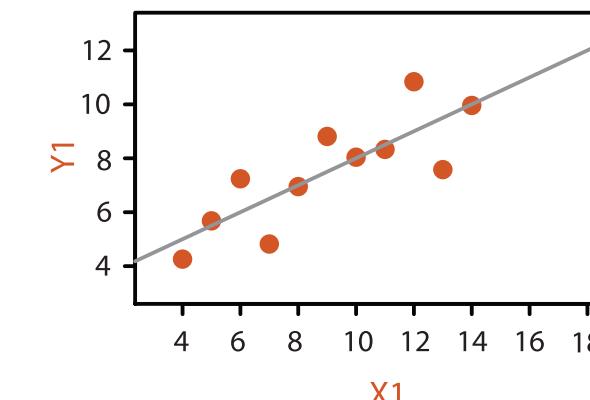
**Visualization is suitable when there is a need to augment human capabilities rather than replace people with computational decision-making methods.**

- many analysis problems ill-specified, not clear what questions to ask in advance
  - don't need vis when fully automatic solution exists and is trusted

## Anscombe's Quartet

### Identical statistics

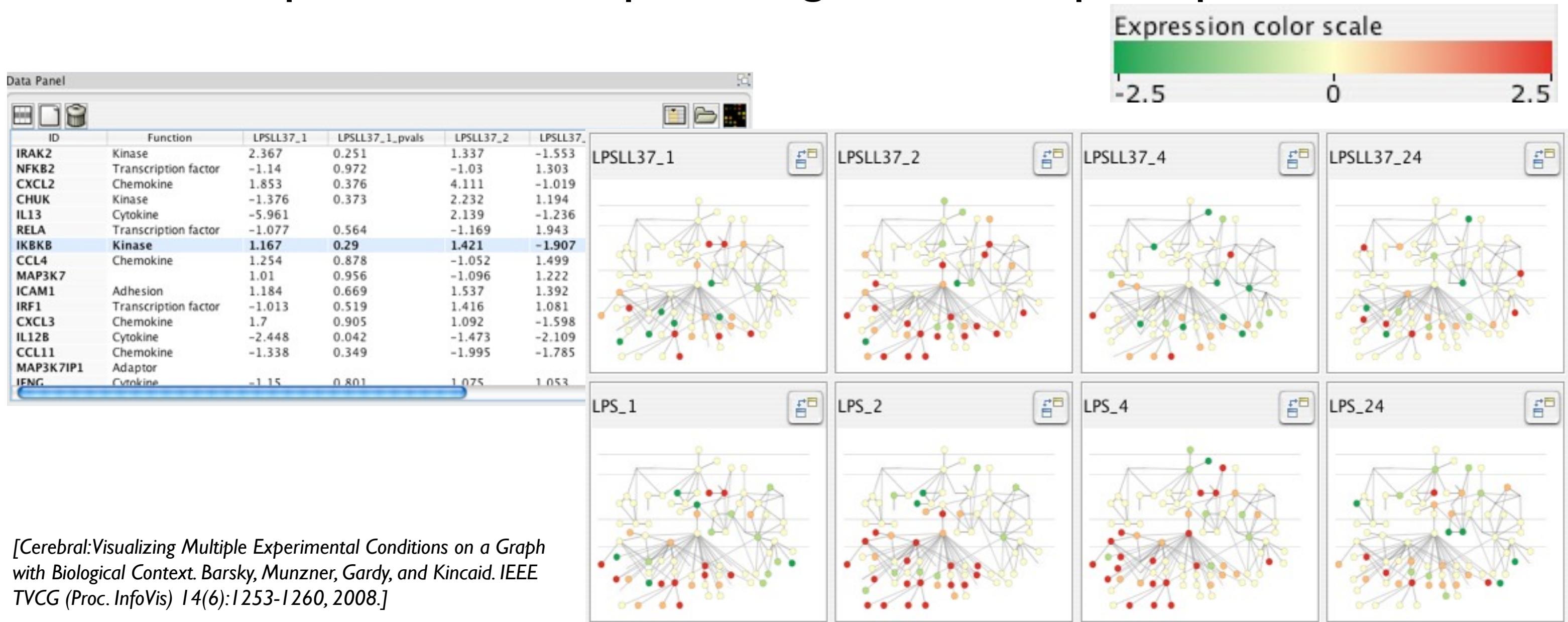
x mean	9
x variance	10
y mean	8
y variance	4
x/y correlation	1



# Why use an external representation?

**Computer-based visualization systems provide visual representations of datasets designed to help people carry out tasks more effectively.**

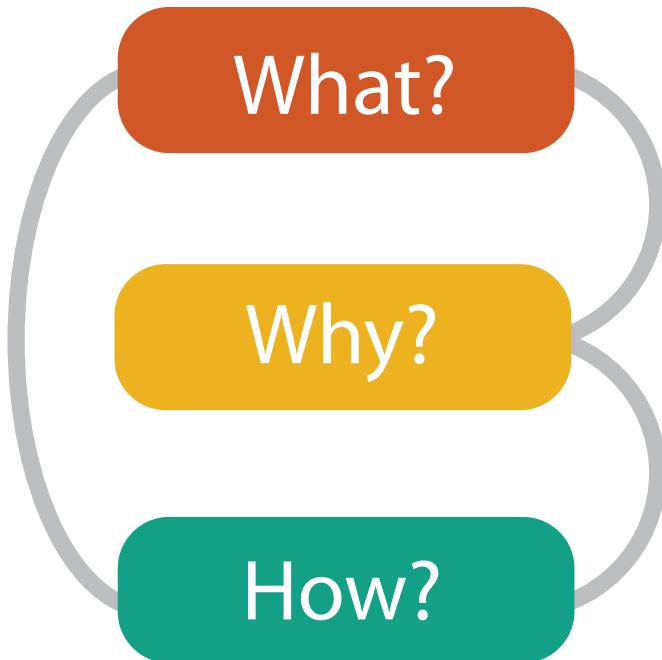
- external representation: replace cognition with perception



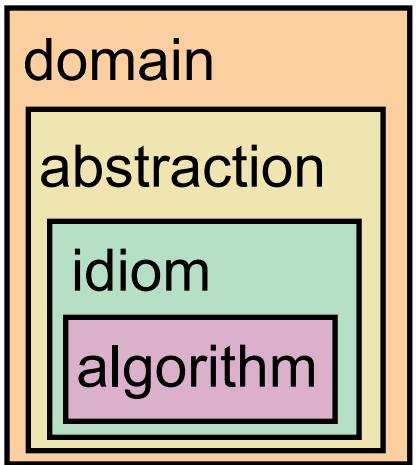
# How to analyze vis design?

Vis usage can be analyzed in terms of what data is shown, why the user needs it, and how the idiom is designed.

- abstractions
  - translate from specifics of domain to vocabulary of vis
    - *data abstraction*: **what** to show
      - might not draw what you're given: transform data into form useful for task
    - *task abstraction*: **why** they're looking at it
- idioms
  - *visual encoding idiom*: **how** to draw
  - *interaction idiom*: **how** to manipulate
- analysis framework: scaffold to think systematically about design space
  - huge, and most possibilities ineffective for particular task/data combination



# How to validate design?



anthropology/  
ethnography

design

computer  
science

cognitive  
psychology

anthropology/  
ethnography

## 👤 Domain situation

Observe target users using existing tools



## Data/task abstraction



## Visual encoding/interaction idiom

Justify design with respect to alternatives



## Algorithm

Measure system time/memory

Analyze computational complexity

Analyze results qualitatively

Measure human time with lab experiment (*lab study*)

Observe target users after deployment (*field study*)

Measure adoption



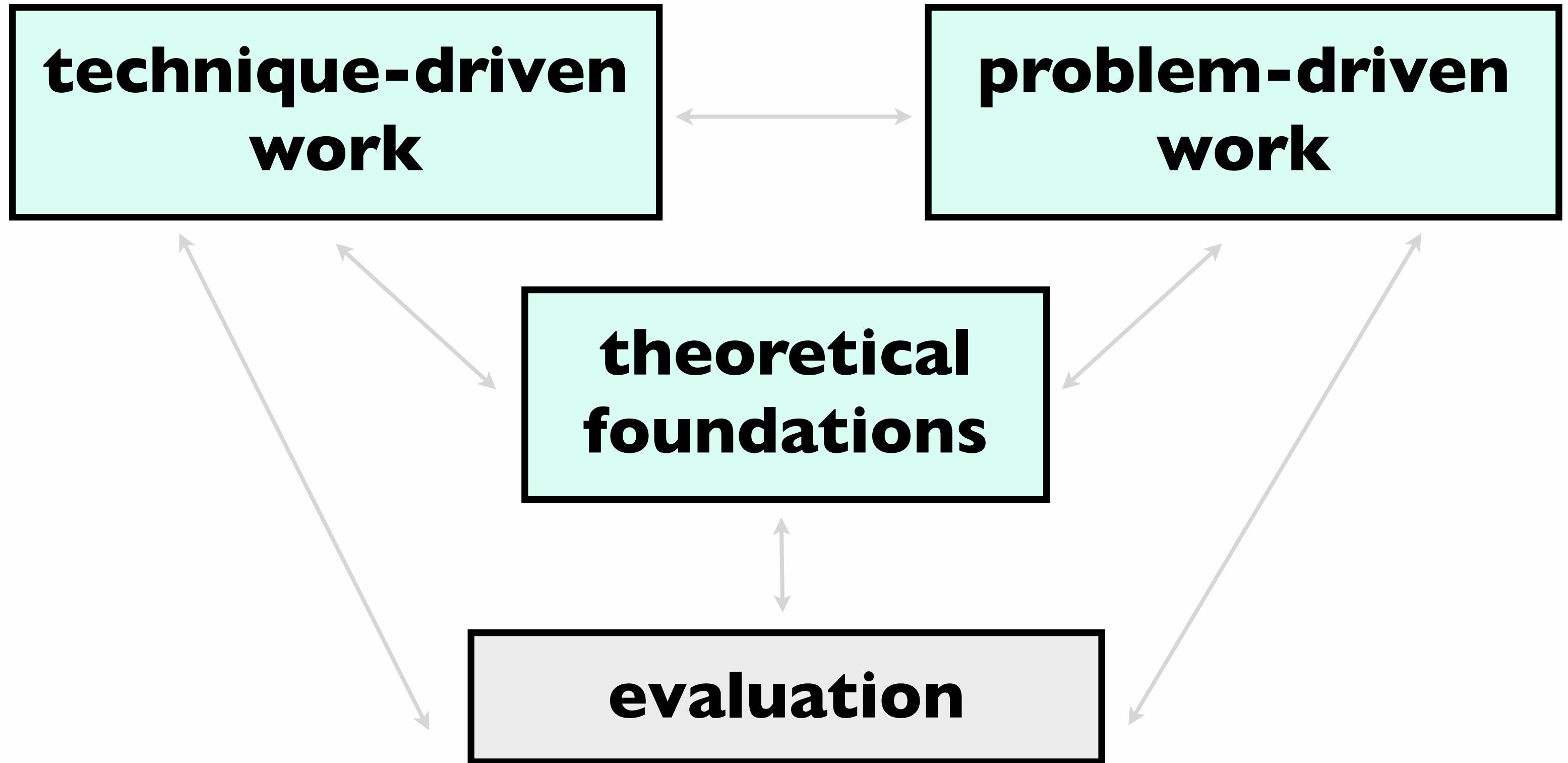
problem-driven  
work

technique-driven  
work

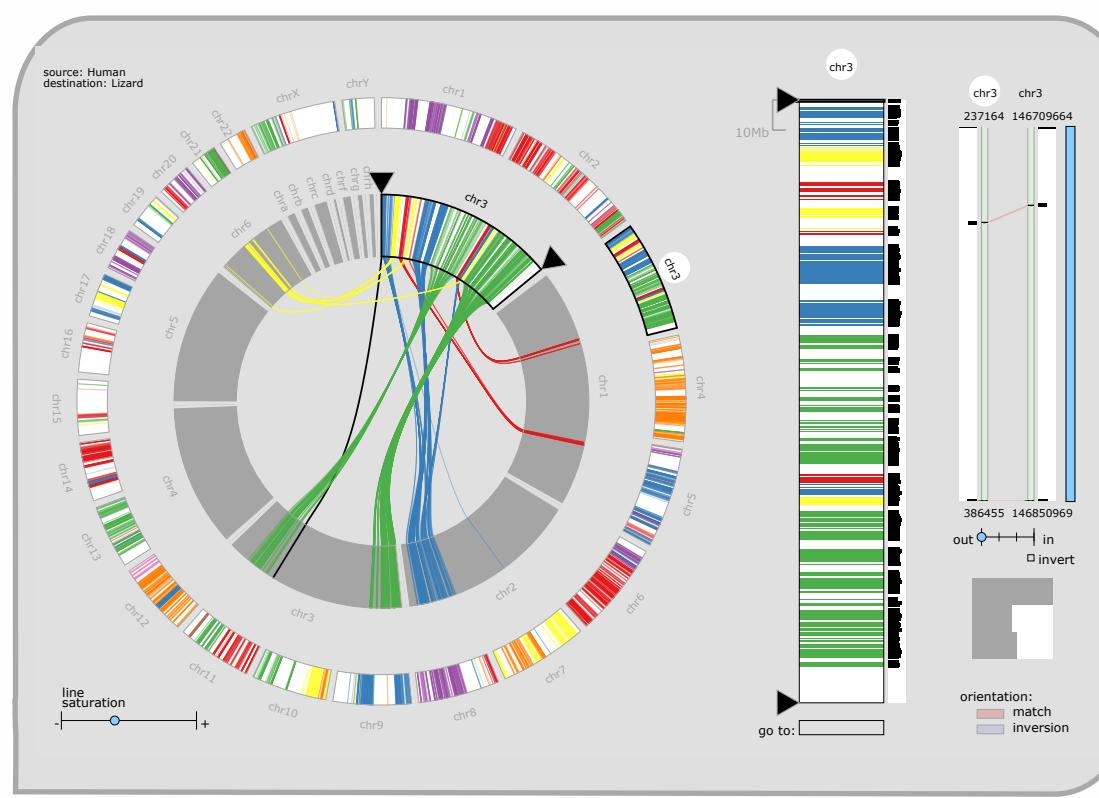
[A Nested Model of Visualization Design and Validation.

Munzner. *IEEE TVCG* 15(6):921-928, 2009 (Proc. InfoVis 2009). ]

# Angles of attack

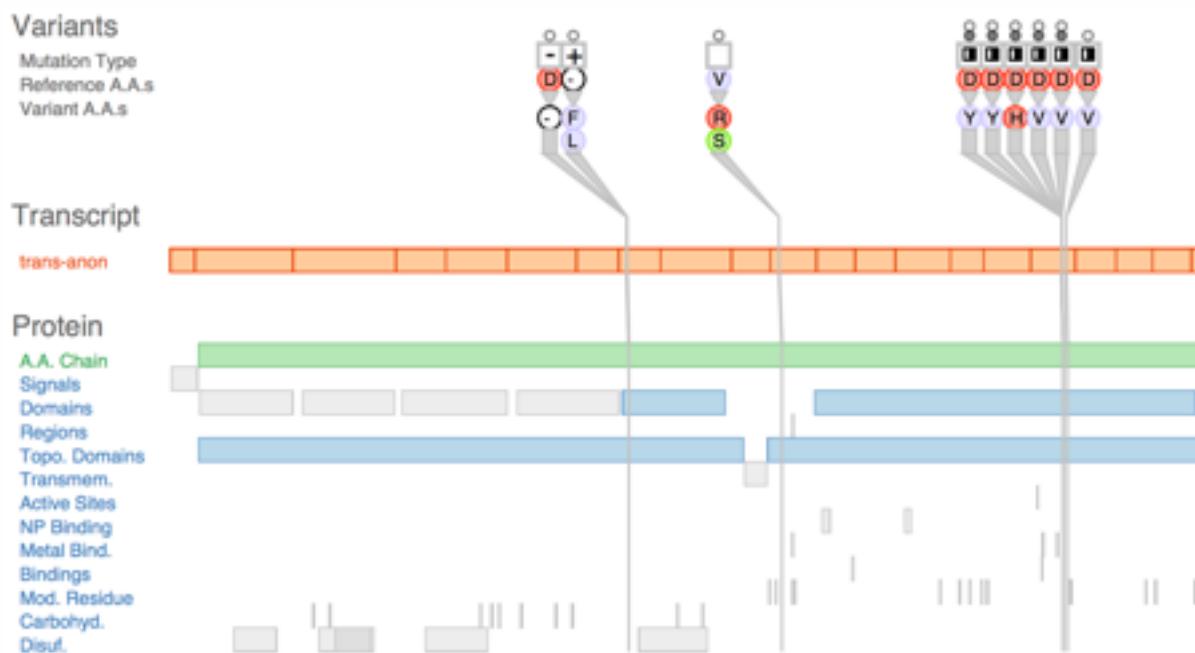


# Problem-driven work: Genomics

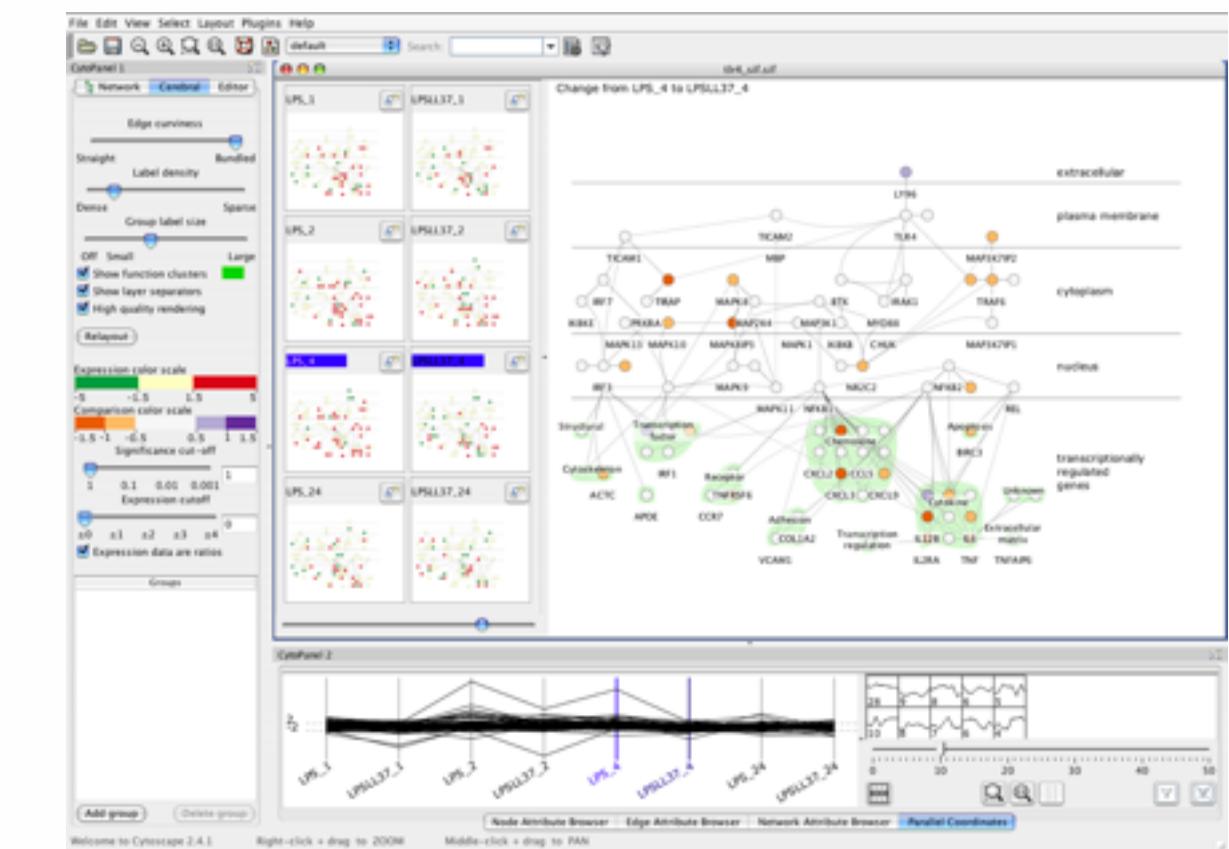


**MizBee**

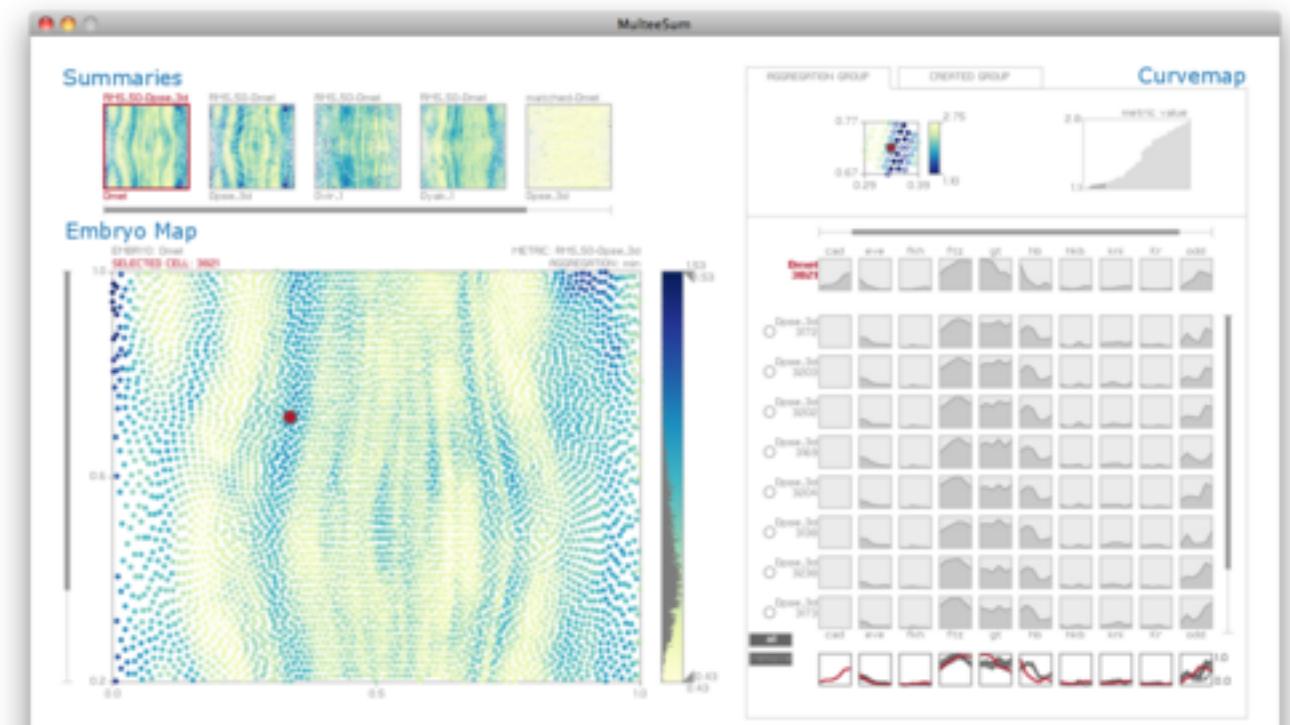
<http://youtu.be/86p7brwuz2>



# Variant View

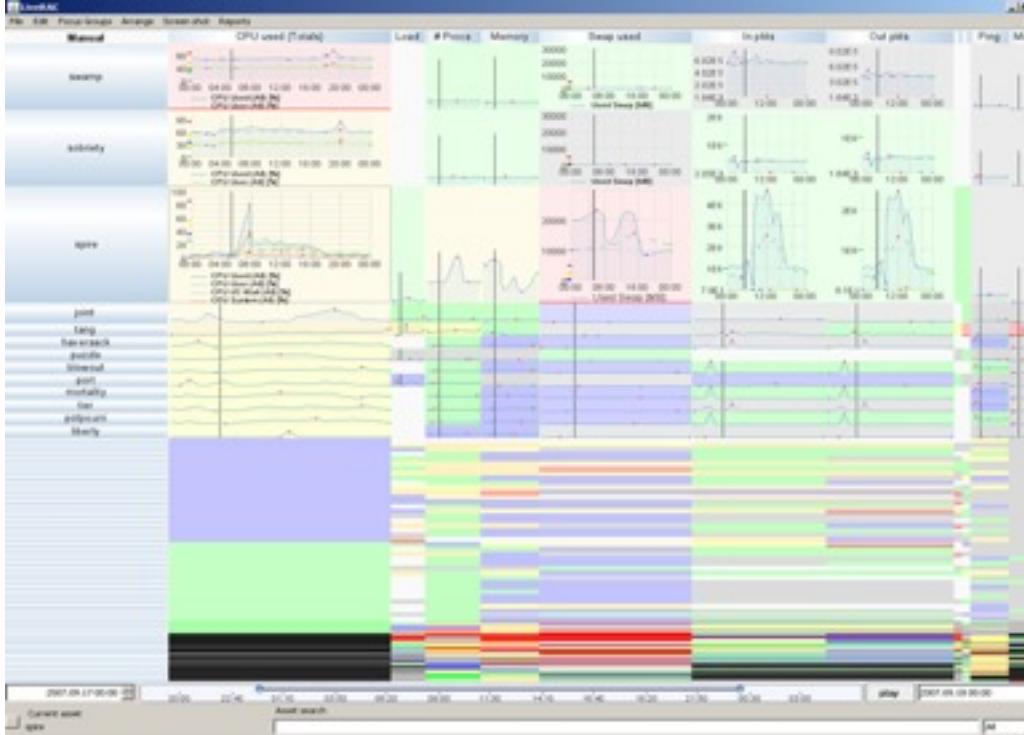


Cerebra

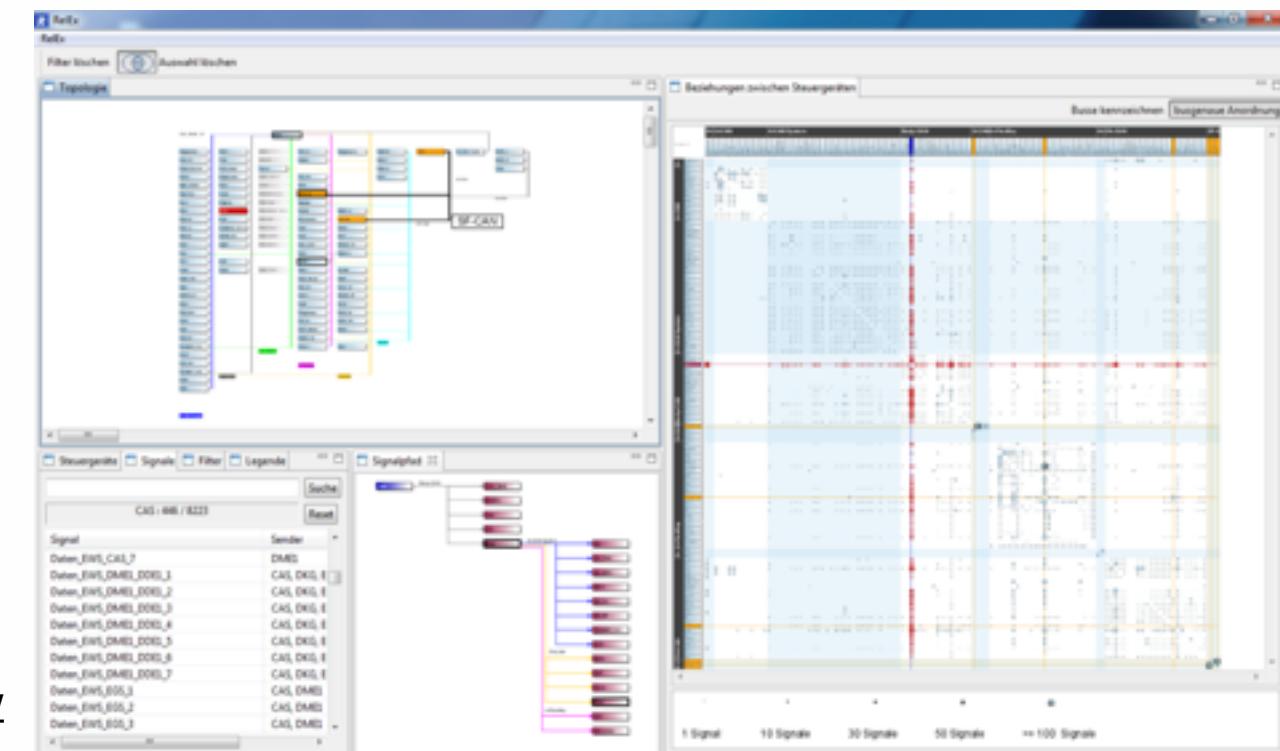


# MulteeSum Pathline

# Problem-driven work: Many other domains



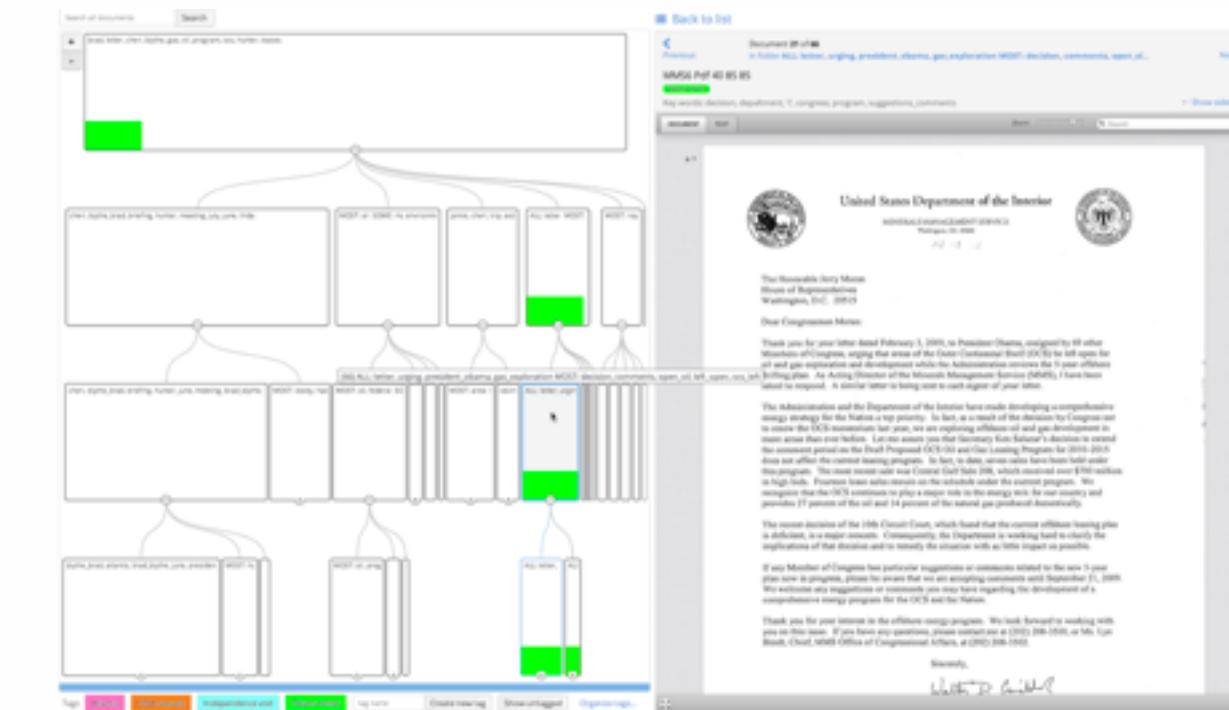
<http://youtu.be/l0c3H0VSkw>



# LiveRAC: system management time-series



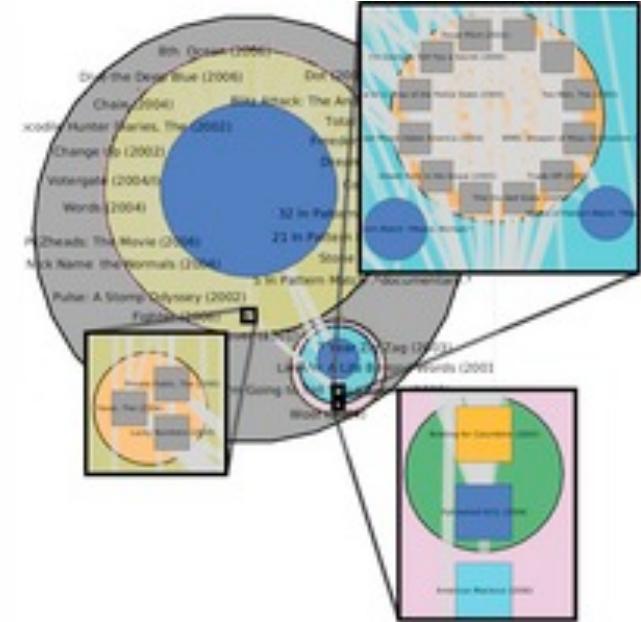
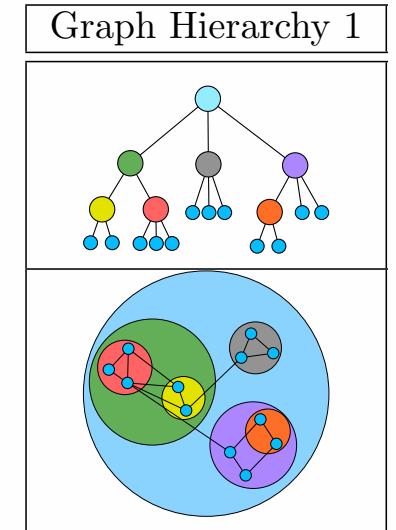
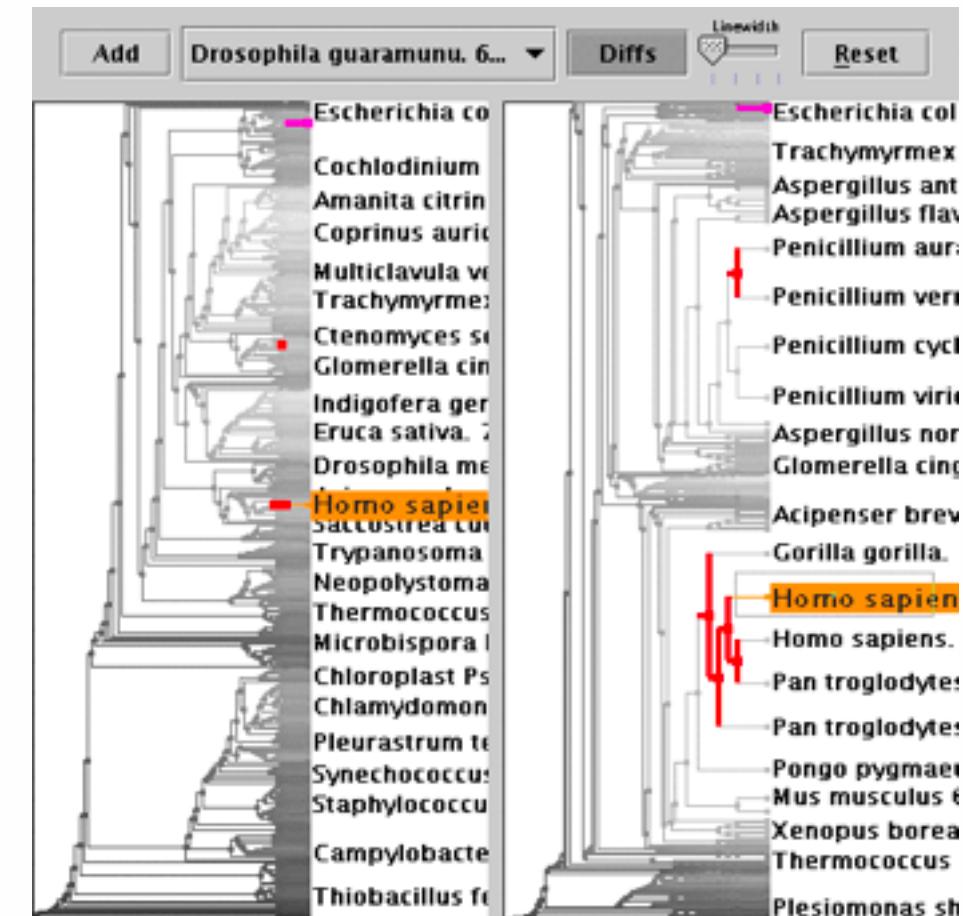
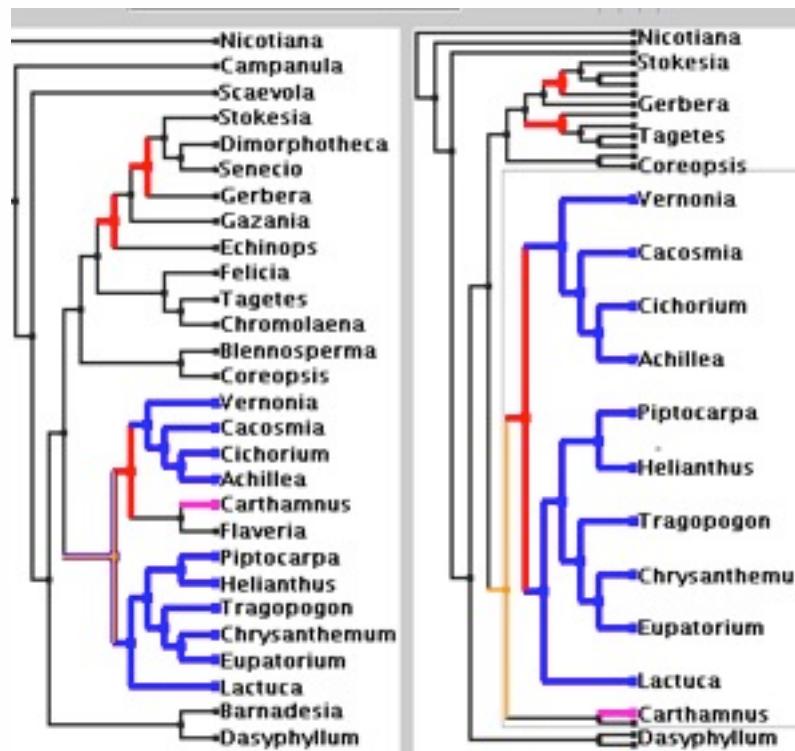
**Vismon: fisheries management** <http://youtu.be/h0kHoS4VYmk>



**Overview: investigative journalism** <http://vimeo.com/71483614>

# Technique-driven work: Networks

- scaling up networks
  - multilevel networks, 10K-100K nodes
    - topologically aware decomposition, layout, browsing
  - trees, millions of nodes
    - guaranteed visibility of semantically meaningful marks



**TopoLayout**  
**Smashing Peacocks Further**  
**Grouse**  
**GrouseFlocks**  
**TugGraph**

<http://youtu.be/t1Xbt6XOWp8>

<http://youtu.be/AWXAe8zvkt8>

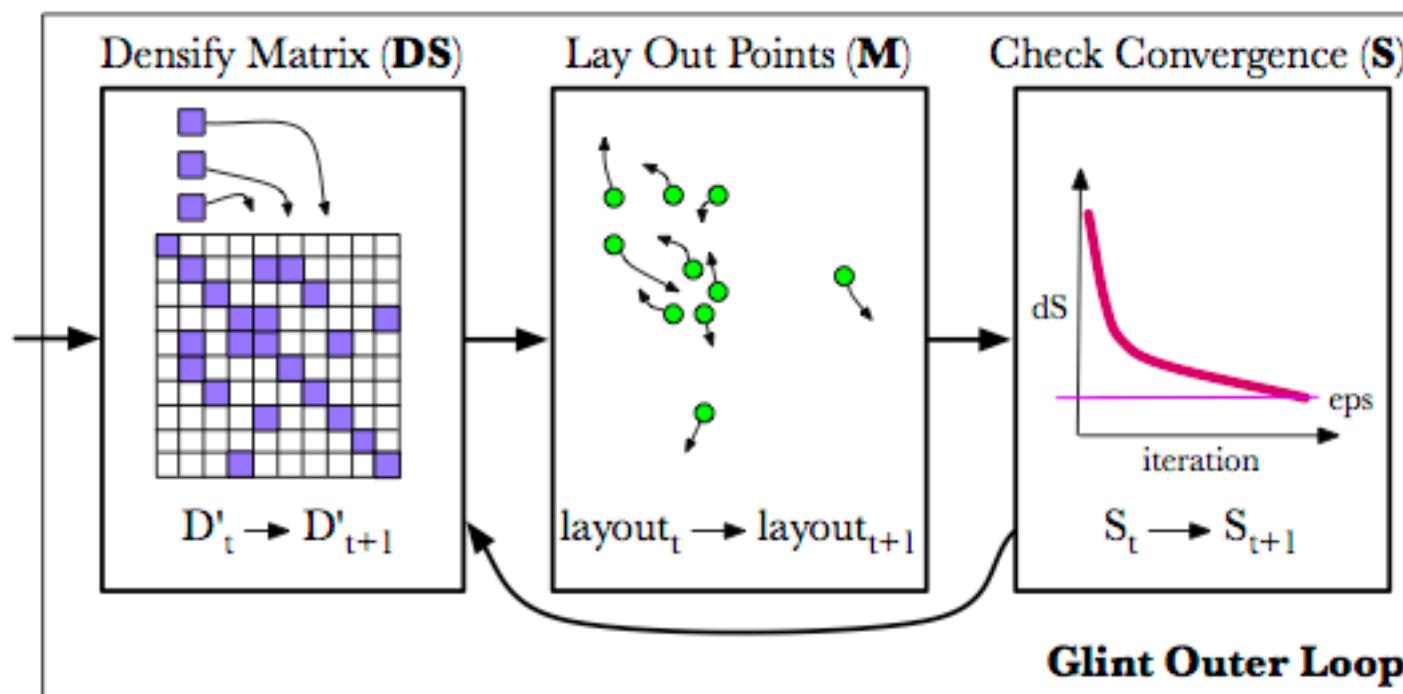
**TreeJuxtaposer**  
**PRISAD**

<http://youtu.be/fq8EIAOutvs>

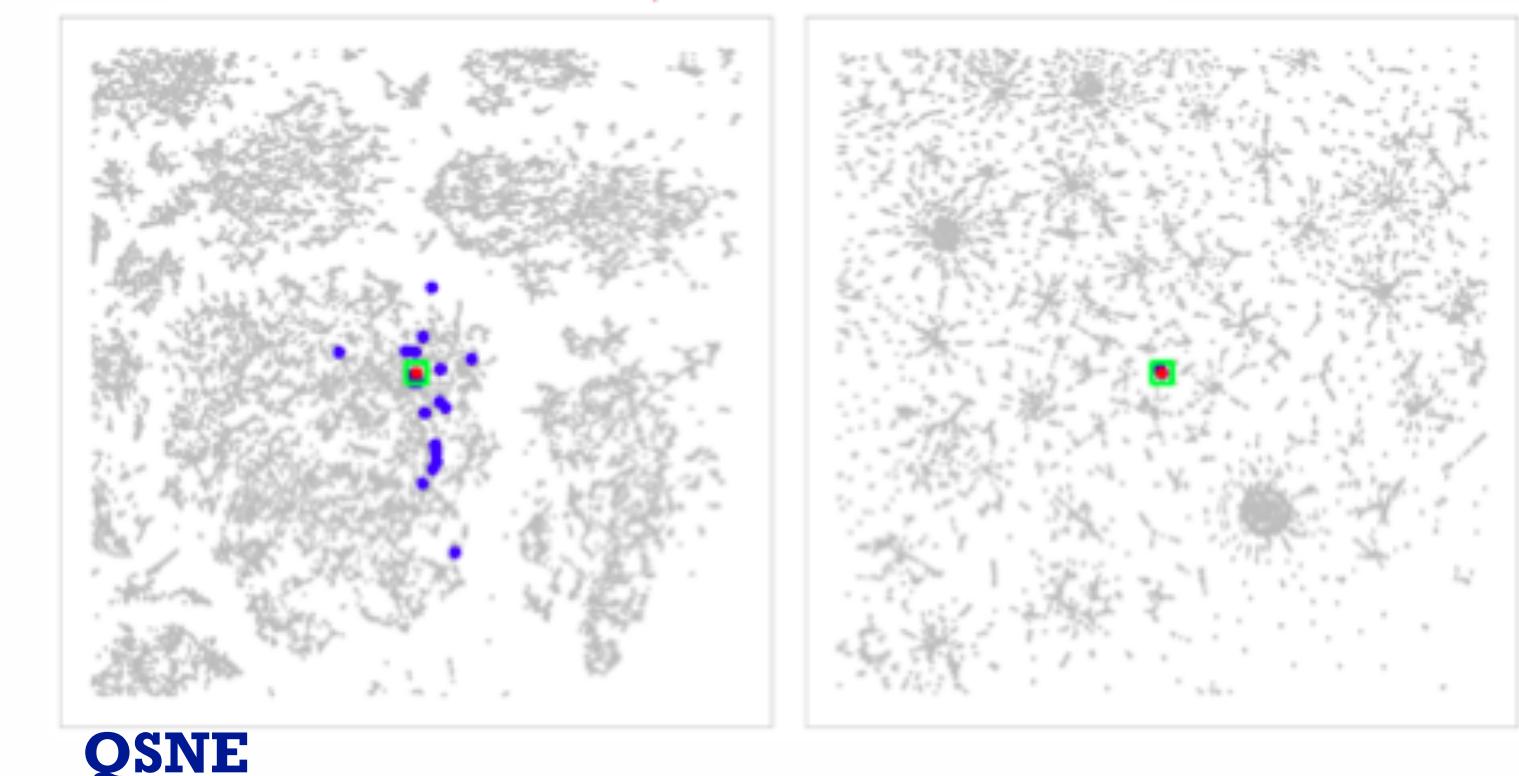
<http://youtu.be/GdaPj8a9QEo>

# Technique-driven work: Dimensionality reduction

- close overlap with machine learning
  - Glimmer: MDS on the GPU
  - Glint: DR for costly distances
  - QSNE: sparse documents
    - high quality for millions of items



**Glint**



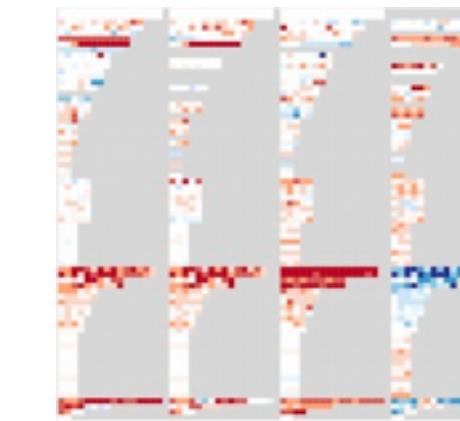
**Glimmer**

<http://youtu.be/PLaBAPM6qLI>



# Evaluation: Dimensionality Reduction

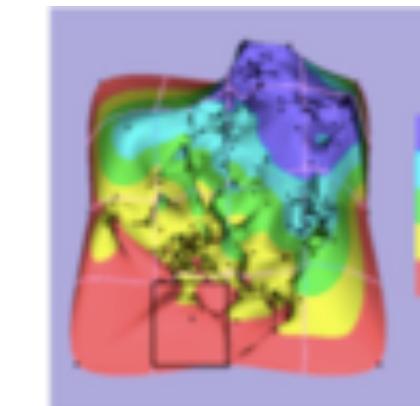
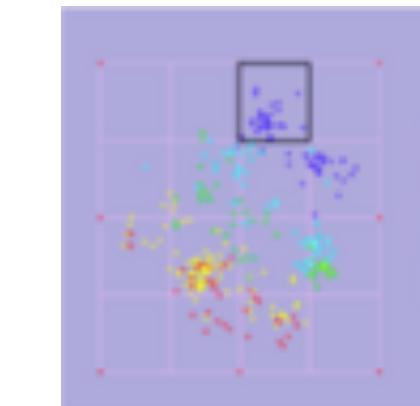
- guidance on scatterplot/DR choices



- taxonomy of cluster separation factors

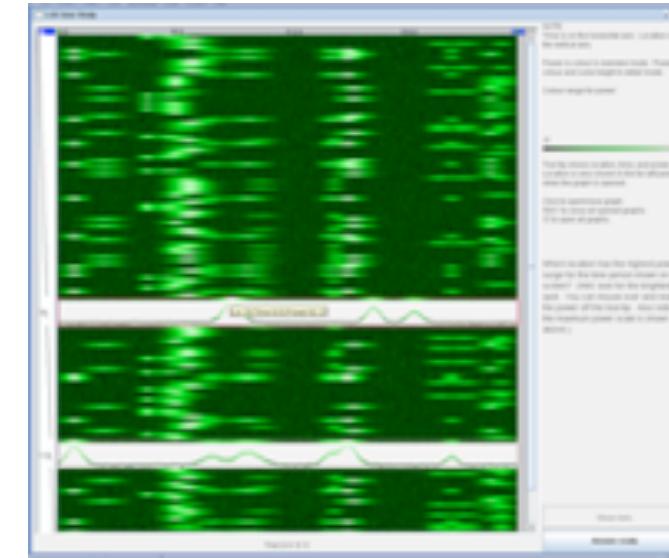


- 2D points vs 3D landscapes

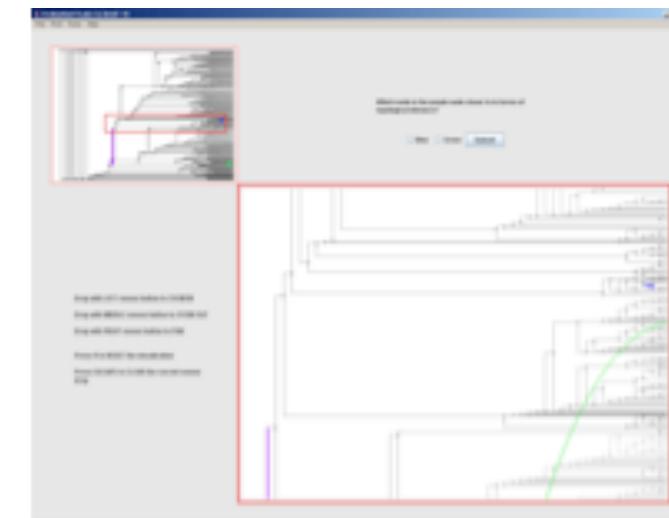


# Evaluation: Focus+Context

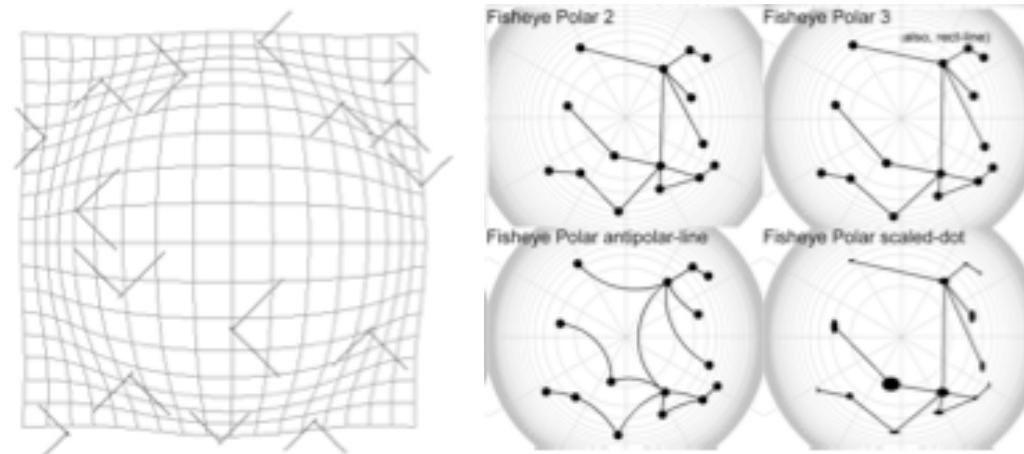
- overviews: separate vs. integrated views



- navigation: stretch and squish vs. pan/zoom navigation

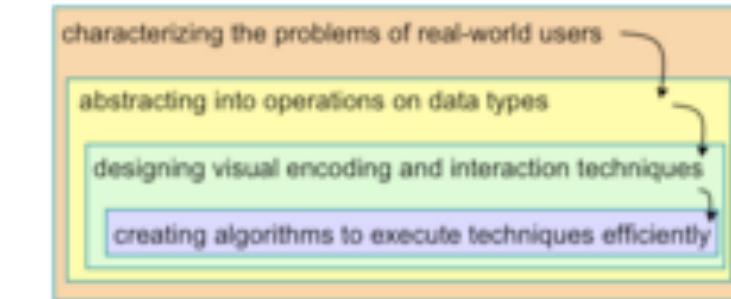
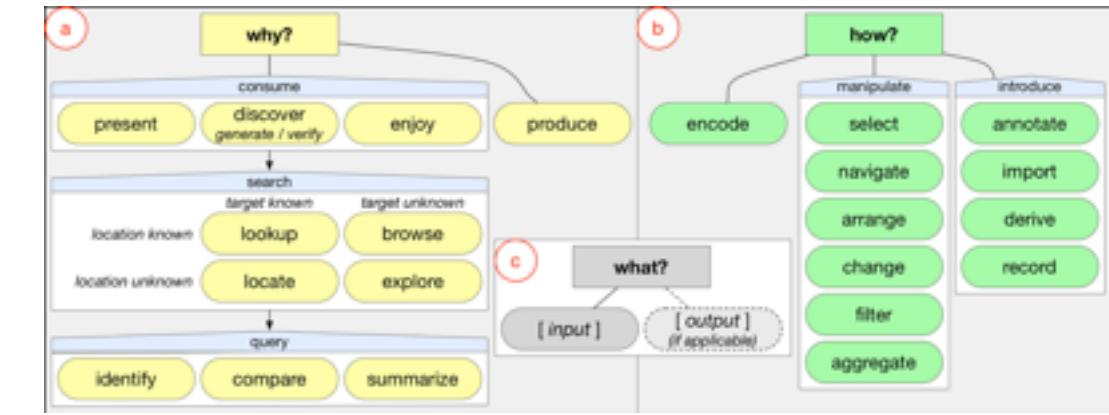


- impact of distortion on visual search, visual memory

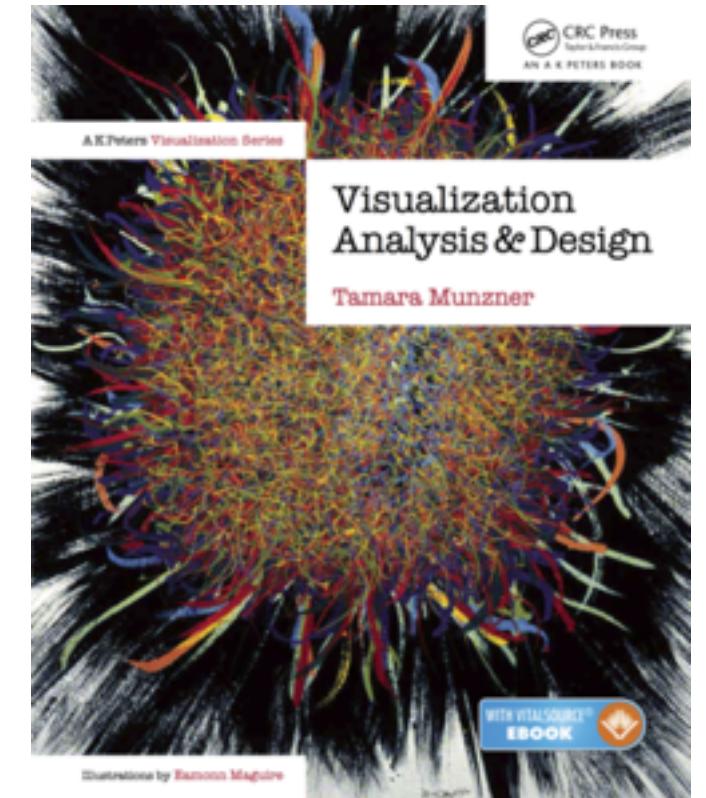


# Theory/Models

- multi-level typology of abstract visualization tasks
- design study methodology
- nested model for vis design and validation
- papers process and pitfalls
- book: Visualization Analysis and Design

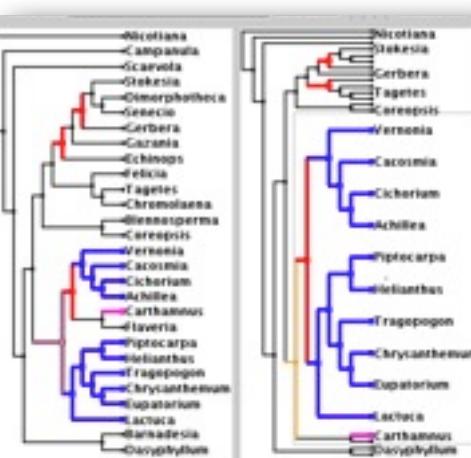


- Type Pitfalls
  - + Design in Technique's Clothing
  - + Application Blings versus Design Study
  - + All That Coding Means I Deserve A Systems Paper
  - + Neither Fish Nor Fowl
- Visual Encoding Pitfalls
  - + Unjustified Visual Encoding
  - + Hammer In Search Of Nail
  - + 2D Good, 3D Better
  - + Color Cacophony



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- wrapup



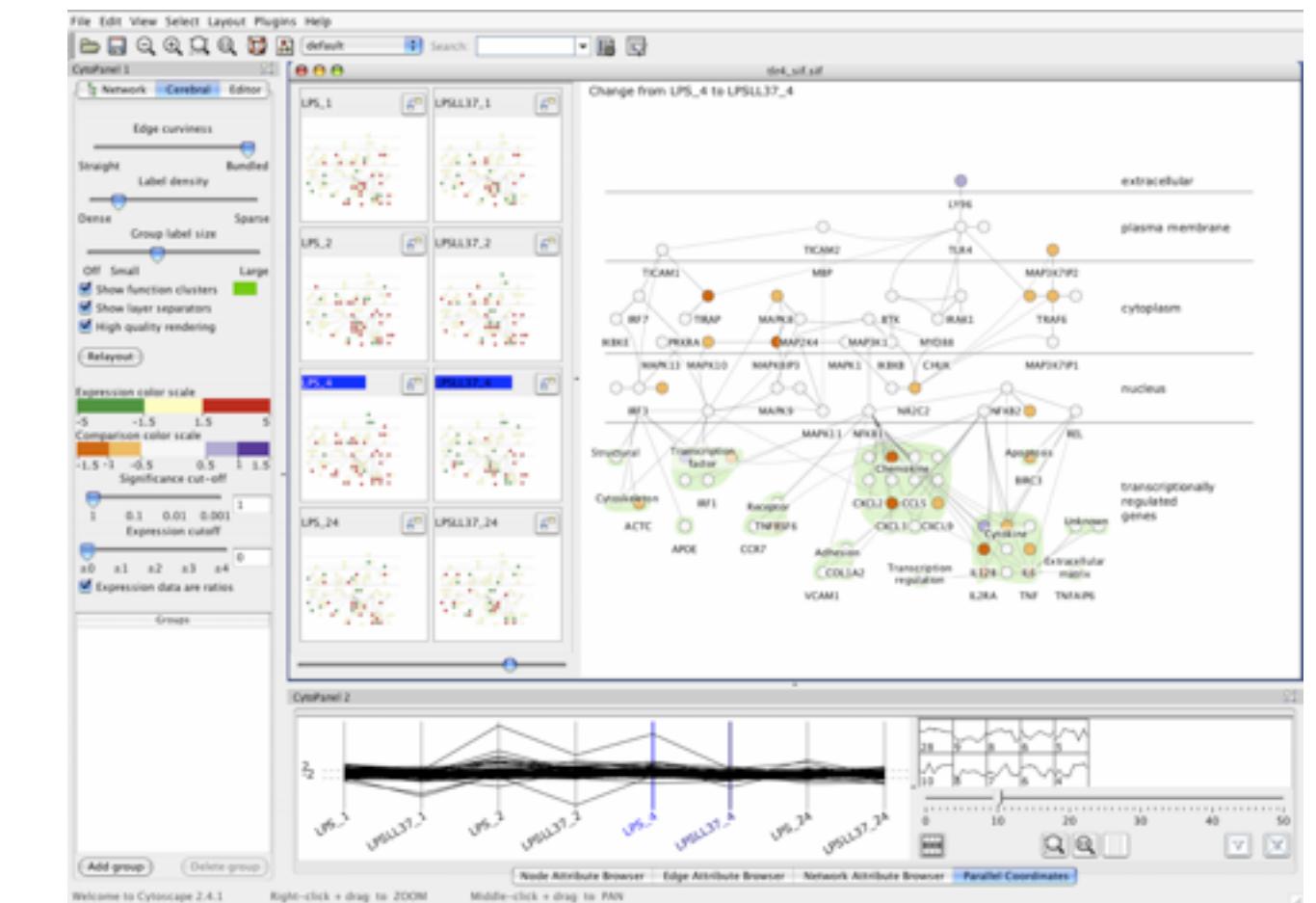
# Cerebral

*Visualizing Multiple Experimental Conditions on a Graph with Biological Context*

**joint work with:**

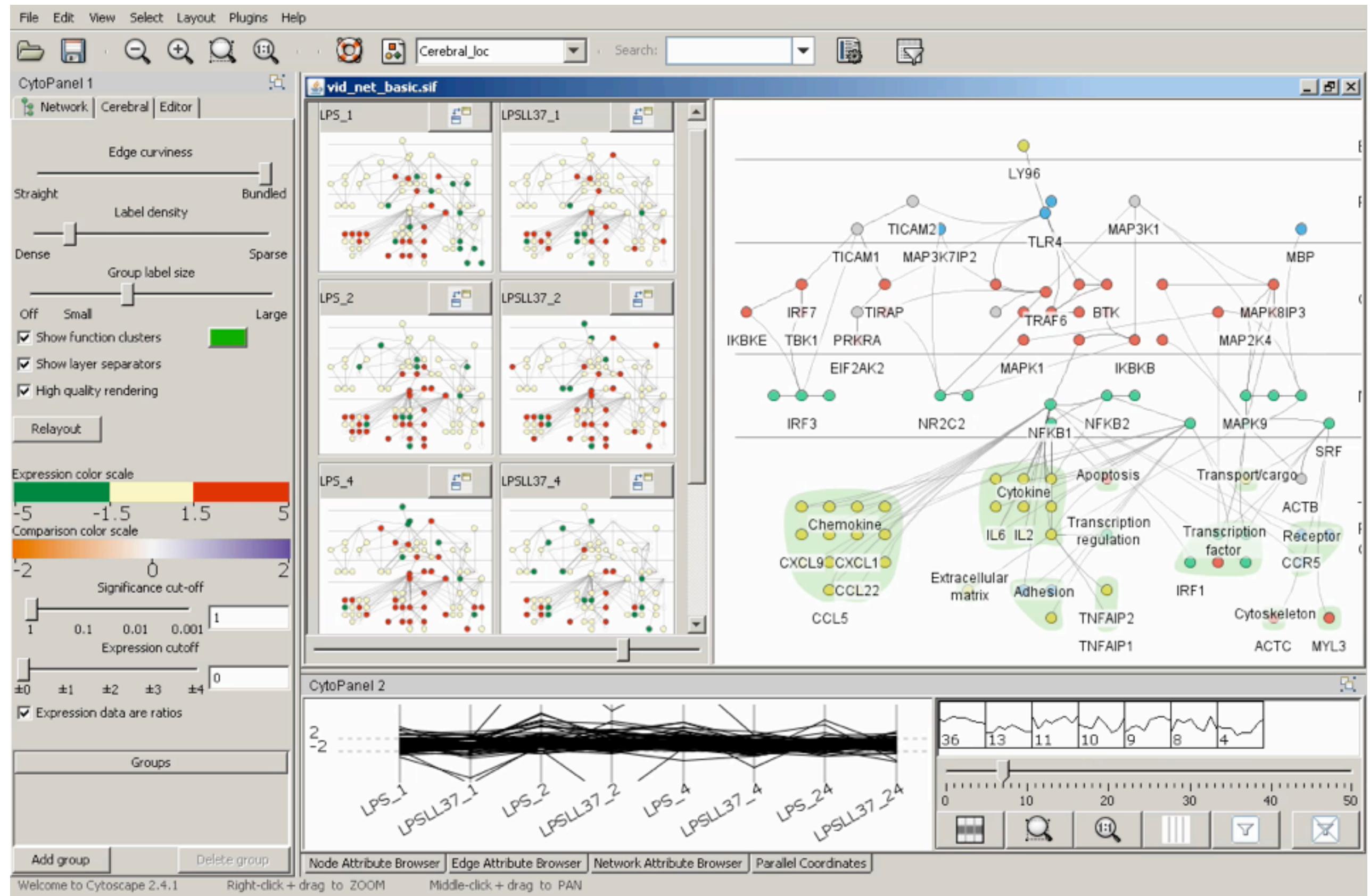
Aaron Barsky, Jennifer Gardy, Robert Kincaid

<http://www.pathogenomics.ca/cerebral/>



Cerebral: Visualizing Multiple Experimental Conditions on a Graph with Biological Context.  
Barsky, Munzner, Gardy, Kincaid. IEEE Trans. Visualization and Computer Graphics 14(6):1253-1260 2008. (Proc. InfoVis 2008).

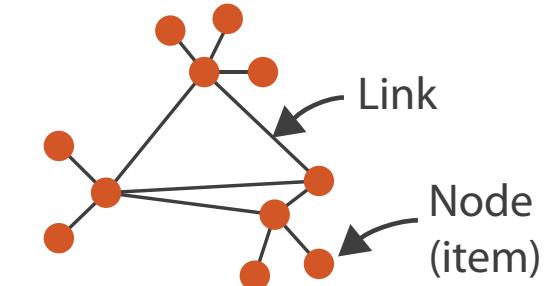
# Cerebral video



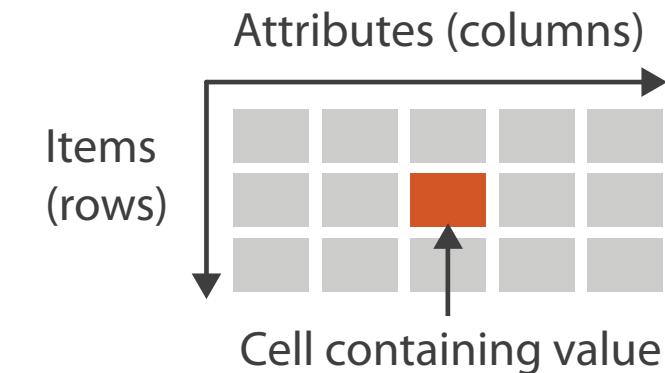
# What: Data abstraction

- dataset types
  - network
    - nodes: genes
    - links: known interactions between genes
  - table
    - quantitative attributes
      - gene expression levels for nodes across different experimental conditions
    - categorical attributes
      - subcellular location of interaction
      - functional groups

→ Networks



→ Tables



➔ Attribute Types

➔ Categorical



➔ Ordered

➔ Ordinal



➔ Quantitative



What?

Why?

How?

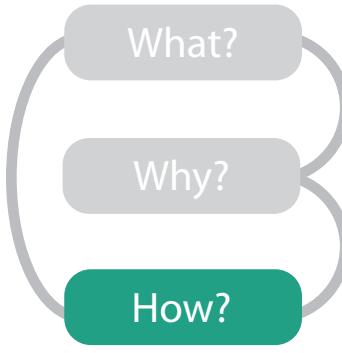
# Why: Task abstraction

- task: interpret microarray experiment results with respect to gene network
  - goal: accelerate existing discovery workflow
  - compare distributions between attributes
    - experimental conditions
  - interpret attributes in context of current network topological structure



# How: Idiom design decisions

- arrange space for networks
  - custom node-link diagram layout
    - points for nodes
    - connection marks for links
  - vertical compartment according to subcellular location attribute
  - cluster according to functional grouping



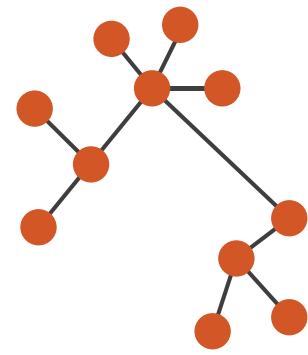
## Arrange Networks And Trees

### → Node-link Diagrams

Connections and Marks

NETWORKS

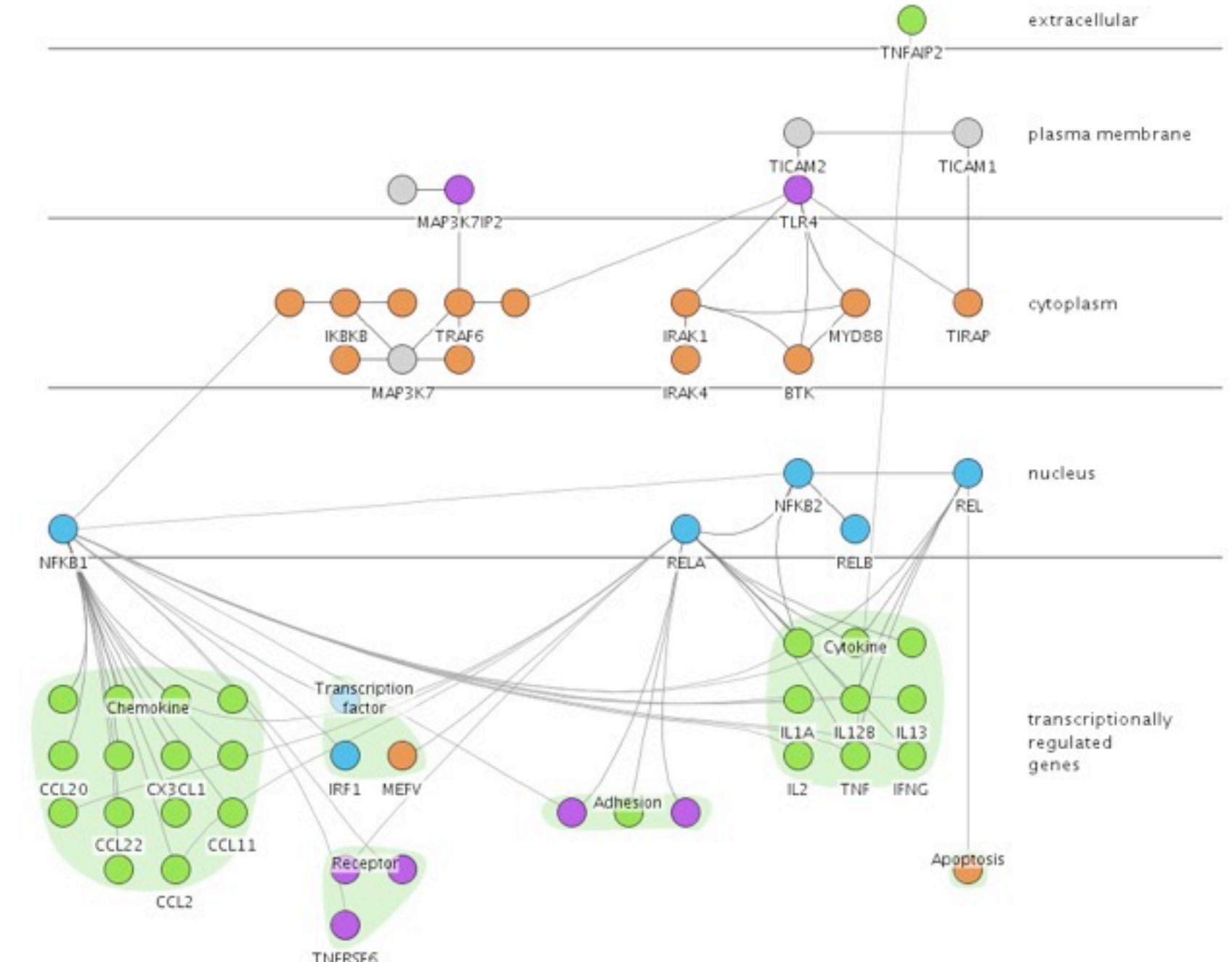
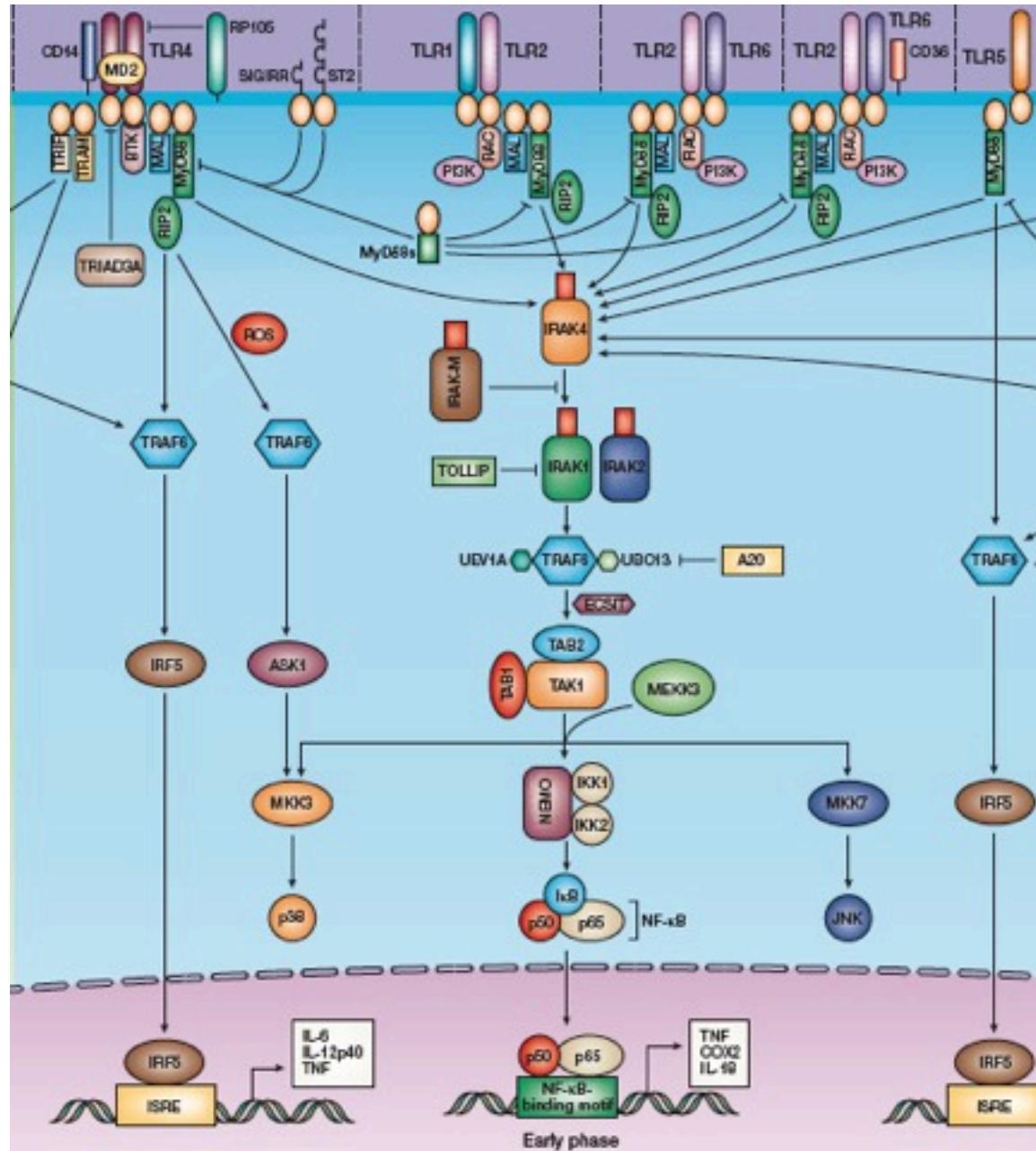
TREES



# How: Arrange space

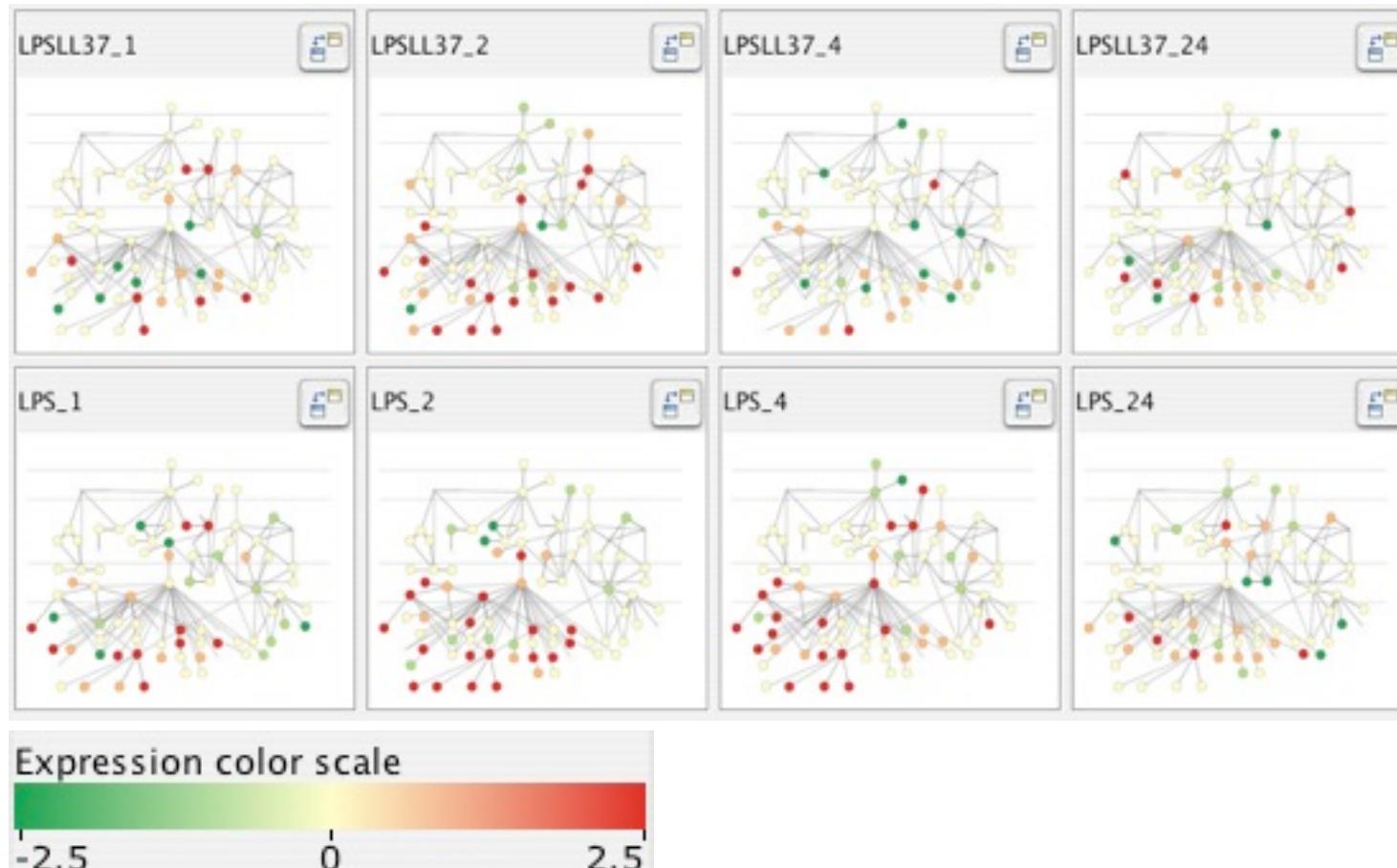
- automatic layout similar to hand-drawn diagrams
  - vertical compartment according to subcellular location attribute

What?  
Why?  
How?



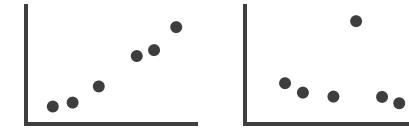
# How: Idiom design decisions

- facet: partition data into multiple views
  - juxtapose views side by side
    - same encoding, different data: *small multiples*
    - nodes in each view colored by expression levels for experimental condition



Facet

→ Juxtapose

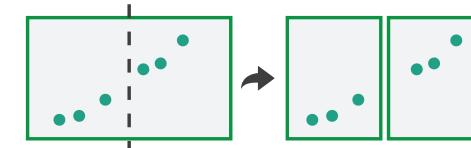


What?

Why?

How?

→ Partition

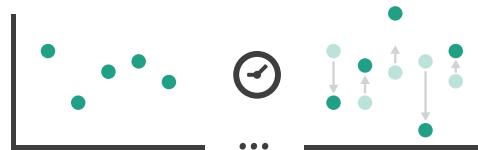


		Data		
		All	Subset	None
Encoding	Same	Redundant	Overview/ Detail	Small Multiples
	Different	Multiform		
		Multiform	Overview/ Detail	No Linkage

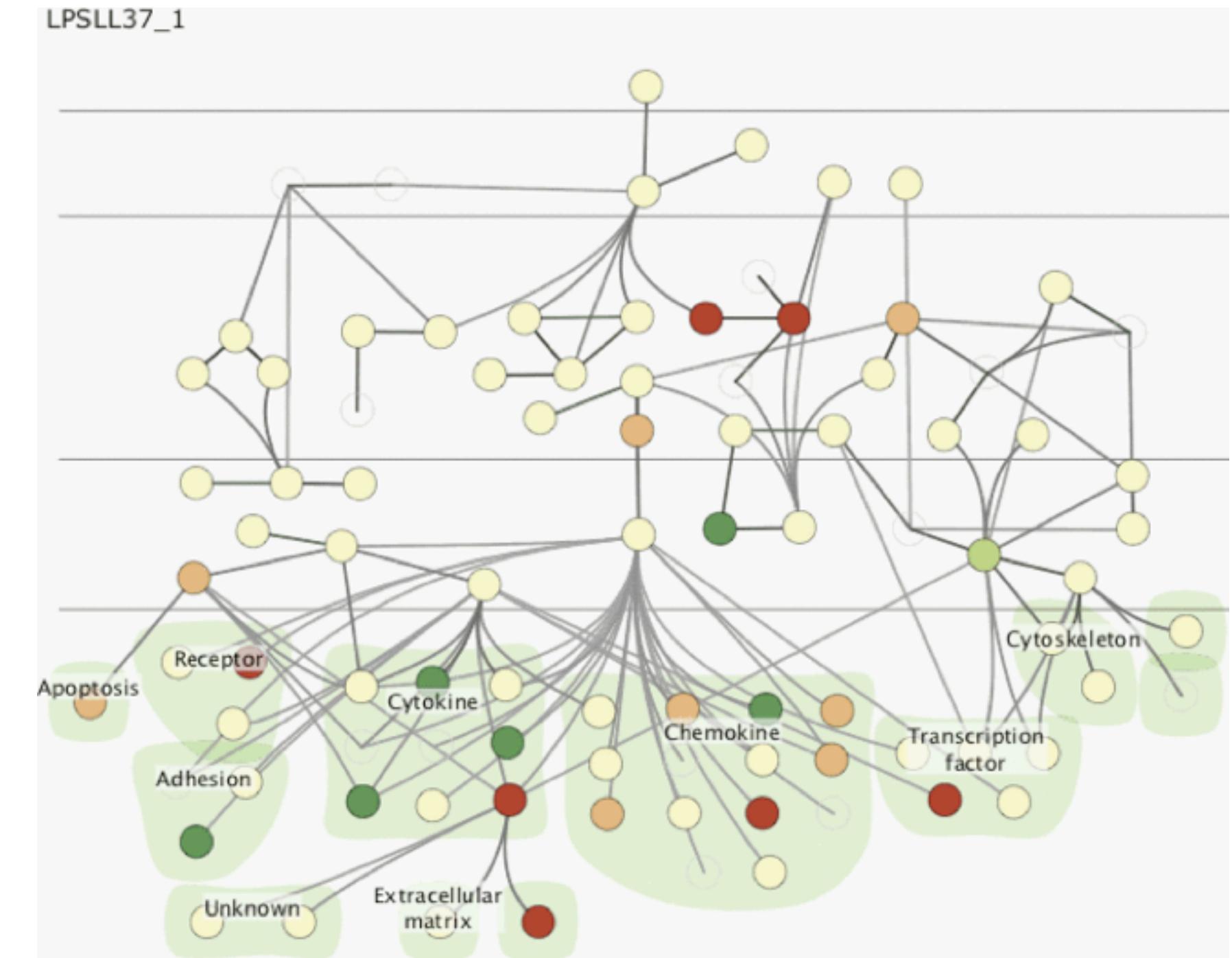
# How: Juxtapose vs. animate

## Manipulate

### → Change



- comparison difficult across many frames with many changes everywhere
- rule of thumb: eyes beat memory
  - principle: external cognition vs. internal memory
    - easy to compare by moving eyes between side-by-side views
    - harder to compare memory of what you saw to visible view



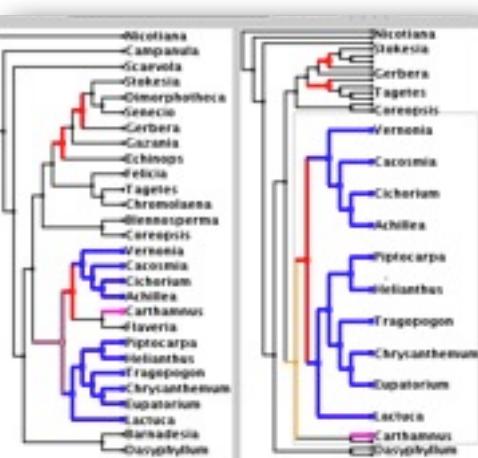
# Cerebral contributions

- multiple juxtaposed views support interactive comparison between gene expression level experimental data and network context
- automatic network layout algorithm in spirit of hand drawn diagrams
  - localization and functional group attributes affect spatial position
- open source
  - Cytoscape plugin
  - InnateDB database integration

<http://www.pathogenomics.ca/cerebral/>

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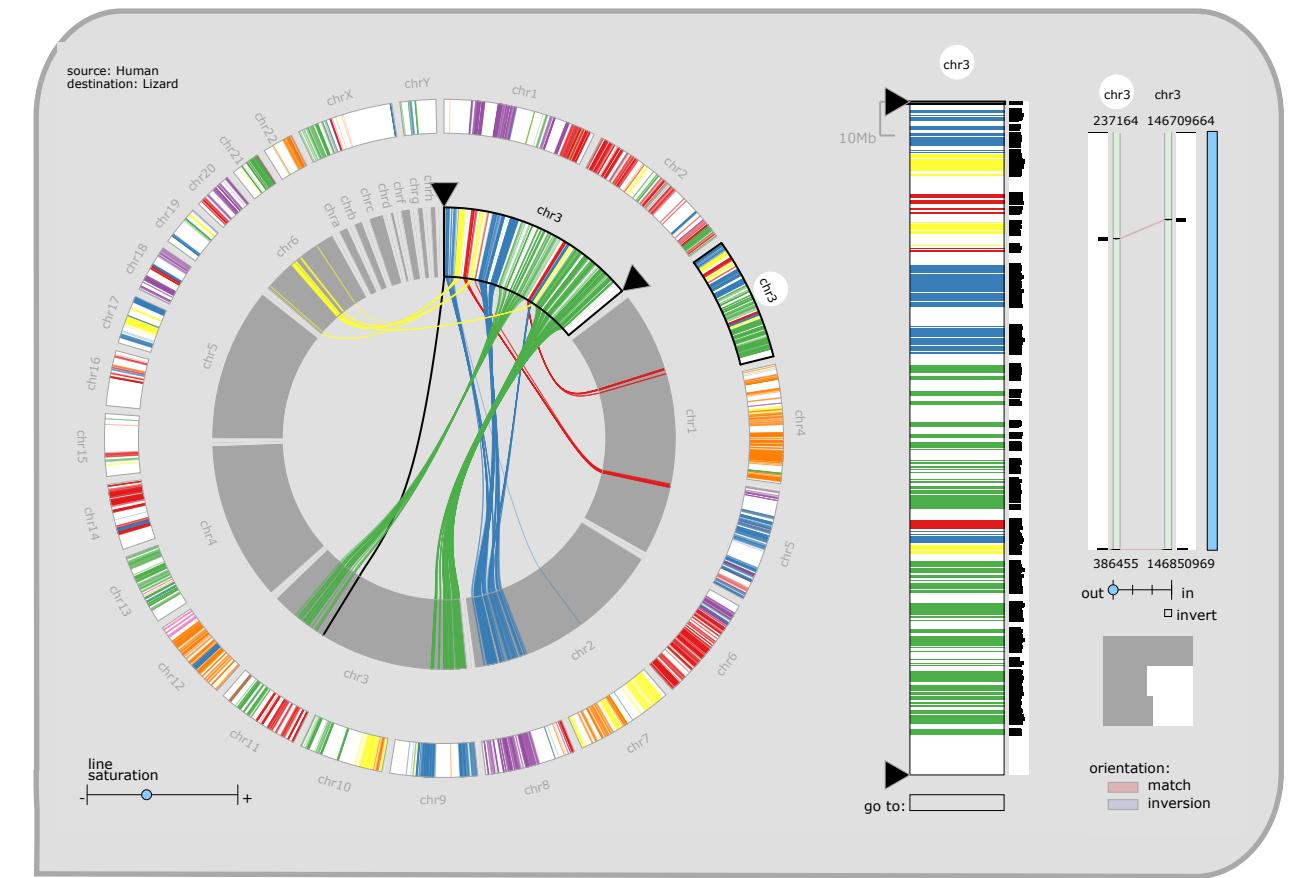
# MizBee

## A Multiscale Synteny Browser

**joint work with:**

Miriah Meyer, Hanspeter Pfister

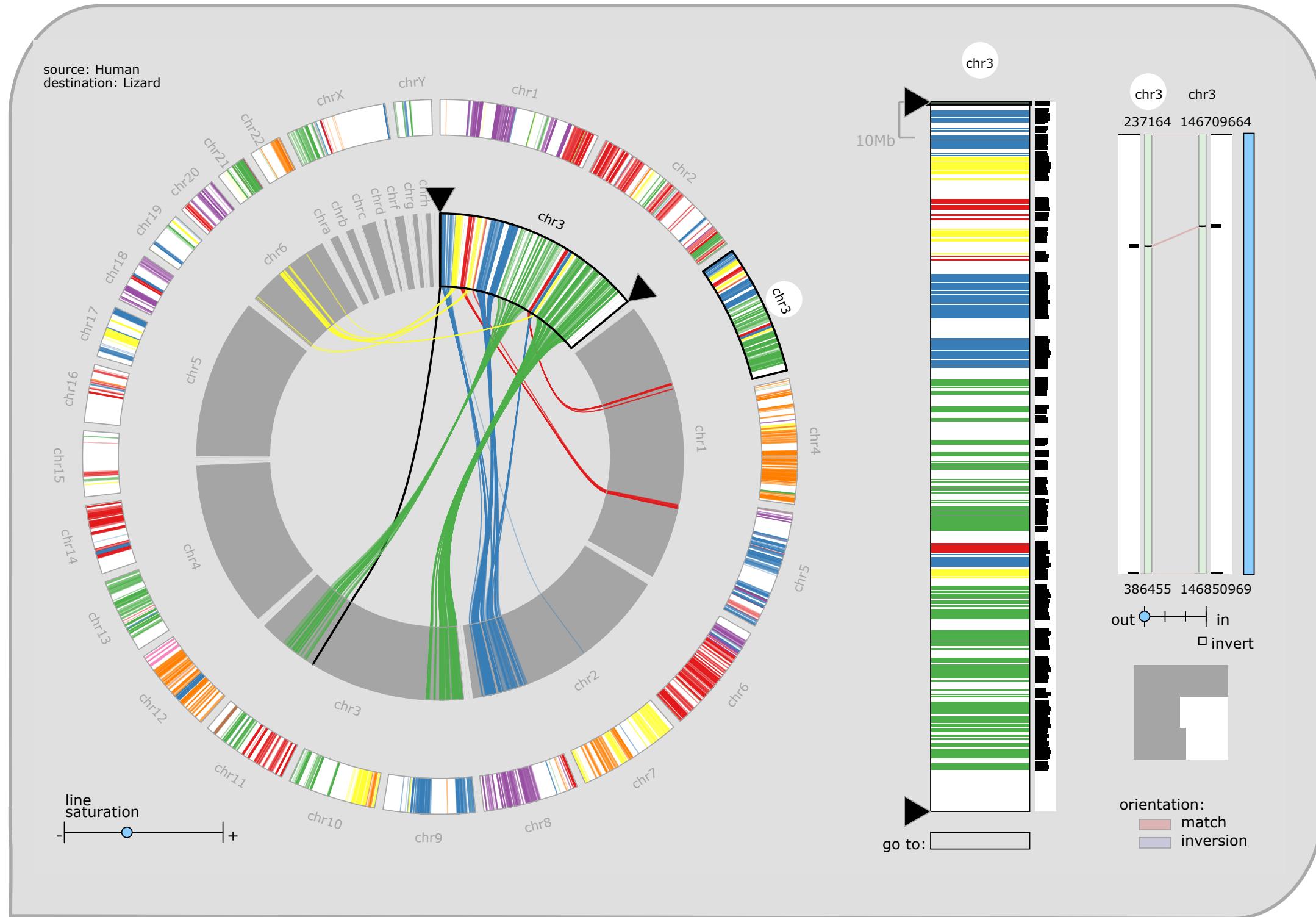
<http://www.cs.utah.edu/~miriah/mizbee>



MizBee: A Multiscale Synteny Browser.

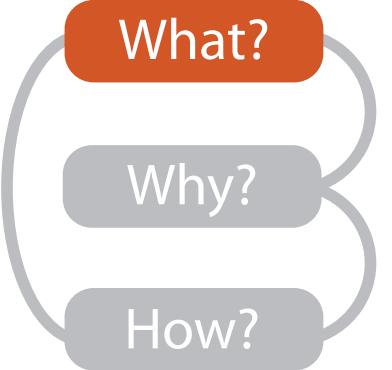
Meyer, Munzner, Pfister. IEEE Trans. Visualization and Computer Graphics 15(6):897-904, 2009 (Proc. InfoVis 2009).

# MizBee video

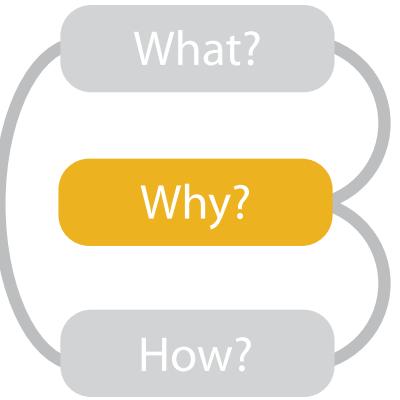


# What: Data abstraction

- data: multiscale lists
  - features: hundreds of thousands
    - ordered attribute: position in chromosome sequence coordinates
    - categorical attributes: orientation, chromosome of matching feature
    - quantitative attributes: length, similarity score
  - syntenic blocks: thousands
    - contiguous sets of features on same chromosome
    - combine thresholded features if
      - destination chromosome and orientation match
      - close together
  - chromosomes: dozens
  - genomes: two



# Why: Tasks in domain language



- analyze conservation (similarity) relationships between genomic features
  - high-level biology questions
    - evolution
      - how long ago did two species share common ancestor?
    - function
      - which segment of the genome is responsible for specific function in the cell?
    - ...
  - low-level data-centric questions
    - algorithm refinement
      - are paired features within a block contiguous?
      - which chromosomes share conserved blocks?
      - are similarity scores alike within block?
      - ...

# Why: Tasks abstraction



relationship scale

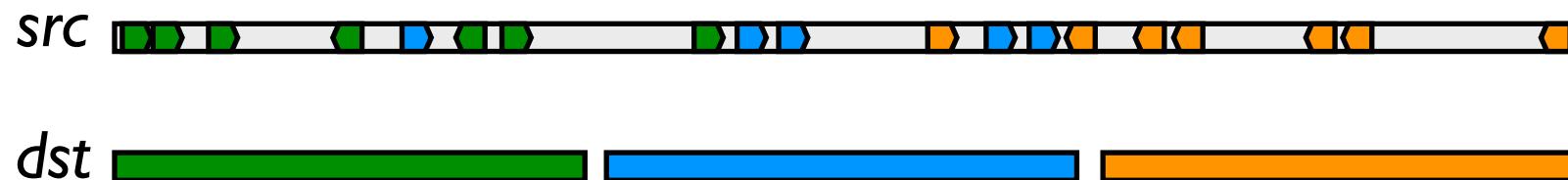
- relationship types: proximity, size, orientation, similarity
- data scales: genome, chromosome, block, feature
- topics: algorithm in/out, block reliability, high-level science

	genome	chromosome	block	feature	proximity / location	size	orientation	similarity
Which chromosomes share conserved blocks?	x				x			
For one chromosome, how many other chromosomes does it share blocks with?	x	x			x			
What is the density of coverage and where are the gaps on: chromosomes? blocks?	x	x	x		x			
Where are the blocks: on chromosomes? around a specific location on a chromosome?	x	x			x			
What are the sizes and locations of other genomic features near a block?			x		x	x		
How large are the blocks?			x				x	
Do neighboring blocks go to the same: chromosomes? relative location on a chromosome?	x	x			x			
Are the orientations matched or inverted for: block pairs? feature pairs?		x	x				x	
Do the orientations match for pairs of: neighboring blocks? features within a block?	x	x					x	
Are similarity scores alike: with respect to neighboring blocks? within a block?	x	x						x
Are the paired features within a block contiguous?			x		x			
How large is a feature relative to other genes within a block?			x				x	
What are the sizes, locations, and names of features within a block?			x	x	x	x		
What are the differences between individual nucleotides of feature pairs?				x				x <sub>31</sub>

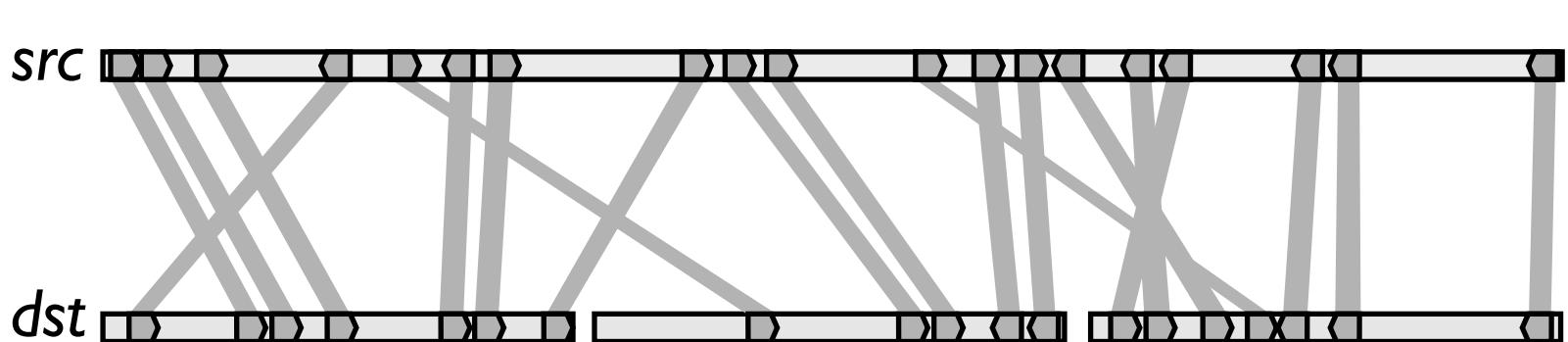
# How: Idiom design choices

- encode match relationships between chromosome segments with both

- color



- connection marks



## → Identity Channels: Categorical Attributes

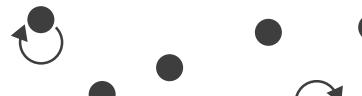
Spatial region



Color hue



Motion

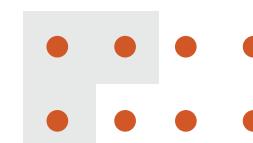


Shape

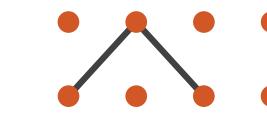


## Marks As Links

### → Containment

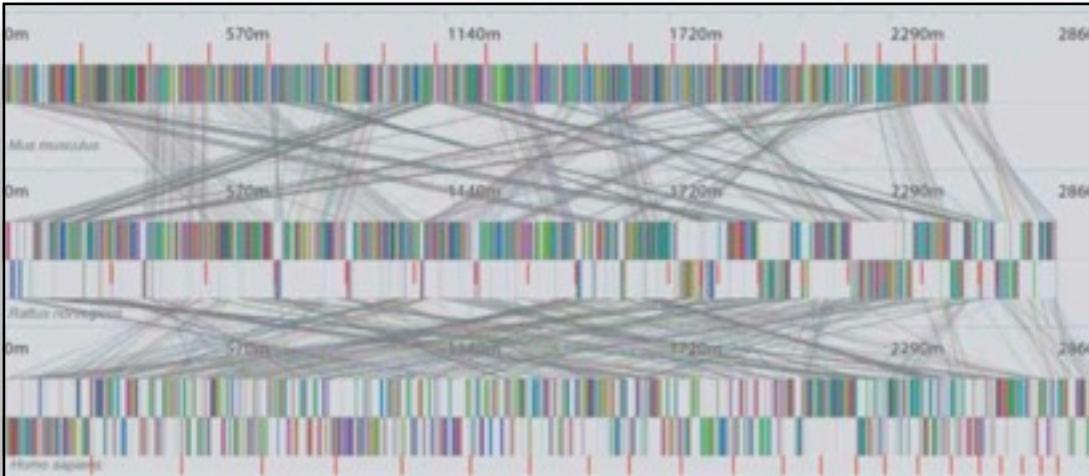


### → Connection

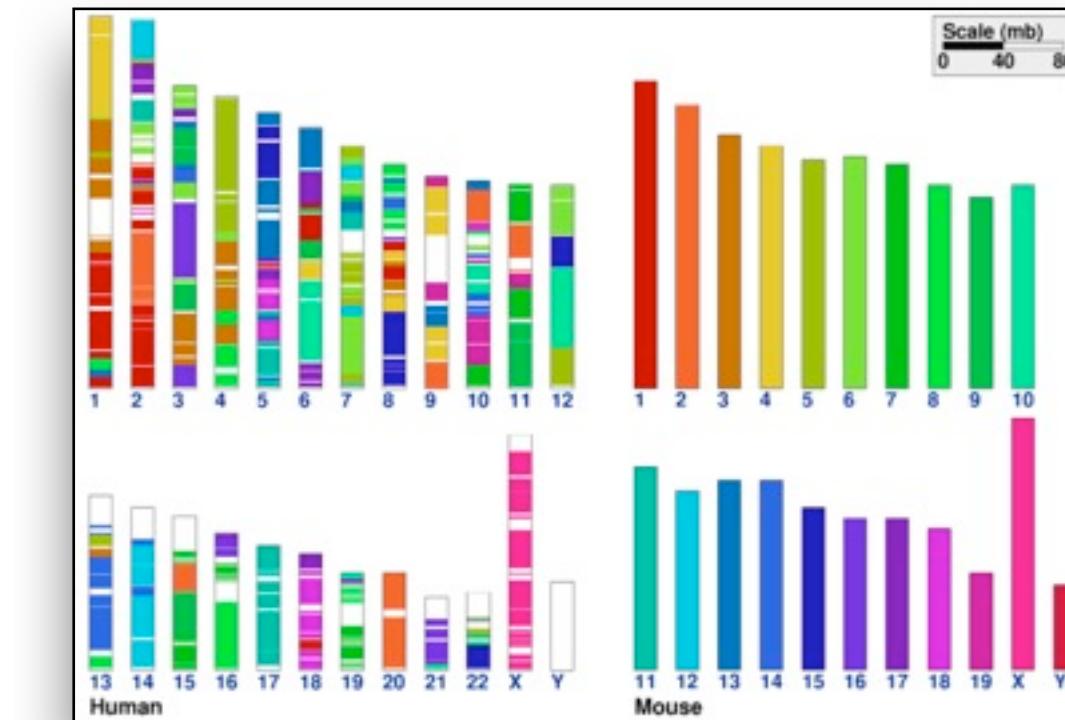


# How: Arrange space

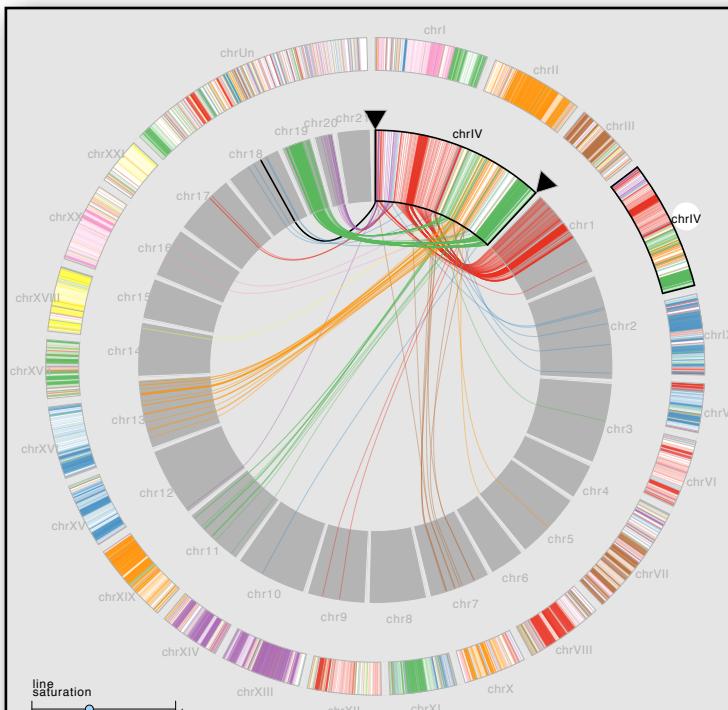
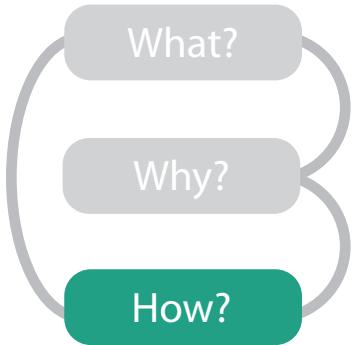
- design space of arrangements



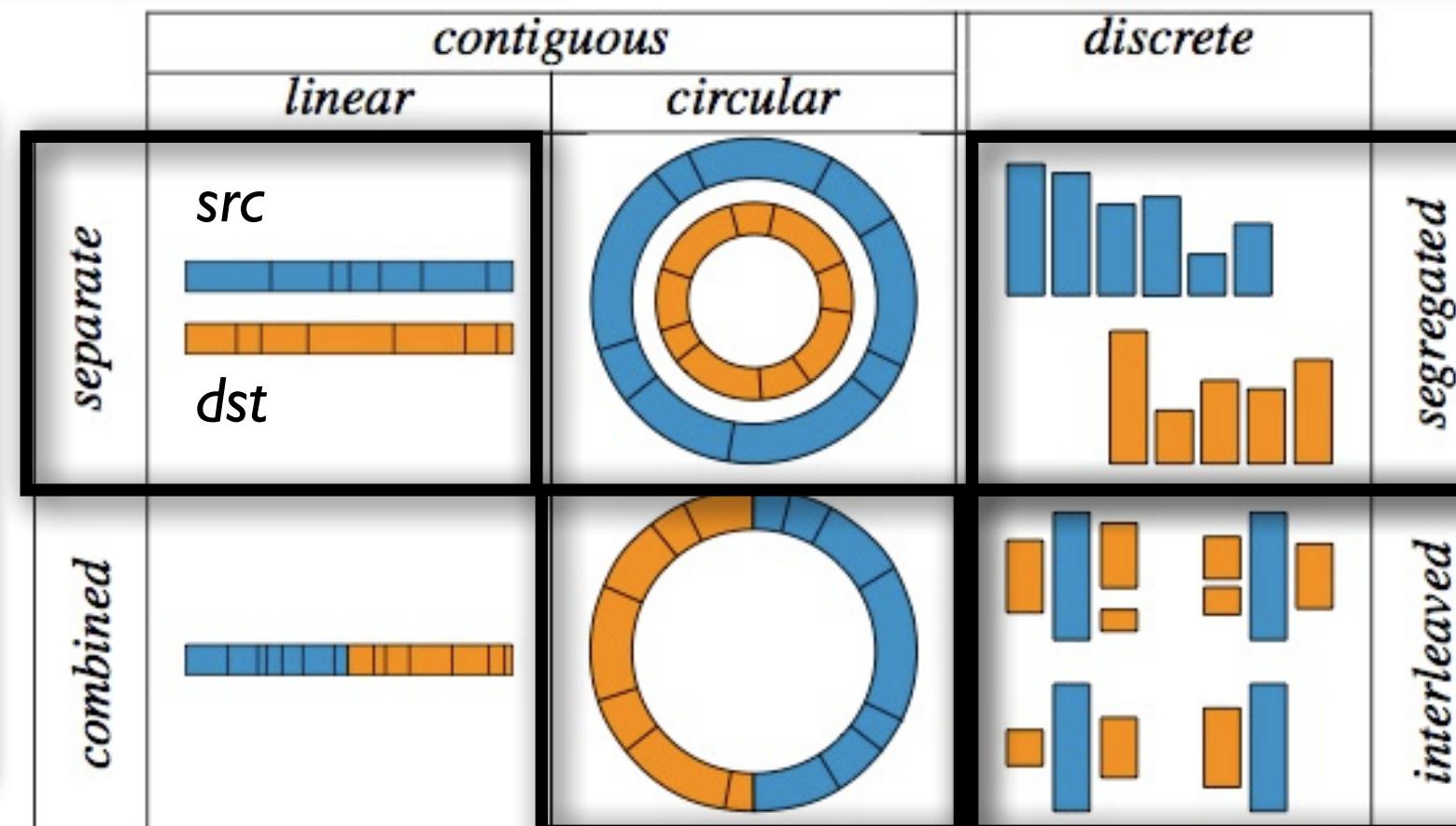
Mauve [Darling04]



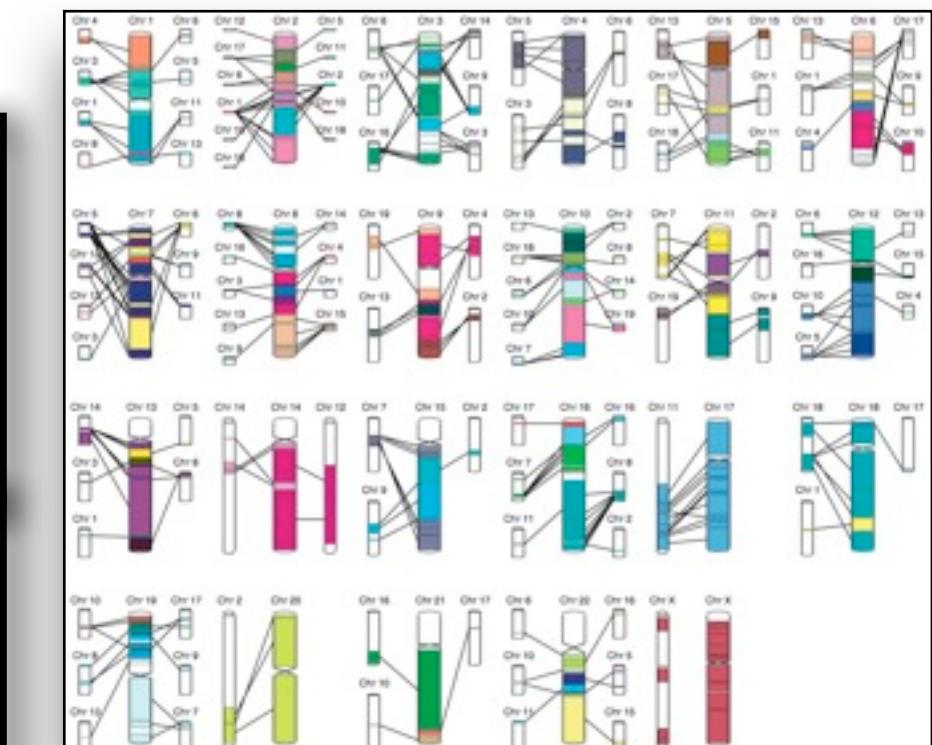
Cinteny



MizBee



Apollo [Lewis02]

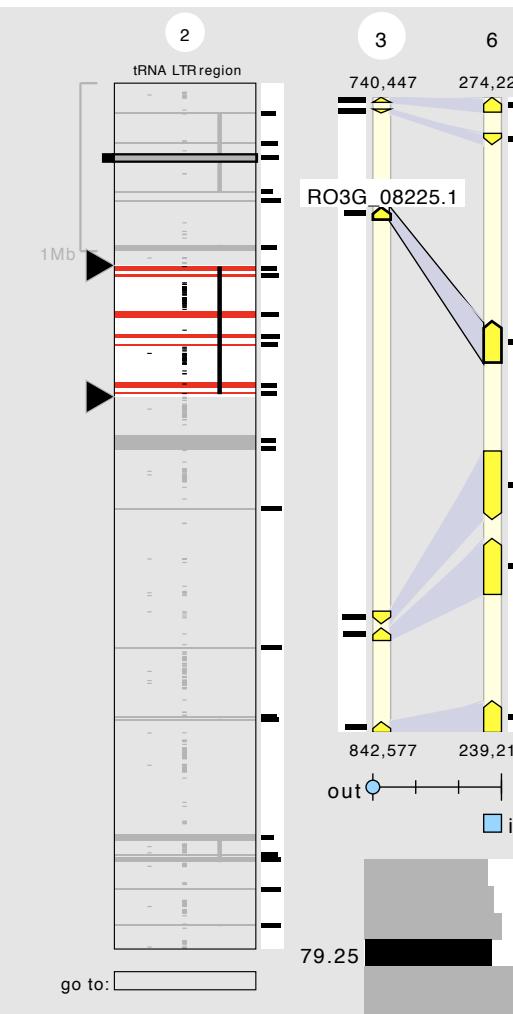
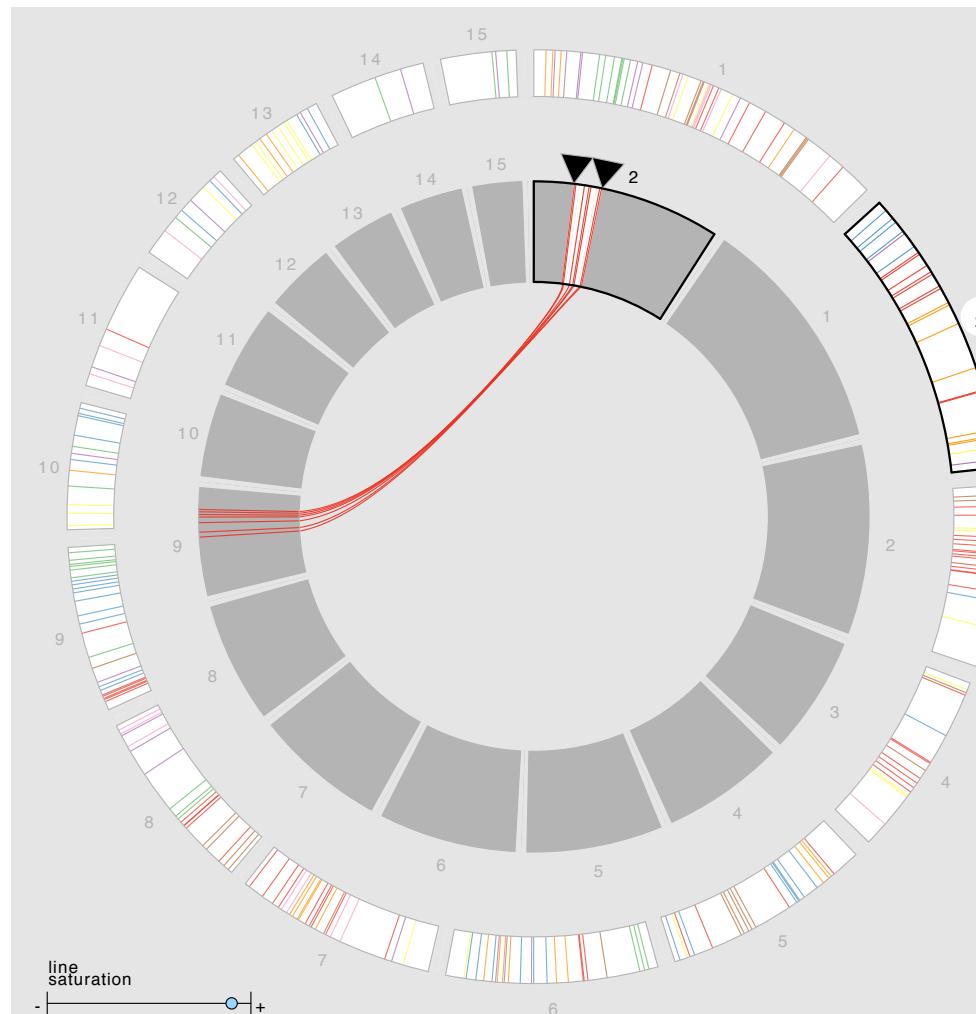


# How: Idiom design choices

- juxtapose linked views

- *multiform overview-detail*

- three views: genome, chromosome, block
    - different visual encoding in each



## Facet

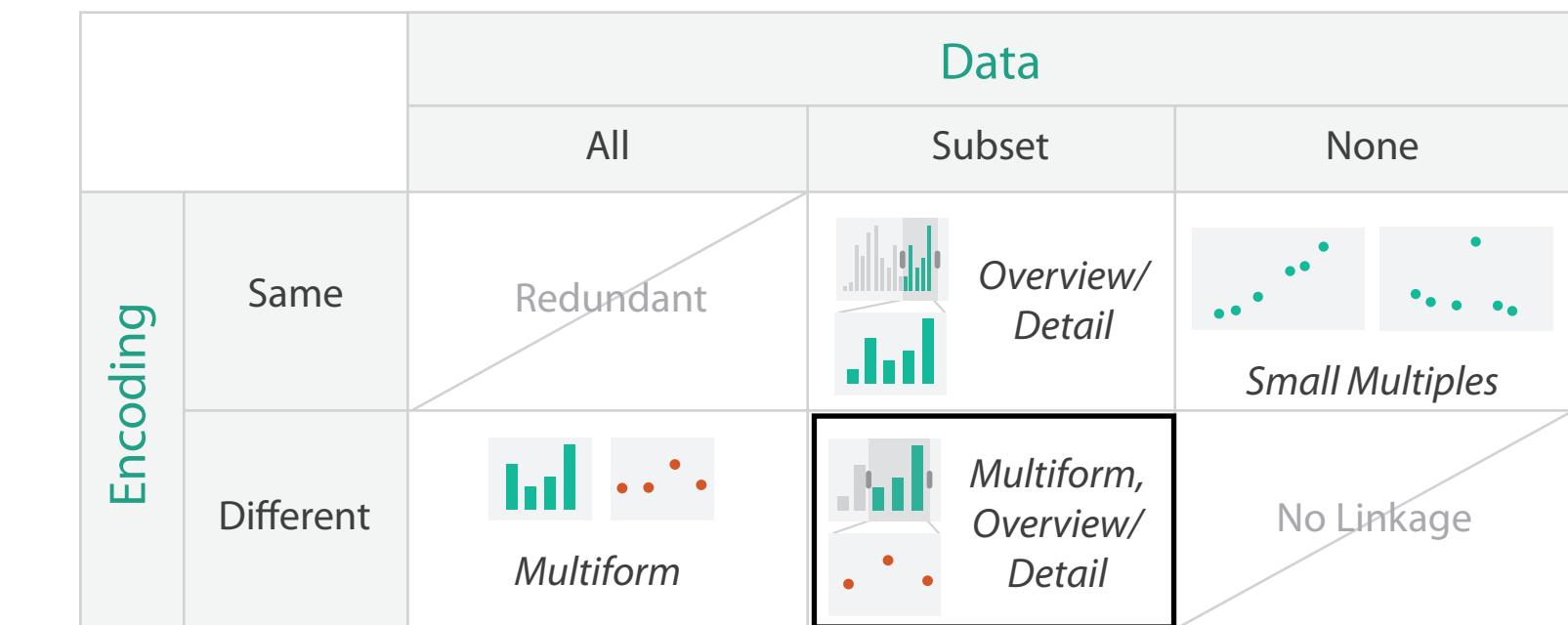
### → Juxtapose



What?

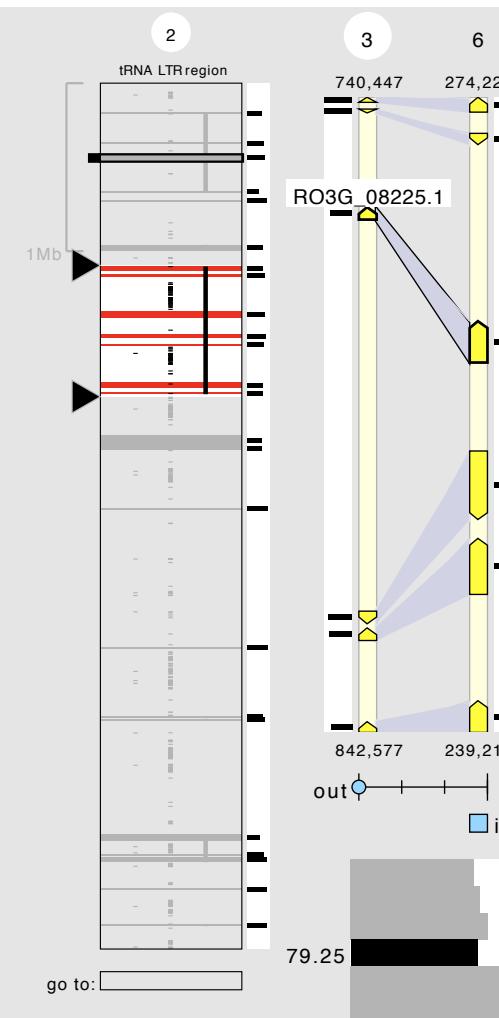
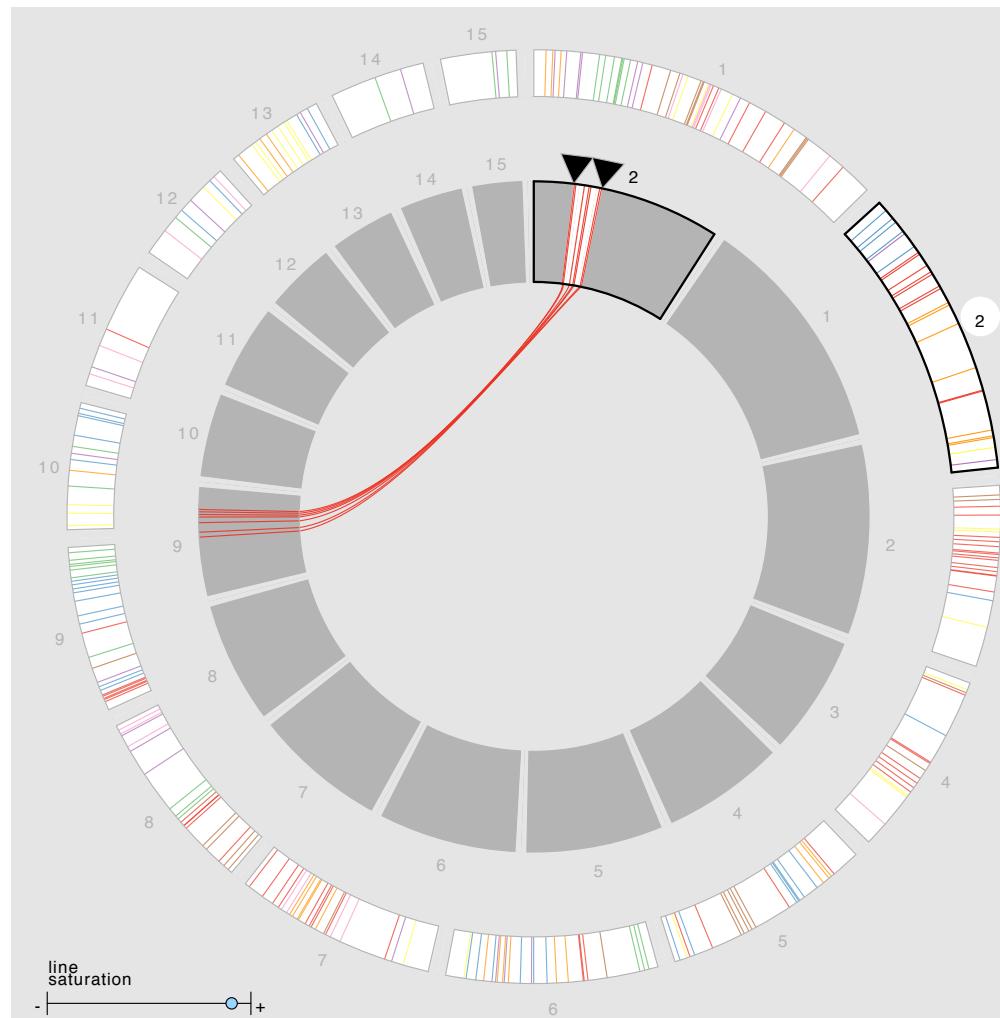
Why?

How?



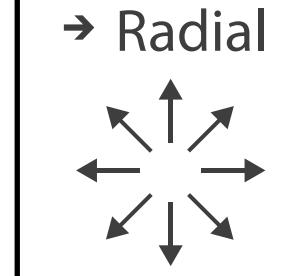
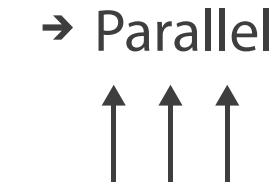
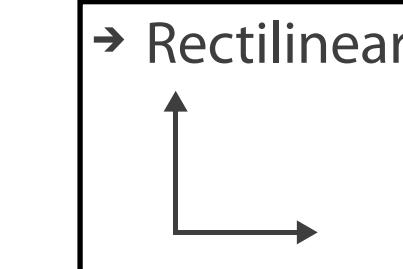
# How: Idiom design choices

- axis orientation
  - radial: genome
  - rectilinear: chromosome, block
    - aligned position more accurate than angle



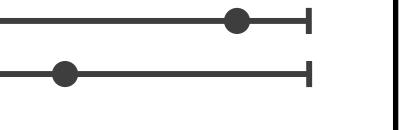
## Arrange

### → Axis Orientation



### → Magnitude Channels: Ordered Attributes

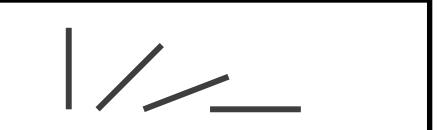
Position on common scale



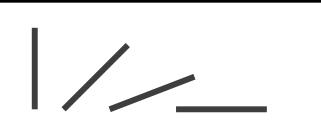
Position on unaligned scale



Length (1D size)



Tilt/angle



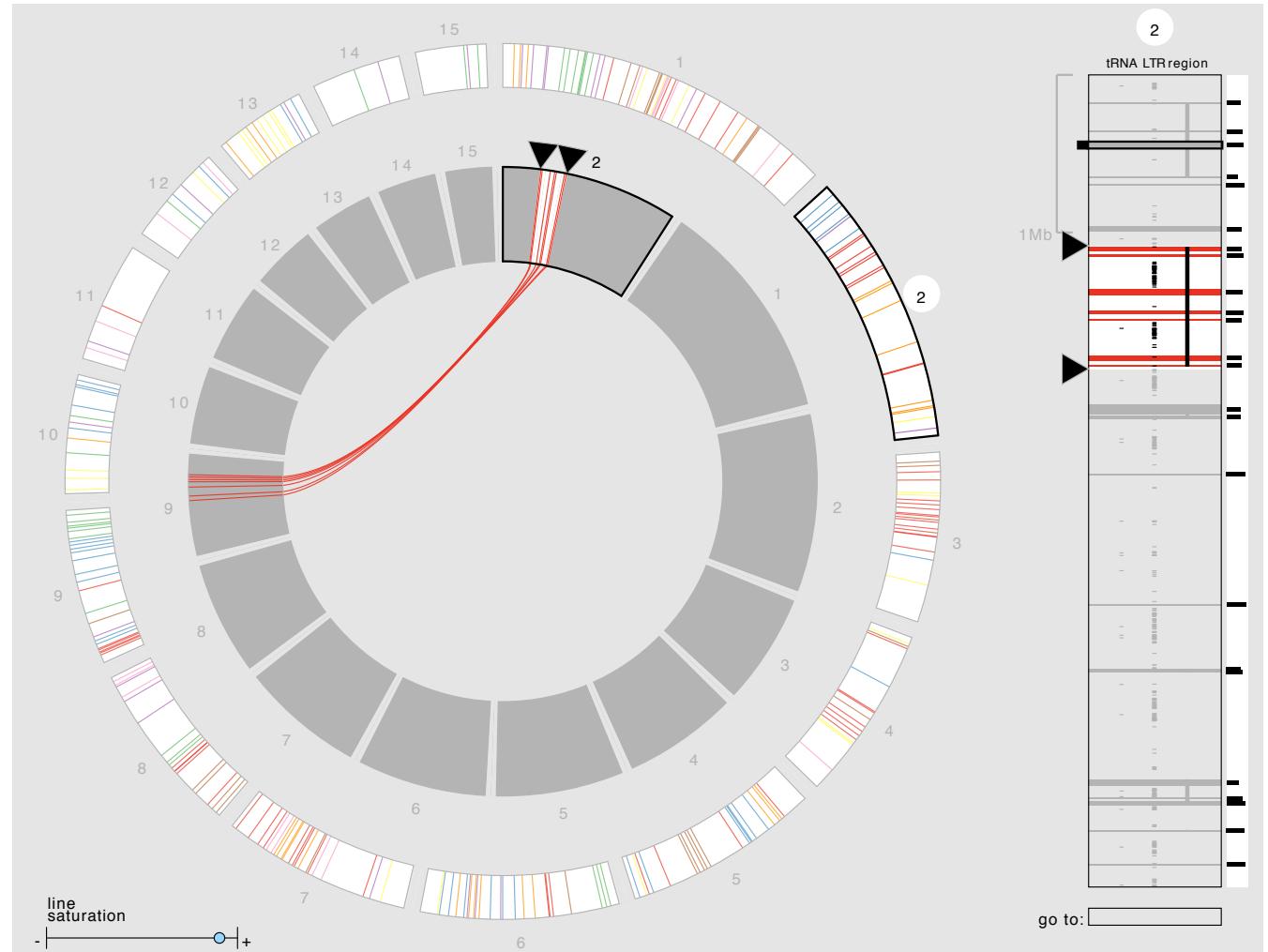
What?

Why?

How?

# How: Idiom design choices

- filter



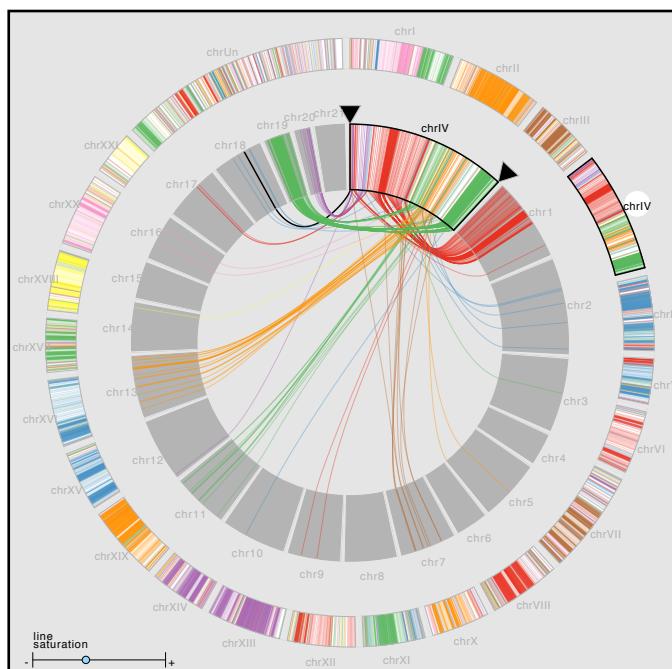
Reduce

Filter



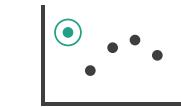
# How: Idiom design choices

- outer ring: summarize relationships with color
  - select one chromosome from set of source chromosomes
- inner ring:
  - destination chromosomes around copy of selected source chromosome
  - show relationship details with connection marks as well as color



Manipulate

→ Select



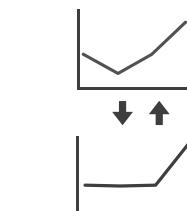
Actions

→ Query

→ Identify



→ Compare



→ Summarise

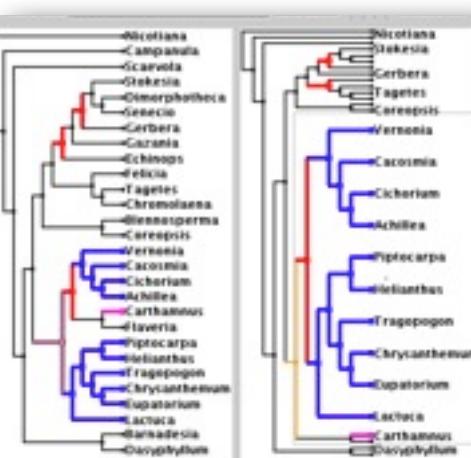


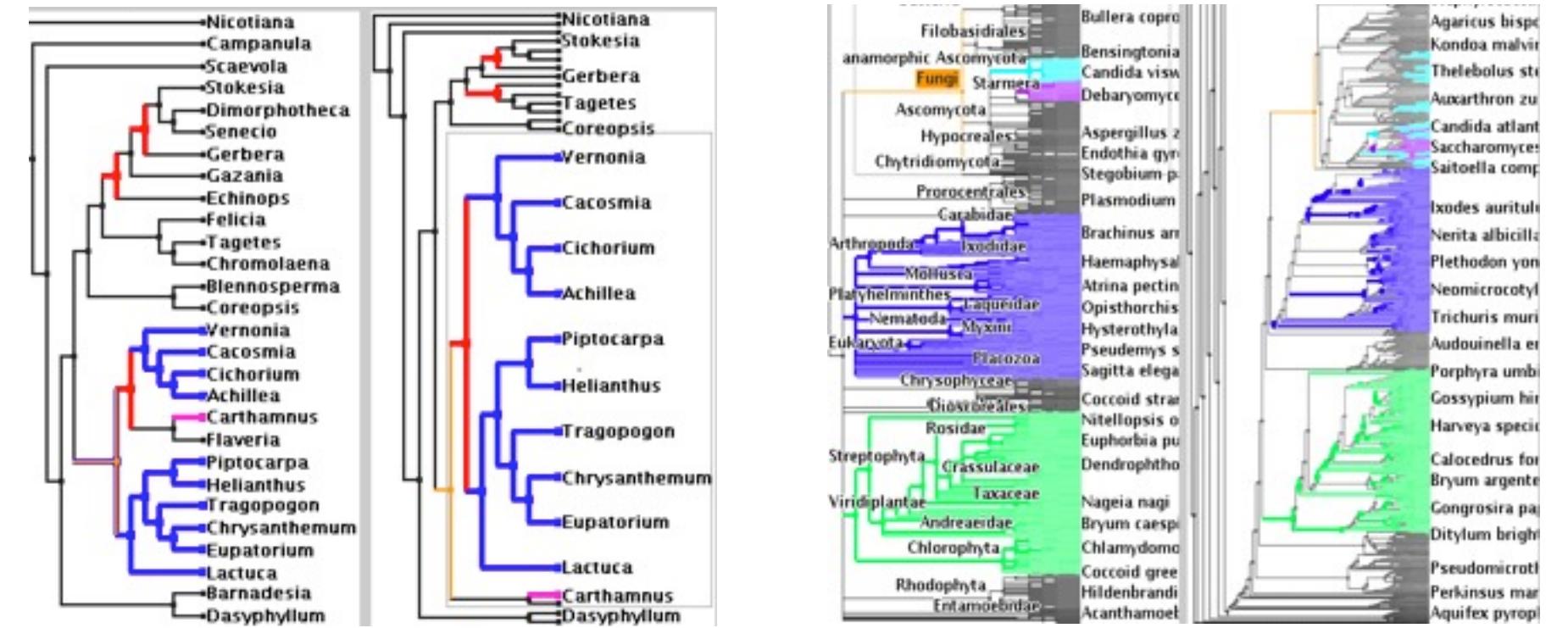
# MizBee contributions

- first synteny browser with side-by-side linked views
  - across the range of scales
  - encoding all four conservation relationship types
    - proximity, size, orientation, similarity
- open source  
<http://www.cs.utah.edu/~miriah/mizbee>

# Outline

- introduction
- Cerebral
- MizBee
- TreeJuxtaposer
- wrapup





# TreeJuxtaposer

*Scalable Tree Comparison using Focus+Context with Guaranteed Visibility*

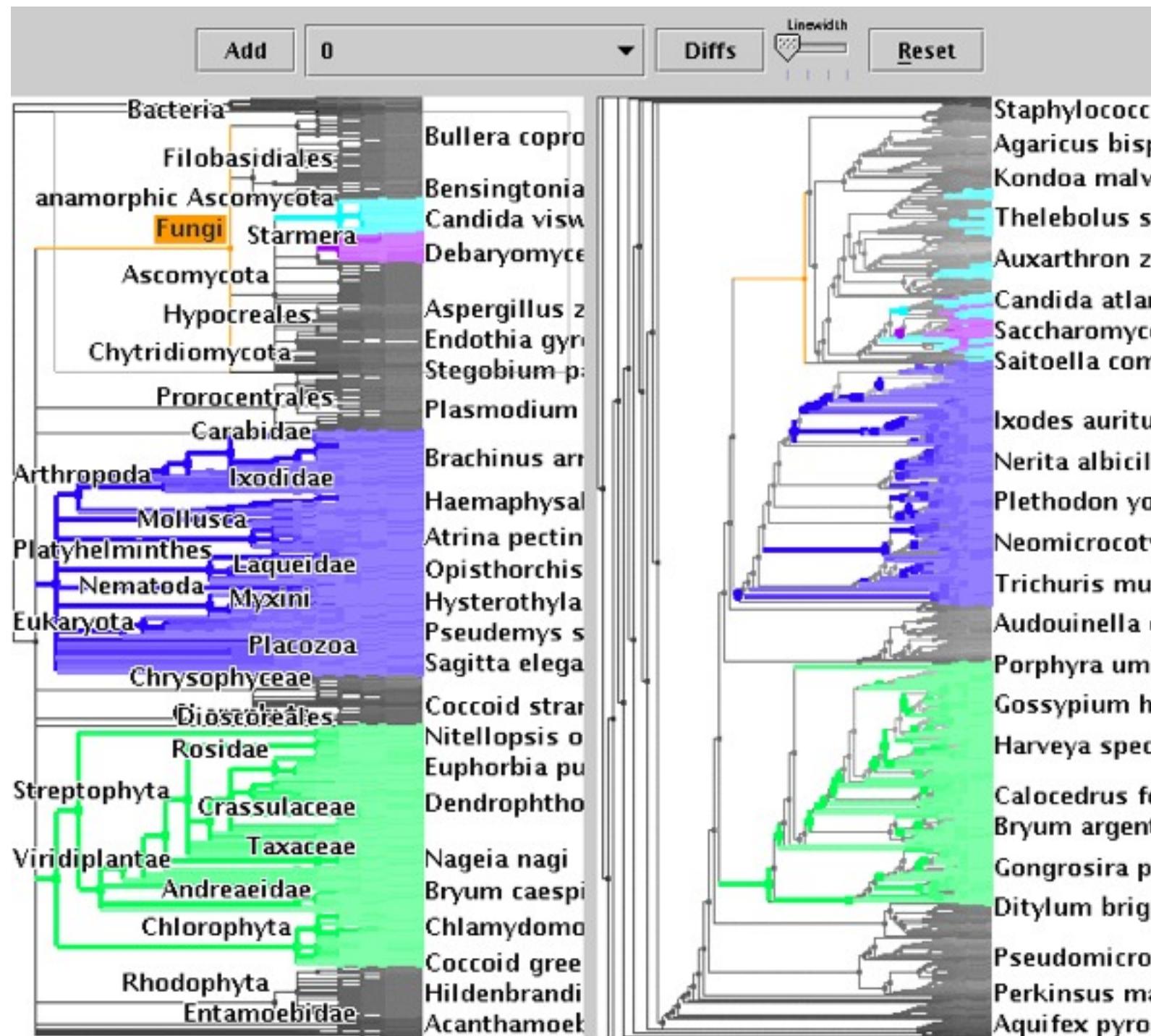
## joint work with:

François Guimbretière, Serdar Tasiran, Li Zhang, Yunhong Zhou

<http://www.cs.ubc.ca/labs/imager/tr/2003/tj/>

TreeJuxtaposer: Scalable Tree Comparison using Focus+Context with Guaranteed Visibility.  
Munzner, Guimbretière, Tasiran, Zhang, Zhou. ACM SIGGRAPH 2003.

# TreeJuxtaposer video



# What and why: Data and task abstraction

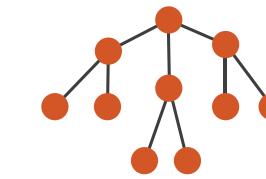
- data: trees
  - phylogenetic tree reconstruction
    - siblings unordered, interior nodes inferred
- task: compare topological structure
  - larger query scopes require more explicit tool support
    - compare several is more difficult than identify/inspect one
      - even trickier: summarize all
- derived data: structural differences
  - best corresponding node in other tree

→ *Derive*



→ Dataset Types

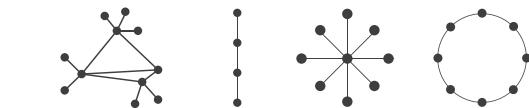
→ Trees



Targets

→ Network Data

→ Topology



→ Paths



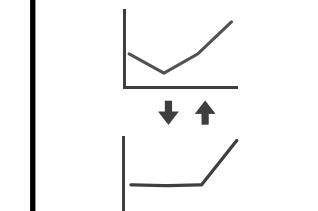
Actions

→ Query

→ Identify



→ Compare



→ Summarise



What?

Why?

How?

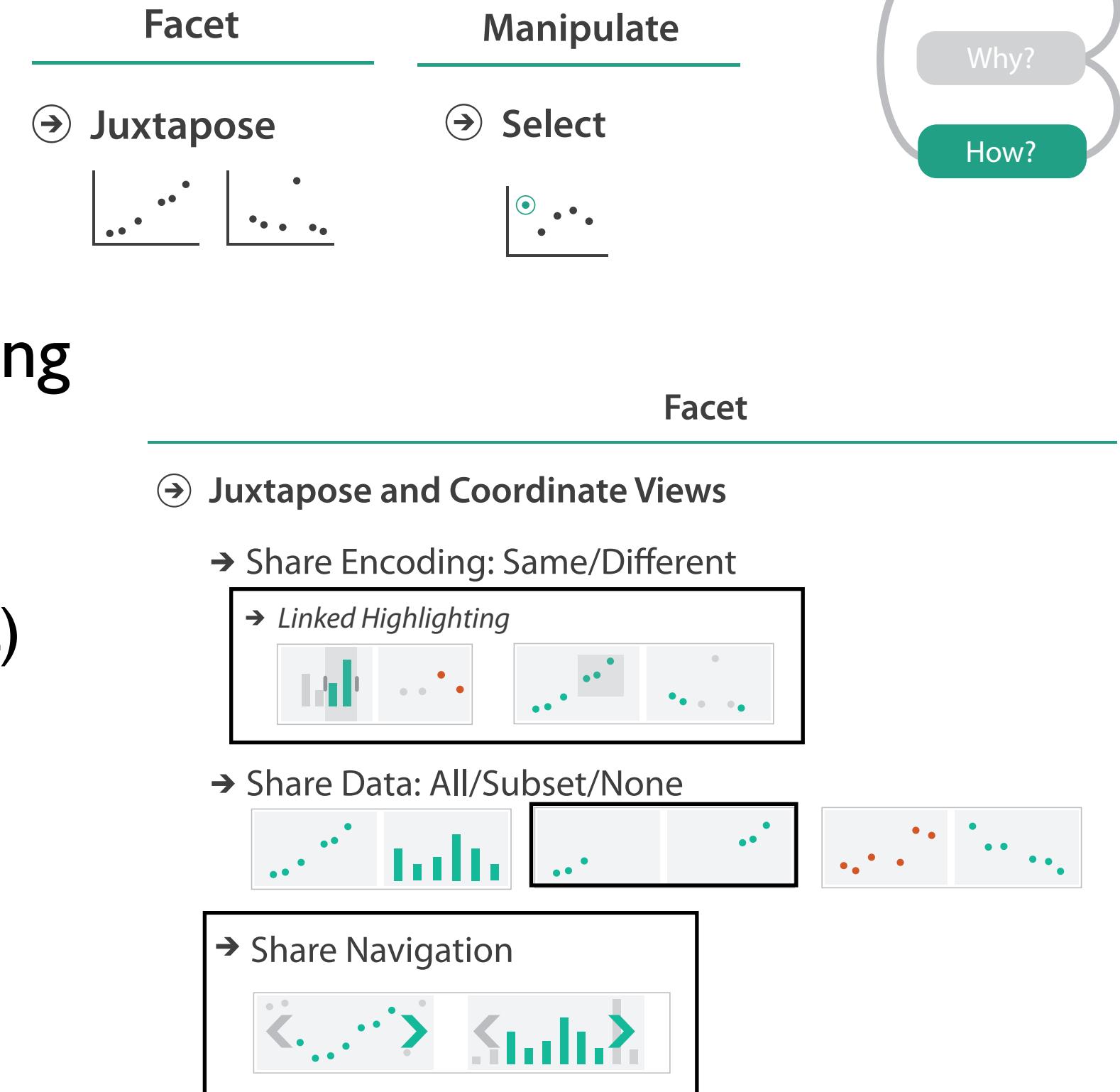
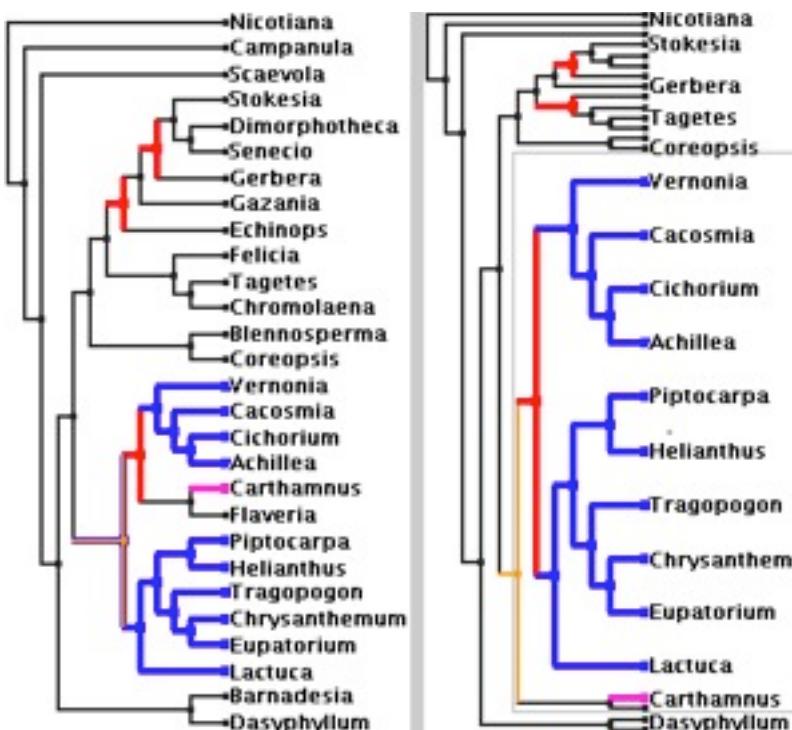
What?

Why?

How?

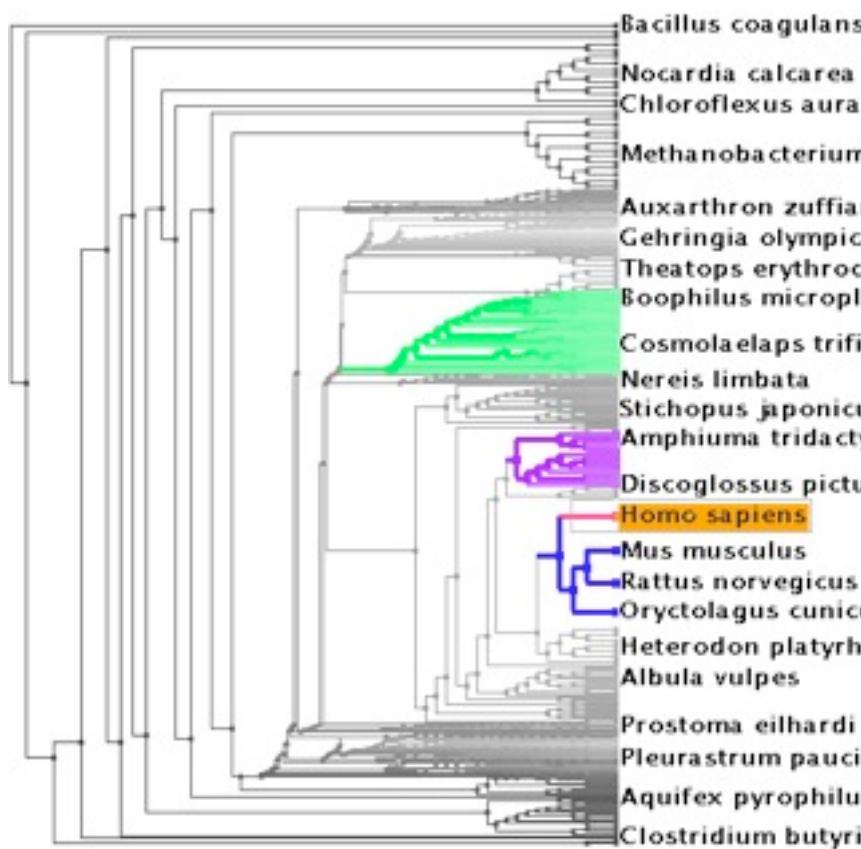
# How: Idiom design decisions

- juxtapose linked views
  - show two tree layouts side by side
  - linked navigation
- encode with color: linked highlighting
  - structural differences
  - corresponding subtree (click select)
  - best corresponding node (hover select)



# How: Idiom design decisions

- embed focus+context in single view
  - reduce with complex combination of filtering and aggregation
- distort geometry
  - metaphor: stretch and squish navigation
  - shape: rectilinear
  - foci: multiple
  - impact: global



Reduce

→ Filter



What?

Why?

How?

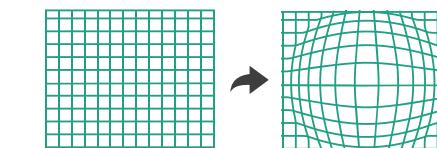
→ Aggregate



→ Embed



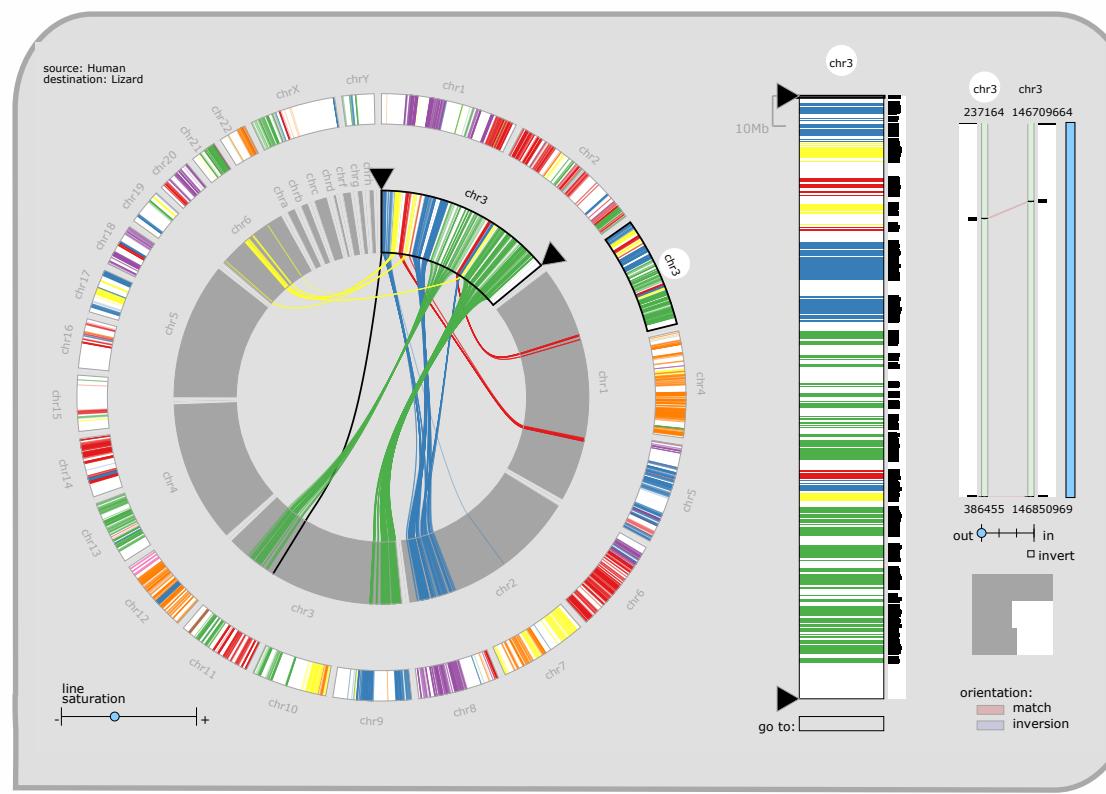
→ Distort Geometry



# TreeJuxtaposer contributions

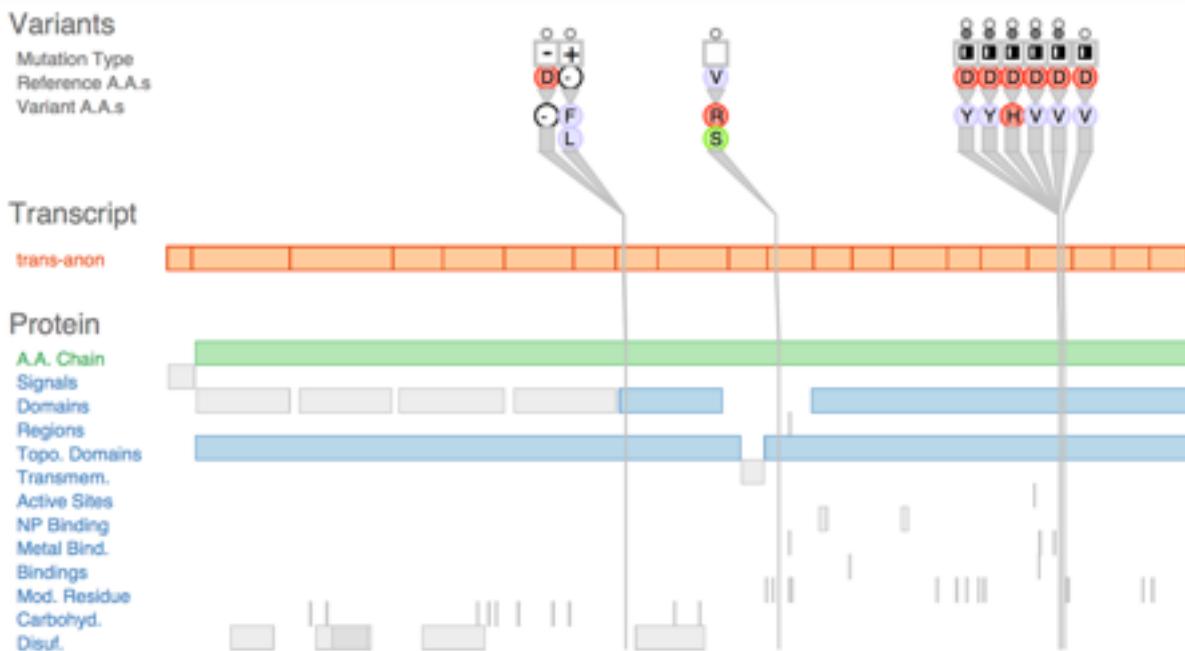
- first interactive tree comparison system
  - derive structural difference data to support comparison task
    - subquadratic algorithm: best corresponding node
  - juxtapose views with cross-dataset linked highlighting
- embed focus+context information in single view with stretch and squish navigation
  - sublinear algorithm: guaranteed visibility of structure marks even when squished
- open source  
<http://olduvai.sf.net/tj>

# Problem-driven work: Genomics



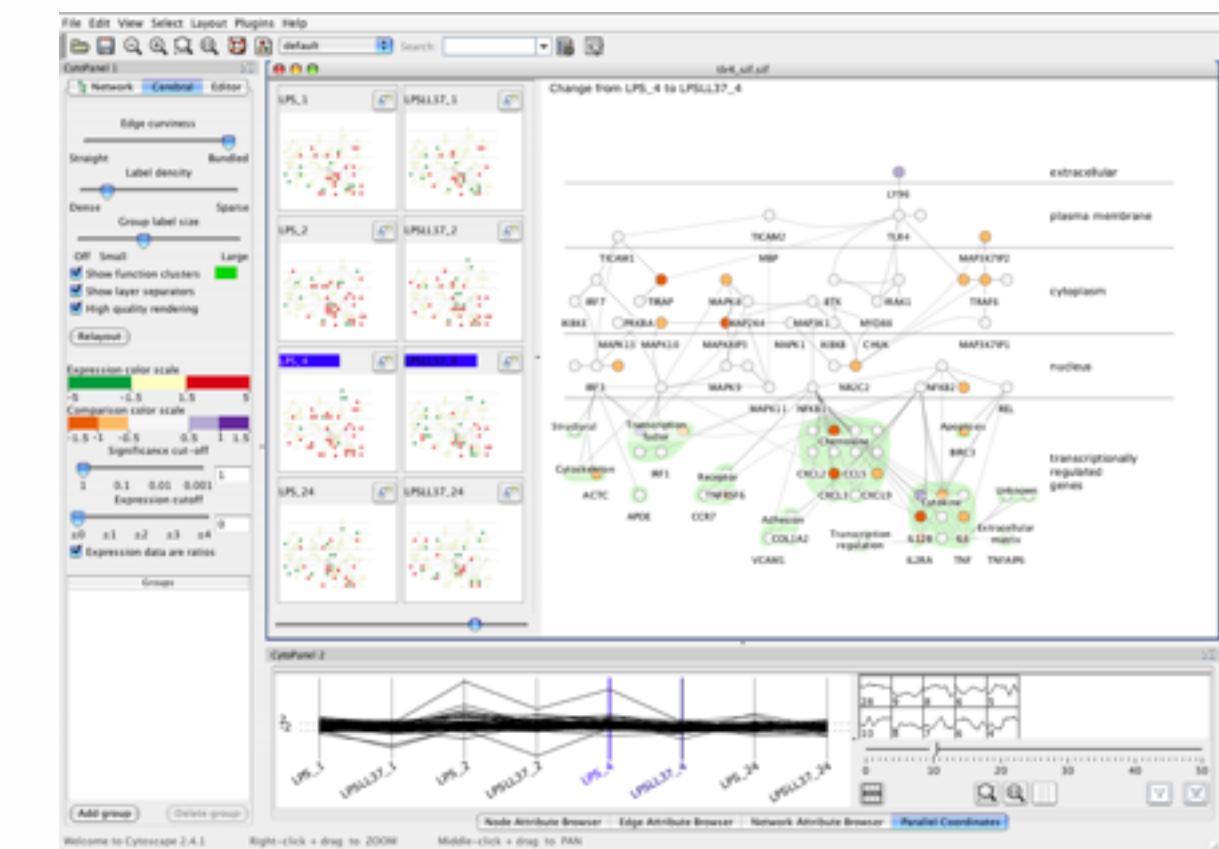
**MizBee**

<http://youtu.be/86p7brwuz2g>



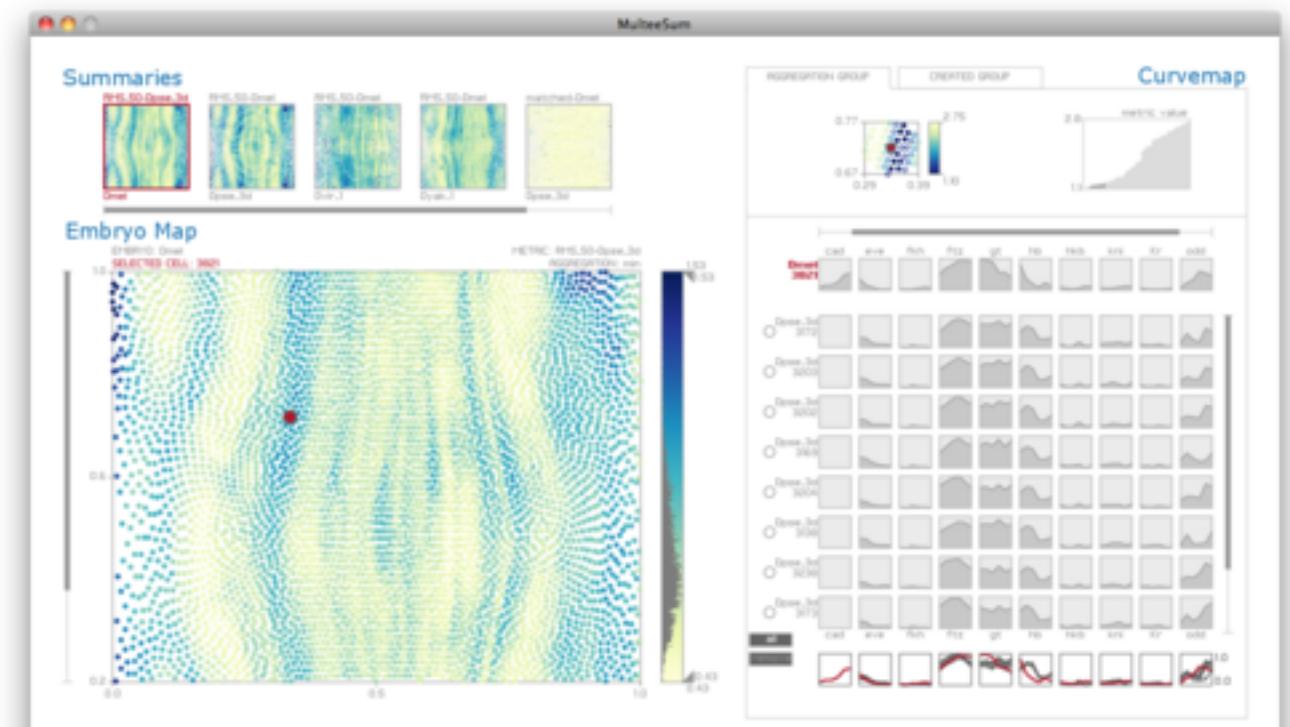
**Variant View**

[http://youtu.be/AHDnv\\_qMXxQ](http://youtu.be/AHDnv_qMXxQ)



**Cerebral**

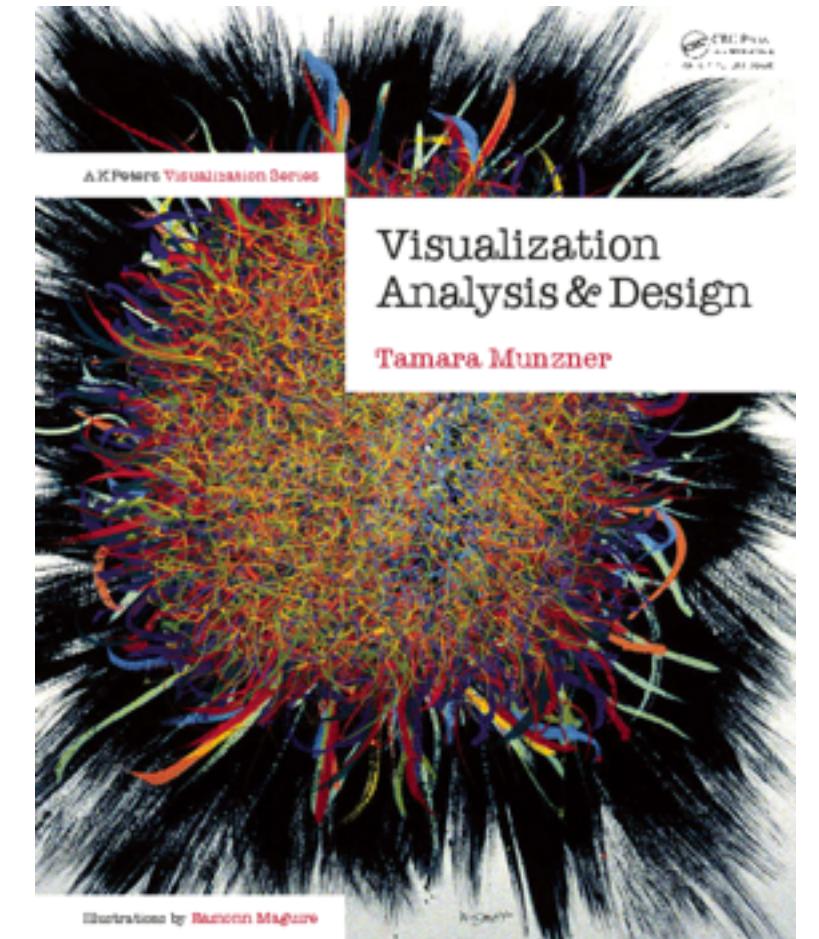
<http://youtu.be/76HhG1FQngI>



**MulteeSum  
Pathline**

# More Information

- this talk  
<http://www.cs.ubc.ca/~tmm/talks.html#daley15>
- papers, videos, software, talks, courses  
<http://www.cs.ubc.ca/group/infovis>  
<http://www.cs.ubc.ca/~tmm>
- book  
<http://www.cs.ubc.ca/~tmm/vadbook>
- acknowledgements
  - funding: Agilent, NSERC, NSF



@tamaramunzner