CURRENT CHALLENGES IN GENOMIC DATA VISUALIZATION

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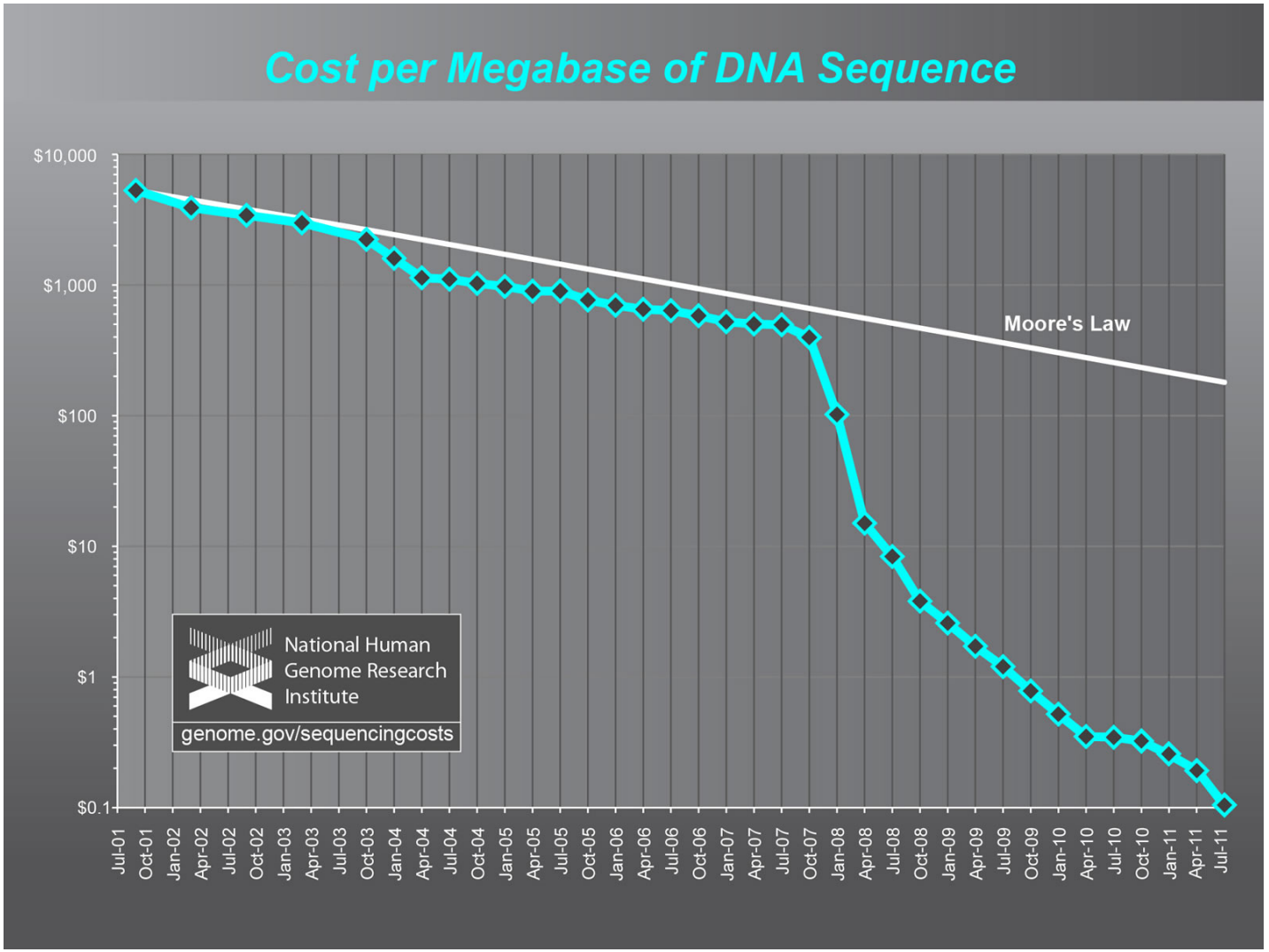
The Data Deluge

~$5,000
in 2001

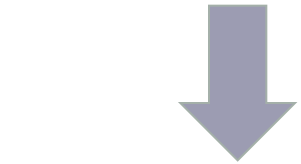


~10¢
in 2011

Sequencing Experiments

De novo assembly

AGCTTCAGATGGACAGATAA
GGCATACAGACTTAGACATA
CCAGACAAGACAGACACAGTA
TACAAGACATAAGCAATACAGA
CCAGACAAGACAGACACAGTA

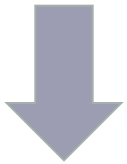


Genome Assembly

Re-sequencing

GGCATACAGACTTAGACATA
AGCTTCAGATGGACAGATAA
CCAGACAAGACAGACACAGTA
CCAGACAAGACAGACACAGTA
TACAAGACATAAGCAATACAGA

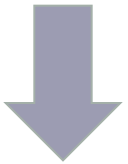
Reference Genome



Enrichment

CCAGACAAGACAGACACAGTA
AGCTTCAGATGGACAGATAA
GGCATACAGACTTAGACATA
CCAGACAAGACAGACACAGTA
TACAAGACATAAGCAATACAGA

Reference Genome



Drew Sheneman, New Jersey - The Newark Star Ledger

Challenge 1

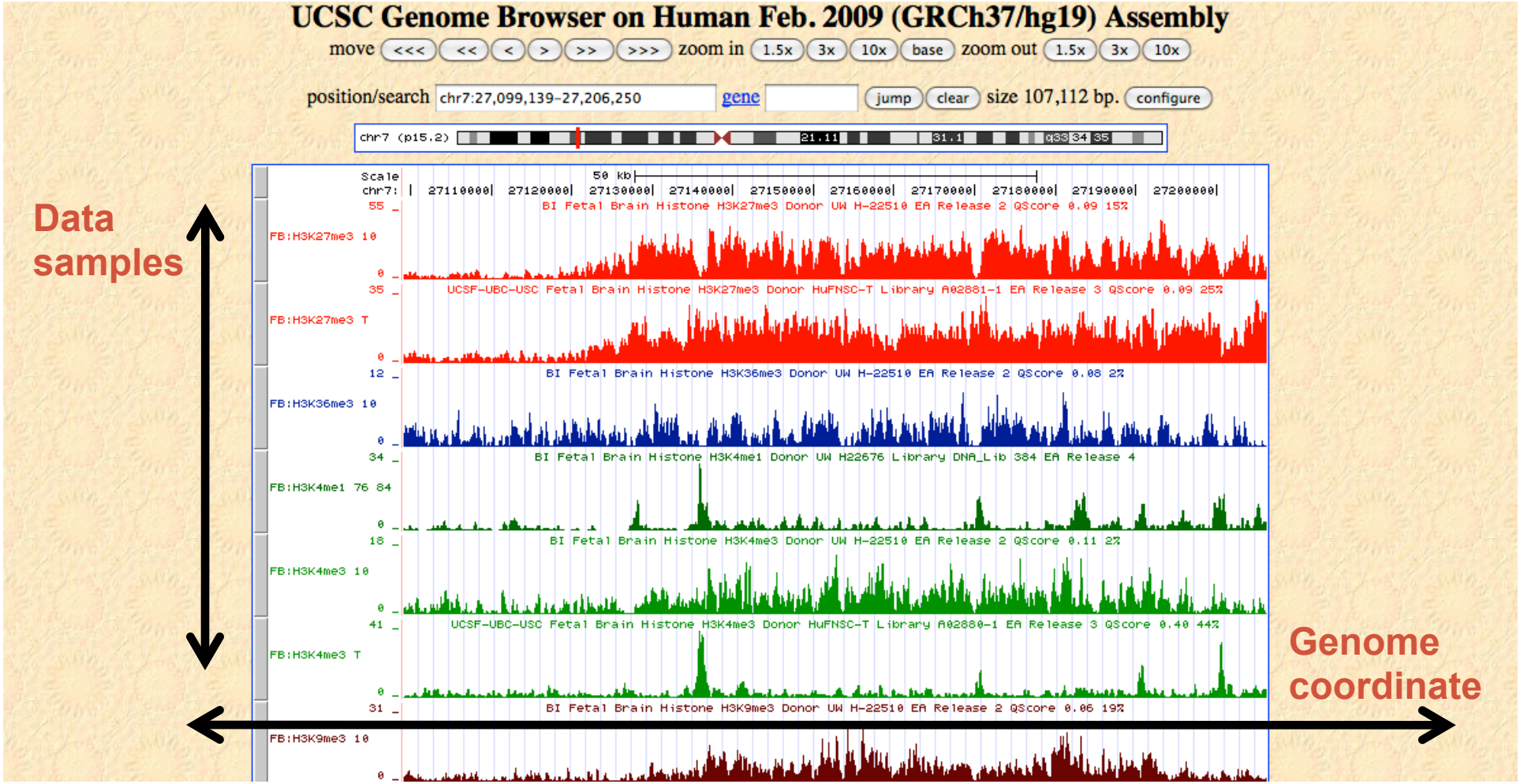
Large number of samples for comparison

“To systematically characterize the genomic changes in hundreds of tumors…and thousands of samples over the next five years”

The Cancer Genome Atlas
www.cancergenome.nih.gov

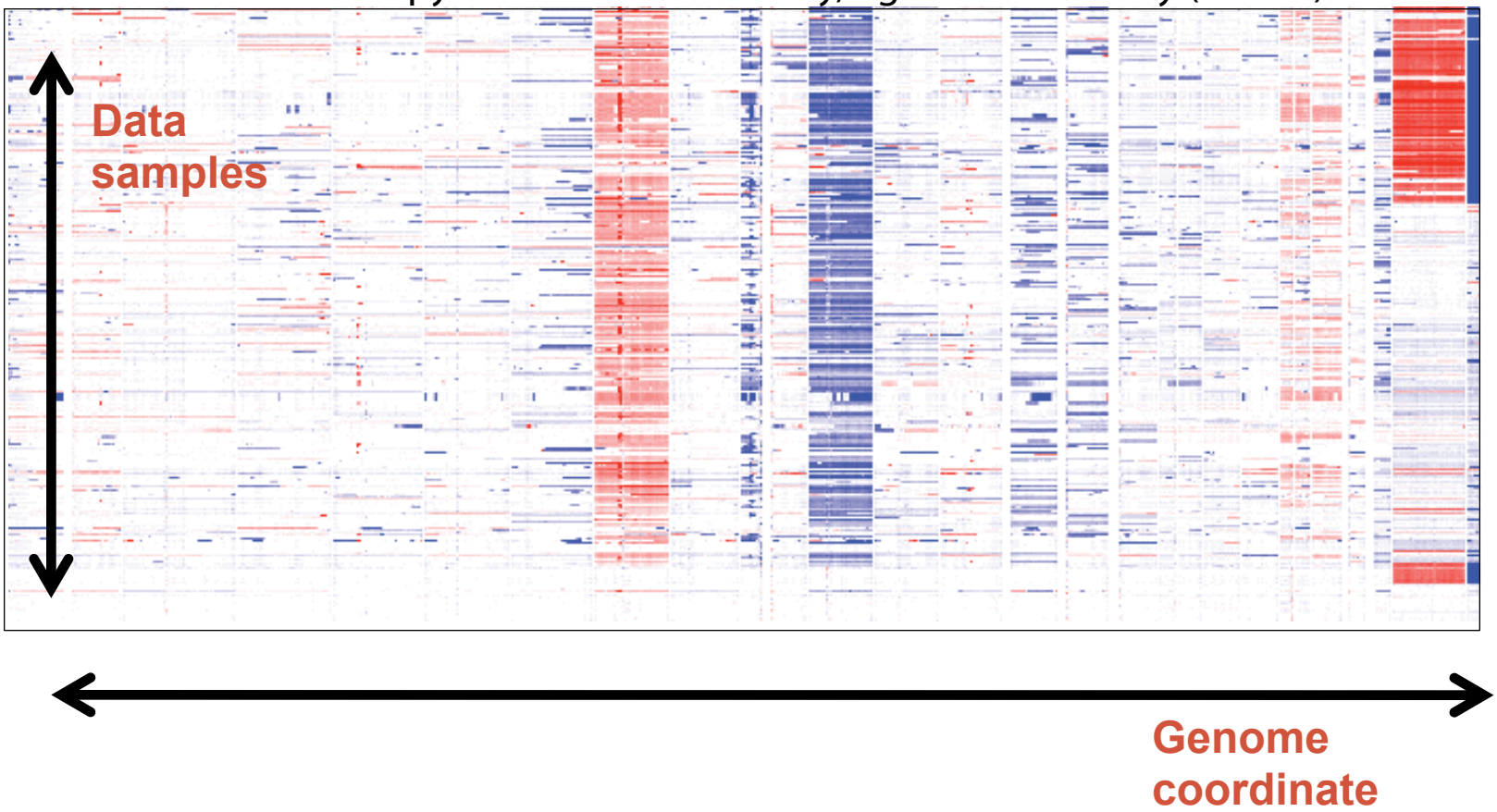
Genome Browsers

Stacked data tracks along a common genome x-axis



UCSC Cancer Genomics Heatmaps

Glioblastoma Copy Number Abnormality, Agilent 244A array (n=200)



Heatmap provides a more condensed view

Zhu *et al*., Nature Methods, 2009

Challenge 1

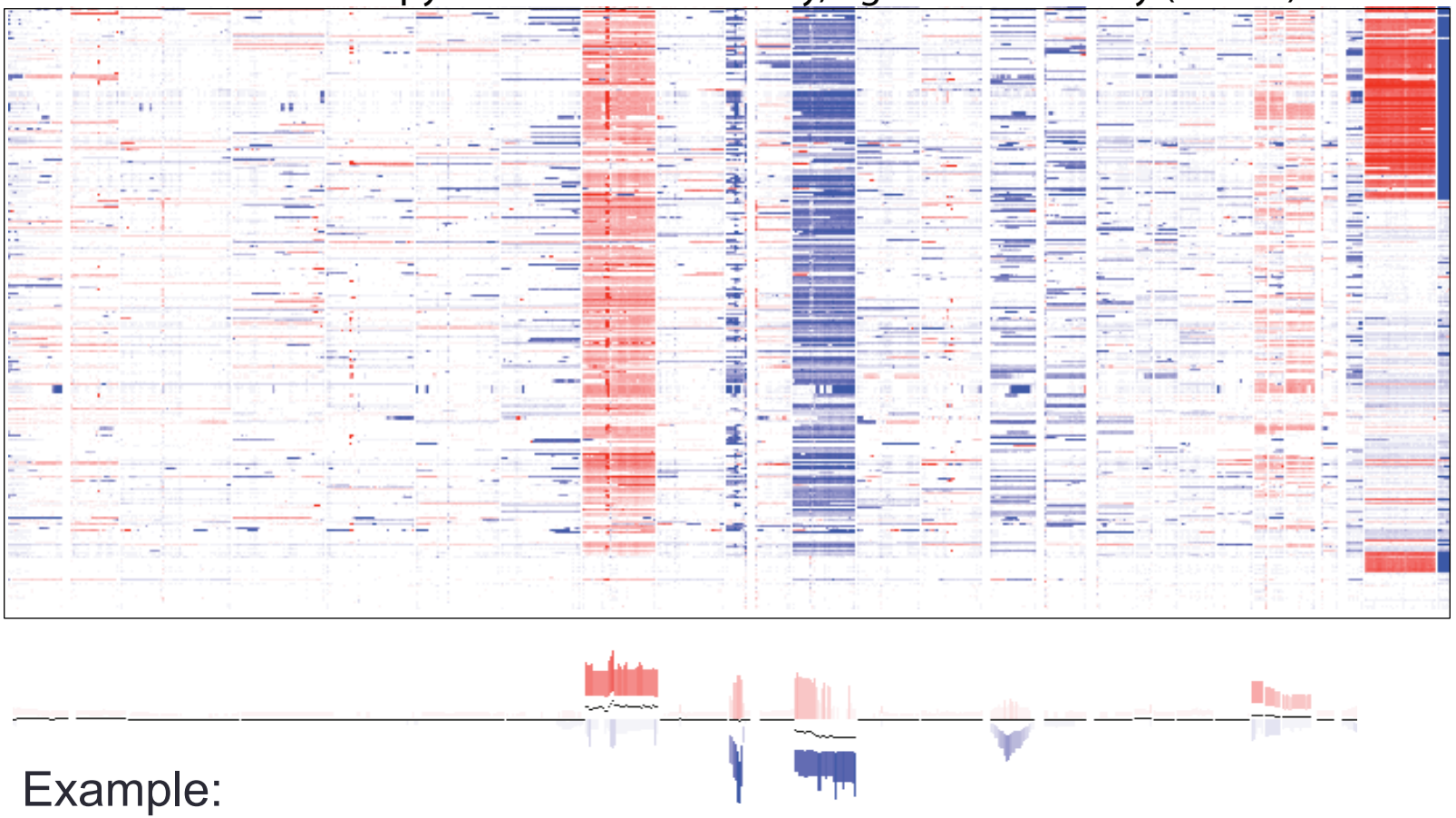
Large number of samples for comparison

- *Consider what information is needed*

  e.g. replace with biologically meaningful summary, such as significant change between samples

UCSC Cancer Genomics Heatmaps

Glioblastoma Copy Number Abnormality, Agilent 244A array (n=200)



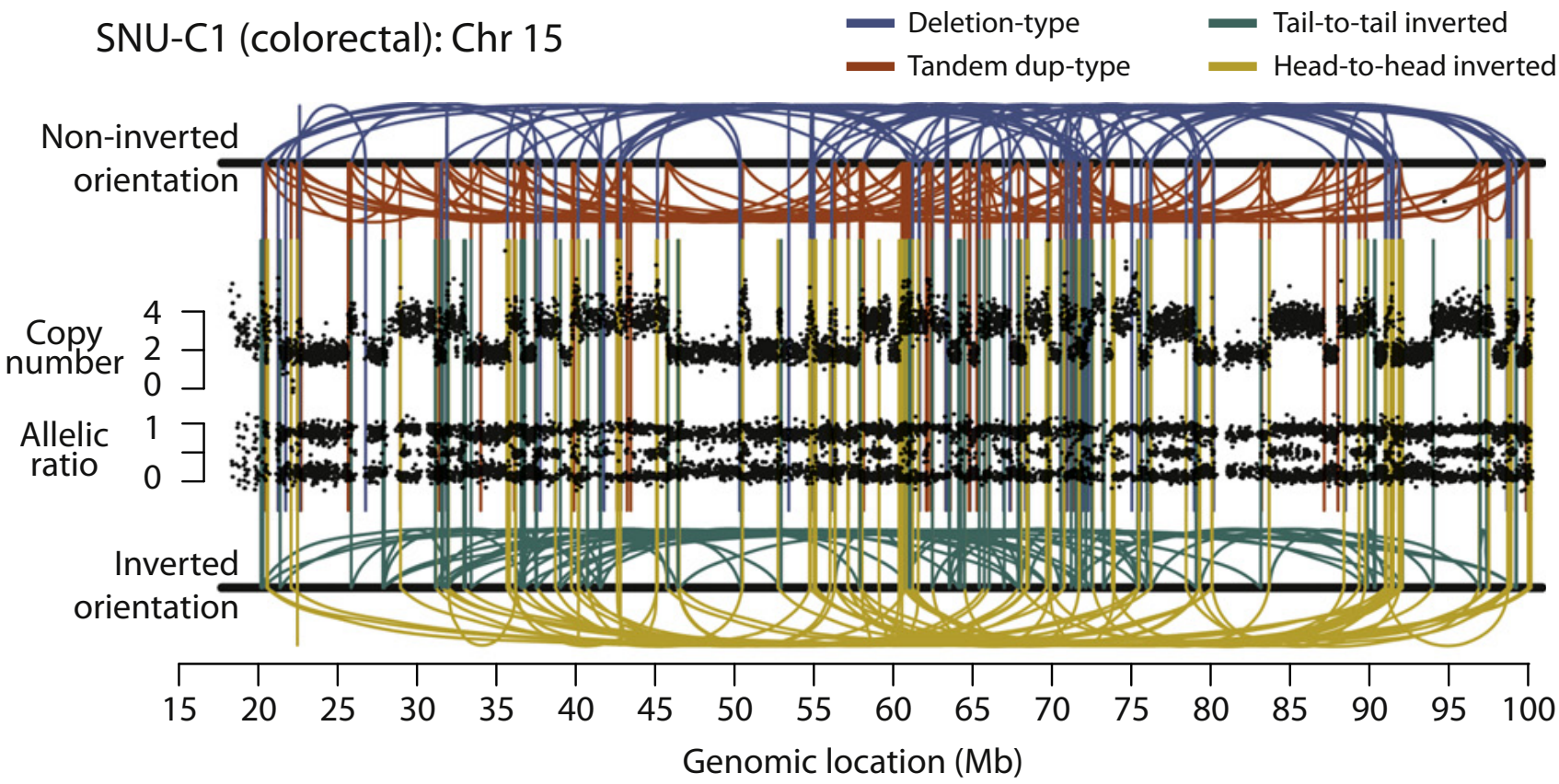
Example:
Summary view (column averages)

Zhu *et al*., Nature Methods, 2009

Challenge 2

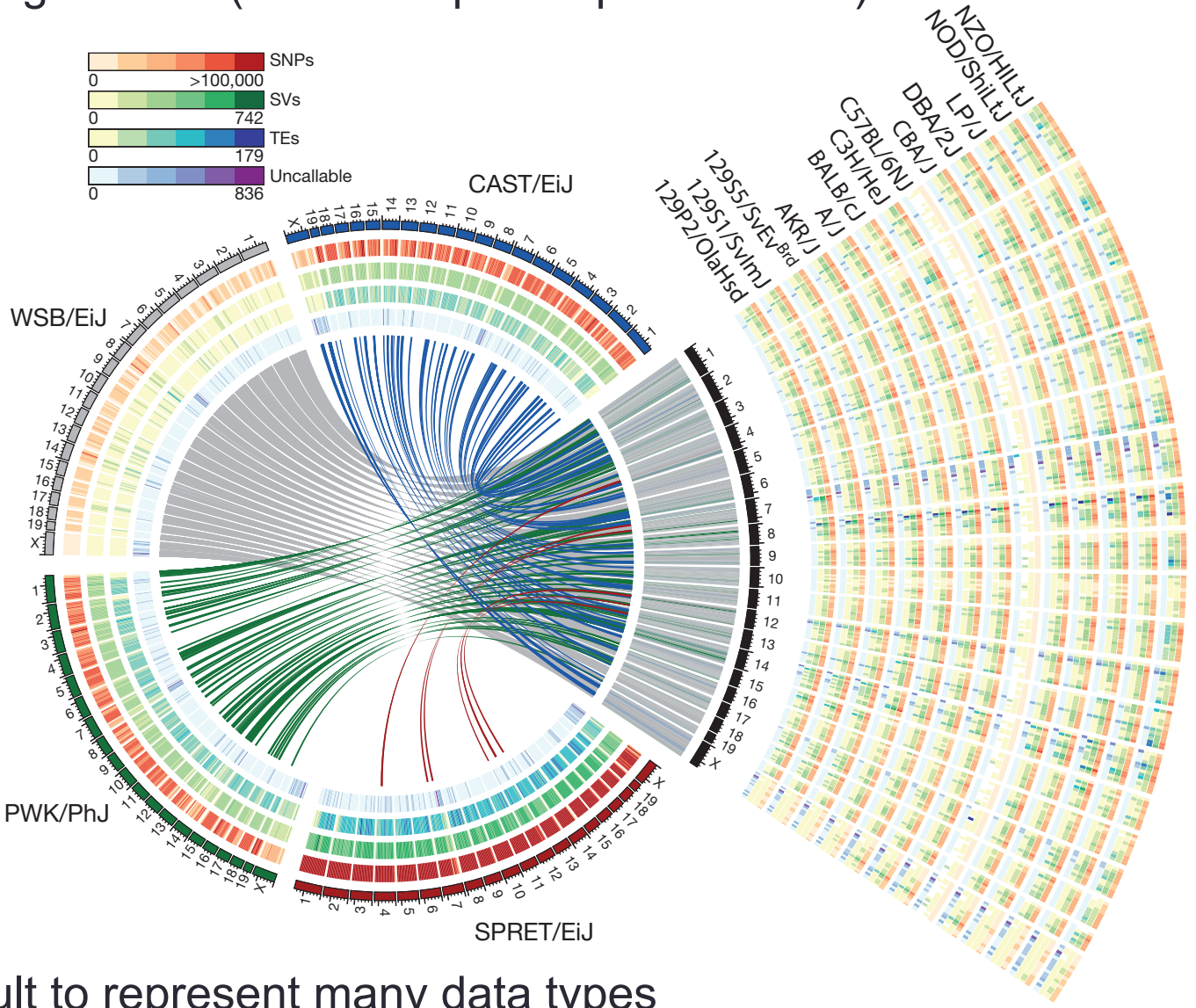
Large number of data types

Genomic rearrangements in cancer (complex representation)



Stephens *et al.*, Cell, 2011

17 mouse genomes (more compact representation)



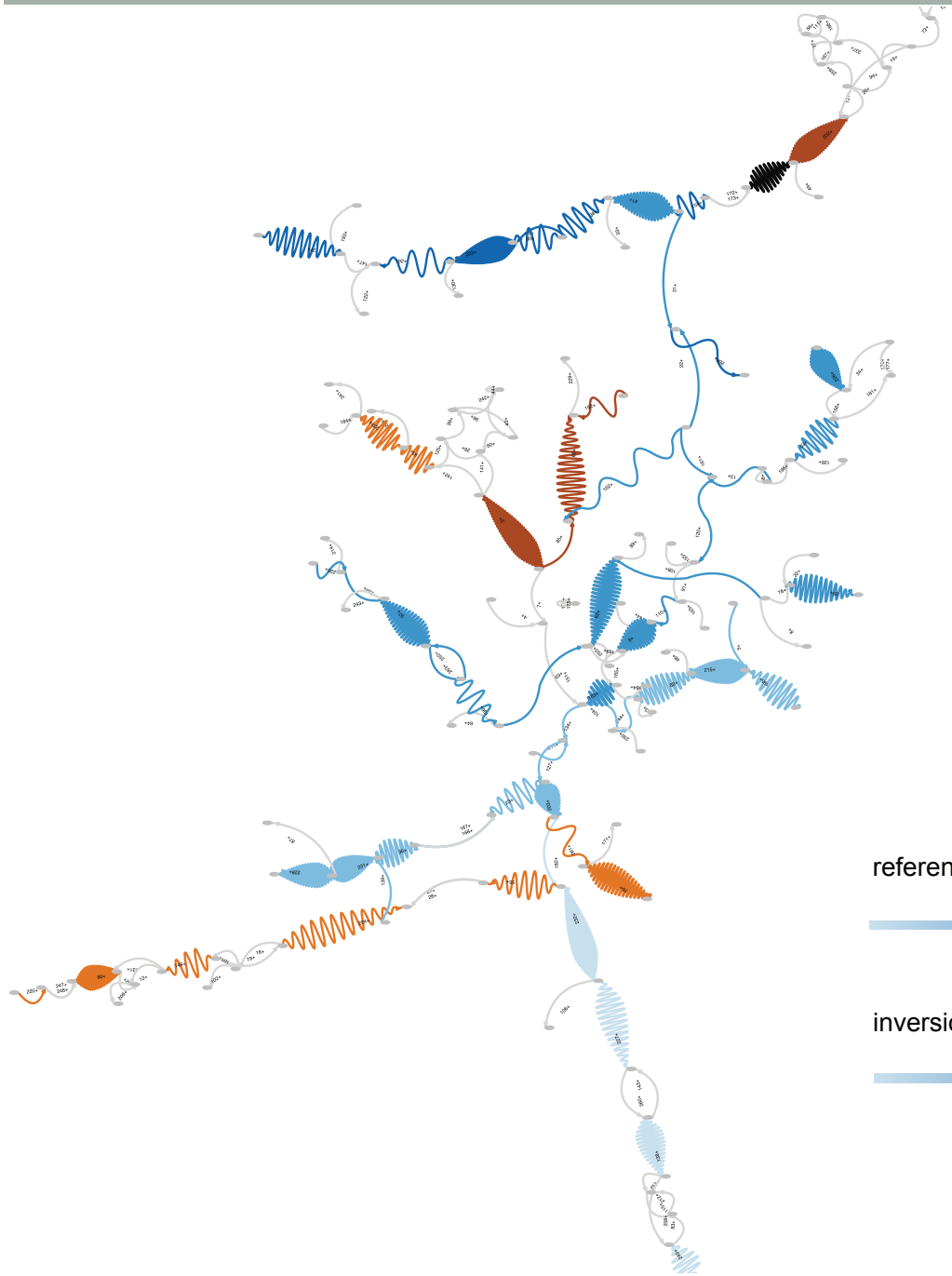
Still difficult to represent many data types in a general tool

Keane *et al.*, Nature, 2011

Challenge 2

Large number of data types

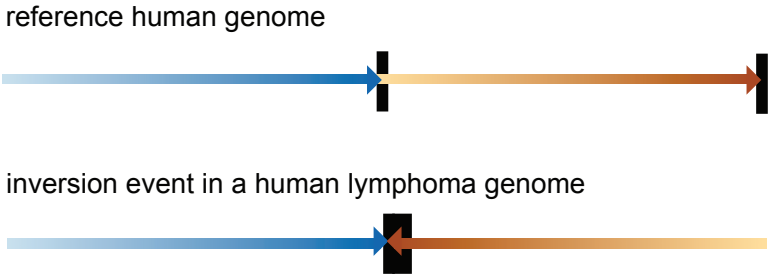
- *Compact, customized data encoding*



ABySS-Explorer

Represents sequence
- connectivity
- strand
- length
- mapping on reference

Interactively access
- sequence coverage
- scaffolding



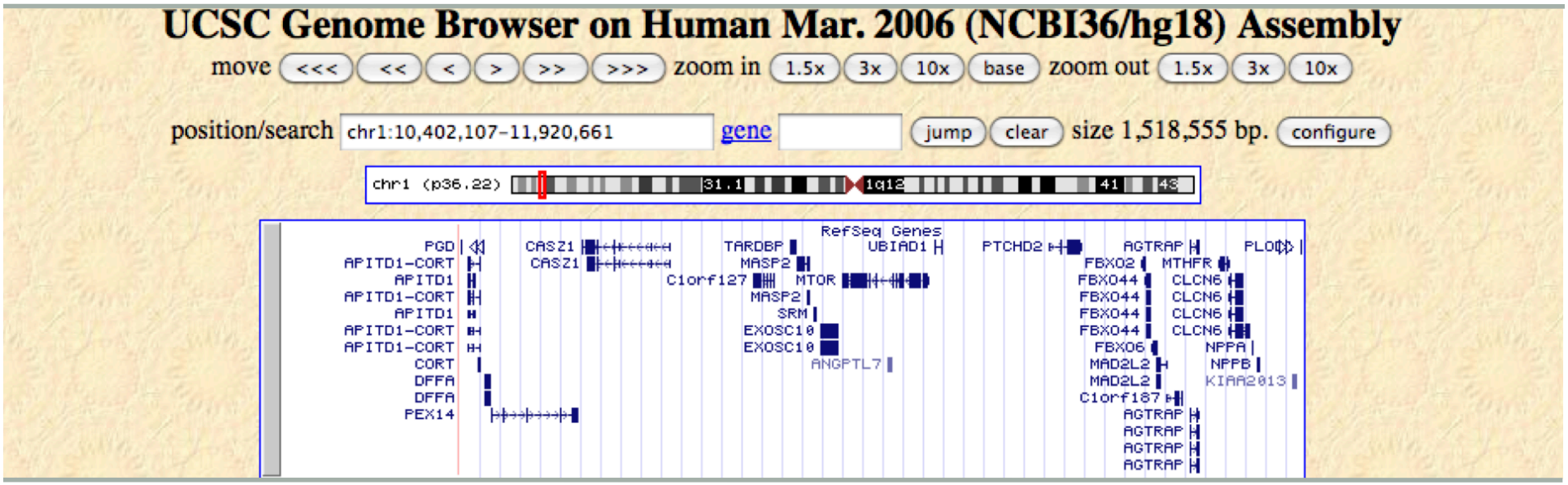
Nielsen *et al.*
Best Paper Award at InfoVis 2009

Challenge 3

Genomic features are sparse

Genome Browsers

LOCAL VIEW

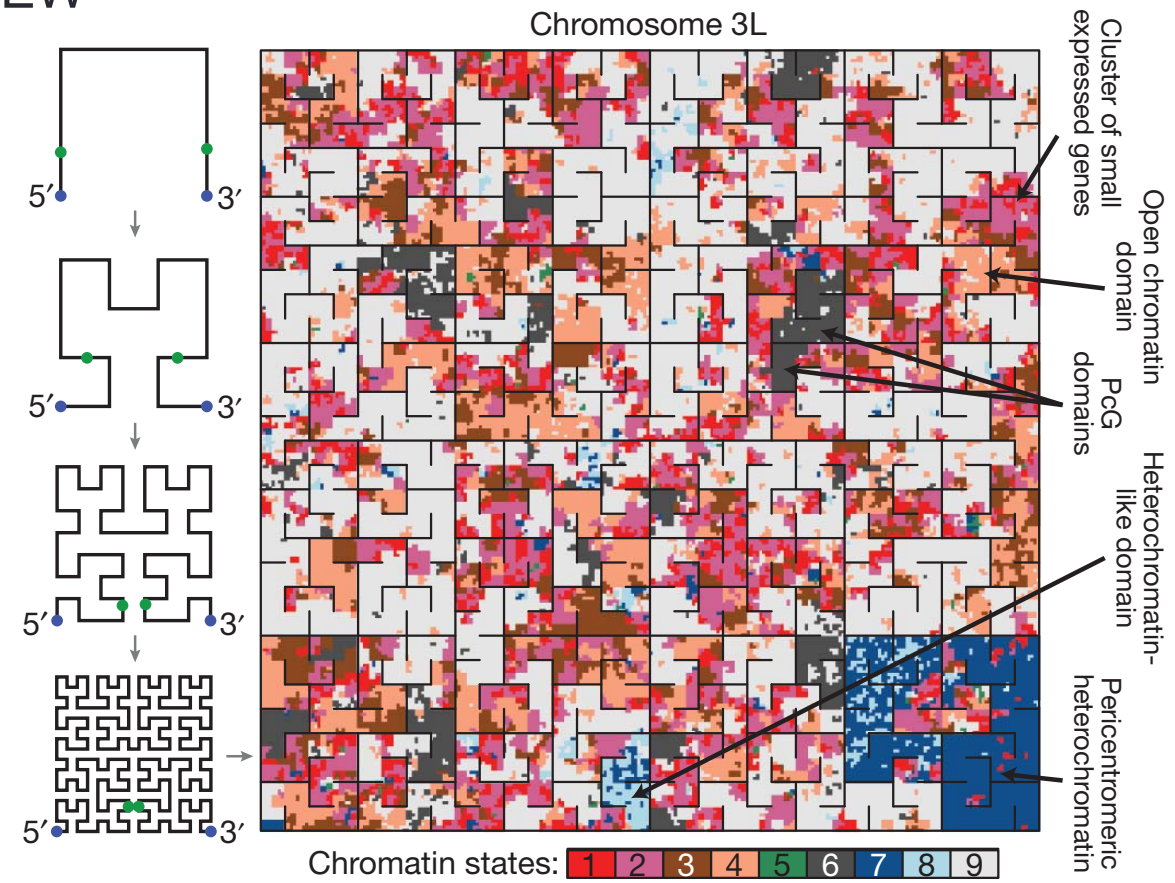


Human chr1, 1 pt corresponds to 480 kb, which is larger than 98% of all human genes!

- Martin Krzywinski

Hilbert Curve

GLOBAL VIEW



Kharchenko *et al*., Nature, 2011
Anders, Bioinformatics, 2009

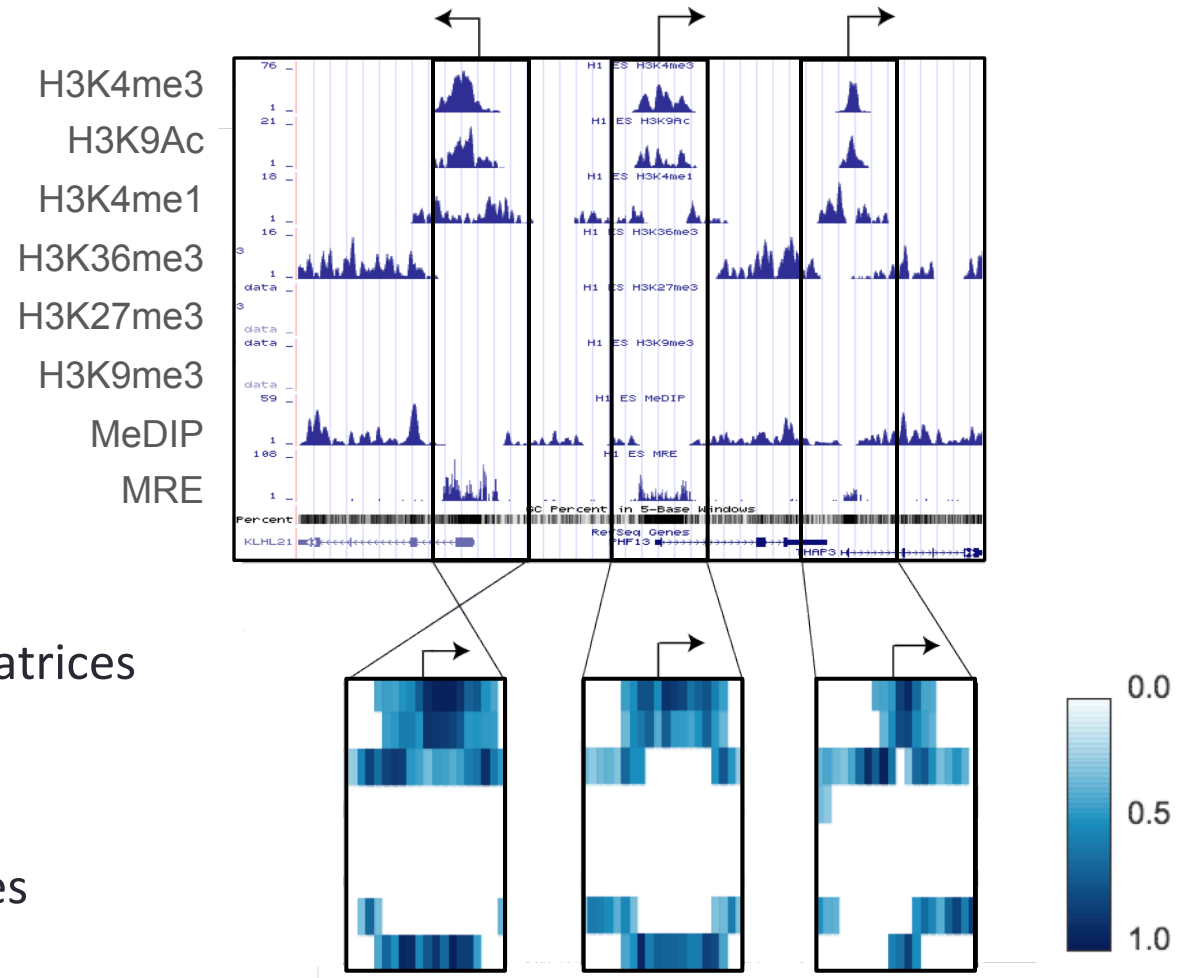
Challenge 3

Genomic features are sparse

- *Need both overview and detail*
  Functional axis (perhaps not full genome)

Spark – a genomic data exploration tool

1. Focus on regions of interest (e.g. transcriptional start sites)



2. Extract data matrices

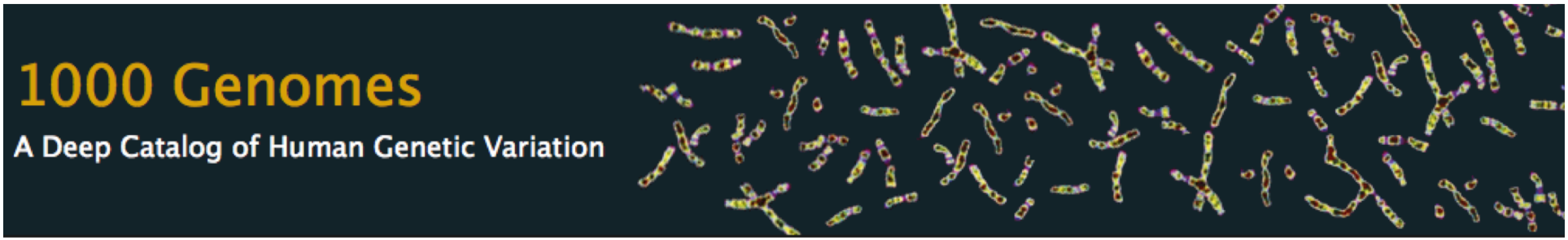
3. Cluster matrices

4. Interactive cluster visualization

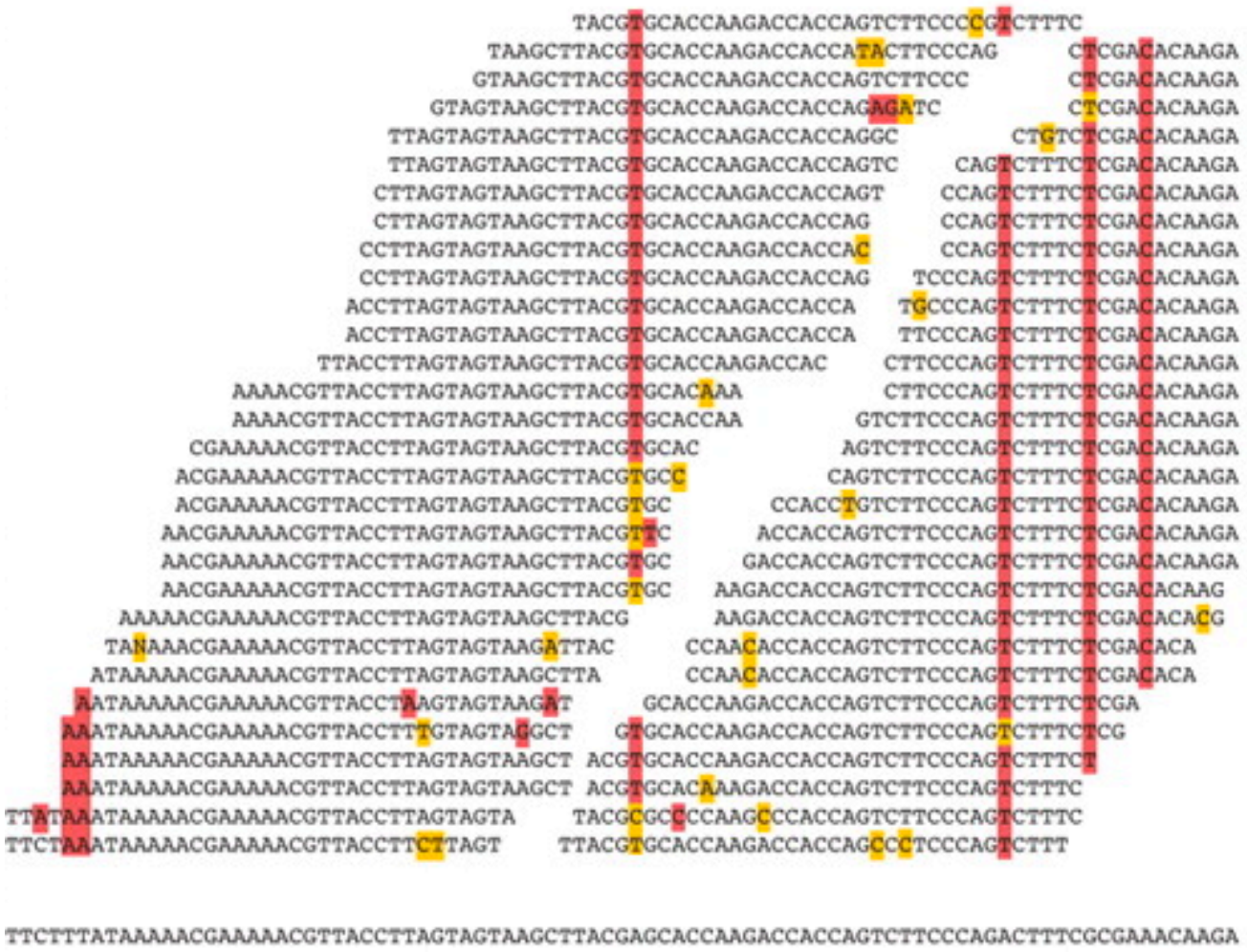
Nielsen *et al*. in preparation

Challenge 4

No longer one genome but many



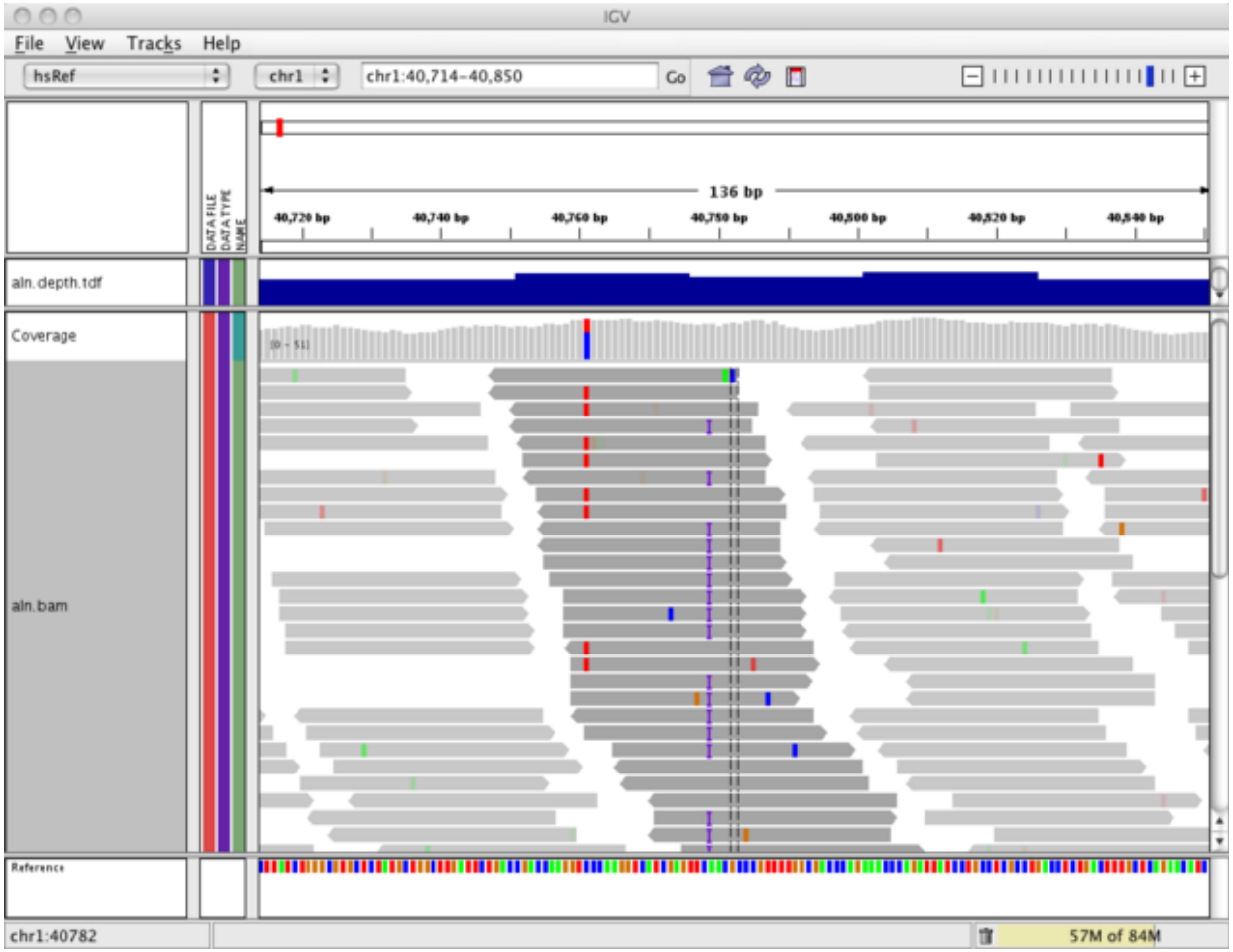
Single nucleotide variation



Ossowski *et al*. Genome Research, 2008

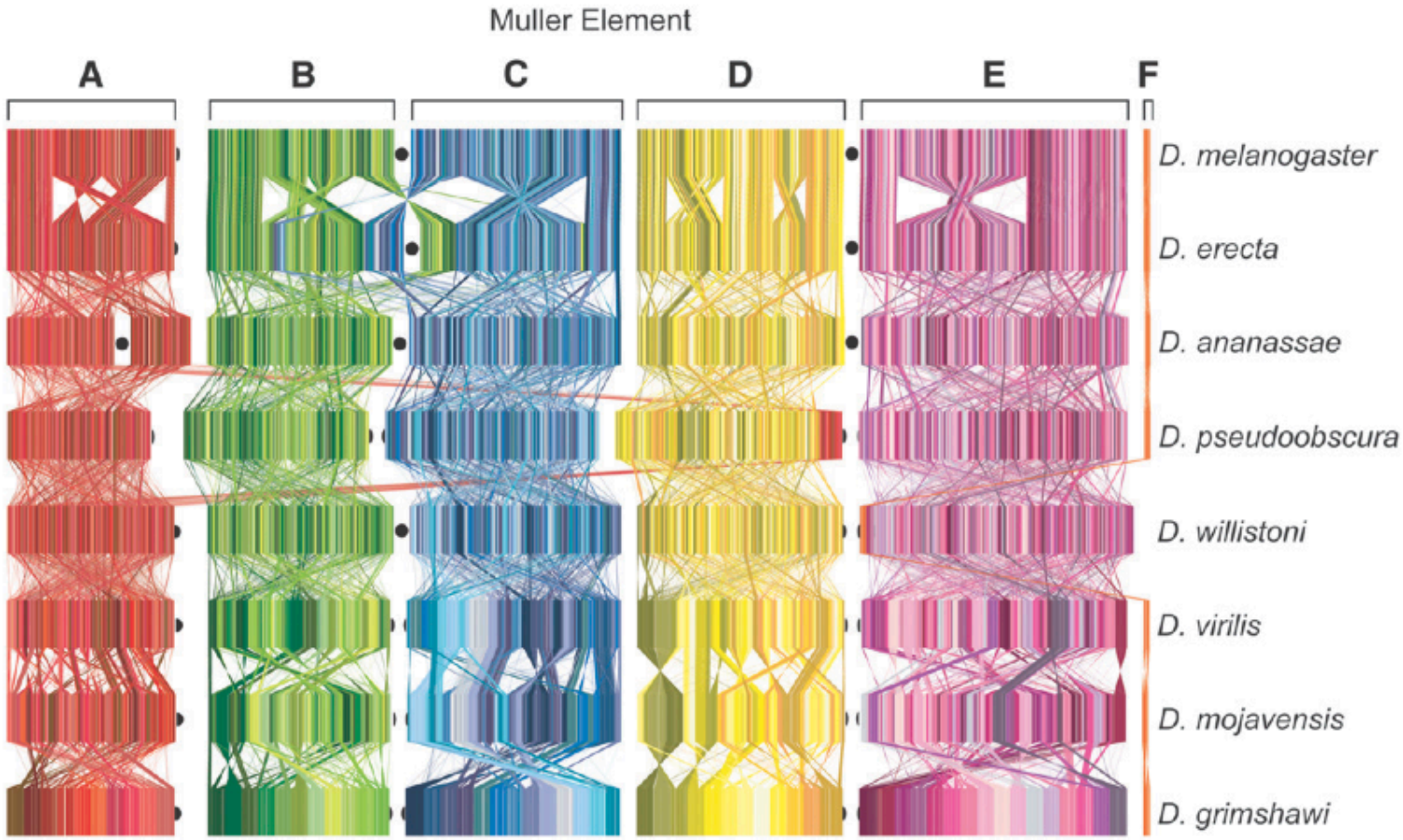
Single nucleotide variation

Integrative Genomics Viewer (IGV)



Robinson *et al*. Nature Biotechnology, 2011

Structural variation



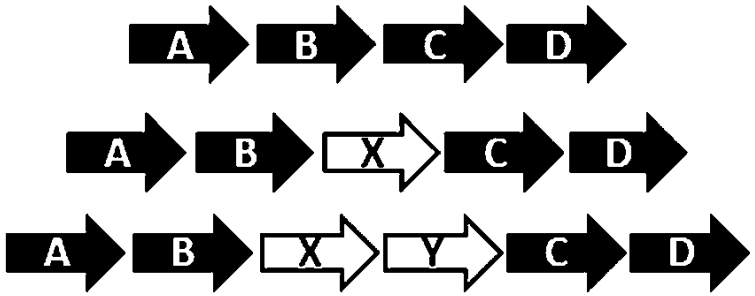
Bhutkar *et al*., Genetics, 2008

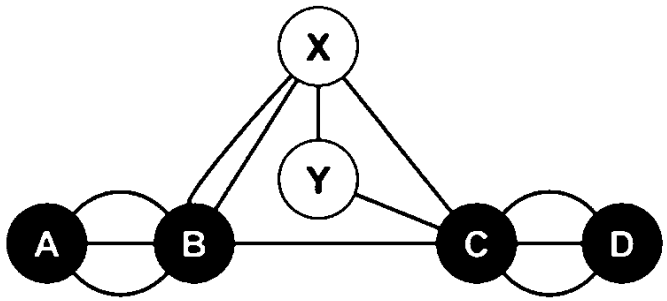
Challenge 4

No longer one genome but many

- *Capture variation on a graph*

Sequence variation on a graph

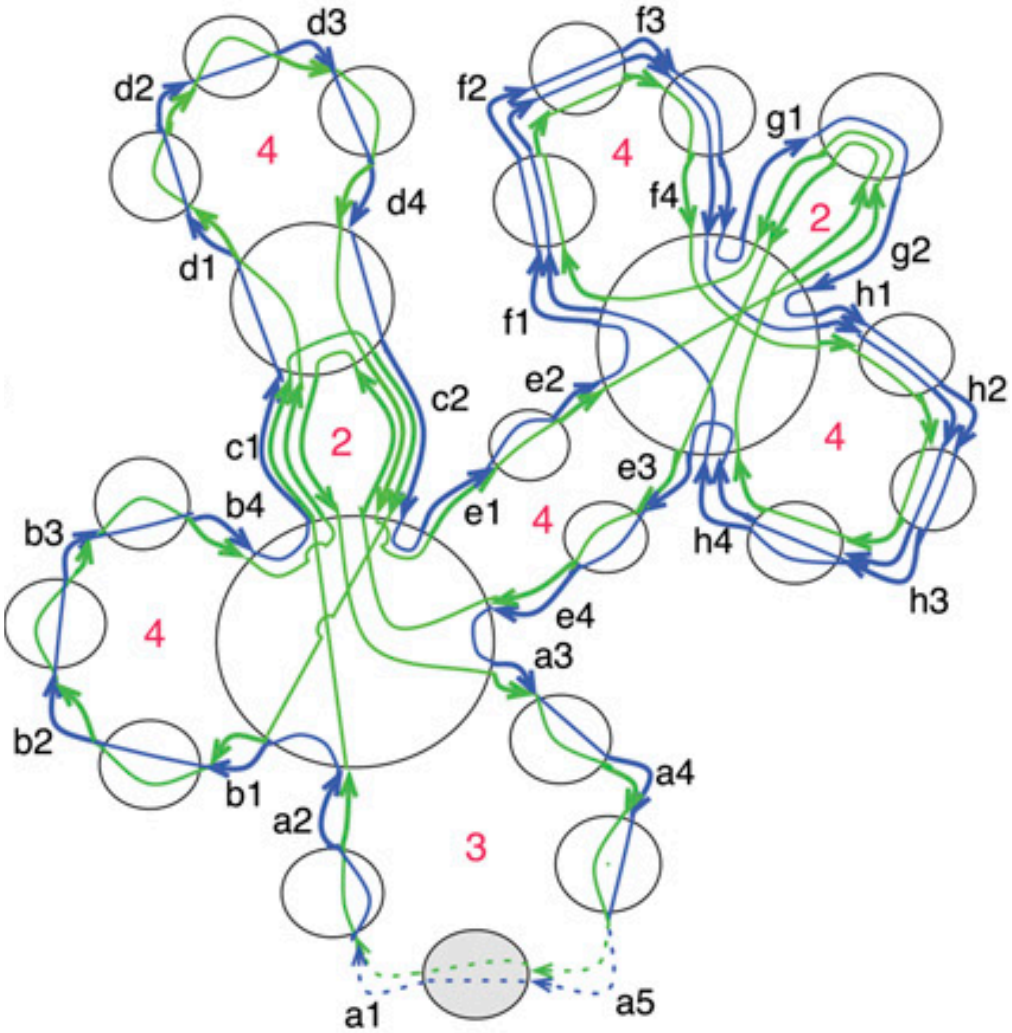




Comeau *et al*., Mol. Biol. Evol., 2010

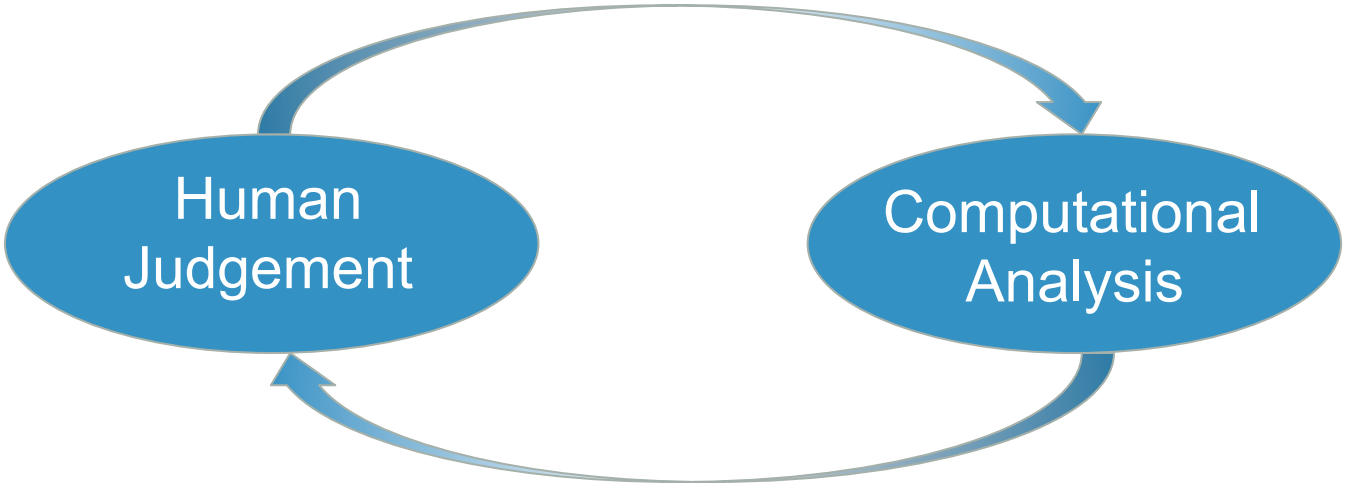
Users may require more time to learn how to interpret graph representations, but such graphs are likely to scale better and may prove more powerful for analysis

Sequence variation on a graph

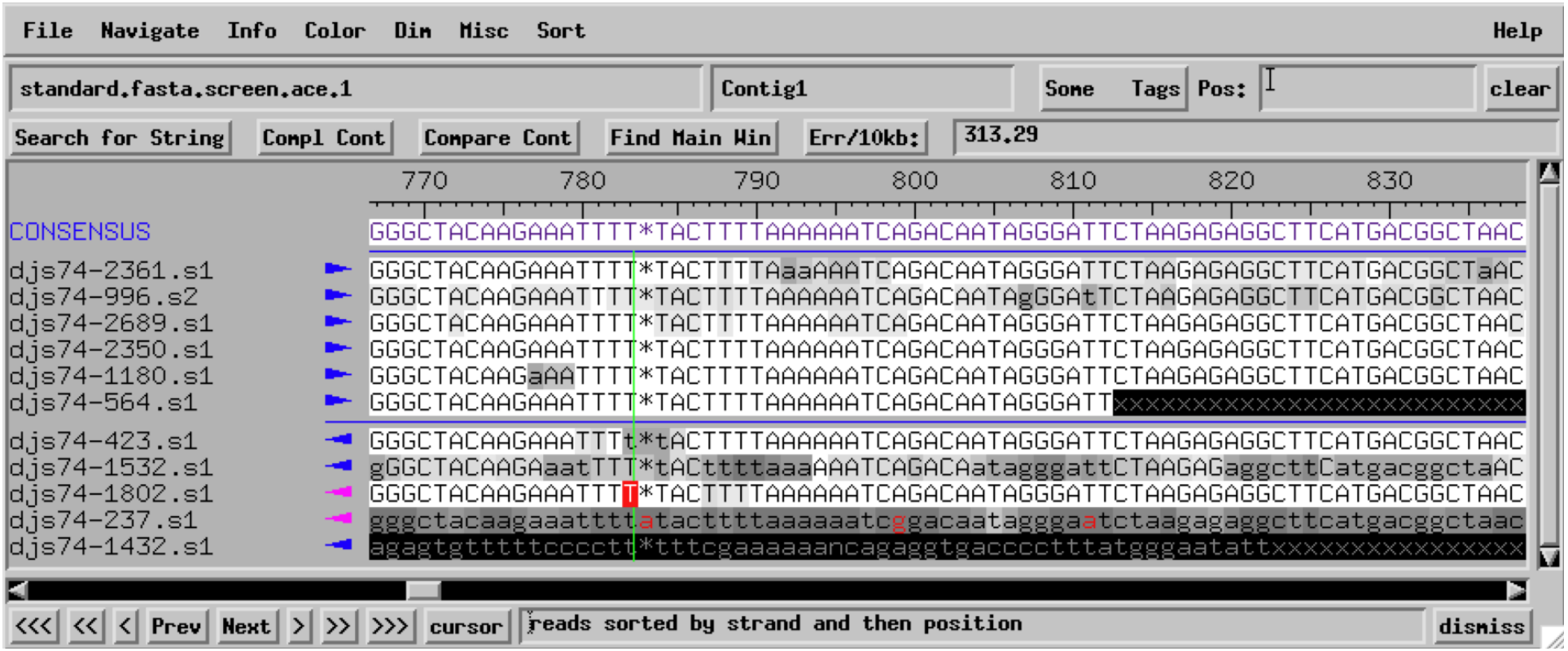


Paten *et al*., Genome Research, 2011

Challenge 5



Consed Genome Assembly and Finishing Tool



David Gordon and Phil Green

Good example of integrated visualization and computational analysis functionality

Challenge 5

Need to integrate computation

- *High interactivity, low memory overhead*

  Avoid storing large data sets locally

  Popularity of web-based tools

  Evolving sequencing technologies

Summary

1. Large number of samples for comparison
2. Large number of data types
3. Genomic features are sparse
4. No longer one genome but many
5. Need to integrate computational analysis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ROADMAP
epigenomics
PROJECT