

TECH DEV

GENOMICS

SEQUENCING

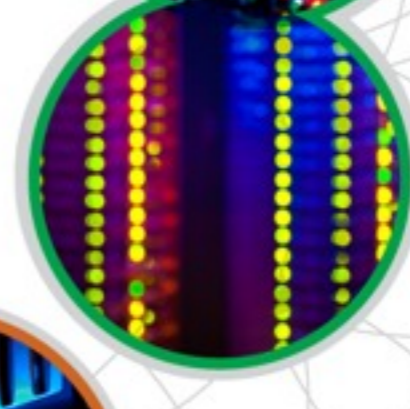
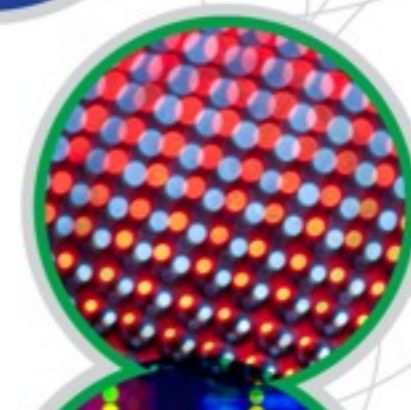
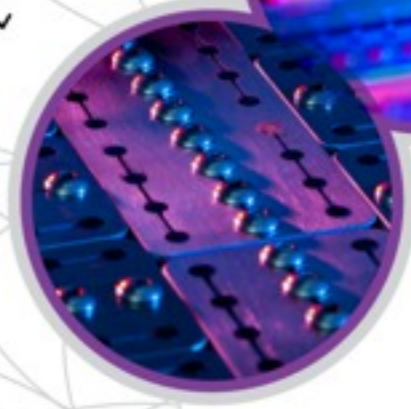
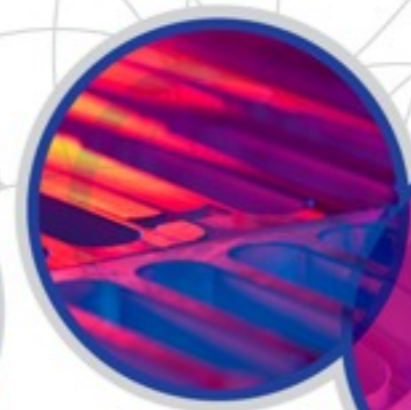
INFORMATICS

COMPUTING



CANADA'S MICHAEL SMITH  
**GENOME  
SCIENCES**  
CENTRE

WWW.BCGSC.CA



# designing effective visualizations in the biological sciences

13.00 - 14.00

**MARTIN KRZYWINSKI**

Genome Sciences Center  
BC Cancer Agency  
Vancouver, Canada

PSA ANNUAL MEETING 2011

GENOMICS WORKSHOP

University of Washington  
12 July 2011

# design : effective visual communication

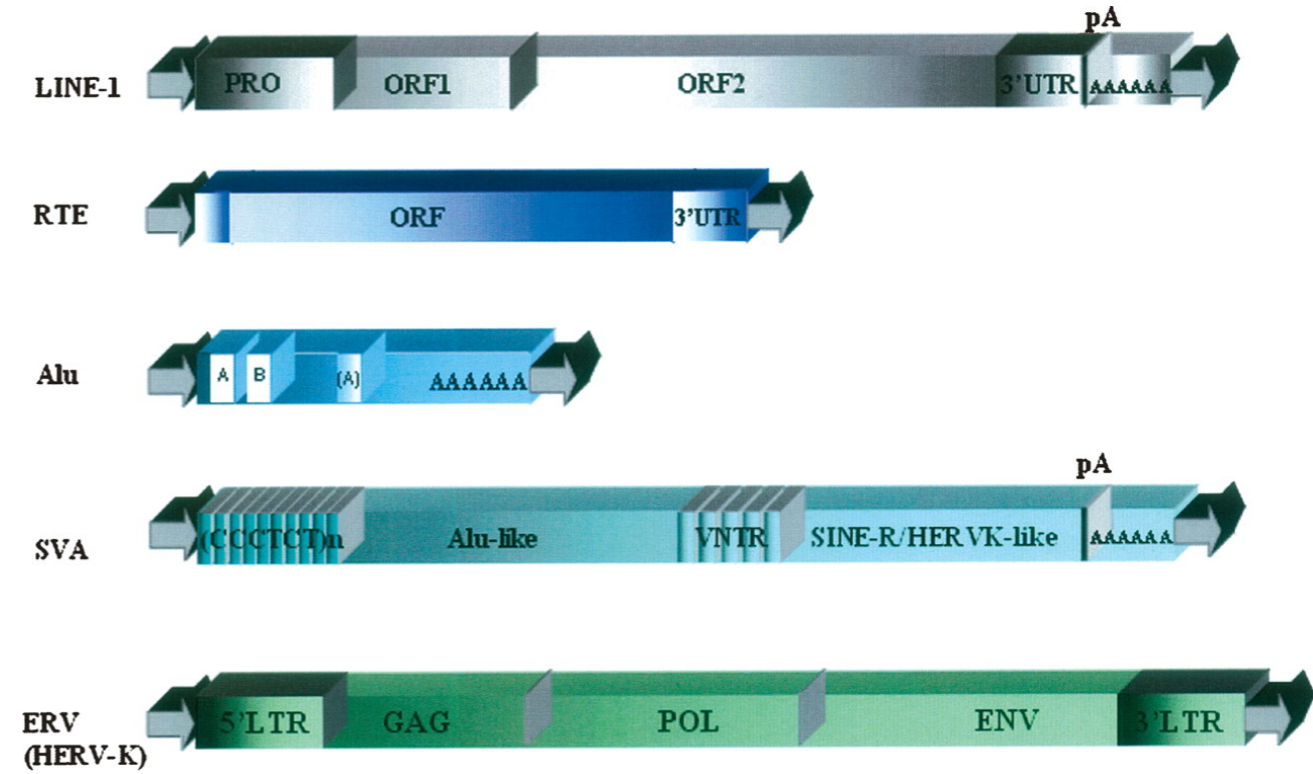
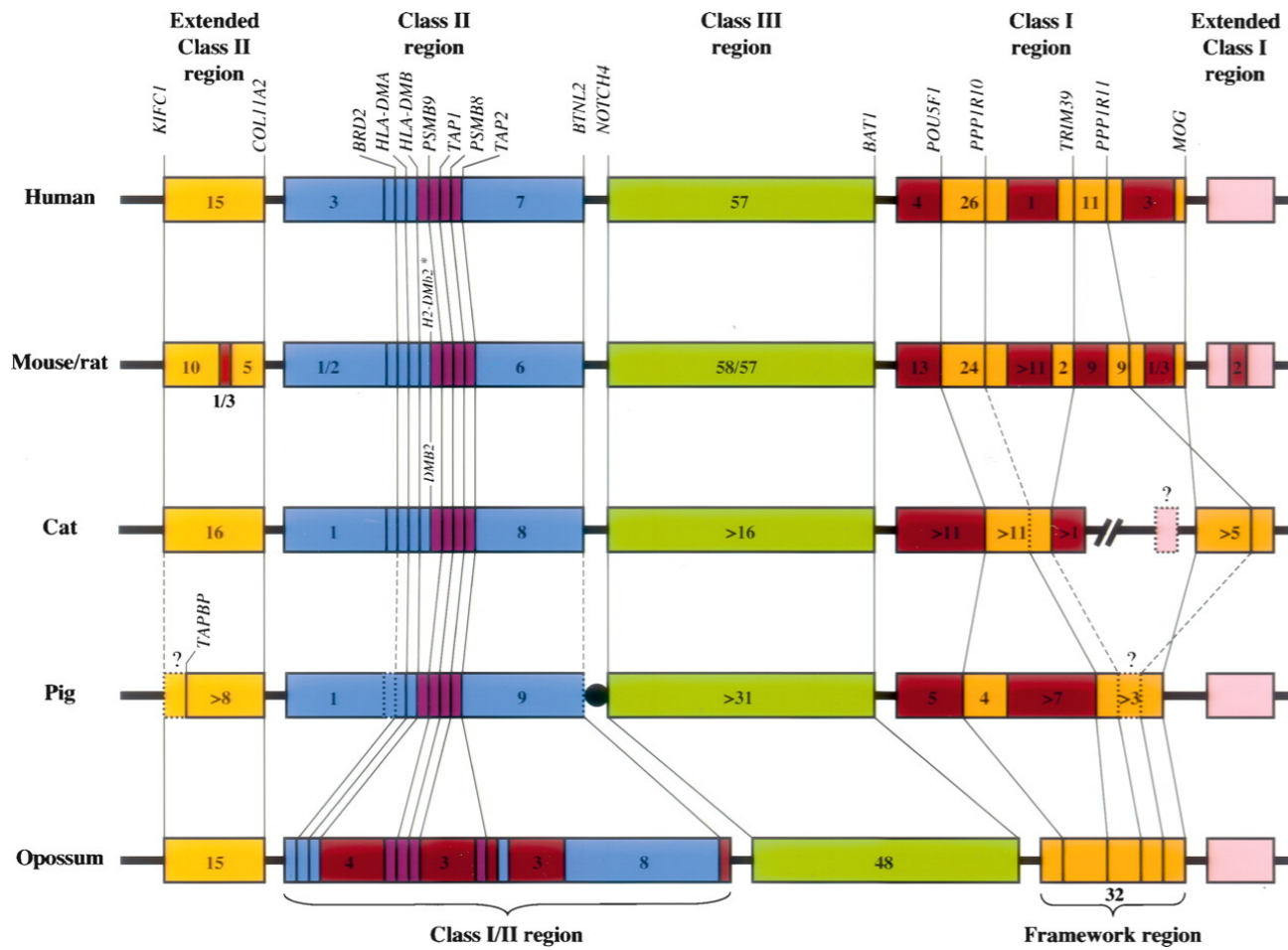
## OBJECTIVE

FOLLOWING GUIDELINES

## SUBJECTIVE

BREAKING GUIDELINES PURPOSEFULLY

# OBJECTIVE ASPECTS OF CLEAR COMMUNICATION



Excellent organization and consistency. Clear use of color. Vertical lines cue continuity.

Samollow, P.B., The opossum genome: insights and opportunities from an alternative mammal. *Genome Res*, 2008. 18(8): p. 1199-215.

Overly ornamental. Illegible and inconsistently formatted text. Redundant elements.

Gentles, A.J., et al., Evolutionary dynamics of transposable elements in the short-tailed opossum *Monodelphis domestica*. *Genome Res*, 2007. 17(7): p. 992-1004.

# OBJECTIVE ASPECTS OF ATTRACTION

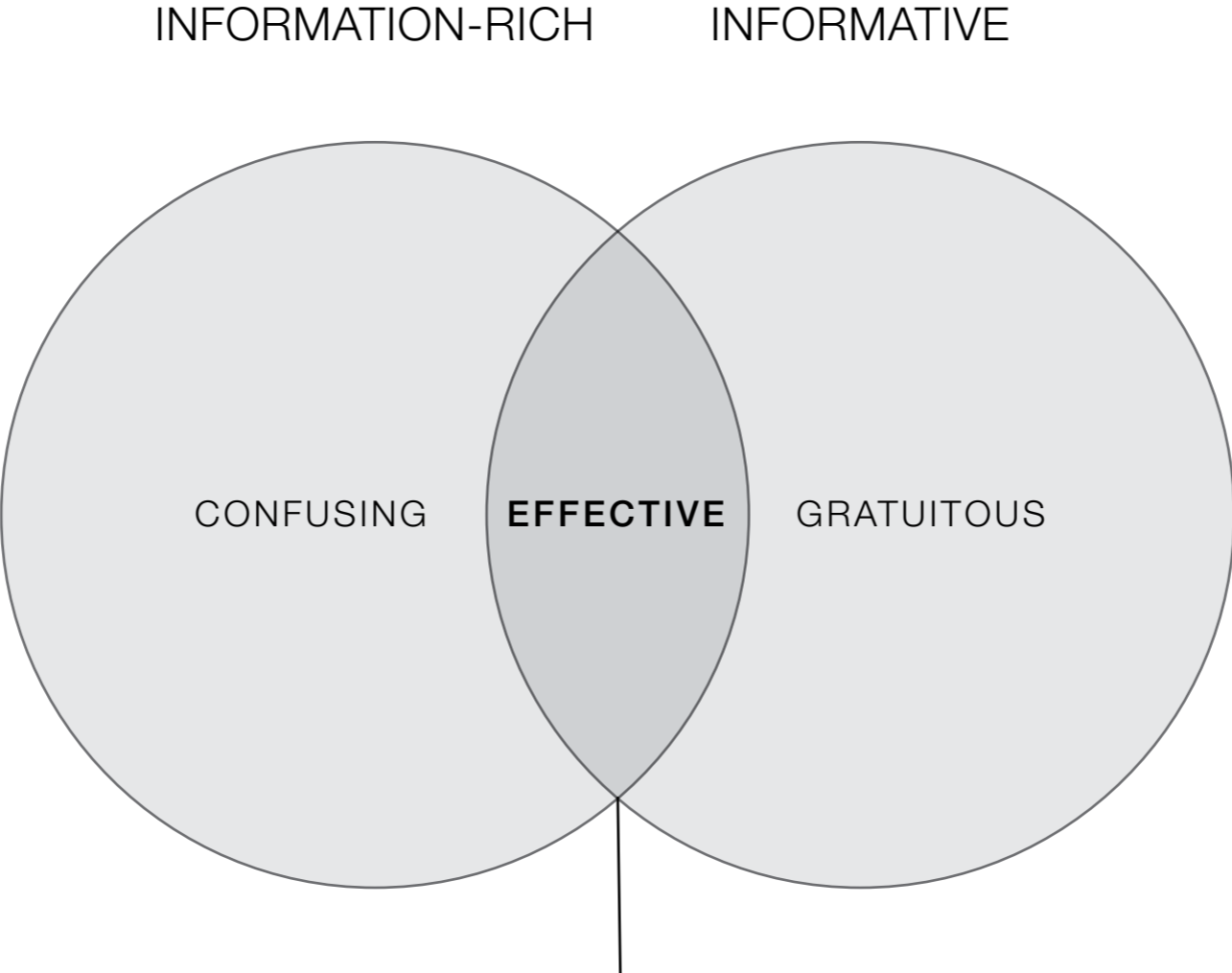


Healthy coat. Pleasant colour scheme. Vigorously adorable proportions. Attentive gaze. Sympathetic eyes.



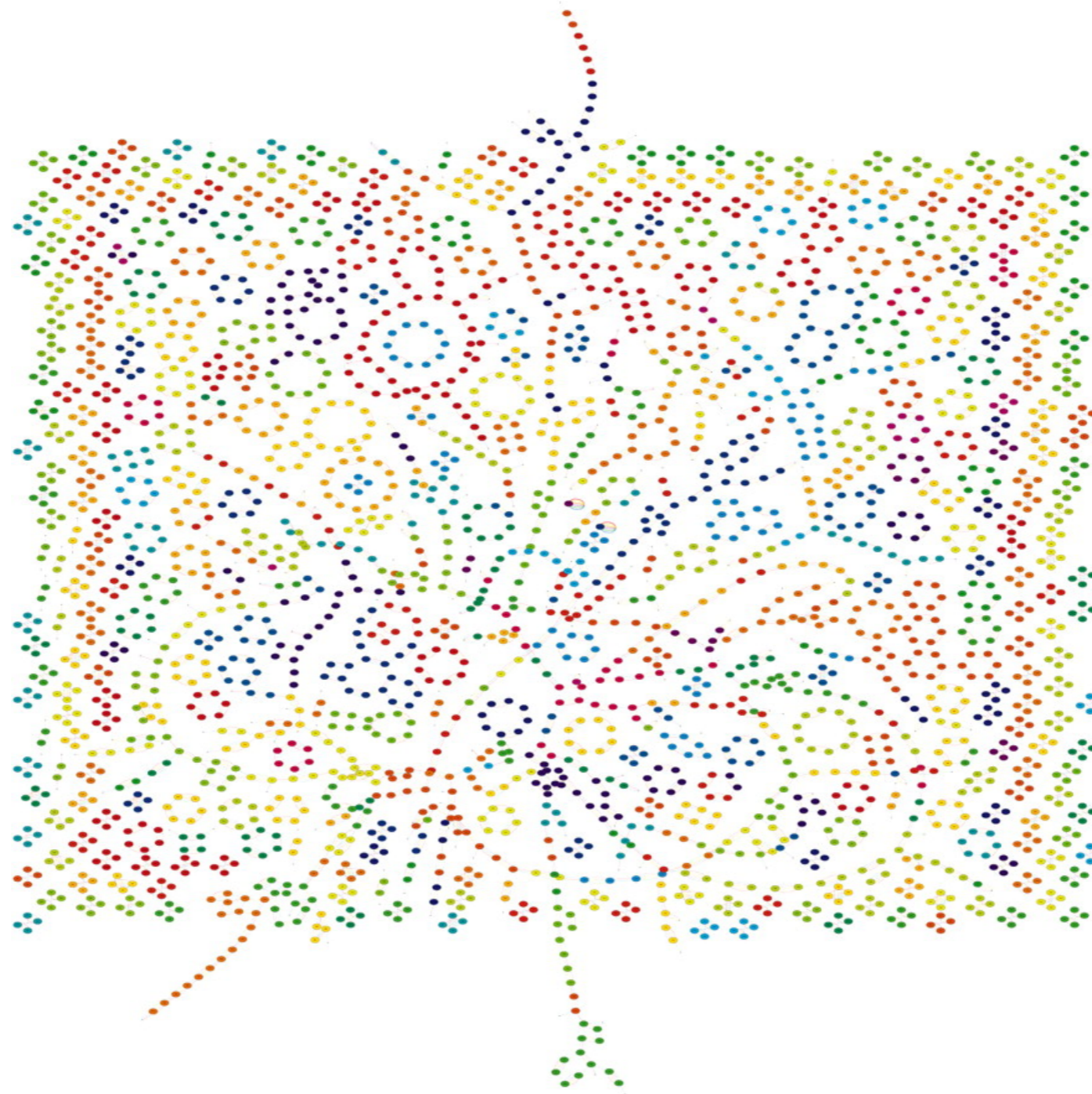
Where do I start?

# WHAT IS AN EFFECTIVE VISUALIZATION?

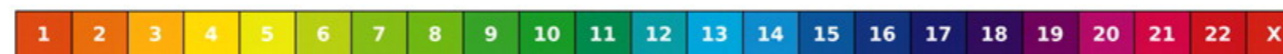


- CLEAR MESSAGE**
- HIGH DATA-TO-INK RATIO**
- ACCESSIBLE COMPLEXITY**

# INFORMATION-RICH, NOT INFORMATIVE

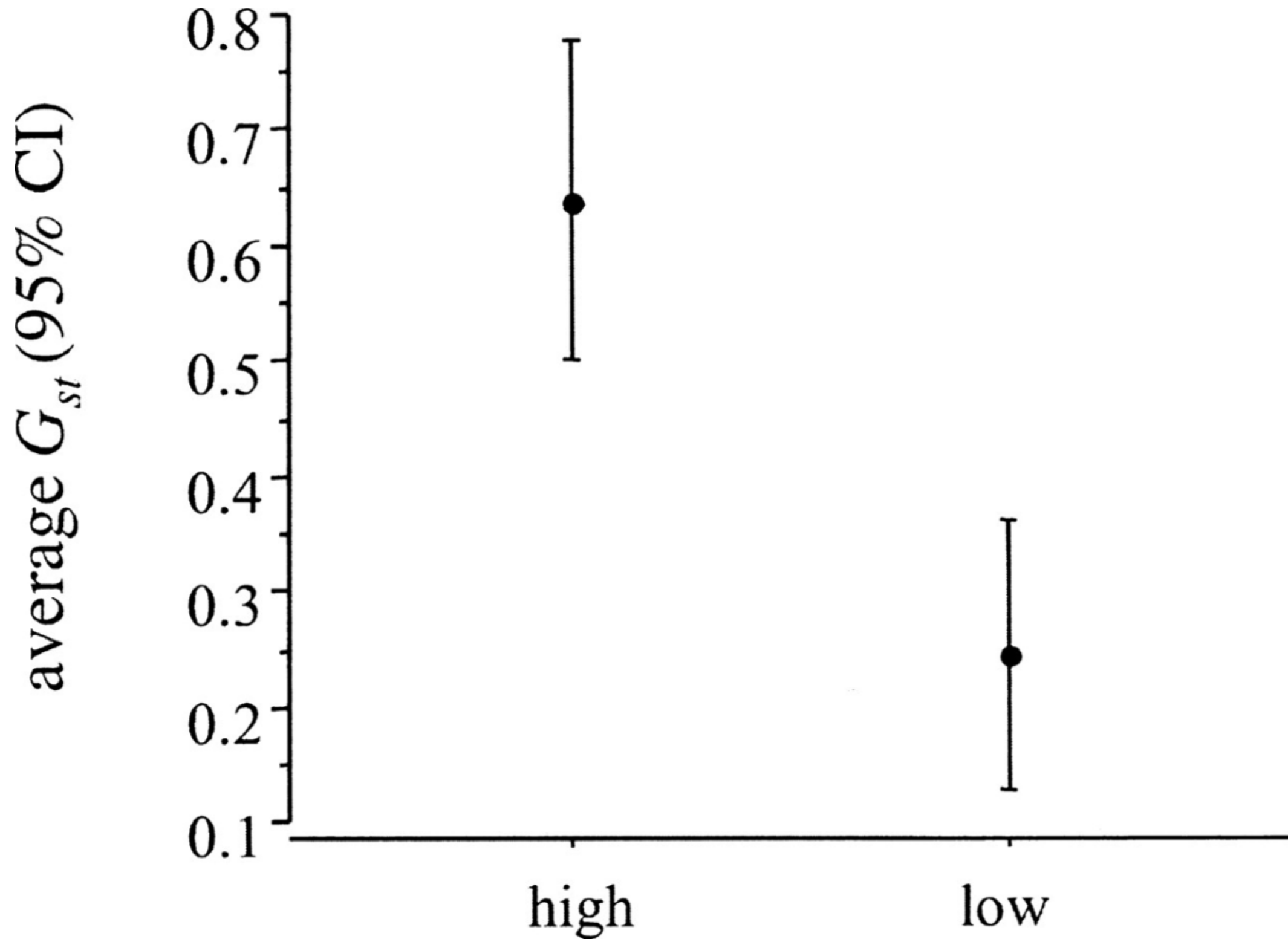


Chromosome colors:



The breakpoint graph  $G(M,R,D,Q,H,C)$  (obverse edges are not shown) of six mammalian genomes. Alekseyev, M.A. and P.A. Pevzner, Breakpoint graphs and ancestral genome reconstructions. *Genome Res*, 2009. 19(5): p. 943-57.

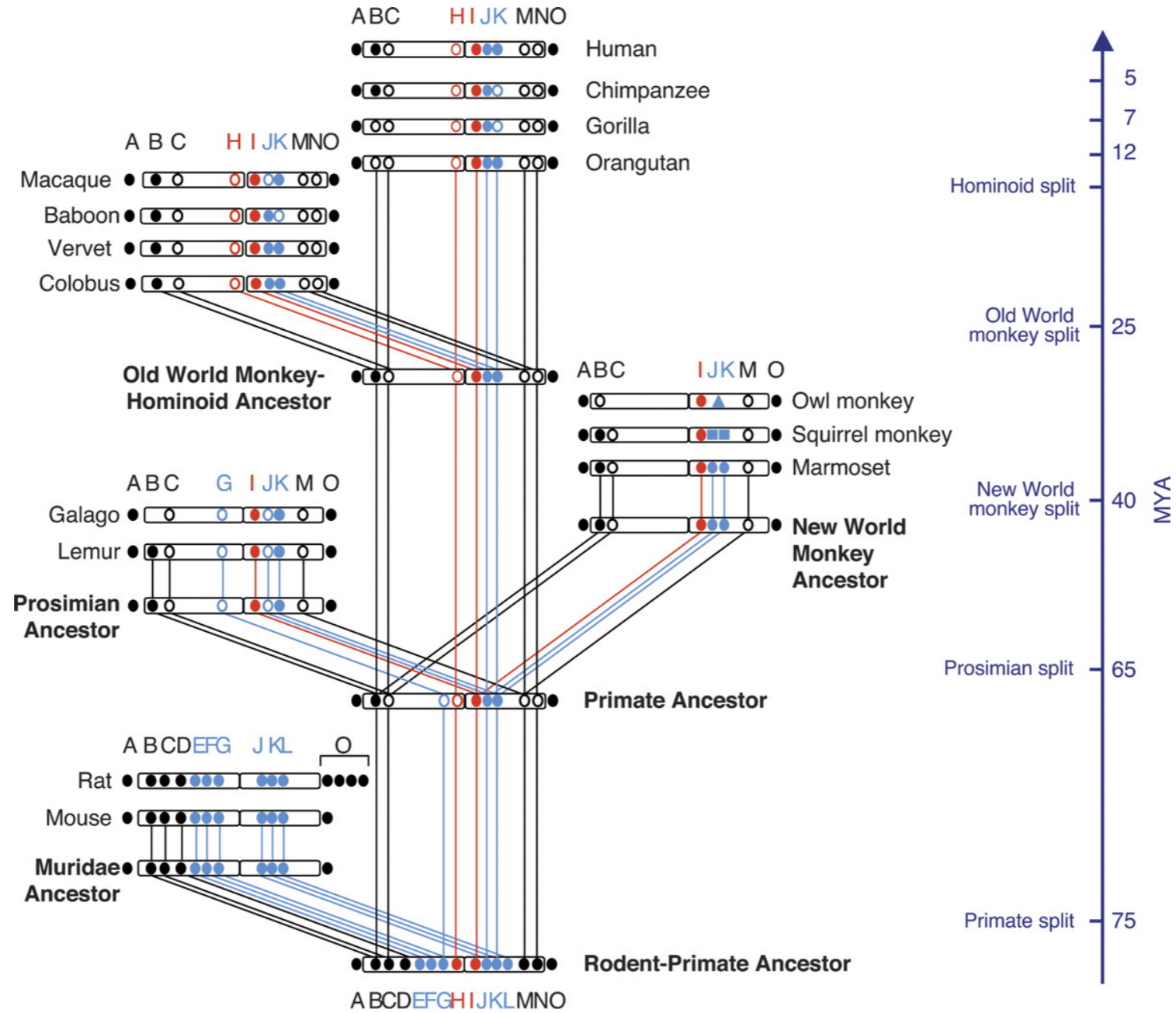
# INFORMATIVE, NOT INFORMATION-RICH



Mean and 95% confidence interval of average  $G_{st}$  based on direct sequencing of fragments within high (N = 8) and low (N = 6) differentiation regions in natural population of house mice. Harr, B., Genomic islands of differentiation between house mouse subspecies. *Genome Res*, 2006. 16(6): p. 730-7.



# INFORMATION-RICH AND INFORMATIVE



<b>A</b> <i>WFDC5/Wfdc5</i>	<b>D</b> <i>WFDC15a/Wfdc15a</i>	<b>M</b> <i>WFDC15c/Wfdc15c</i>
<b>B</b> <i>WFDC12/Wfdc12</i>	<b>E, F, G, J, K, L</b> <i>SEMG/Svs</i> family	<b>N</b> <i>WFDC15d/Wfdc15d</i>
<b>C</b> <i>WFDC15b/Wfdc15b</i>	<b>H, I</b> Trappin ( <i>PI3</i> ) family	<b>O</b> <i>SLPI/Slpi</i> family

Evolutionary history of the centromeric WFDC sublocus. Hurlle, B., W. Swanson, and E.D. Green, Comparative sequence analyses reveal rapid and divergent evolutionary changes of the WFDC locus in the primate lineage. *Genome Res*, 2007. 17(3): p. 276-86.

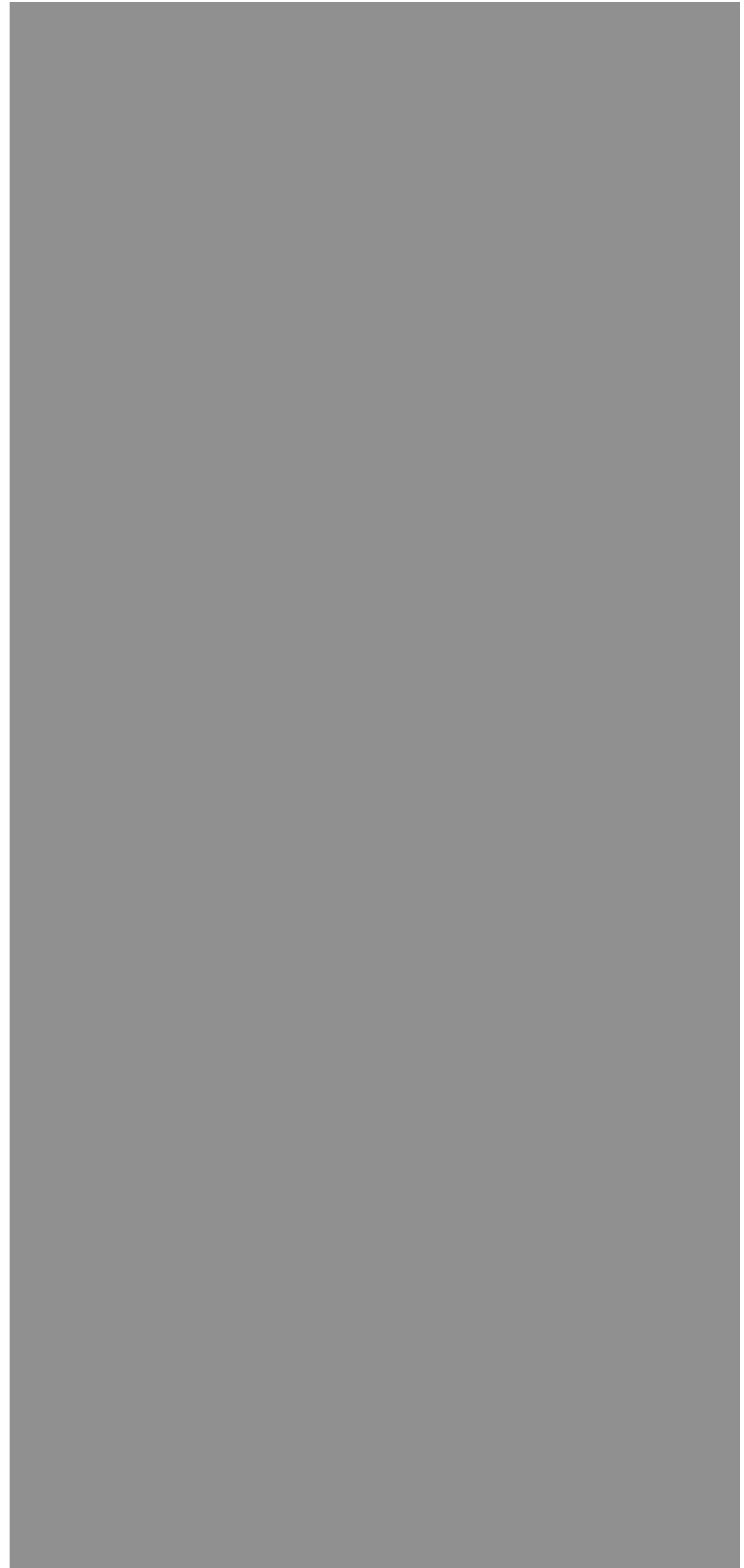
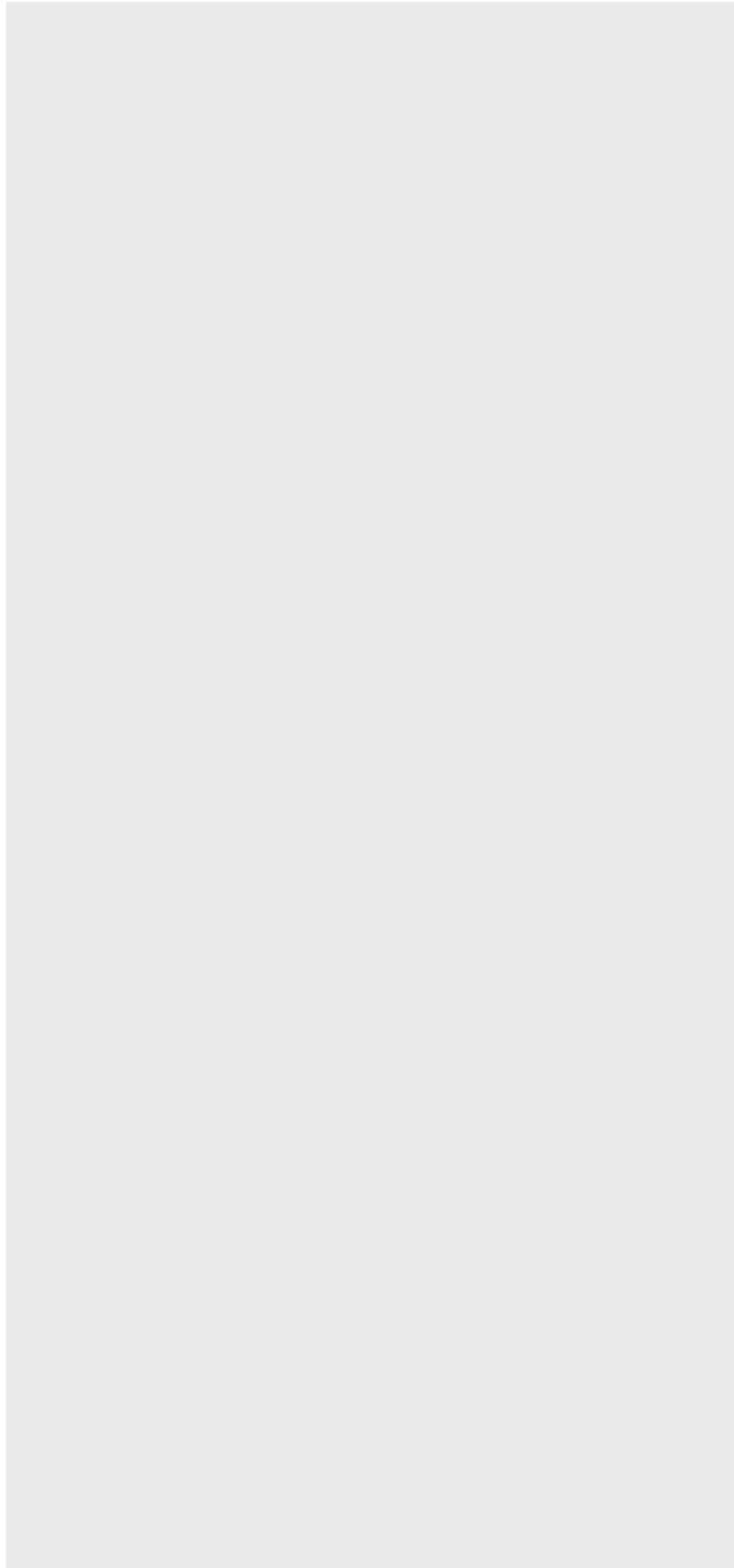
# requirements for effective visual communication

**LEGIBILITY**

**CLARITY**

AND OPTIONALLY,

**ATTRACTIVENESS**



# LEGIBLE

are all elements discernable?

does text contrast well with background?

is there simultaneous contrast?

RESOLUTION  
PARSABILITY  
COLOR

# LEGIBLE

are all elements discernable?

does text contrast well with background?

is there simultaneous contrast?

RESOLUTION

PARSABILITY

COLOR

# CLEAR

does the reader see the noise or trends?

message delivered?

are there redundant or ornamental elements?

STRONG MESSAGE

EXCESS INK

REDUNDANCY

GLYPHS

OUTLIER HIJACK

# LEGIBLE

are all elements discernable?

does text contrast well with background?

is there simultaneous contrast?

RESOLUTION

PARSABILITY

COLOR

# CLEAR

does the reader see the **noise** or **trends**?

message delivered?

are there redundant or ornamental elements?

STRONG MESSAGE

EXCESS INK

REDUNDANCY

GLYPHS

OUTLIER HIJACK

# ATTRACTIVE

are spacing and orientation of visual cues consistent?

GRID LAYOUT

# LEGIBLE

are all elements discernable?

does text contrast well with background?

is there simultaneous contrast?

# CLEAR

does the reader see the noise or trends?

message delivered?

are there redundant or ornamental elements?

# ATTRACTIVE

are spacing and orientation of visual cues consistent?

DO I HAVE A MESSAGE ?    HAVE I COMMUNICATED IT ?

RESOLUTION  
PARSABILITY  
COLOR

STRONG MESSAGE  
EXCESS INK  
REDUNDANCY  
GLYPHS  
OUTLIER HIJACK

GRID LAYOUT

**legibility** clarity attractiveness

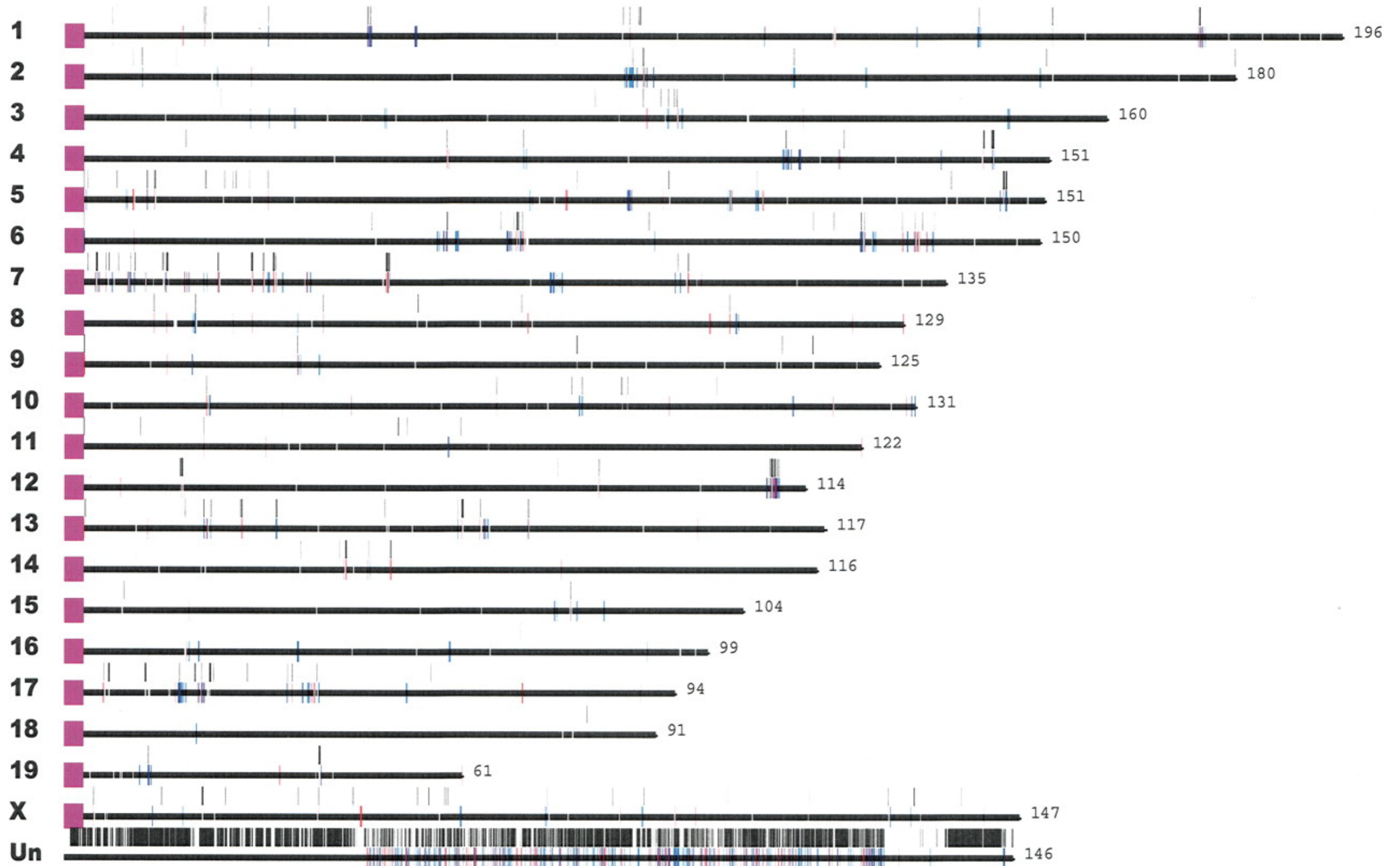
RESOLUTION

PARSABILITY

COLOR

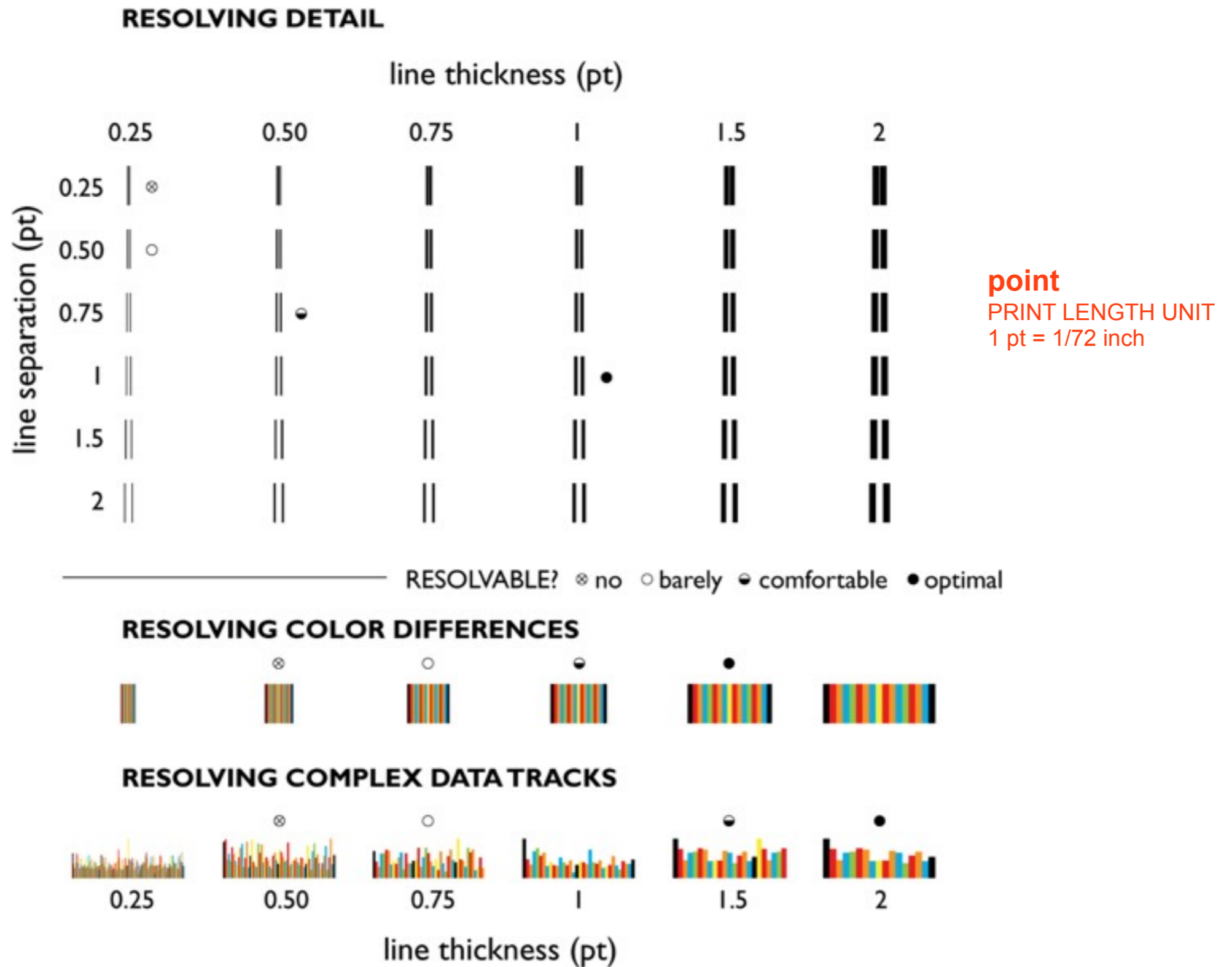


# RESOLUTION & ACUITY LIMITS



Mouse segmental duplications. J. A. Bailey, D. M. Church, M. Ventura, M. Rocchi, E. E. Eichler, Genome Res 14, 789 (May, 2004).

# RESOLUTION & ACUITY LIMITS

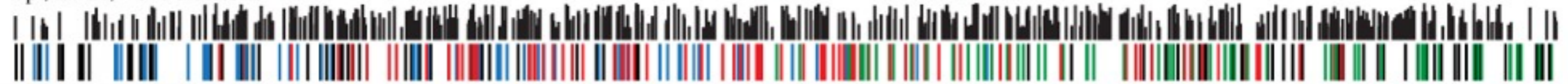


The resolving power of the eye is approximately 50 cycles per degree. This limits us from distinguishing features smaller than 0.1 mm at a reading distance of 30 cm. Larger features must be used to maintain legibility and comprehension. 1 point = 1/72 inch = 0.0353 cm

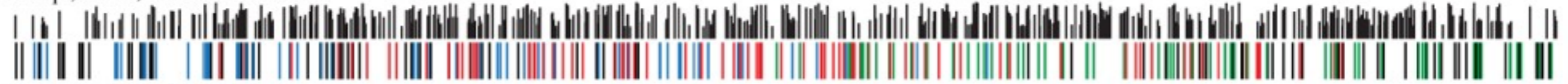
# RESOLUTION & ACUITY LIMITS

**DO NOT DIVIDE YOUR SCALE INTO MORE THAN 500 INTERVALS**

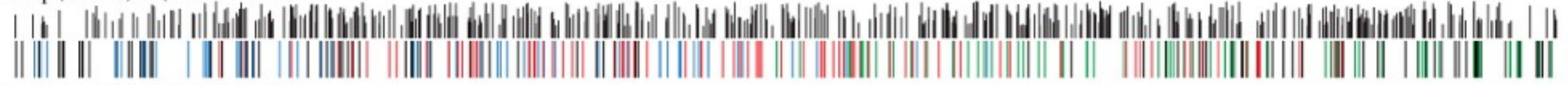
1 pt, 2.9 kb, ~ 500 divisions



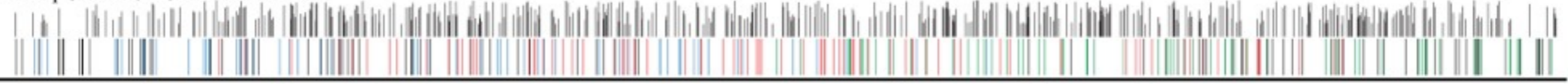
0.75 pt, 2.2 kb, ~ 700 divisions



0.5 pt, 1.5 kb, ~ 1,050 divisions



0.25 pt, 0.7 kb, ~2,100 divisions



*S. cerevisiae* chrIV, 1,531 kb scale length 183 mm, 7.2 in, 519 pt scale resolution 8.4 kb/mm, 213 kb/in, 2.9 kb/pt

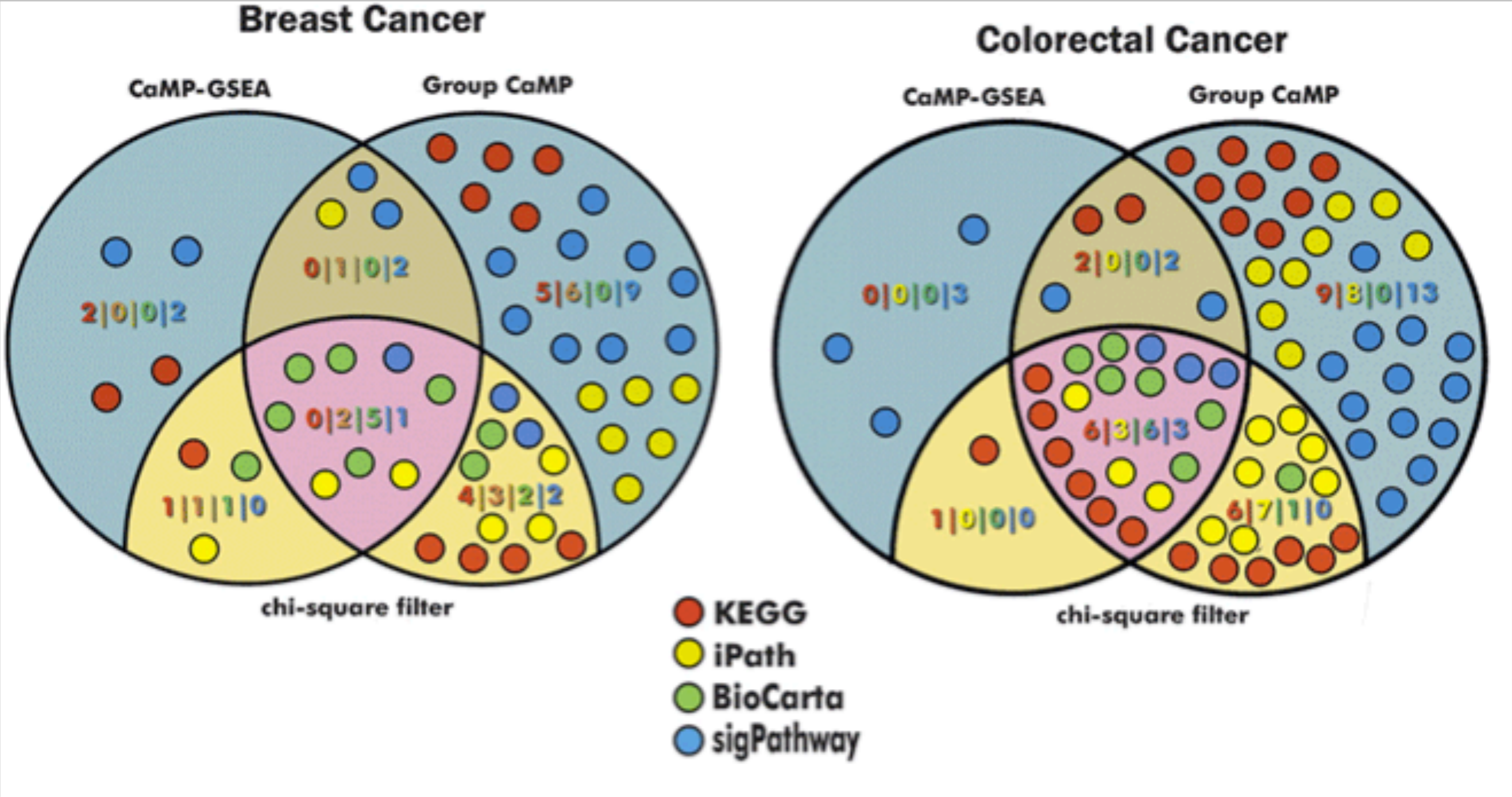
As a rule of thumb, you should not divide your scale into more than 500 intervals per 8.5 inch (216mm, US letter size). This corresponds to 1 pt on a 183 mm figure, 4 pixels on a 1920 horizontal resolution display, or 2 pixels on a typical LCD projector.

# PARSABILITY LIMITS



The specific domains and combinations in the domain graph. Ye, Y. and A. Godzik, Comparative analysis of protein domain organization. Genome Res, 2004. 14(3): p. 343-53.

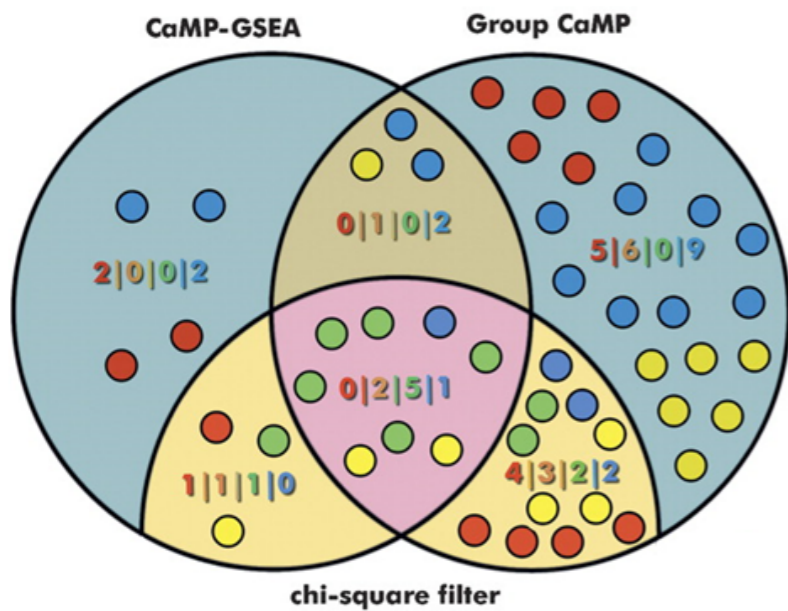
# PARSABILITY LIMITS



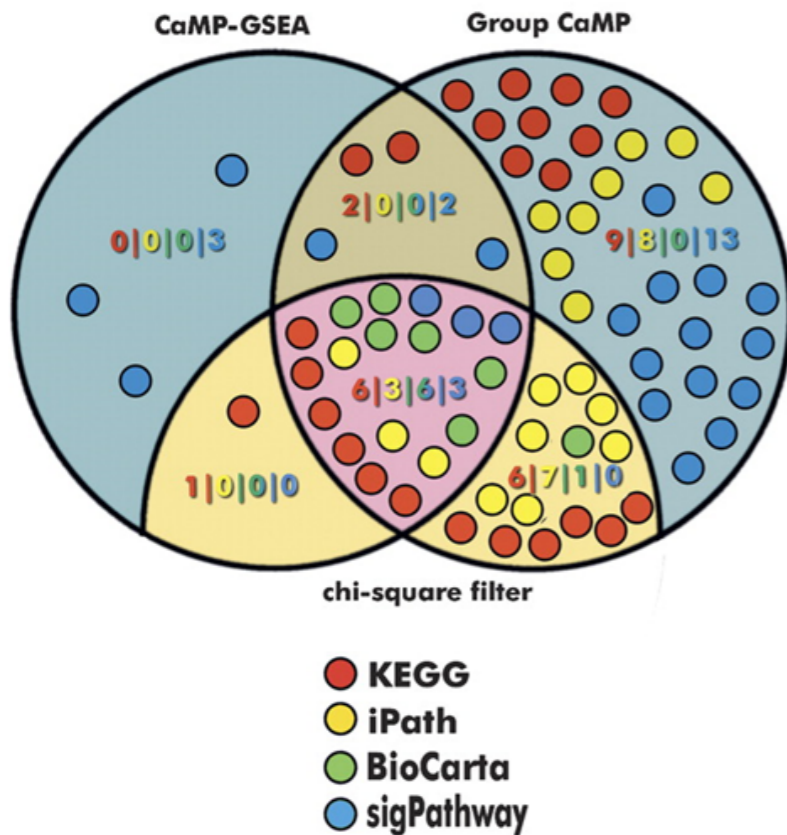
Comparison of mutation enrichment in cellular pathways using complementary statistical approaches. Lin, J., et al., A multidimensional analysis of genes mutated in breast and colorectal cancers. Genome Res, 2007. 17(9): p. 1304-18.

# REFACTORIZING COMPLEXITY

## Breast Cancer

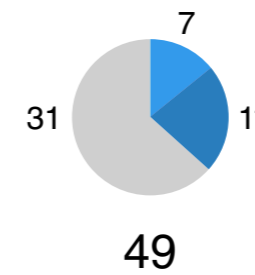


## Colorectal Cancer

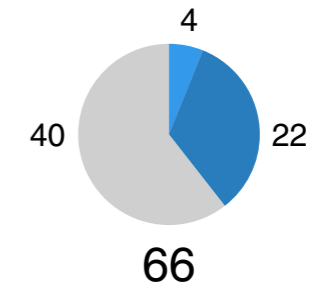


A

BREAST



COLORECTAL

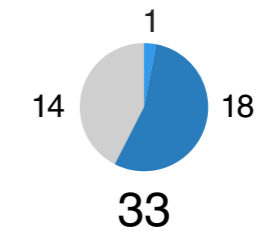


ALL PATHWAYS

CaMP-GSEA      CaMP



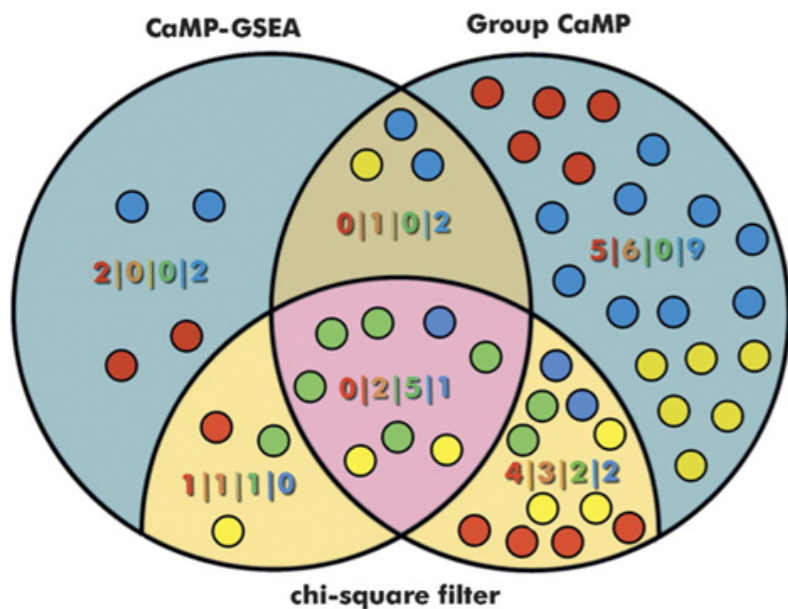
$\chi^2$



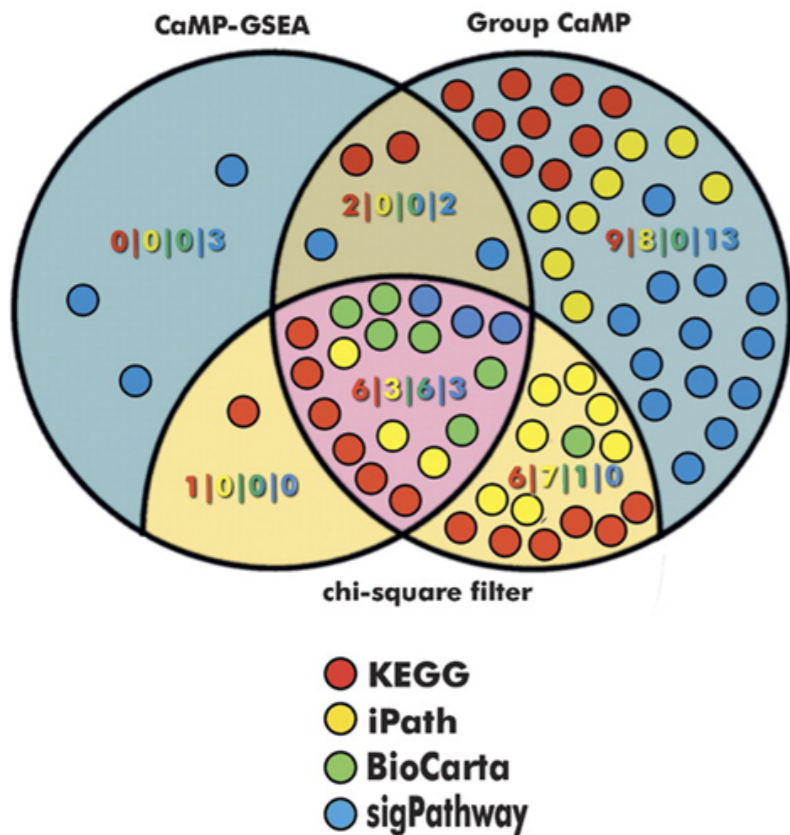
Comparison of mutation enrichment in cellular pathways using complementary statistical approaches. Lin, J., et al., A multidimensional analysis of genes mutated in breast and colorectal cancers. Genome Res, 2007. 17(9): p. 1304-18.

# REFACTORIZING COMPLEXITY

## Breast Cancer

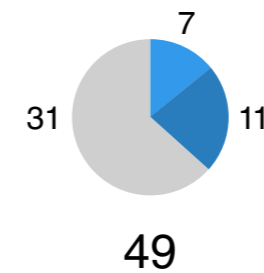


## Colorectal Cancer

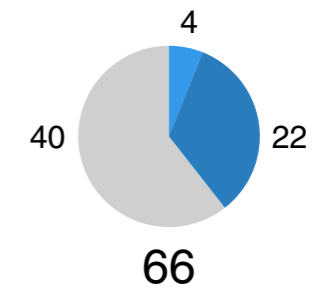


A

BREAST

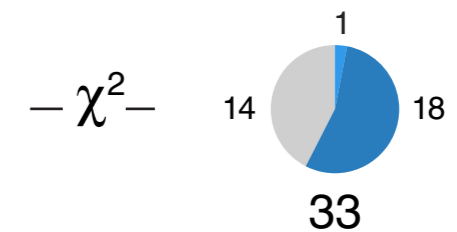


COLORECTAL

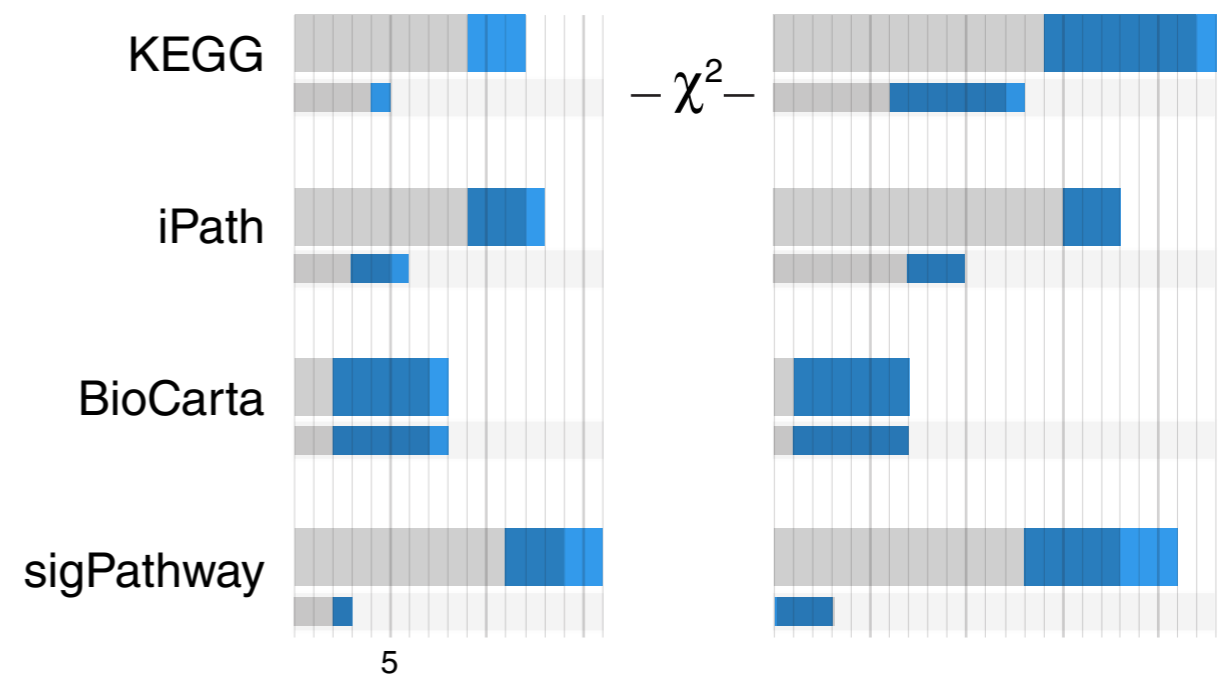


ALL PATHWAYS

CaMP-GSEA      CaMP



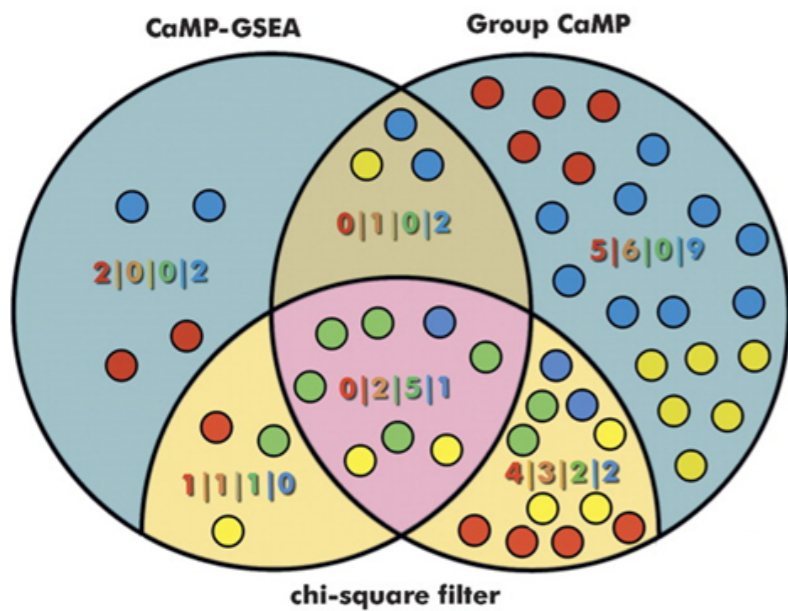
B



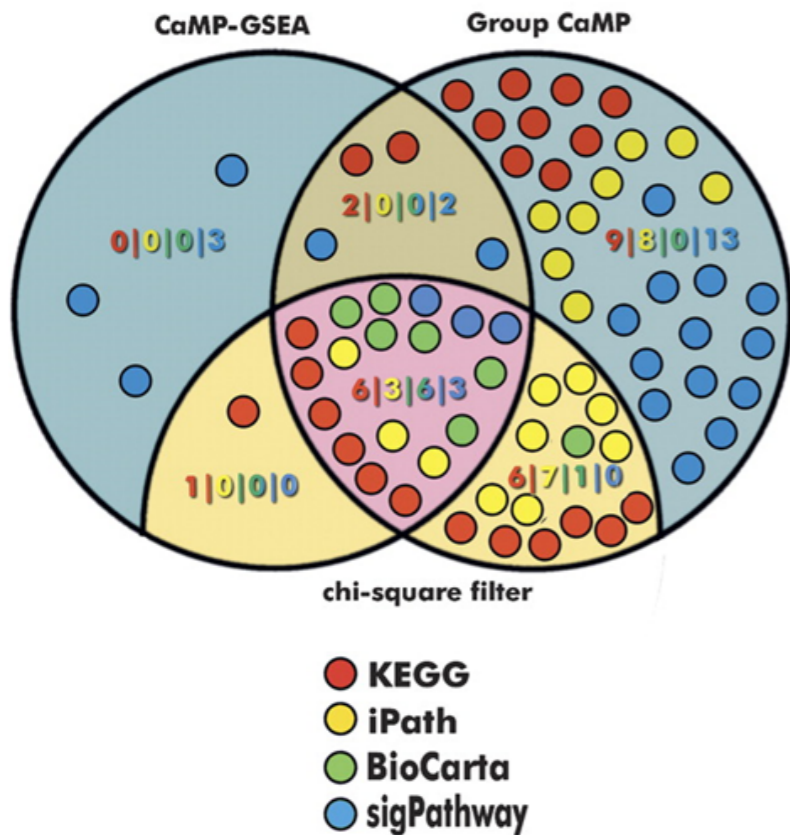
Comparison of mutation enrichment in cellular pathways using complementary statistical approaches. Lin, J., et al., A multidimensional analysis of genes mutated in breast and colorectal cancers. Genome Res, 2007. 17(9): p. 1304-18.

# REFACTORIZING COMPLEXITY

## Breast Cancer

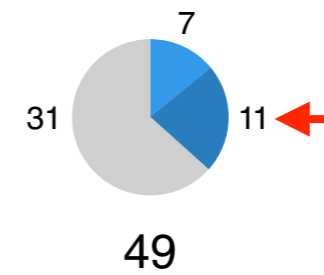


## Colorectal Cancer

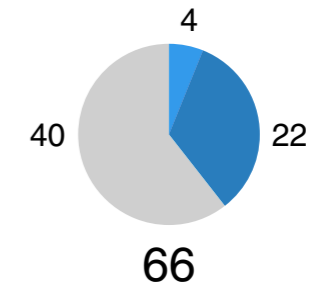


A

BREAST

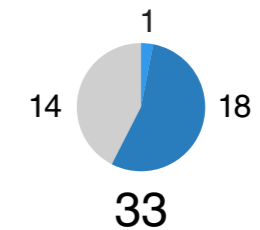
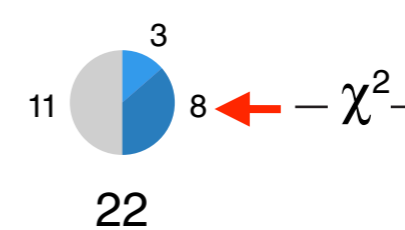


COLORECTAL

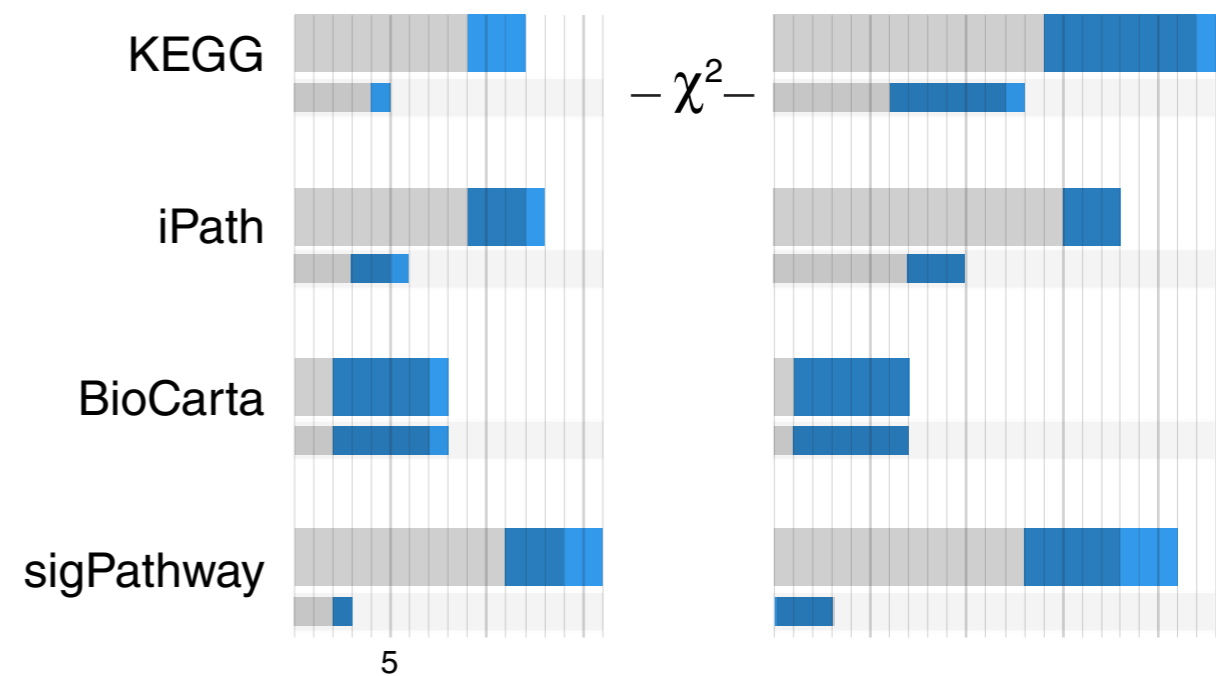


ALL PATHWAYS

CaMP-GSEA      CaMP



B

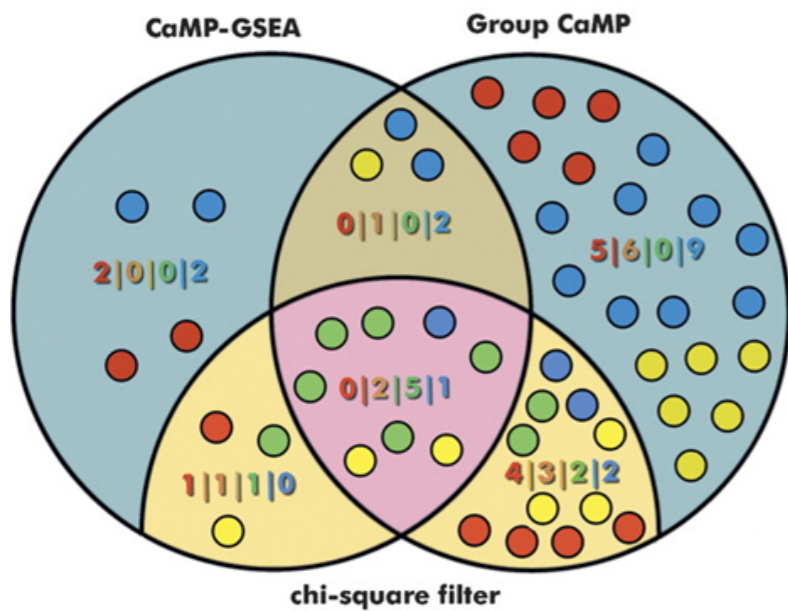


Comparison of mutation enrichment in cellular pathways using complementary statistical approaches. Lin, J., et al., A multidimensional analysis of genes mutated in breast and colorectal cancers. Genome Res, 2007. 17(9): p. 1304-18.

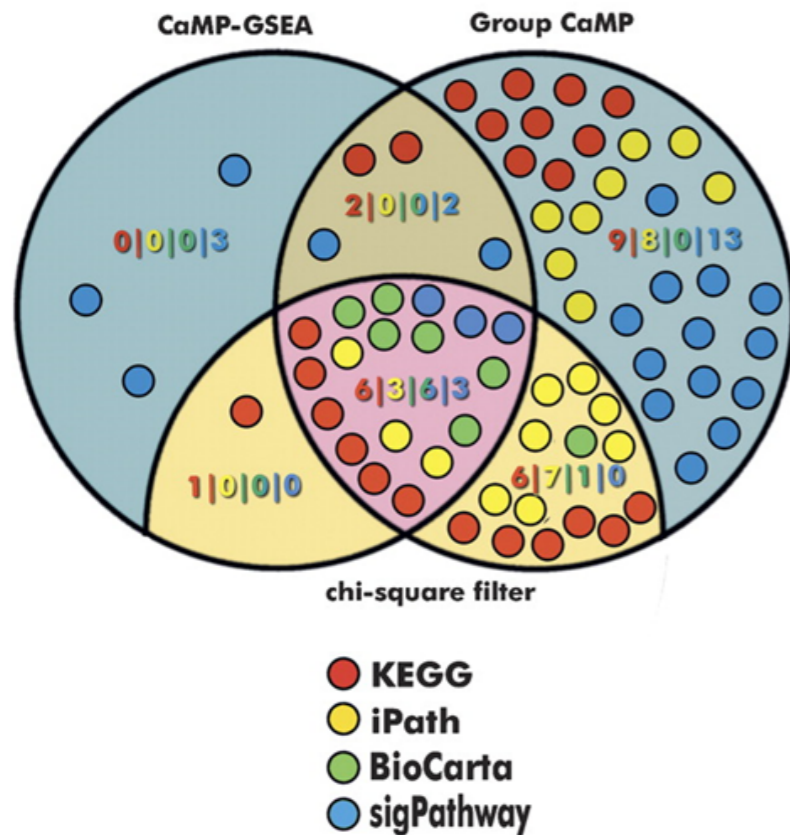


# REFACTORIZING COMPLEXITY

## Breast Cancer

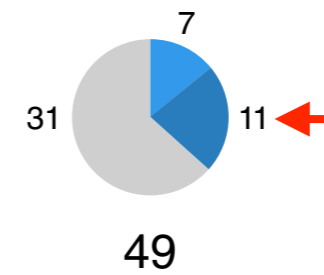


## Colorectal Cancer

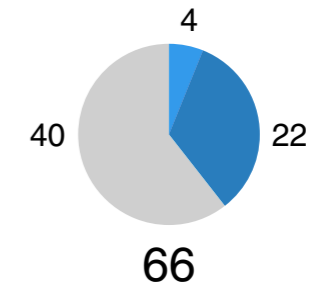


A

BREAST

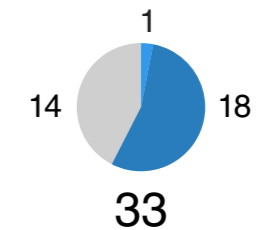
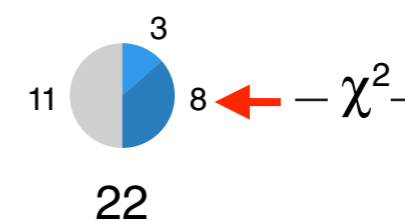


COLORECTAL

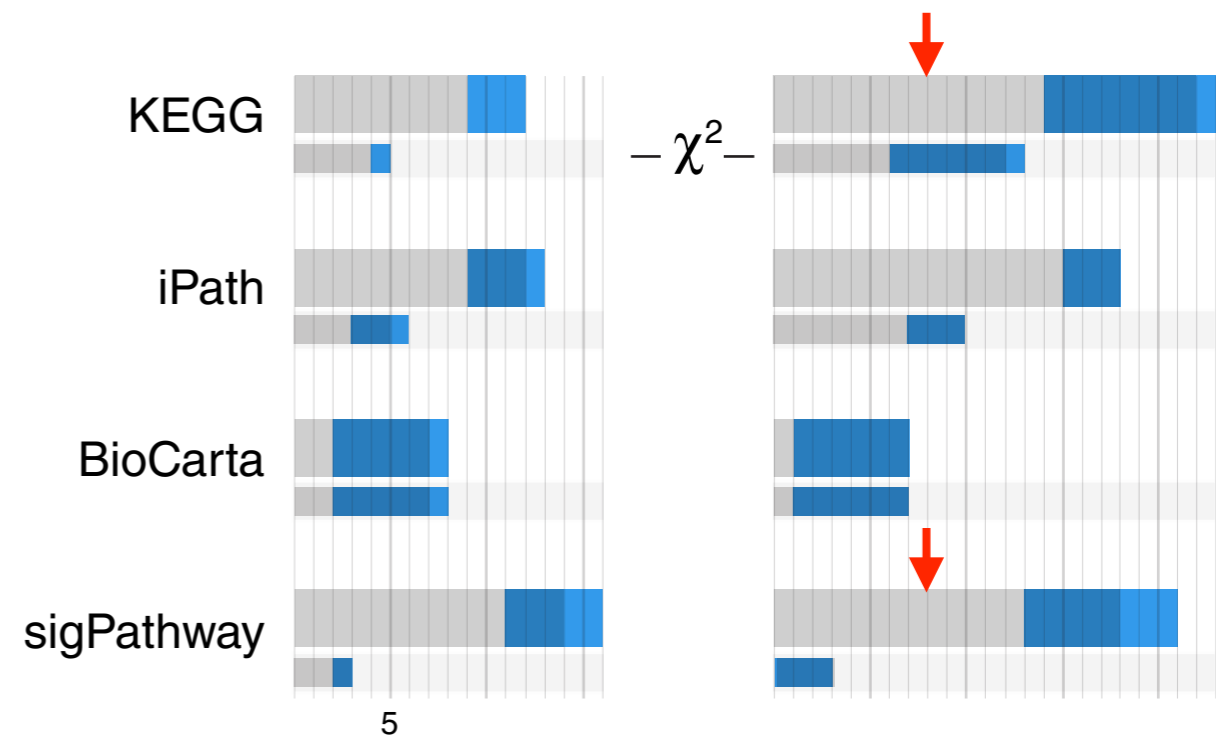


ALL PATHWAYS

CaMP-GSEA      CaMP

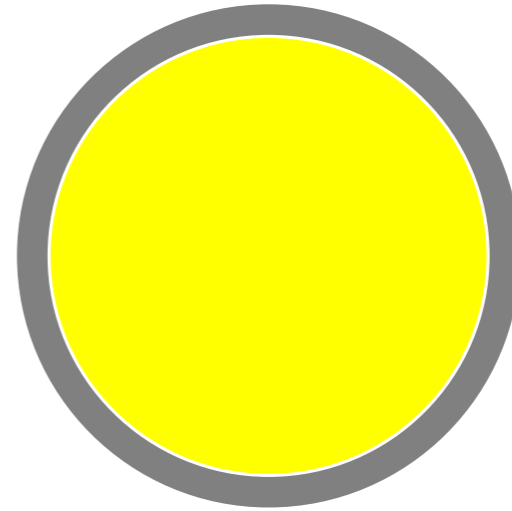
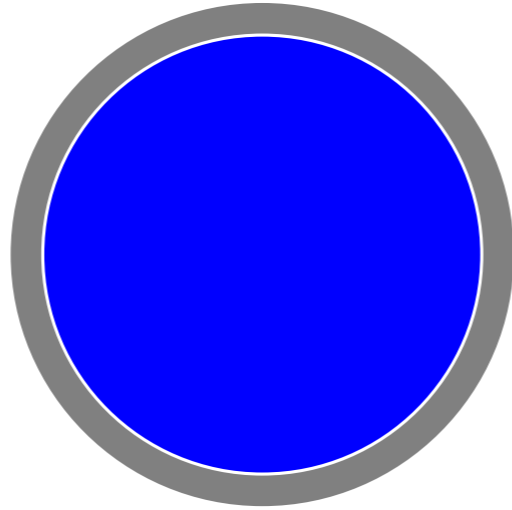


B



Comparison of mutation enrichment in cellular pathways using complementary statistical approaches. Lin, J., et al., A multidimensional analysis of genes mutated in breast and colorectal cancers. Genome Res, 2007. 17(9): p. 1304-18.

# COLOR PERCEPTION



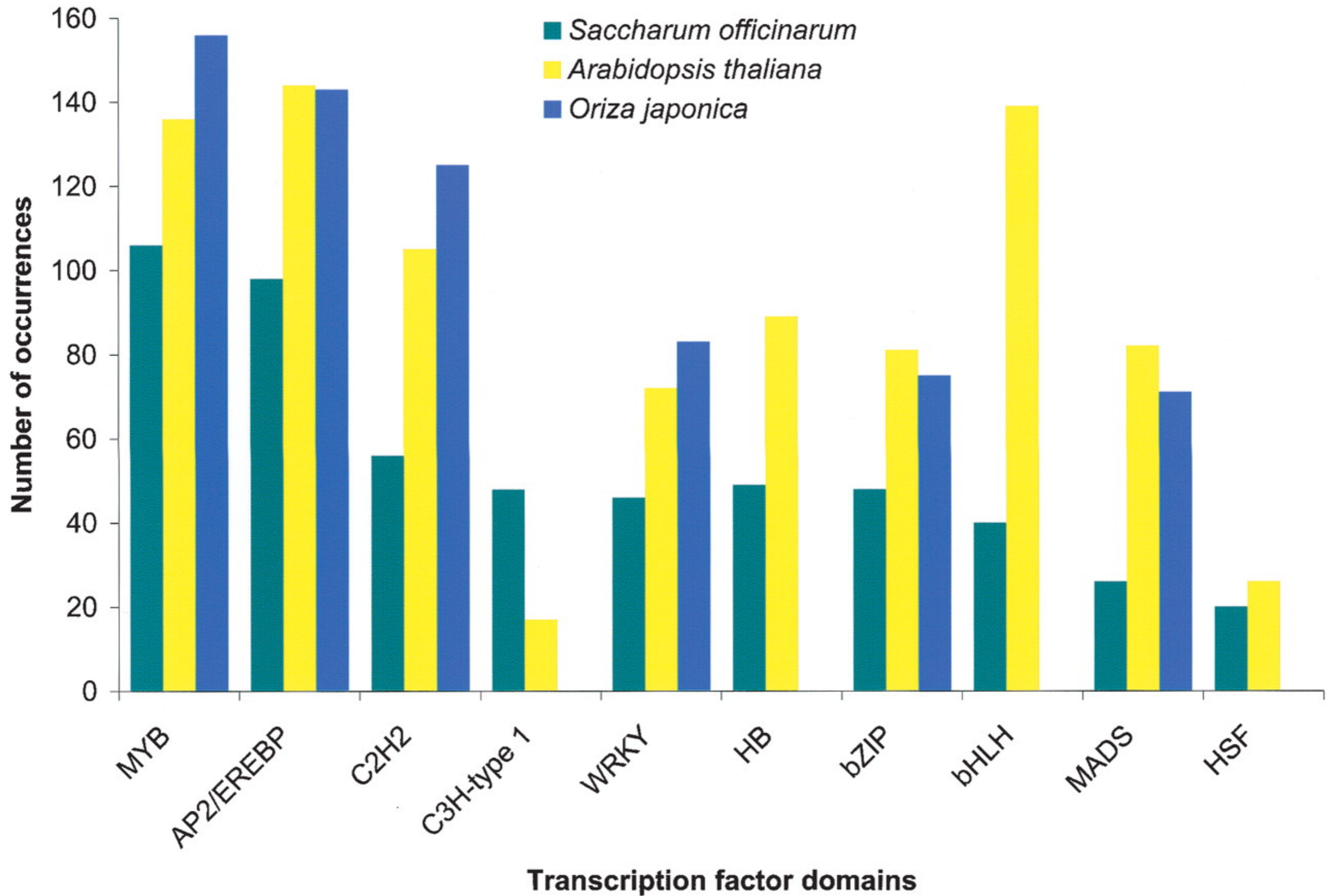
## HSB COLOR SPACE

HUE	240	60
SATURATION	1	1
BRIGHTNESS	1	1

## LCH COLOR SPACE

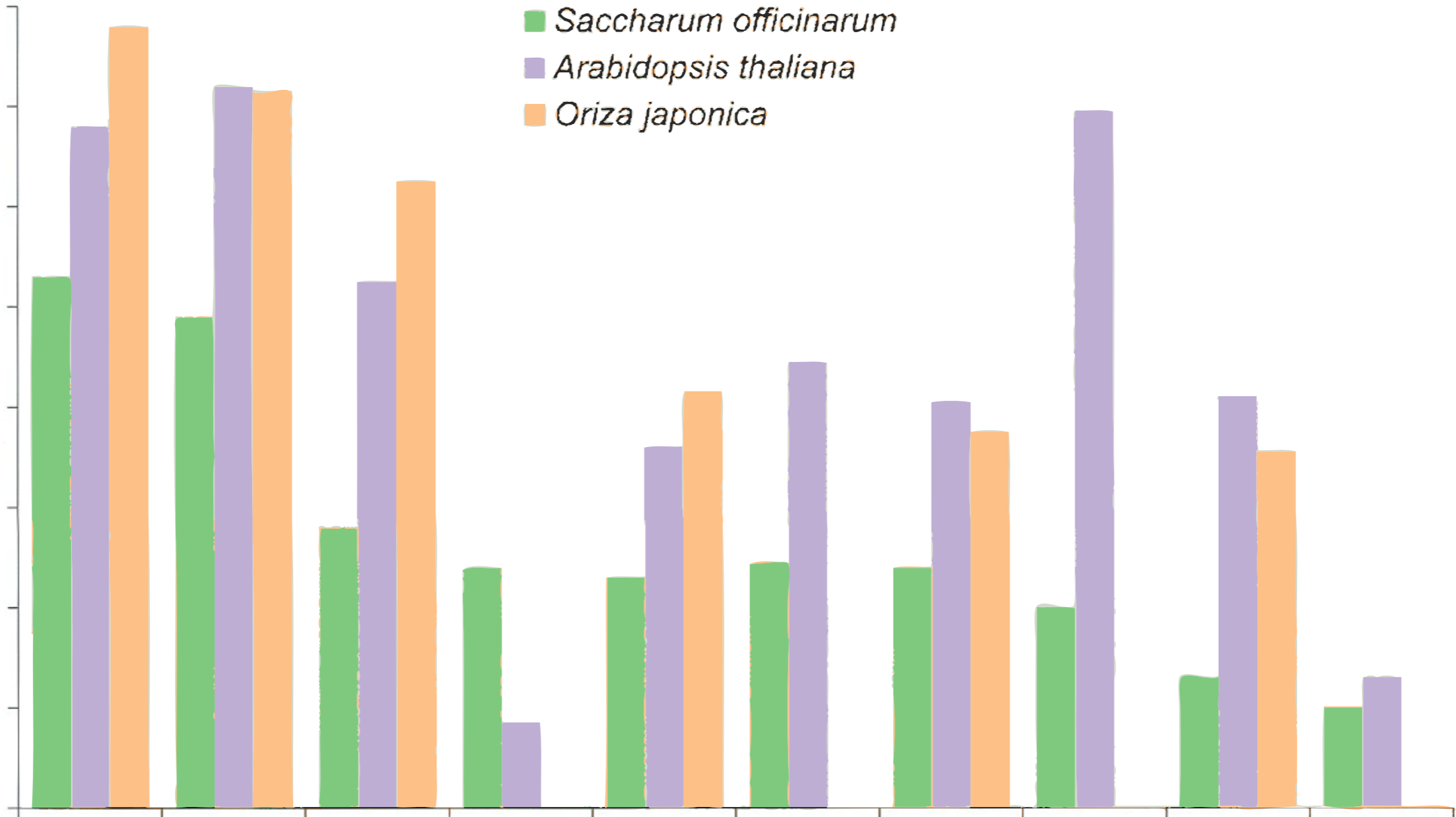
HUE	266	86
CHROMA	130	107
LIGHTNESS	0.32	0.97

# COLOR PERCEPTION



The 10 most common transcription factor Pfam domains in SAS proteins. Vettore, A.L., et al., Analysis and functional annotation of an expressed sequence tag collection for tropical crop sugarcane. *Genome Res*, 2003. 13(12): p. 2725-35.

# COLOR PERCEPTION

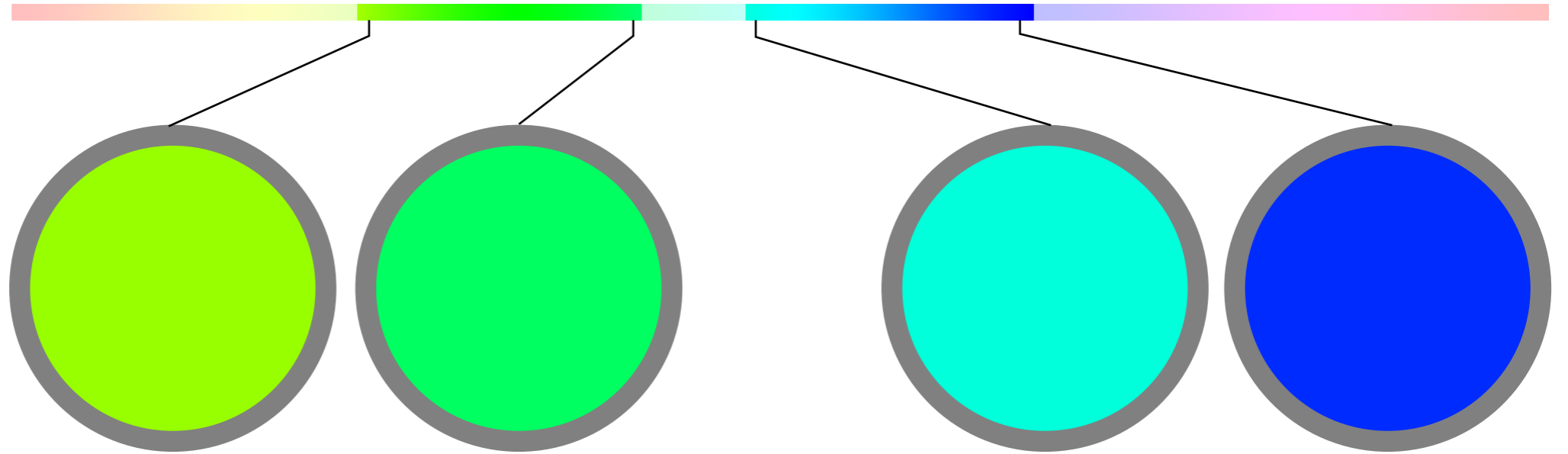


## BREWER QUALITATIVE 3-COLOR PALETTES



In a qualitative palette, colors have no perceived order or importance.

# PERCEPTUAL UNIFORMITY



## HSB COLOR SPACE

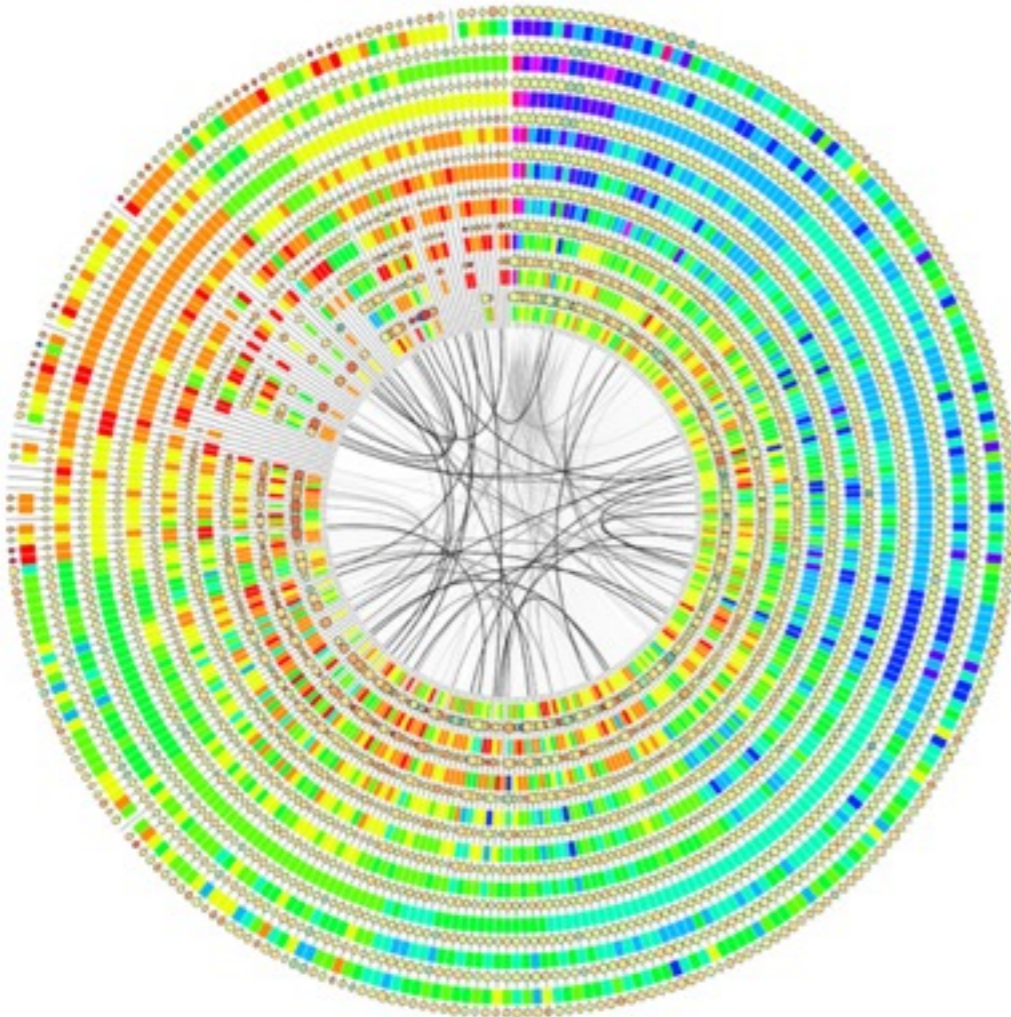
		$\Delta H = 60$		$\Delta H = 60$	
HUE	83		143	171	231
SATURATION	1		1	1	1
BRIGHTNESS	1		1	1	1

## Lab COLOR SPACE

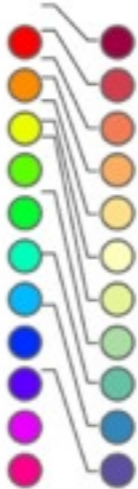
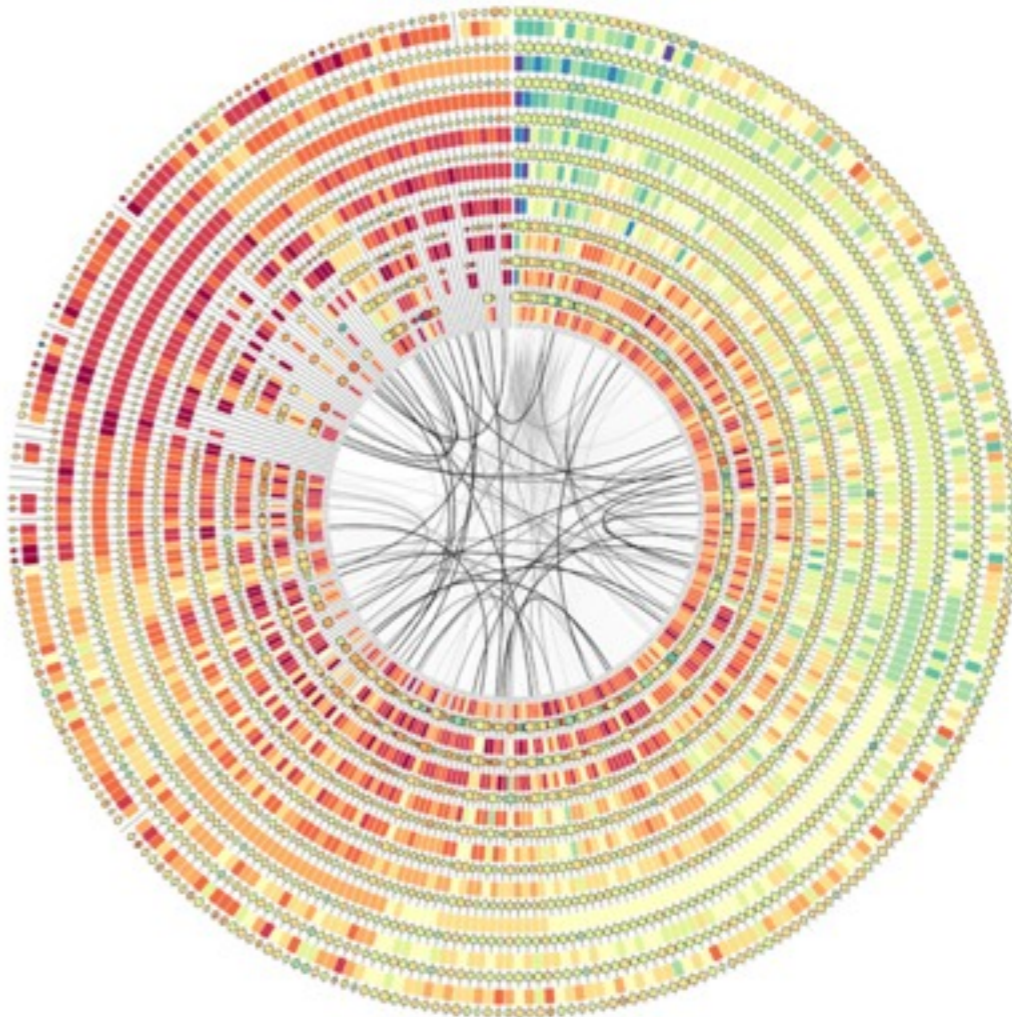
		$\Delta E_{ab} = 35$		$\Delta E_{ab} = 176$	
L	91		88	90	35
a	-59		-81	-58	70
b	87		60	4	-102

# DIVERGING BREWER PALETTES

UNIFORM HSB 11 COLOR PALETTE



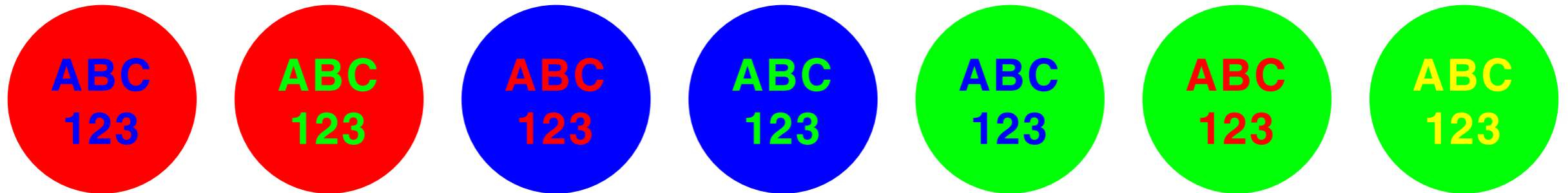
BREWER 11 COLOR SPECTRAL PALETTE



last.fm music listening habits by age.  
<http://mkweb.bcgsc.ca/spl/circos-lastfm-legend.png>

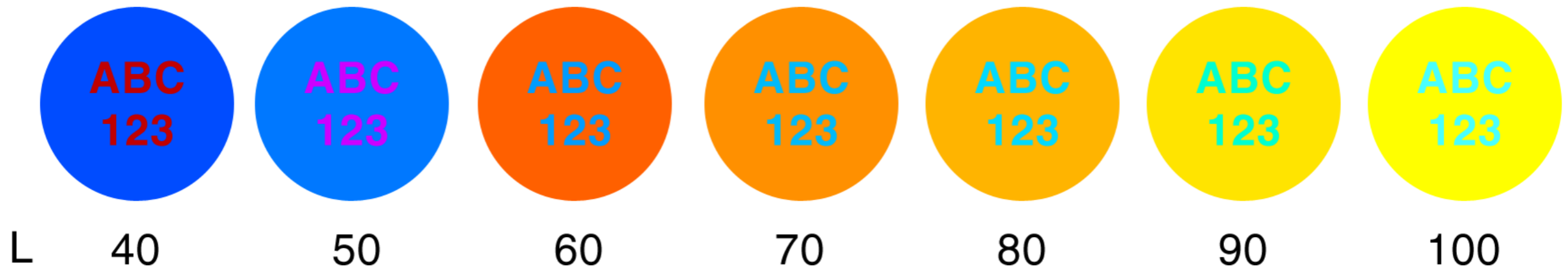
# CONTRAST

## AVOID ADJACENT PURE COLORS



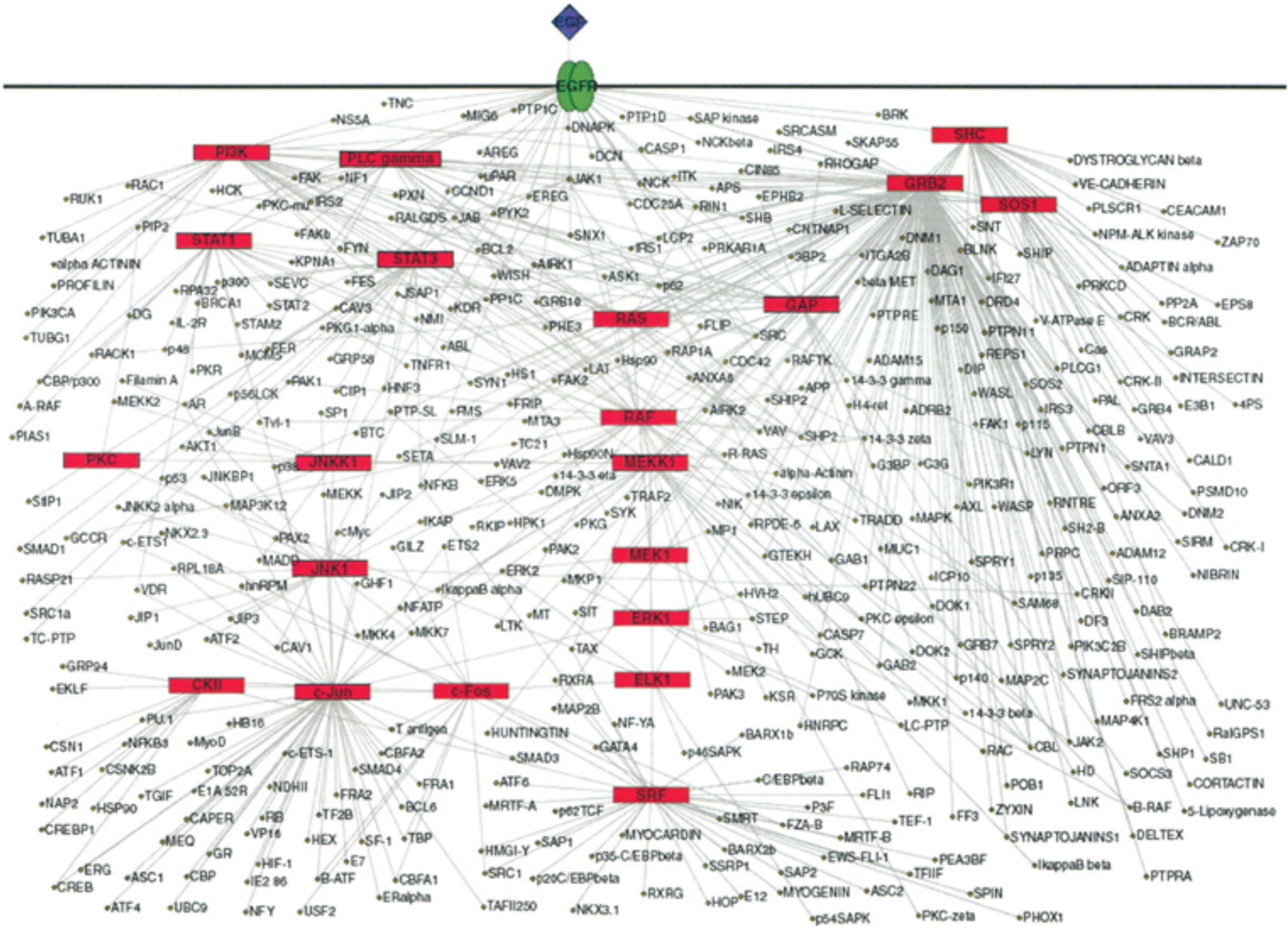
## AVOID ADJACENT COLORS WITH SIMILAR LUMINANCE

$$\Delta L = 0$$



Simultaneous contrast occurs when two pure colors are adjacent.  
Poor contrast occurs when two colors have similar luminance (perceived brightness).

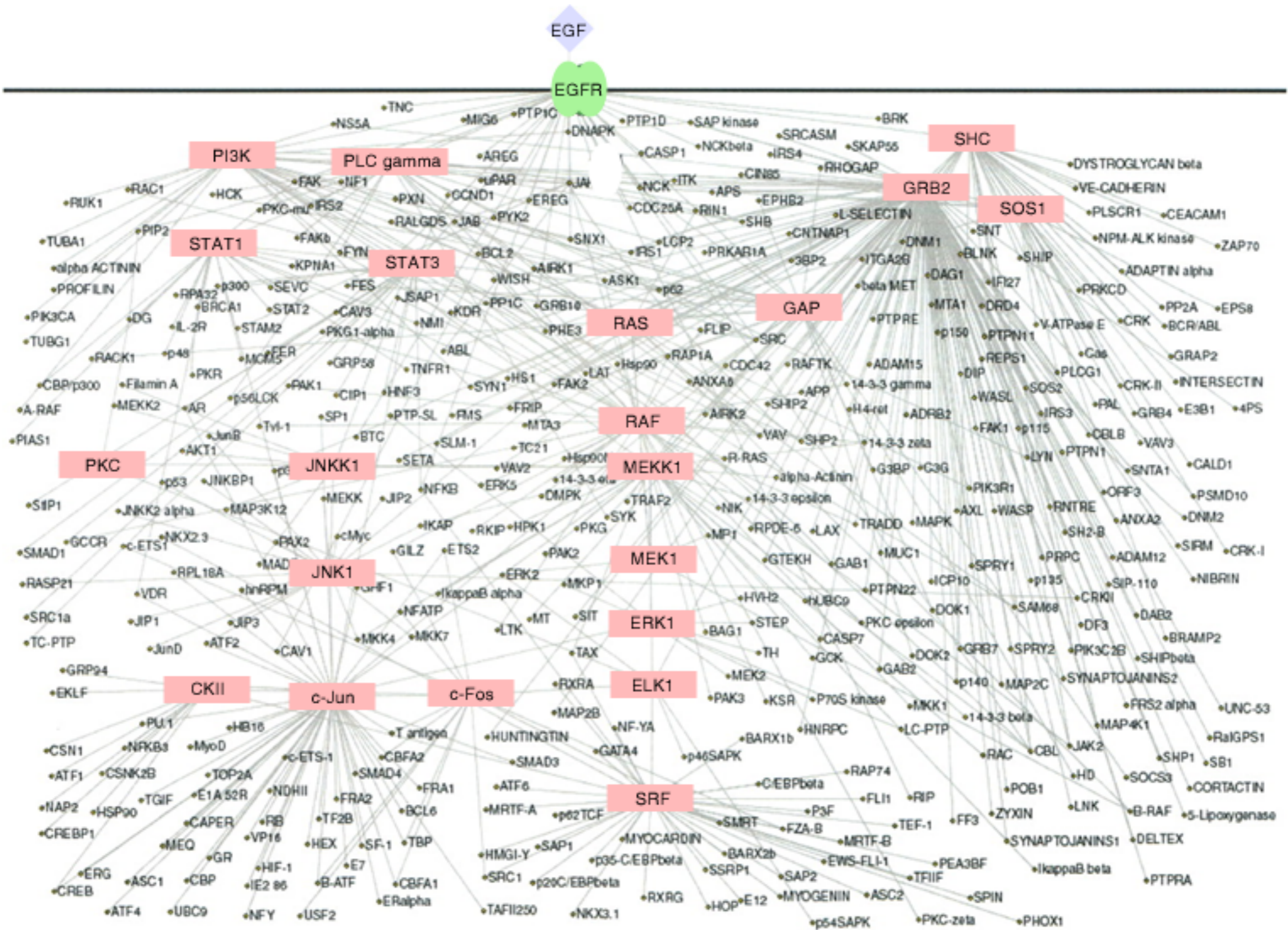
# CONTRAST



Black text on dark colors is illegible due to insufficient luminosity contrast.

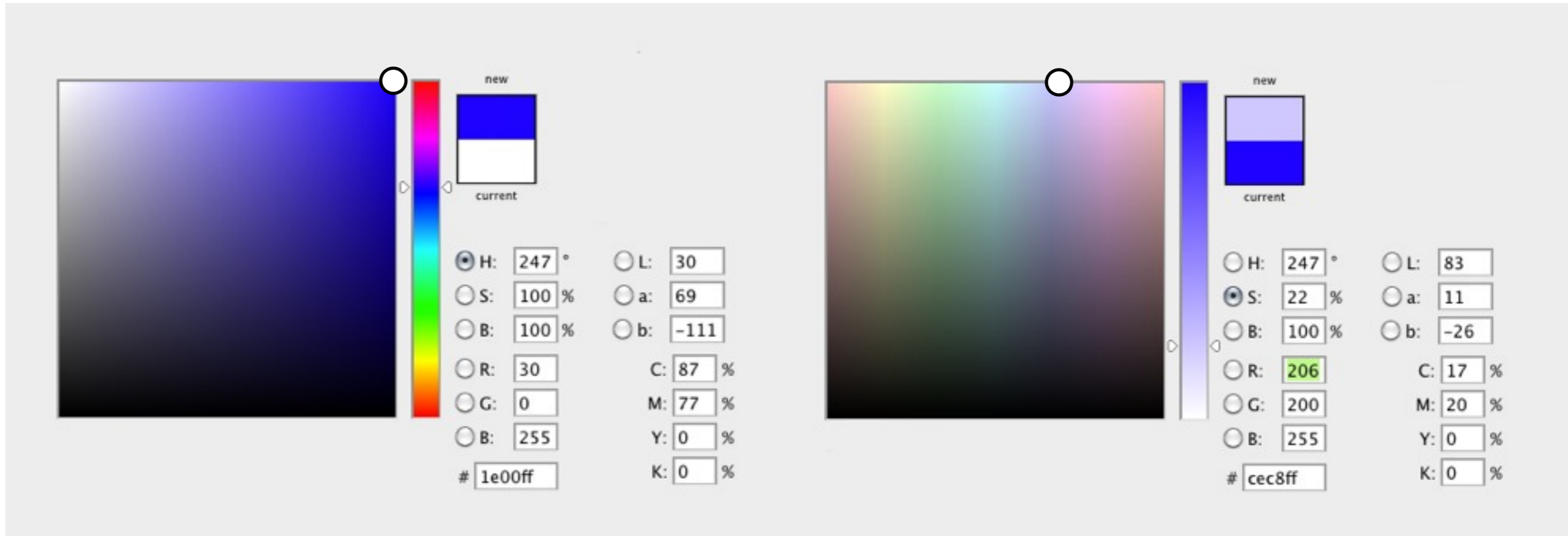


# GOOD CONTRAST



Decreasing the saturation of the background colors improves contrast.

# USING COLOR PICKERS



Photoshop shows a color's coordinates in several color spaces. Decrease saturation to make a color appear lighter. Use the "L" channel of the Lab space to determine a color's perceived brightness (luminance).

legibility **clarity** attractiveness

**STRONG MESSAGE**

**REDUNDANCY**

**EXCESS INK**

**GLYPHS**

**OUTLIER HIJACK**

# IMPORTANCE OF CLARITY



This is fairly clear. Either

- (a) do not poop near the tiny trees, or
- (b) no pooping for giants



This is less clear. Perhaps

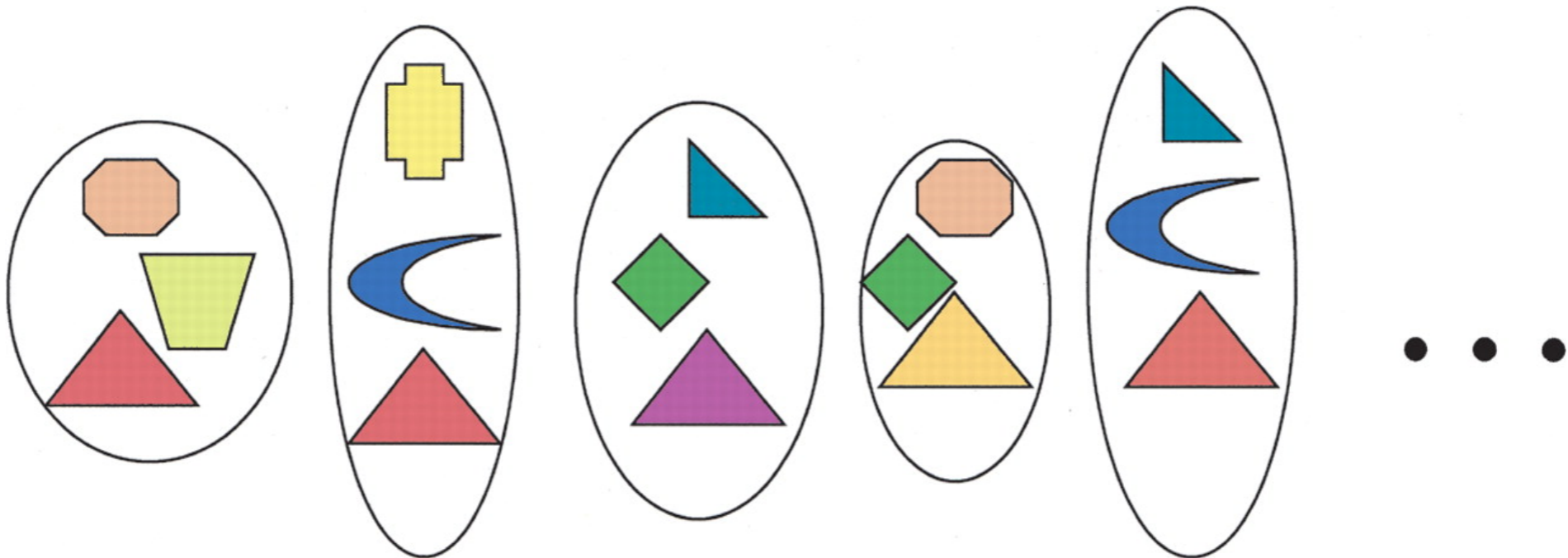
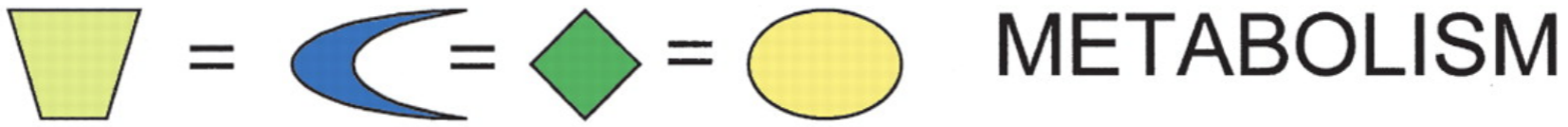
- (a) no talking
- (b) did you remember your black glove?
- (c) stop and wait

# IS YOUR MESSAGE NECESSARY?



<http://lushlush.livejournal.com/190093.html>

# ACADEMIC FIGURE OR MENSA TEST?



A mix-and-match model for prokaryotic genome evolution. Charlebois, R.L. and W.F. Doolittle, Computing prokaryotic gene ubiquity: rescuing the core from extinction. Genome Res, 2004. 14(12): p. 2469-77.

# CRAFTING A MESSAGE

WHAT IS SHOWN?

RAW DATA

12 54 82 29 25 22 67 61 23 79

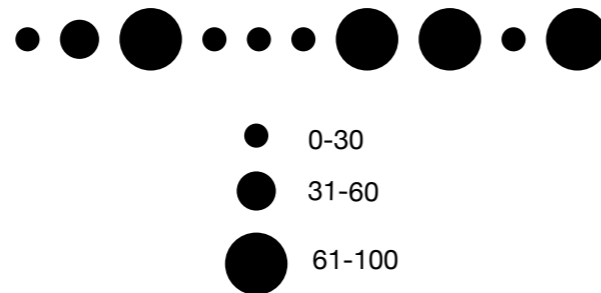
WHAT IS COMMUNICATED?

NO CLEAR MESSAGE.

WHAT IS INTERPRETED?

UNKNOWN. READER IS ON THEIR OWN.

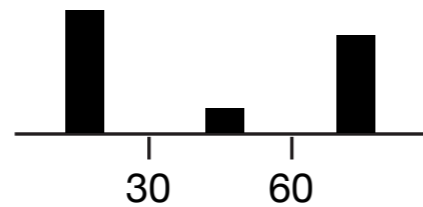
DISCRETIZED



SCALE

THREE RANGES ARE IMPORTANT. INDIVIDUAL VALUES WITHIN A RANGE ARE NOT.

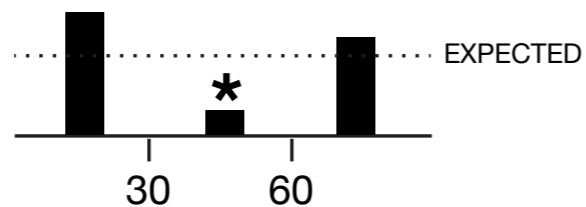
BINNED



DISTRIBUTION

THERE ARE FEWER MEDIUM-SIZED VALUES.

TREND



SIGNIFICANCE

THERE ARE SIGNIFICANTLY FEWER MEDIUM-SIZED VALUES.

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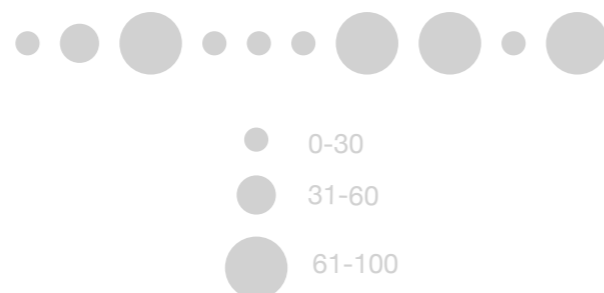
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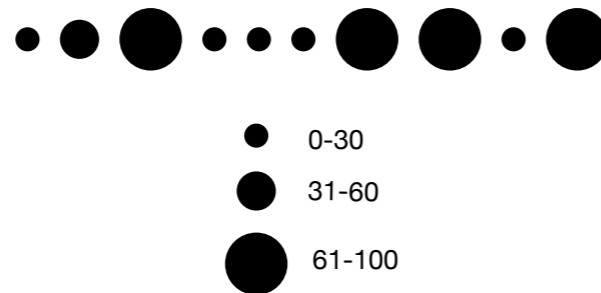
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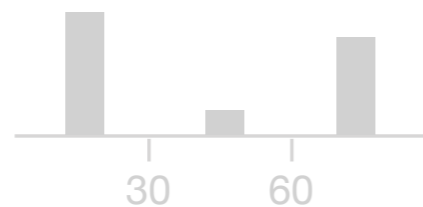
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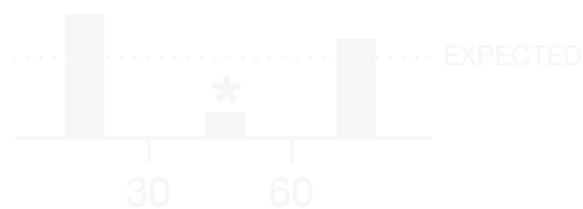
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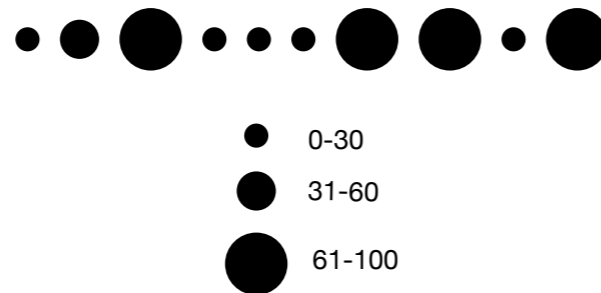
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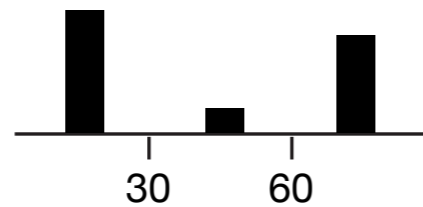
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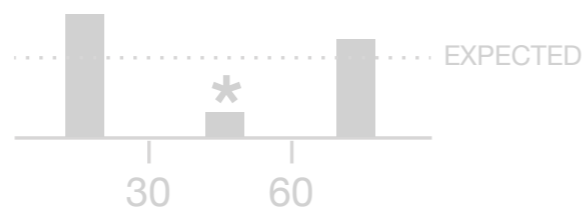
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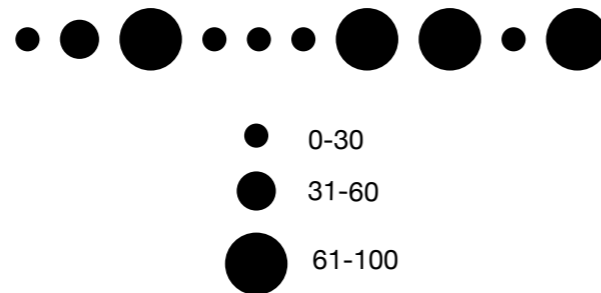
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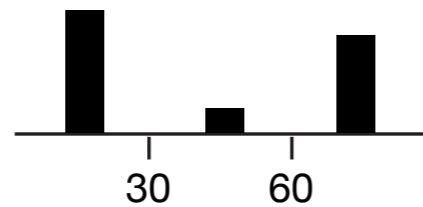
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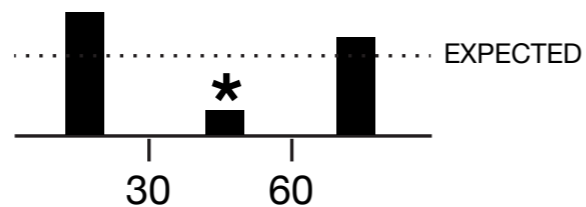
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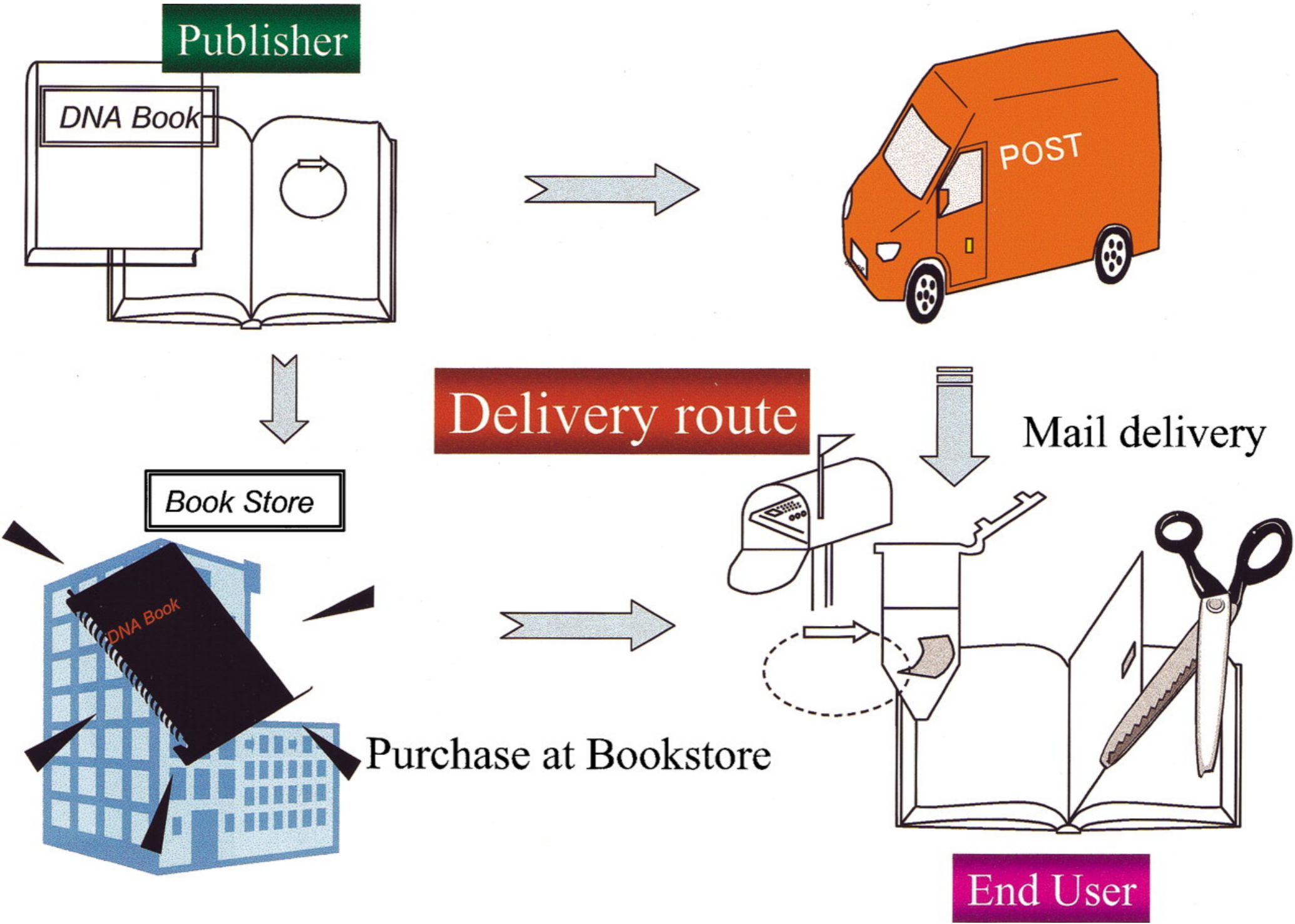
TREND



SIGNIFICANCE

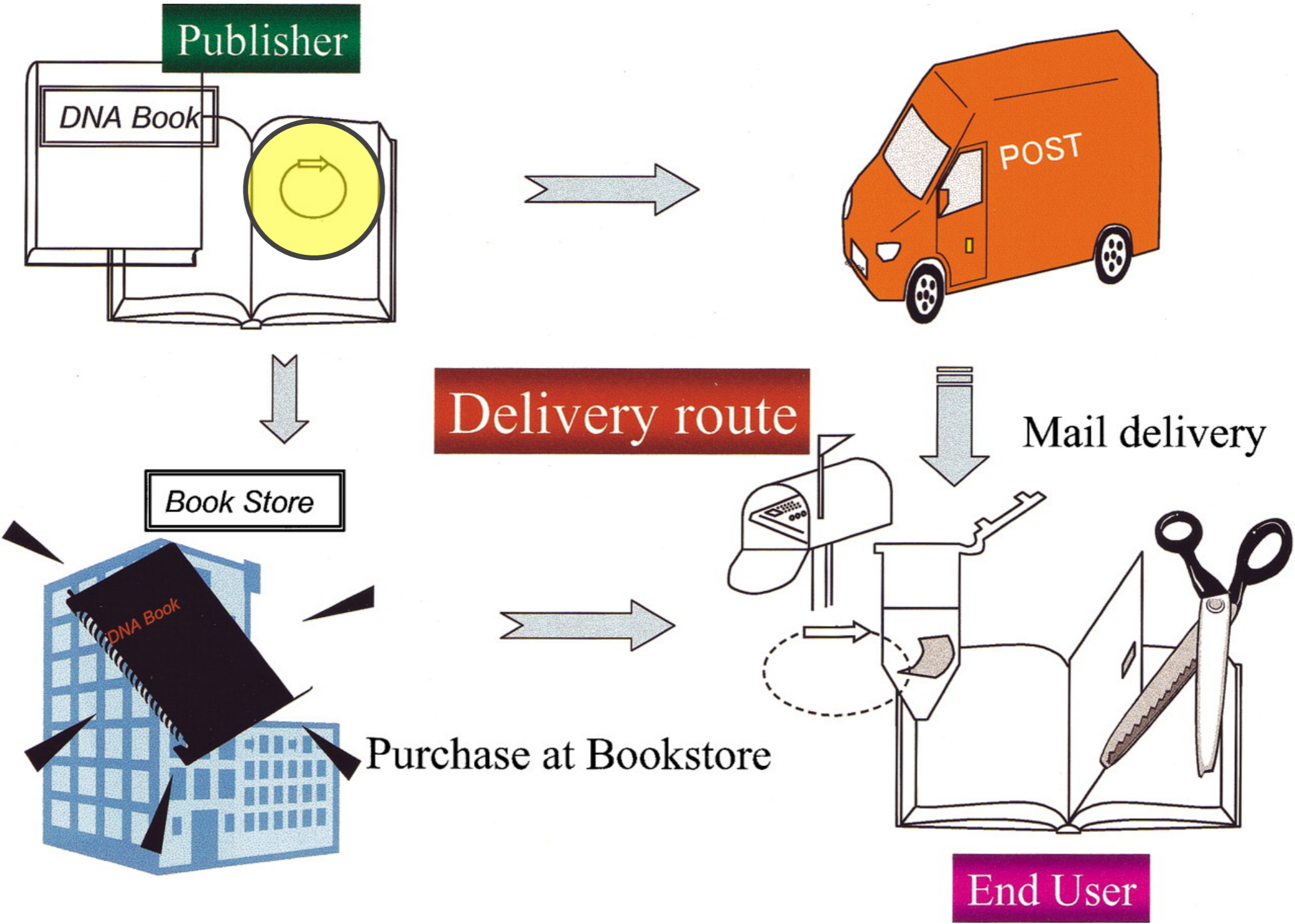
THERE ARE SIGNIFICANTLY FEWER MEDIUM-SIZED VALUES.

# WHAT IS THE CORE MESSAGE?



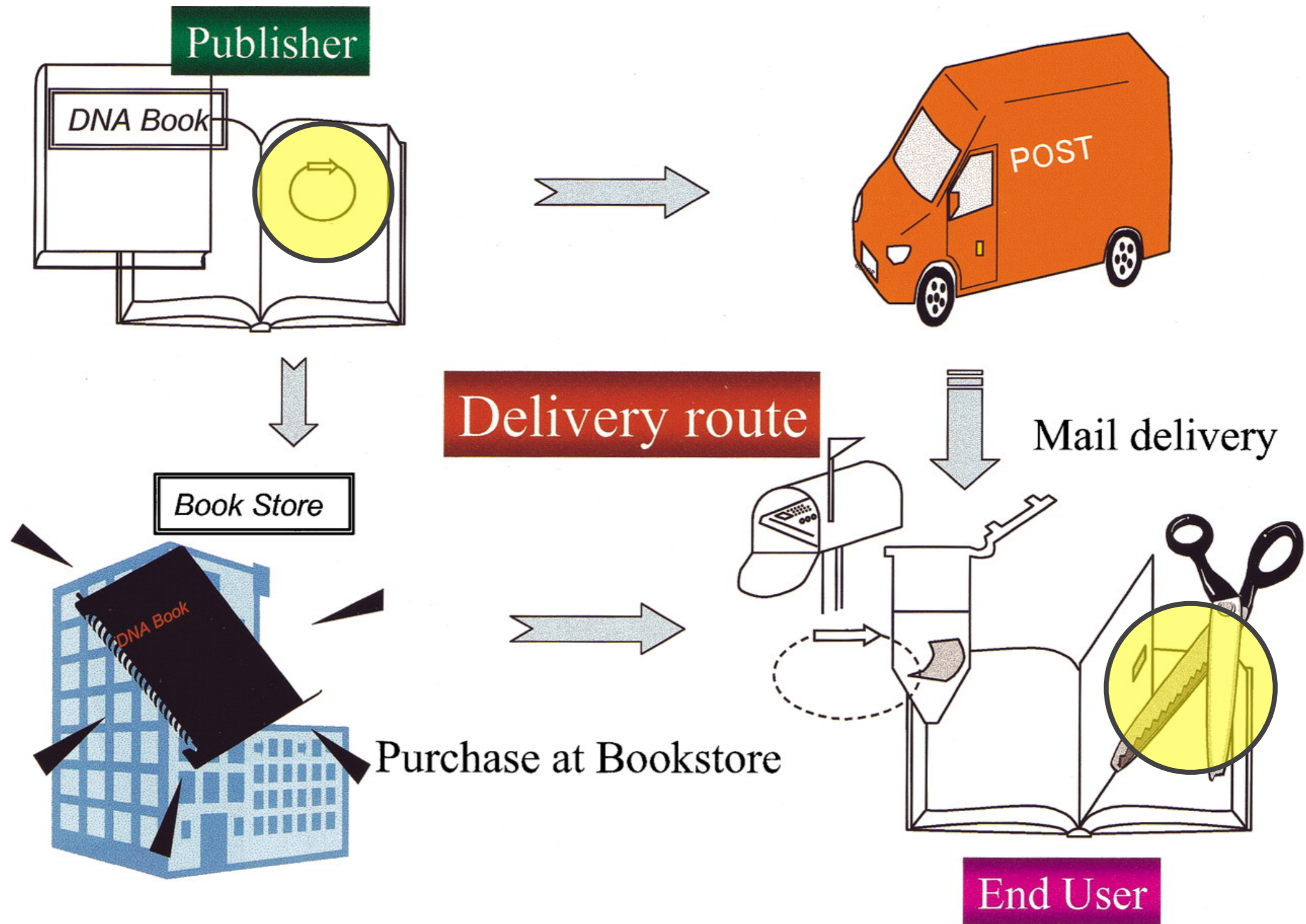
Concept of the "DNA Book." Kawai, J. and Y. Hayashizaki, DNA book. Genome Res, 2003. 13(6B): p. 1488-95.

# WHAT IS THE CORE MESSAGE?



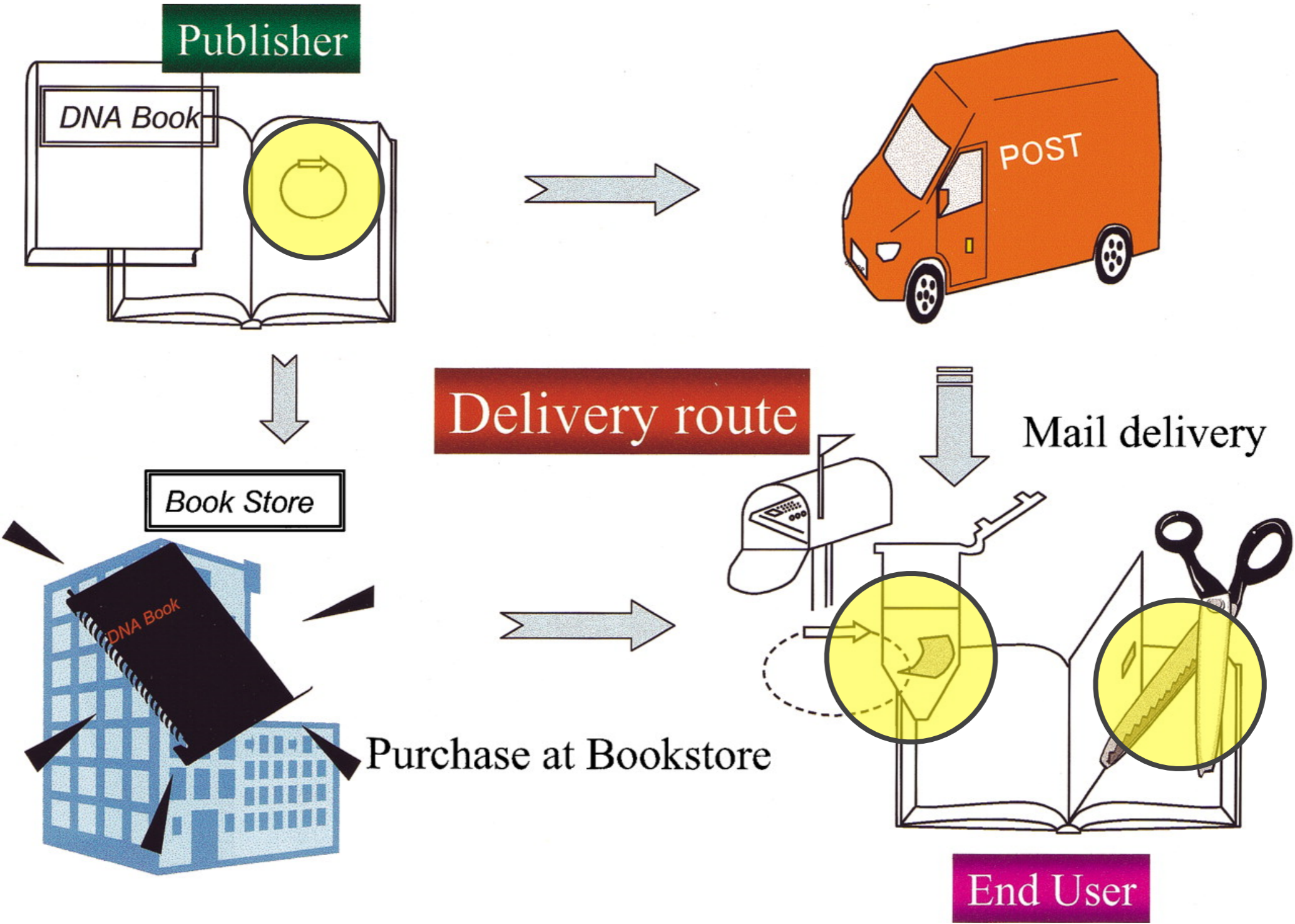
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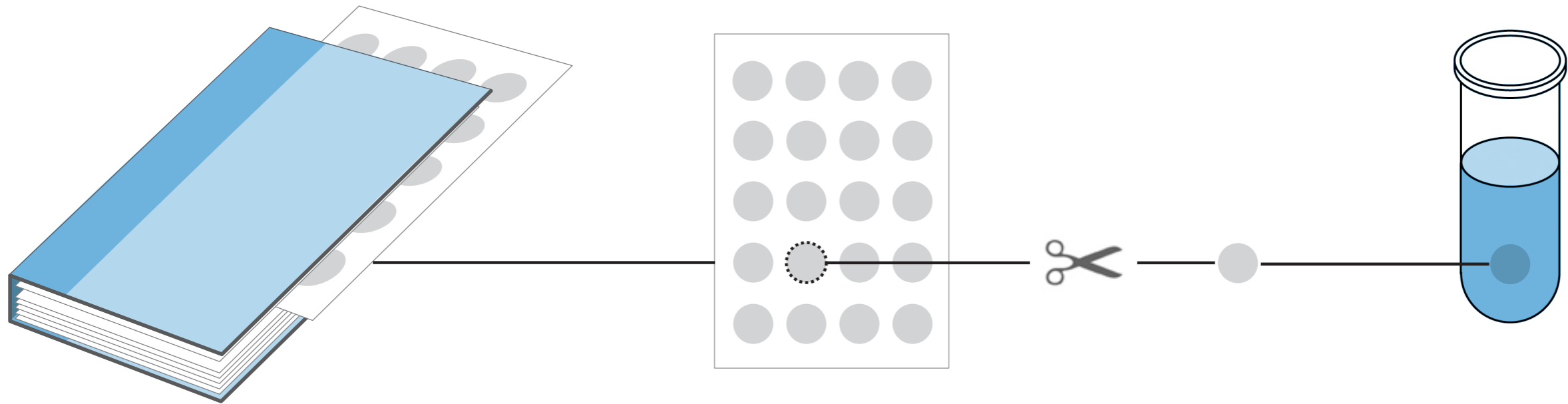
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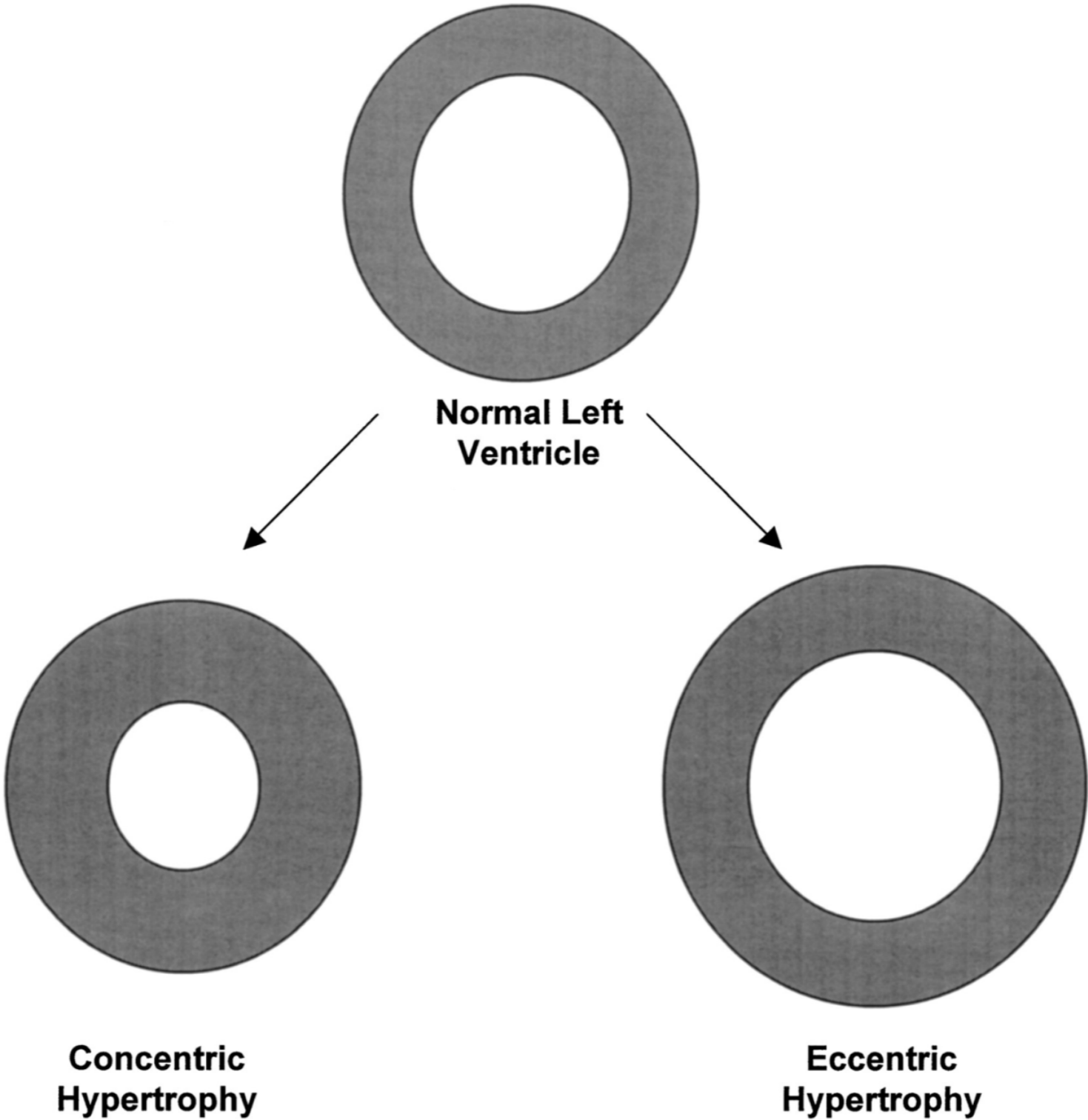
Concept of the "DNA Book." Kawai, J. and Y. Hayashizaki, DNA book. Genome Res, 2003. 13(6B): p. 1488-95.

# EXTRACTING THE CORE MESSAGE



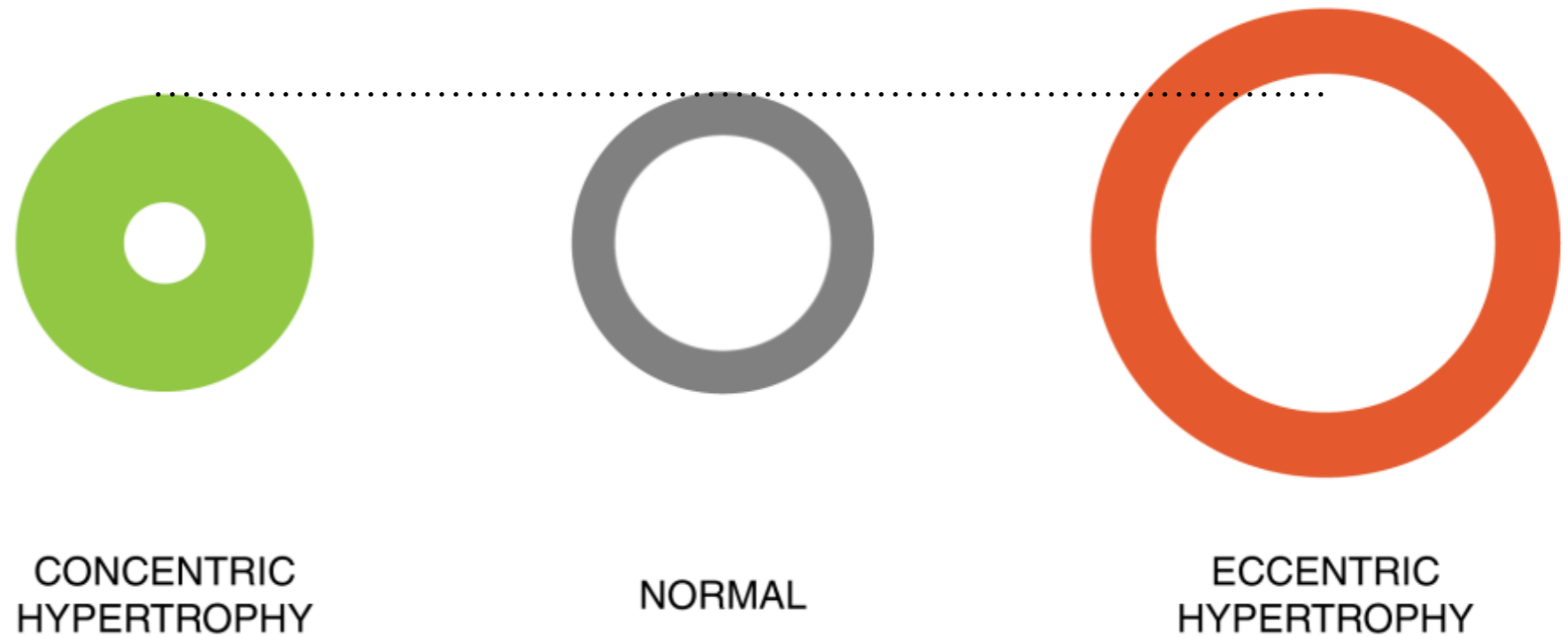
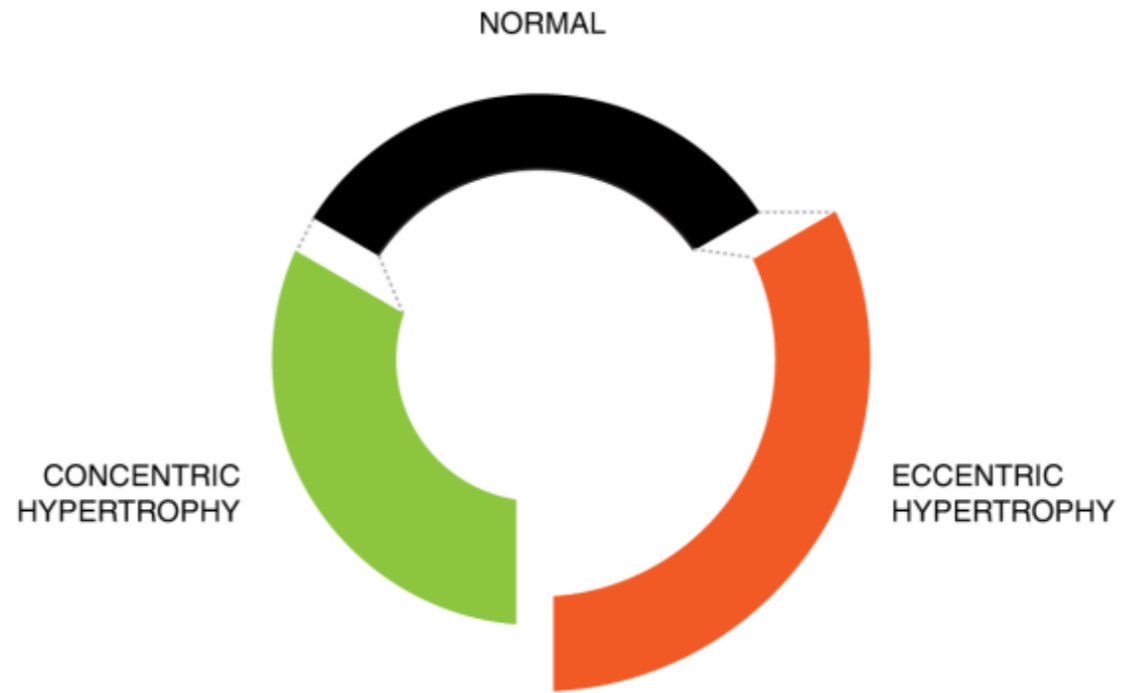
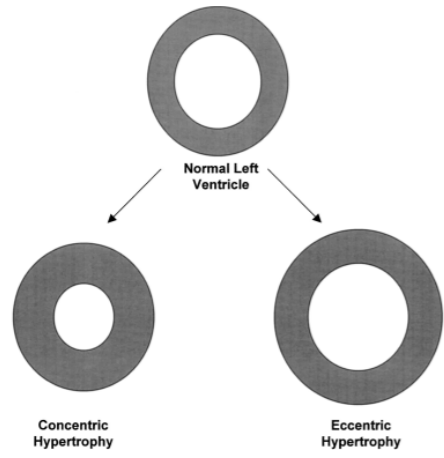


# AMBIGUOUS PROPORTIONS

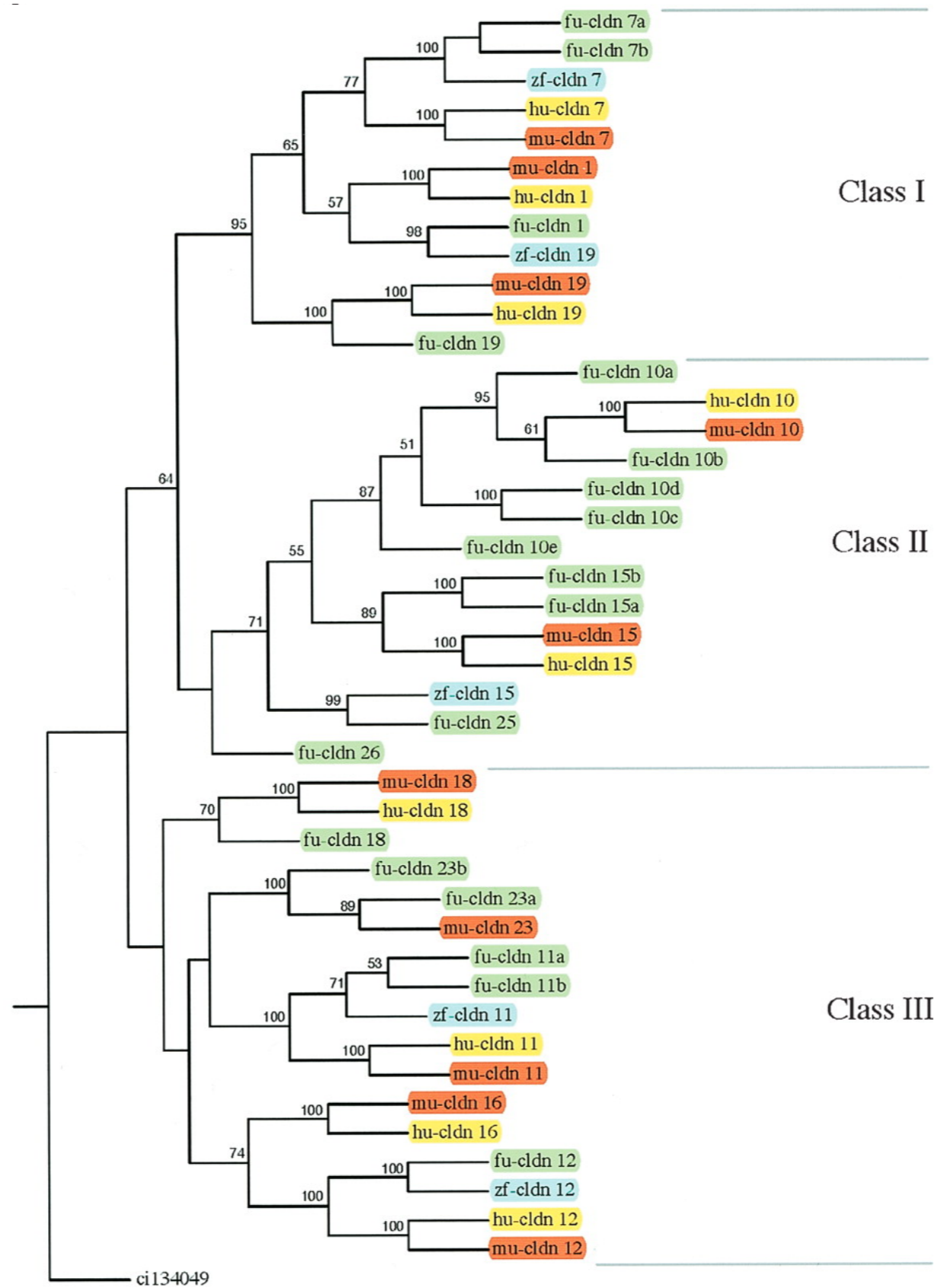


Nadeau, J.H., et al., Pleiotropy, homeostasis, and functional networks based on assays of cardiovascular traits in genetically randomized populations. *Genome Res*, 2003. 13(9): p. 2082-91.

# CLEAR PROPORTIONS



# REDUNDANCY



All labels are of the form

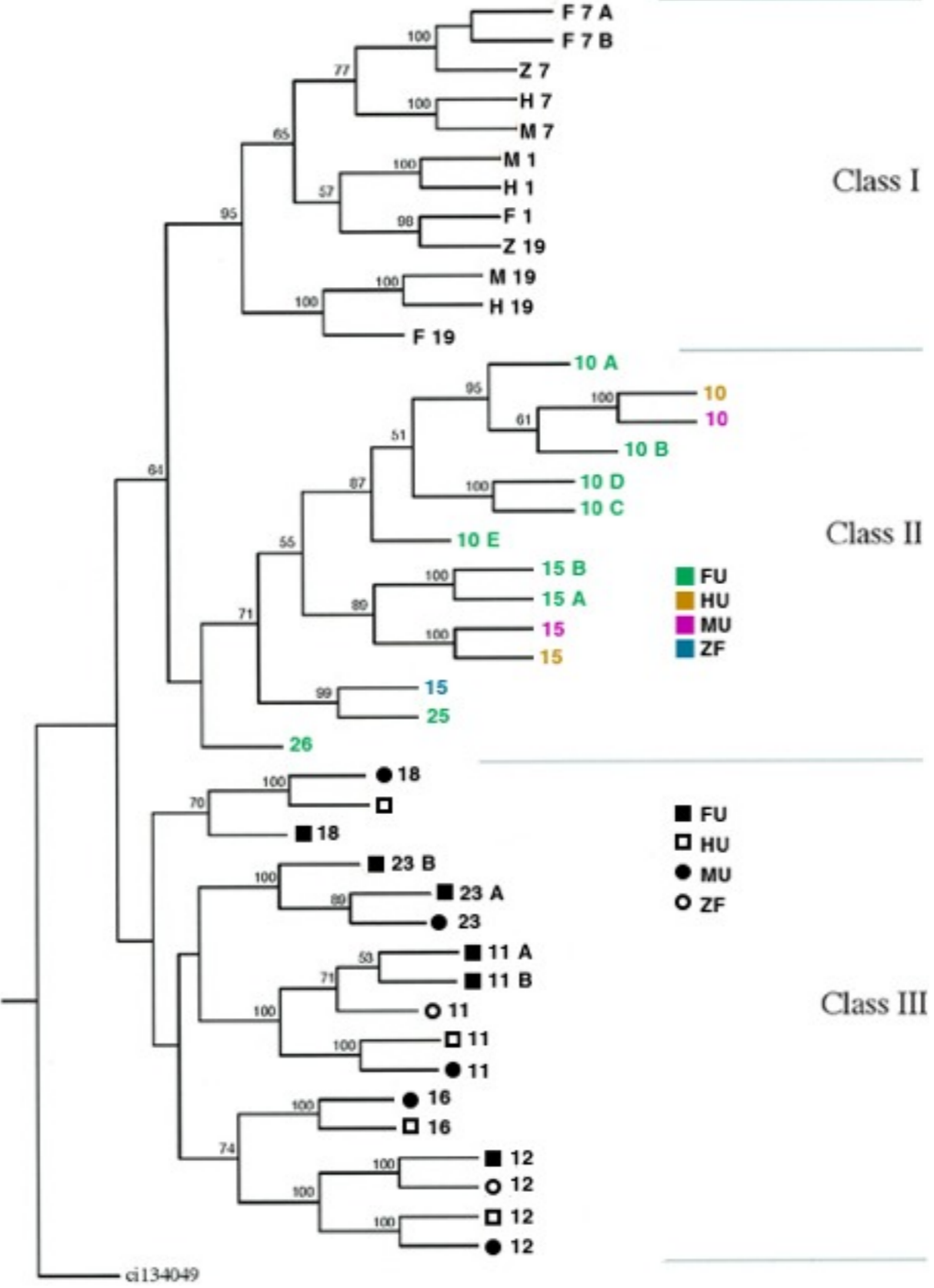
fu-cldn 7a  
 fu-cldn 7b  
 zf-cldn 7  
 mu-cldn 19  
 hu-cldn 15  
 ...

Instead, consider

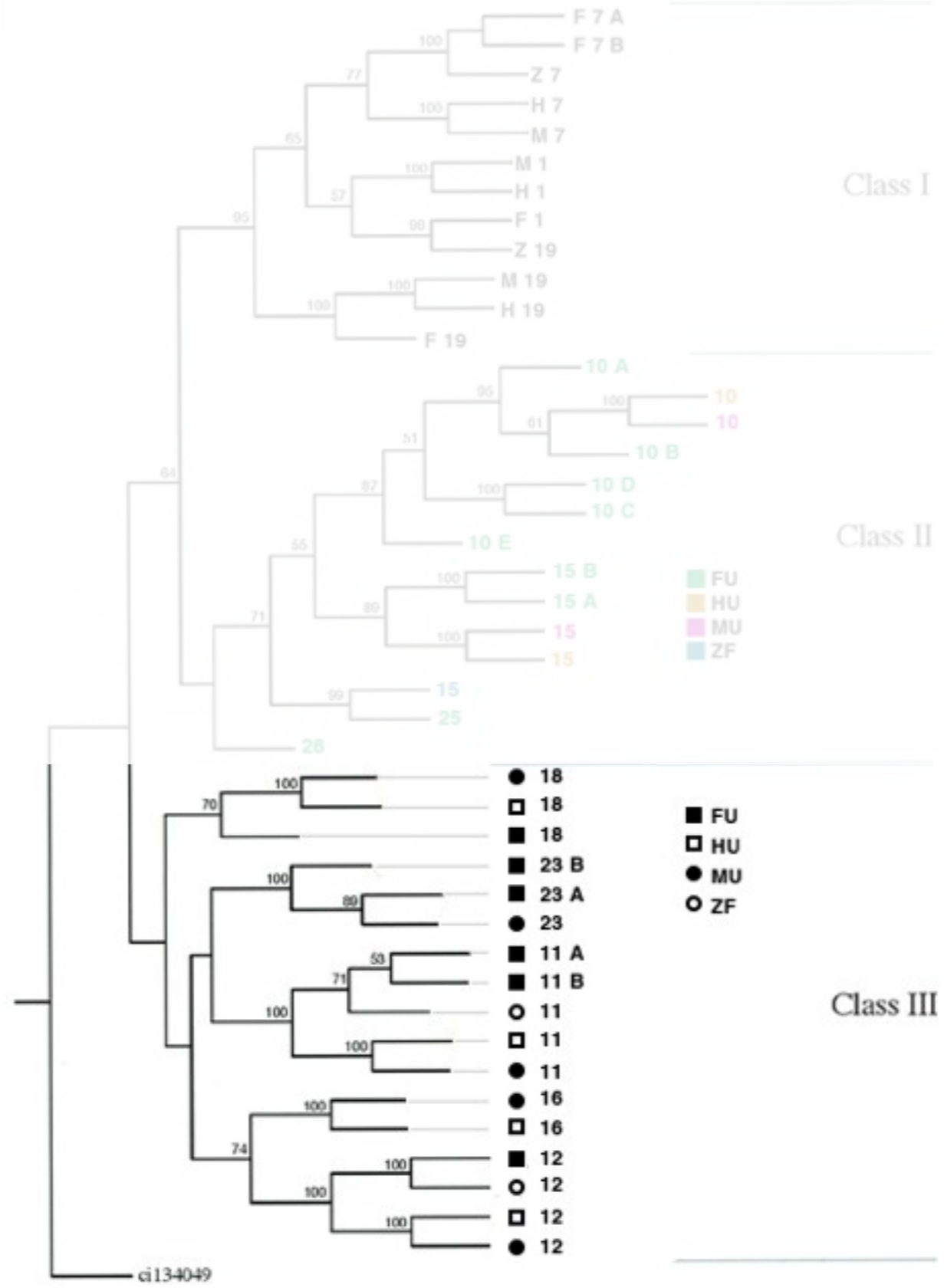
F7a  
 F7b  
 Z7  
 M19  
 H15  
 ...

Y. H. Loh, A. Christoffels, S. Brenner, W. Hunziker, B. Venkatesh, Genome Res 14, 1248 (Jul, 2004).

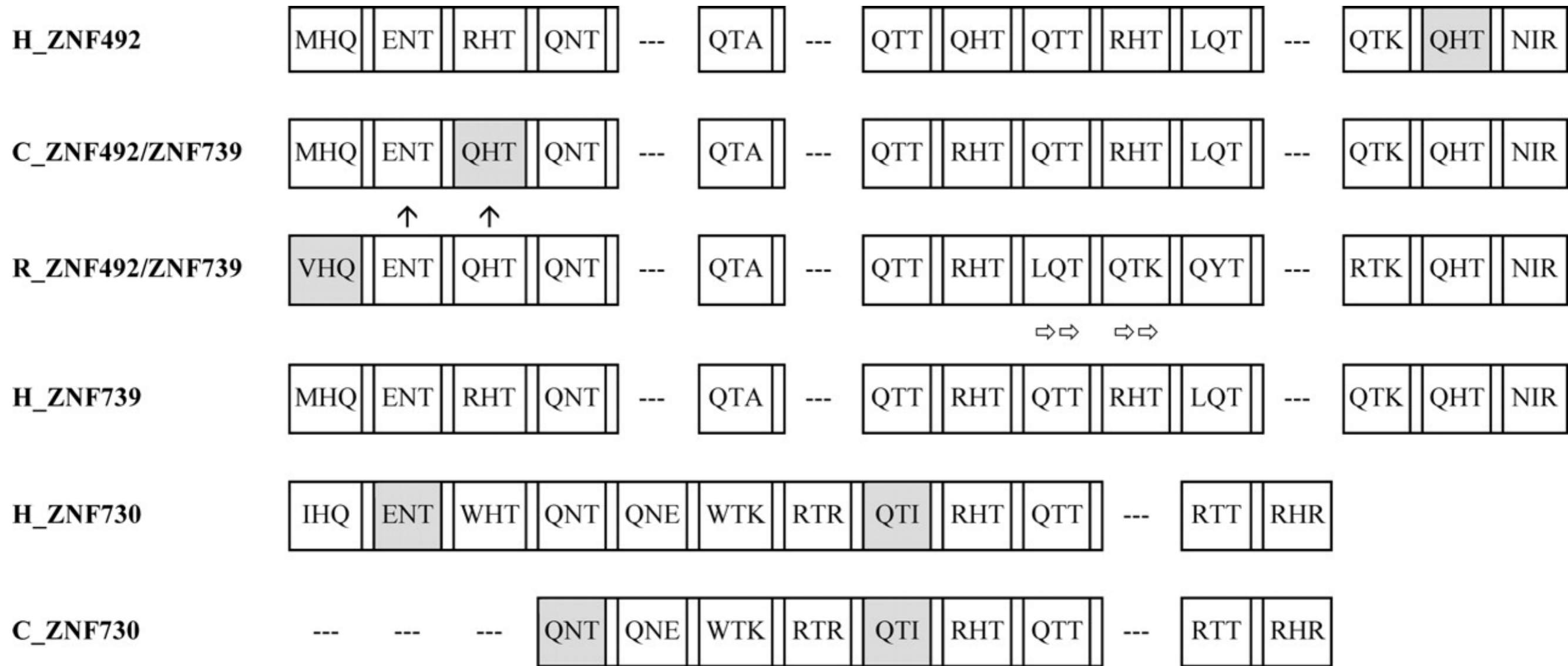
# REFACTORED REDUNDANCY



# REFACTORED REDUNDANCY

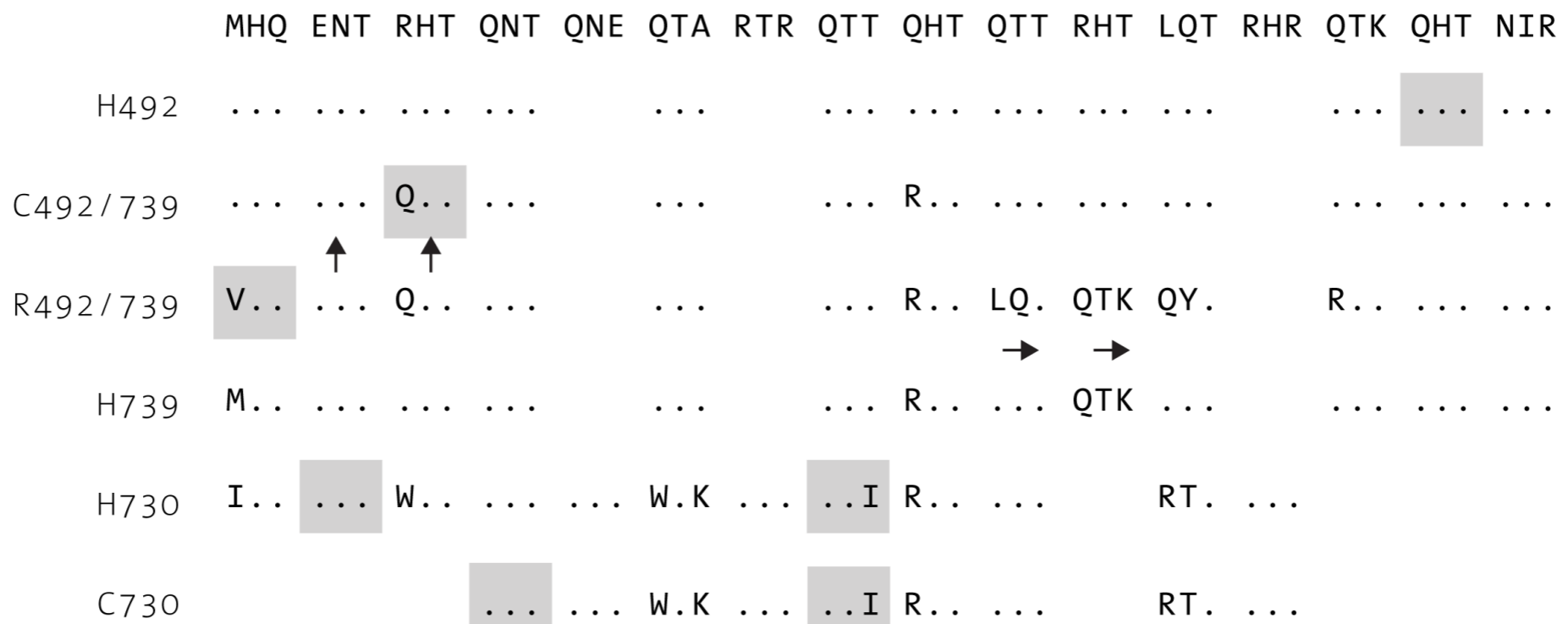
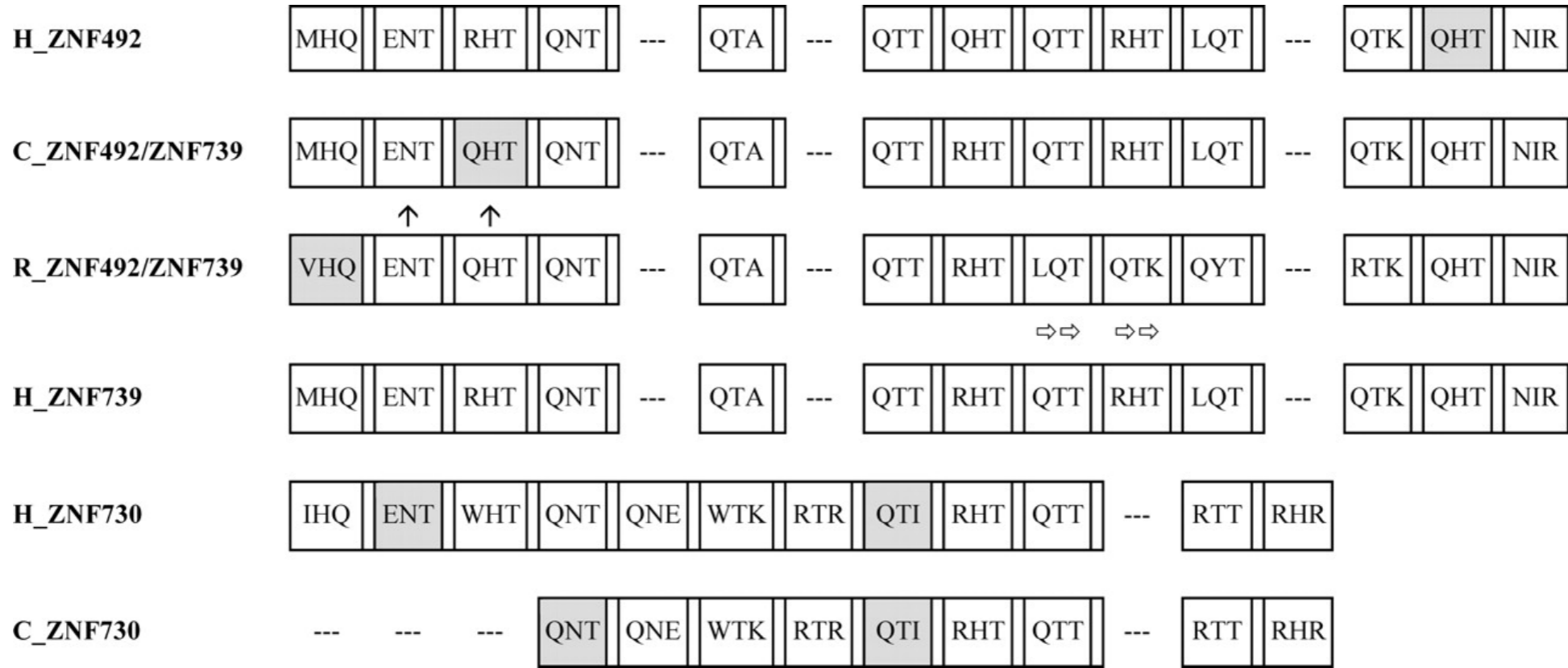


# EXCESS INK



Zinc finger exon analysis for ZNF493 and ZNF738, two divergent genes from the ZNF431 clade. Hamilton, A.T., et al., Evolutionary expansion and divergence in the ZNF91 subfamily of primate-specific zinc finger genes. *Genome Res*, 2006. 16(5): p. 584-94.

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# THE LEGEND IS THE MAP

- all genes
- ▲ specific in situ result
- △ selective in situ result
- ▽ nonselective in situ result
- ▲ specific in literature
- × nonspecific in literature

ISH results in relation to profile similarity to SM-MHC (Y-axis) and to expression level (X-axis). Nelander, S., P. Mostad, and P. Lindahl, Prediction of cell type-specific gene modules: identification and initial characterization of a core set of smooth muscle-specific genes. *Genome Res*, 2003. 13(8): p. 1838-54.



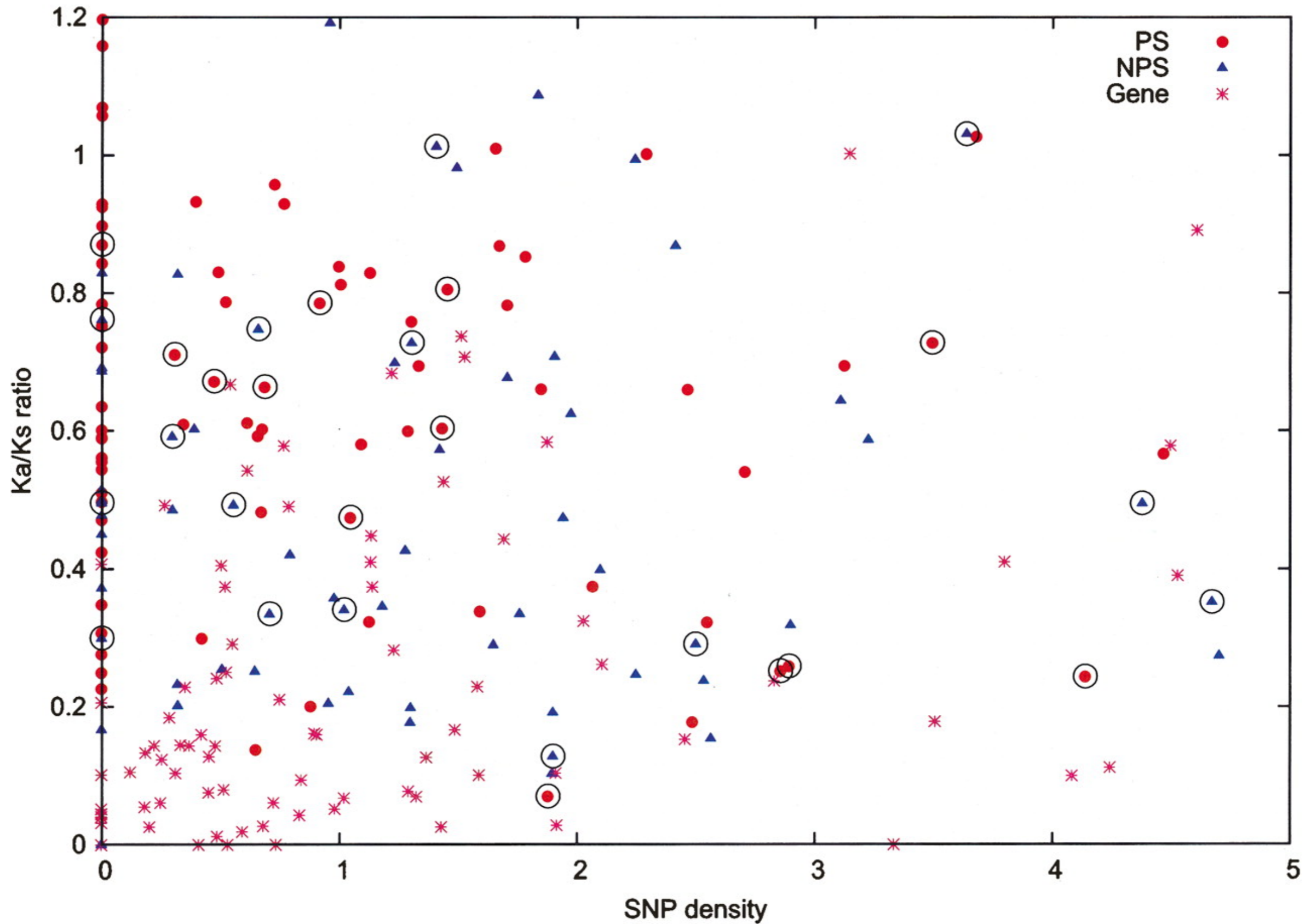
# THE LEGEND IS THE MAP

- all genes
- ▲ specific in situ result
- △ selective in situ result
- ▽ nonselective in situ result
- ▲ specific in litterature
- × nonspecific in litterature

GENE	•	
		IN SITU    IN LIT
SPECIFIC	●	▲
SELECTIVE	●	
NON-SELECTIVE	○	
NON-SPECIFIC		△

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# THE GLYPHS ARE CUES



Comparison of Ka/Ks ratio and SNP density for genes and pseudogenes. Zheng, D., et al., Pseudogenes in the ENCODE regions: consensus annotation, analysis of transcription, and evolution. *Genome Res*, 2007. 17(6): p. 839-51.

# HIERARCHICAL GLYPHS

PS  
NPS  
Gene



PSEUDOGENES

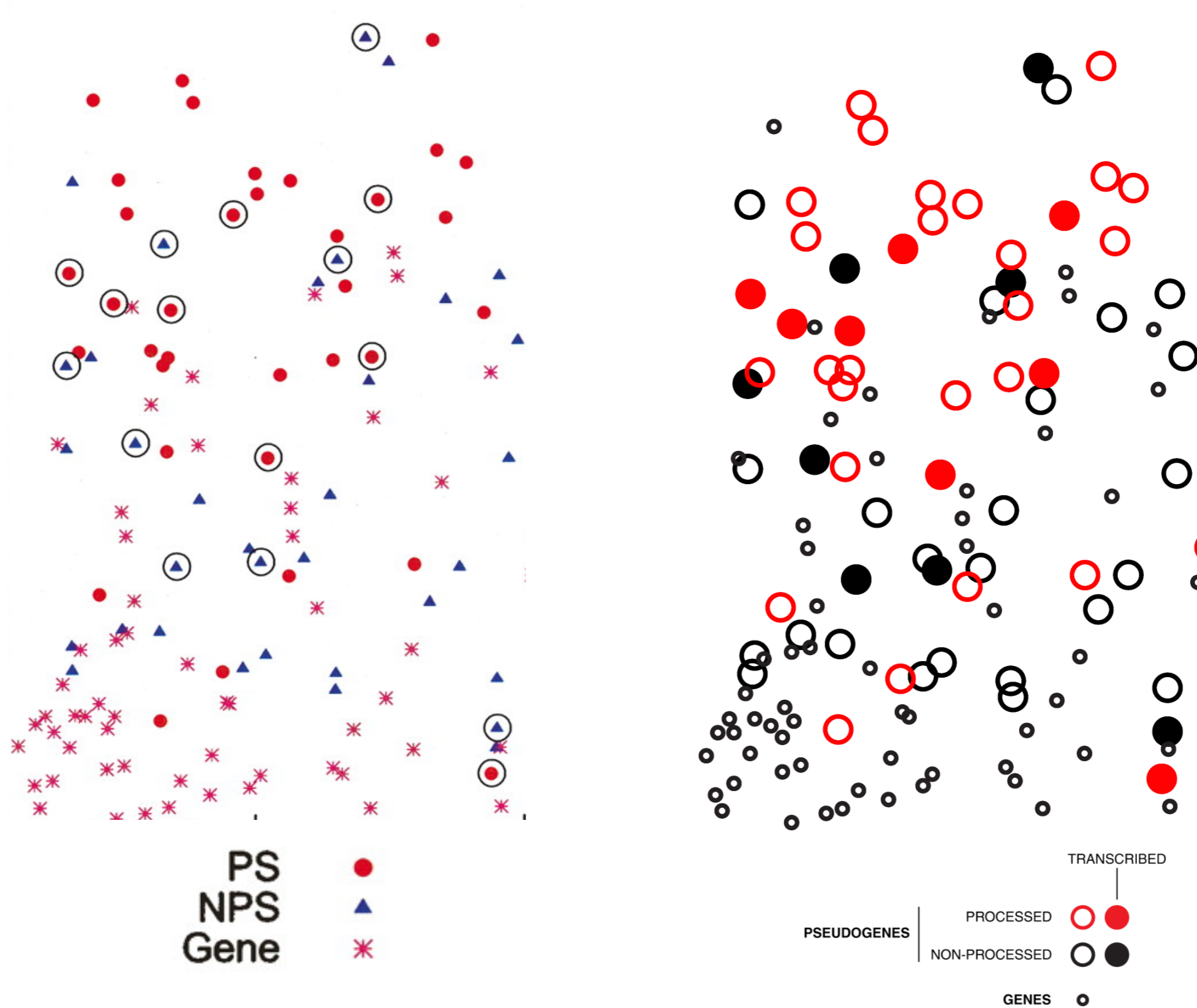
PROCESSED  
NON-PROCESSED

TRANSCRIBED



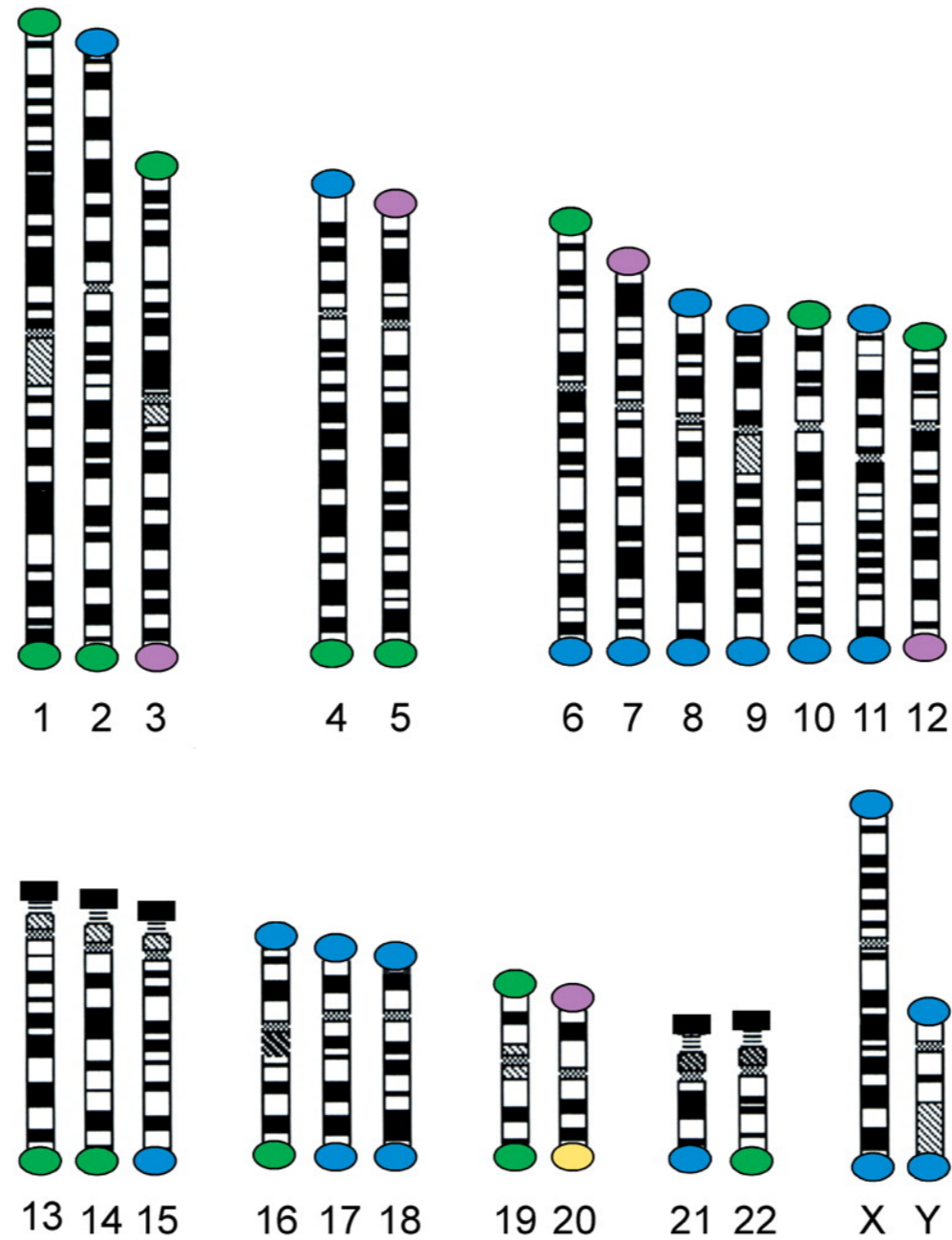
GENES

# HIERARCHICAL GLYPHS



Comparison of Ka/Ks ratio and SNP density for genes and pseudogenes. Zheng, D., et al., Pseudogenes in the ENCODE regions: consensus annotation, analysis of transcription, and evolution. *Genome Res*, 2007. 17(6): p. 839-51.

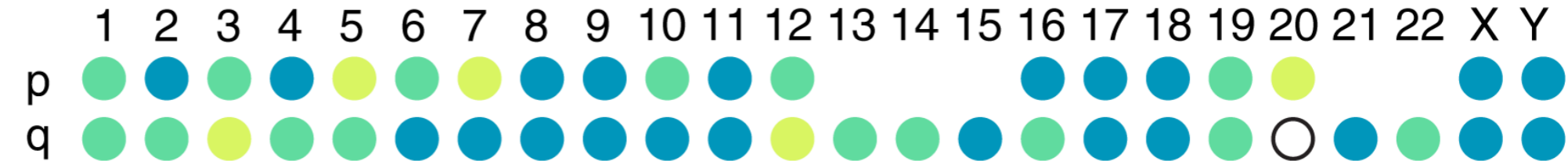
# EXCESS INK



Telomere sequence gaps. Riethman, H., et al., Mapping and initial analysis of human subtelomeric sequence assemblies. Genome Res, 2004. 14(1): p. 18-28.

# REMOVING EXCESS INK

## OPTION 1

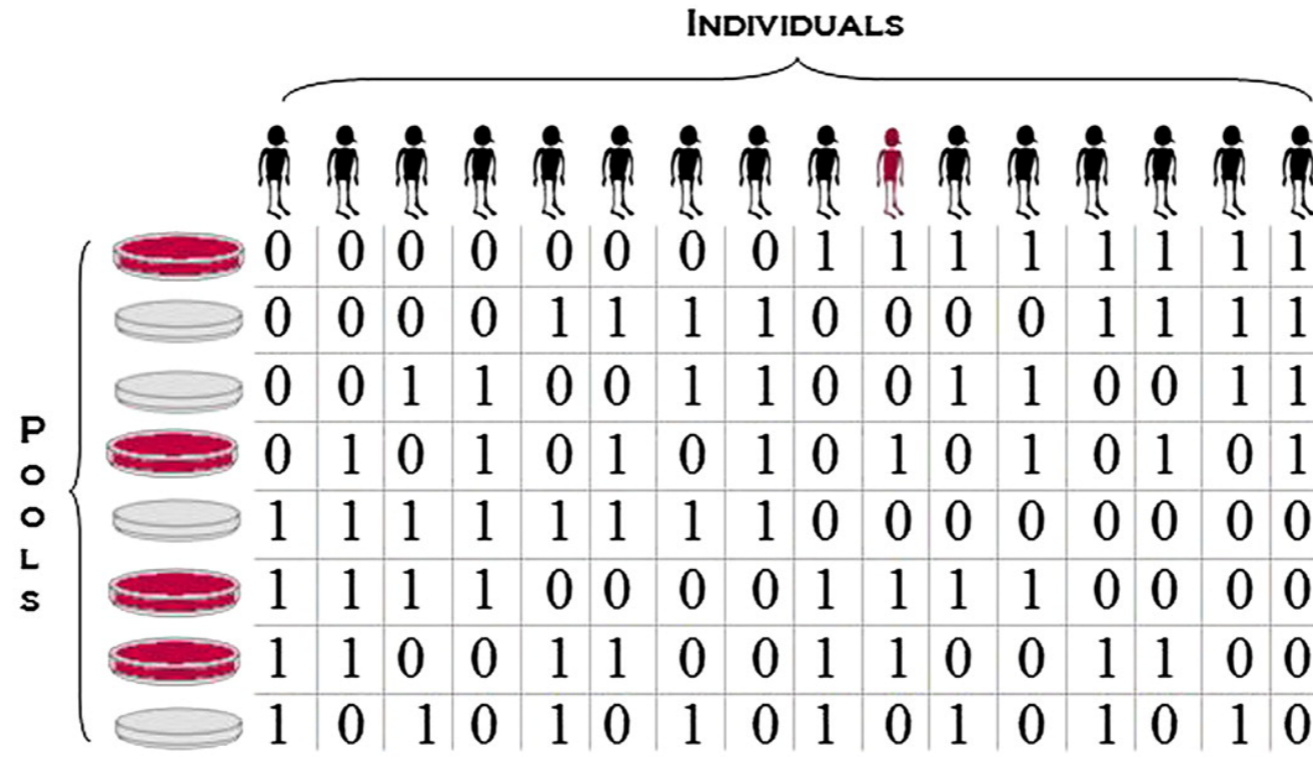
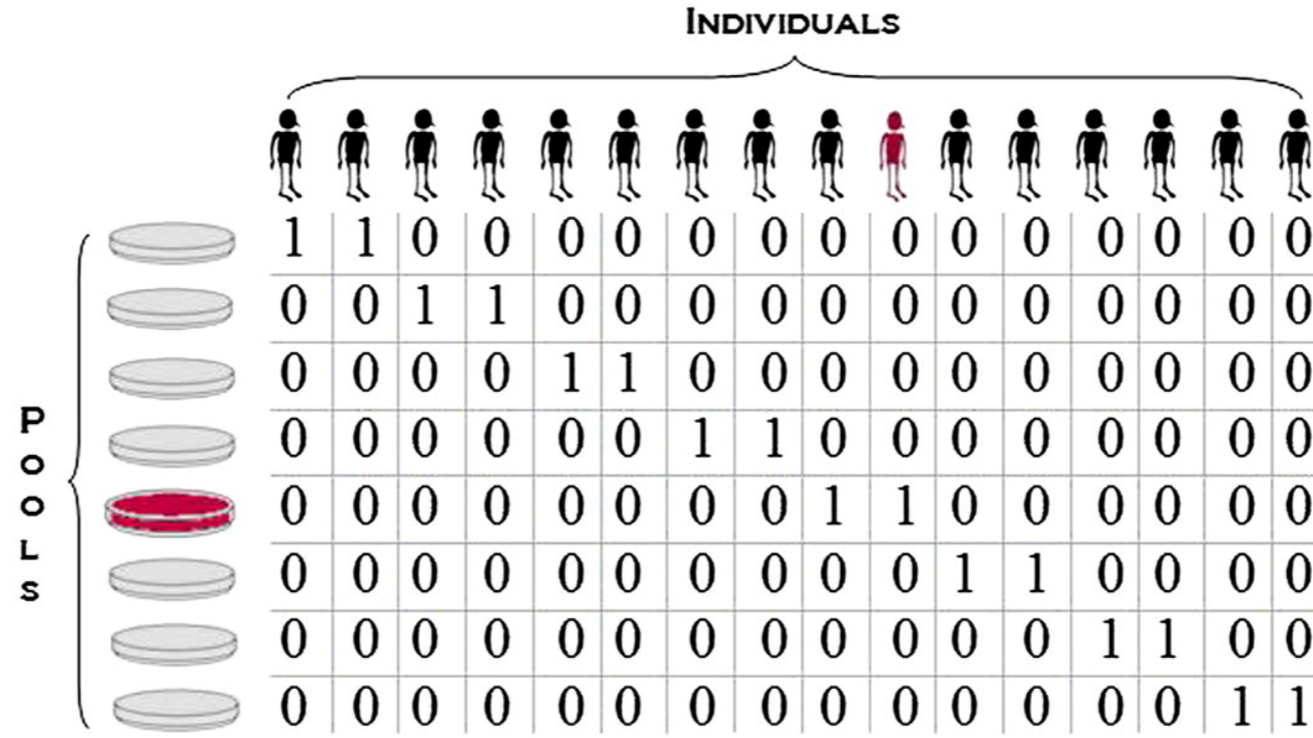


## OPTION 2



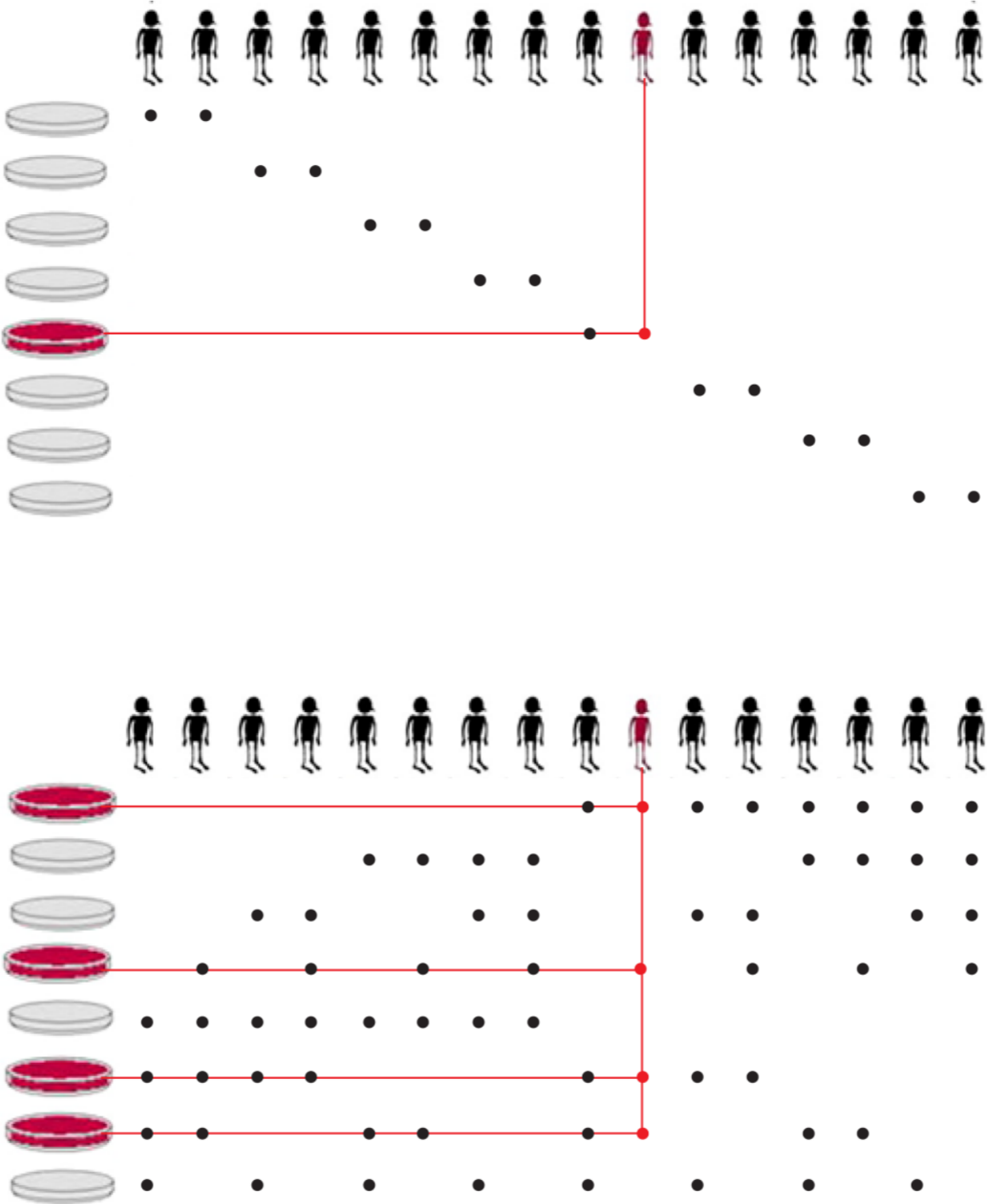
- no gap ●
- <20kb ●
- <20-70kb ●
- unk/srp ○
- acrocentric ●

# EXCESS INK COMPETES WITH MESSAGE



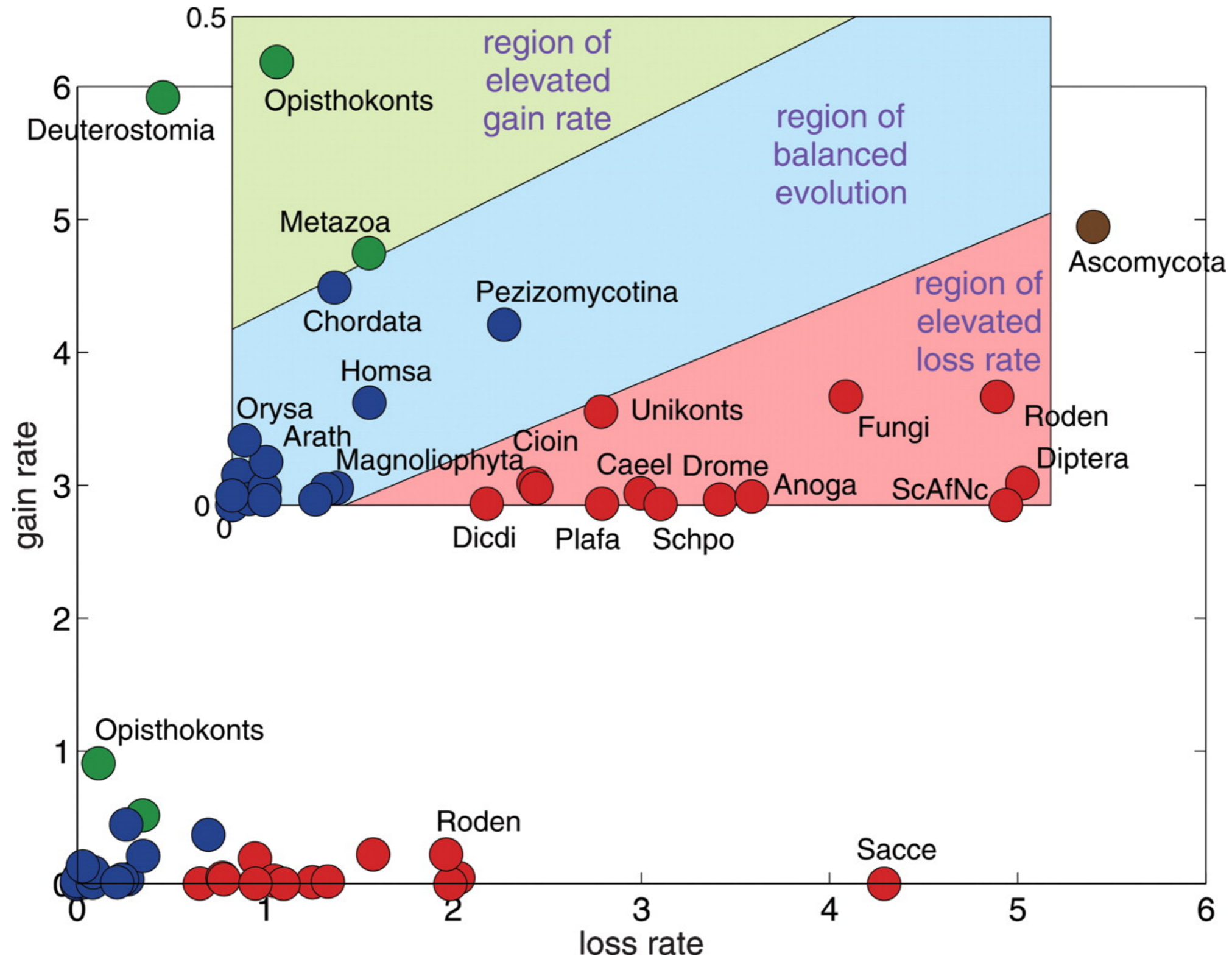
Resequencing with naïve and log pool designs. Prabhu, S. and I. Pe'er, Overlapping pools for high-throughput targeted resequencing. Genome Res, 2009. 19 (7): p. 1254-61.

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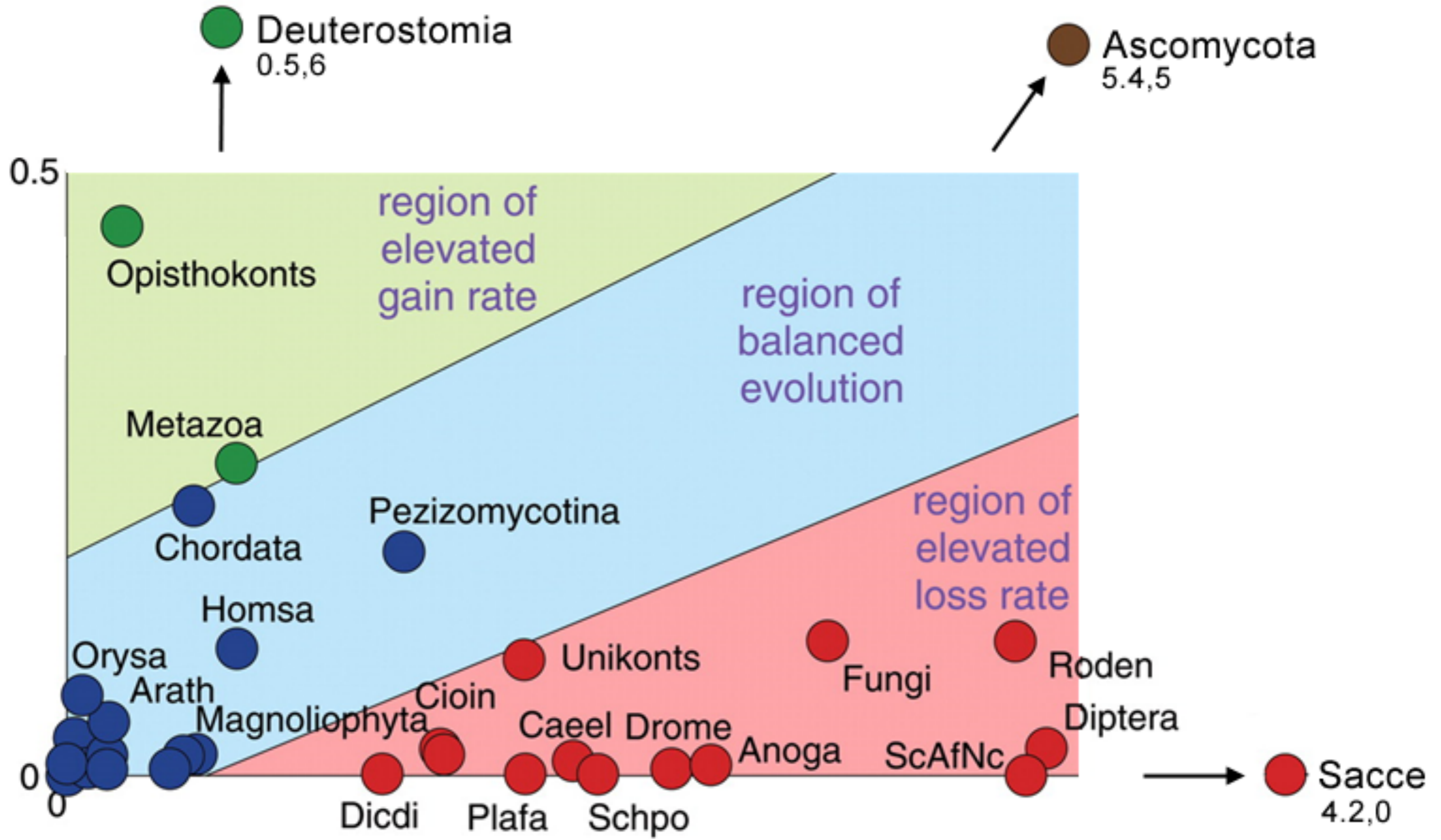


# HIJACKED BY OUTLIERS



Carmel, L., et al., Three distinct modes of intron dynamics in the evolution of eukaryotes. *Genome Res*, 2007. 17(7): p. 1034-44.

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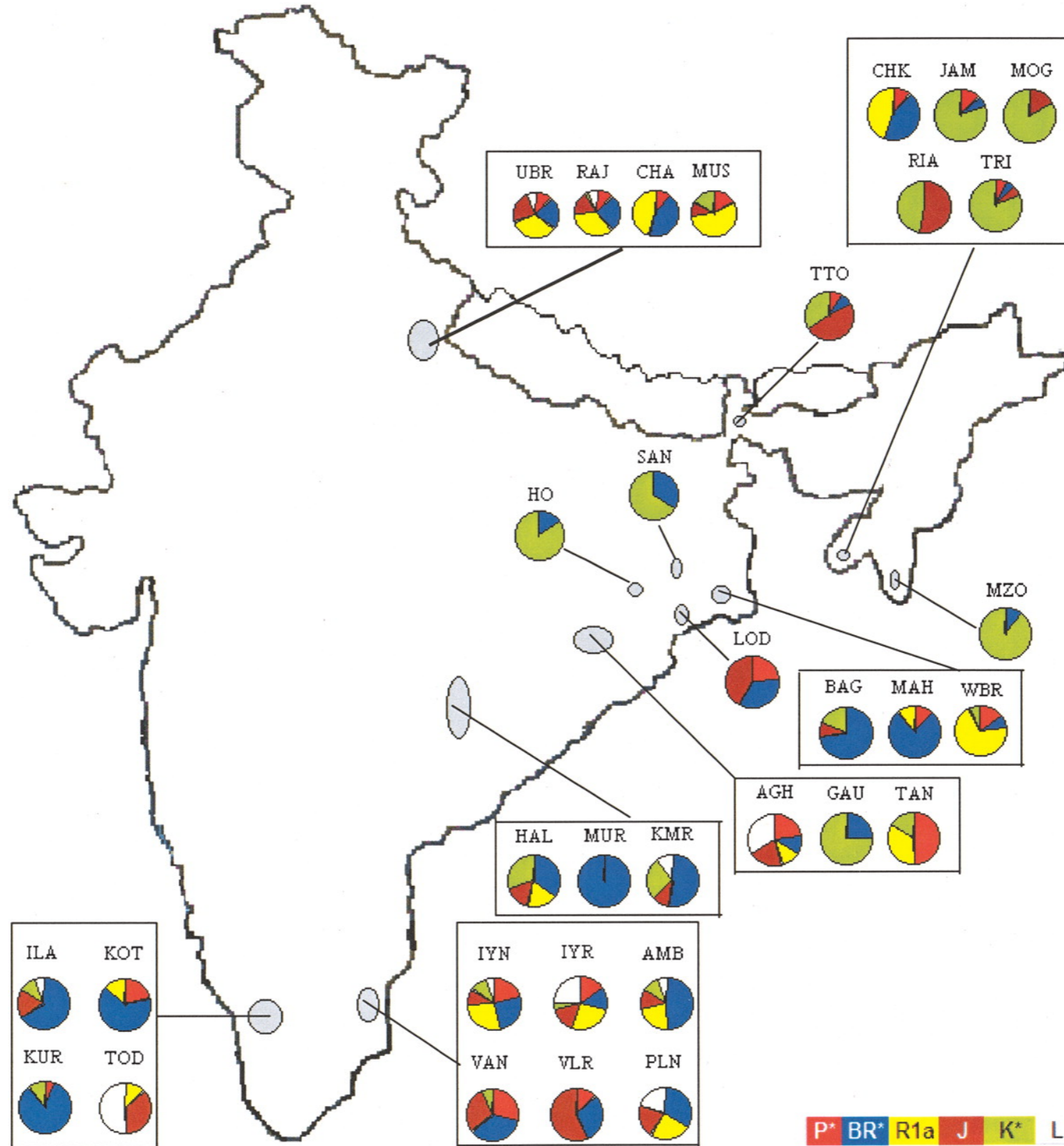
Intron gain and loss rates of eukaryotic lineages.

Carmel, L., et al., Three distinct modes of intron dynamics in the evolution of eukaryotes. *Genome Res*, 2007. 17(7): p. 1034-44.

legibility clarity **attractiveness**

**GRID LAYOUT**

# GRID LAYOUT



Frequencies (%) of Y-chromosomal haplogroups among ethnic populations. Basu, A., et al., Ethnic India: a genomic view, with special reference to peopling and structure. *Genome Res*, 2003. 13(10): p. 2277-90.

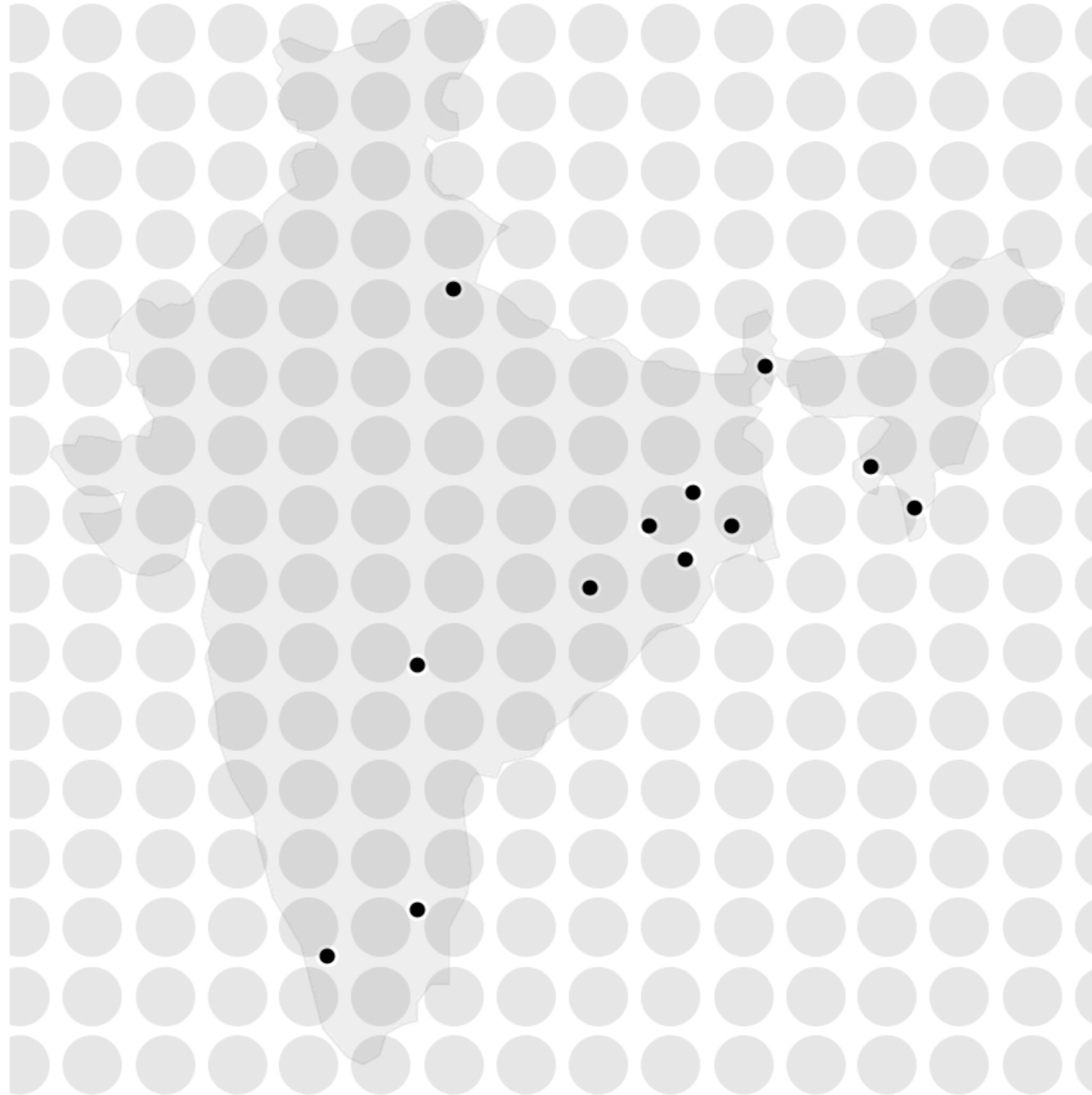
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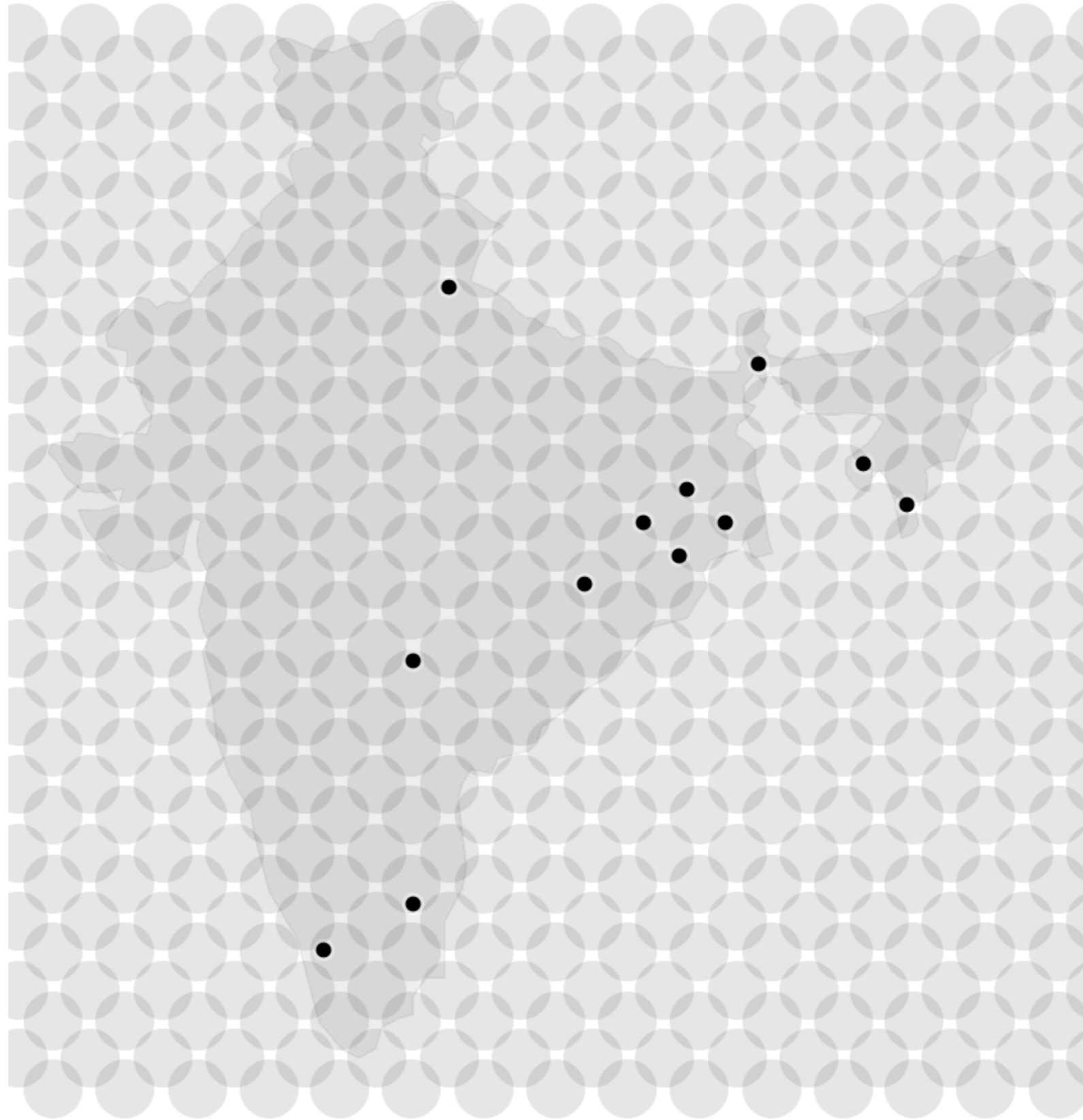
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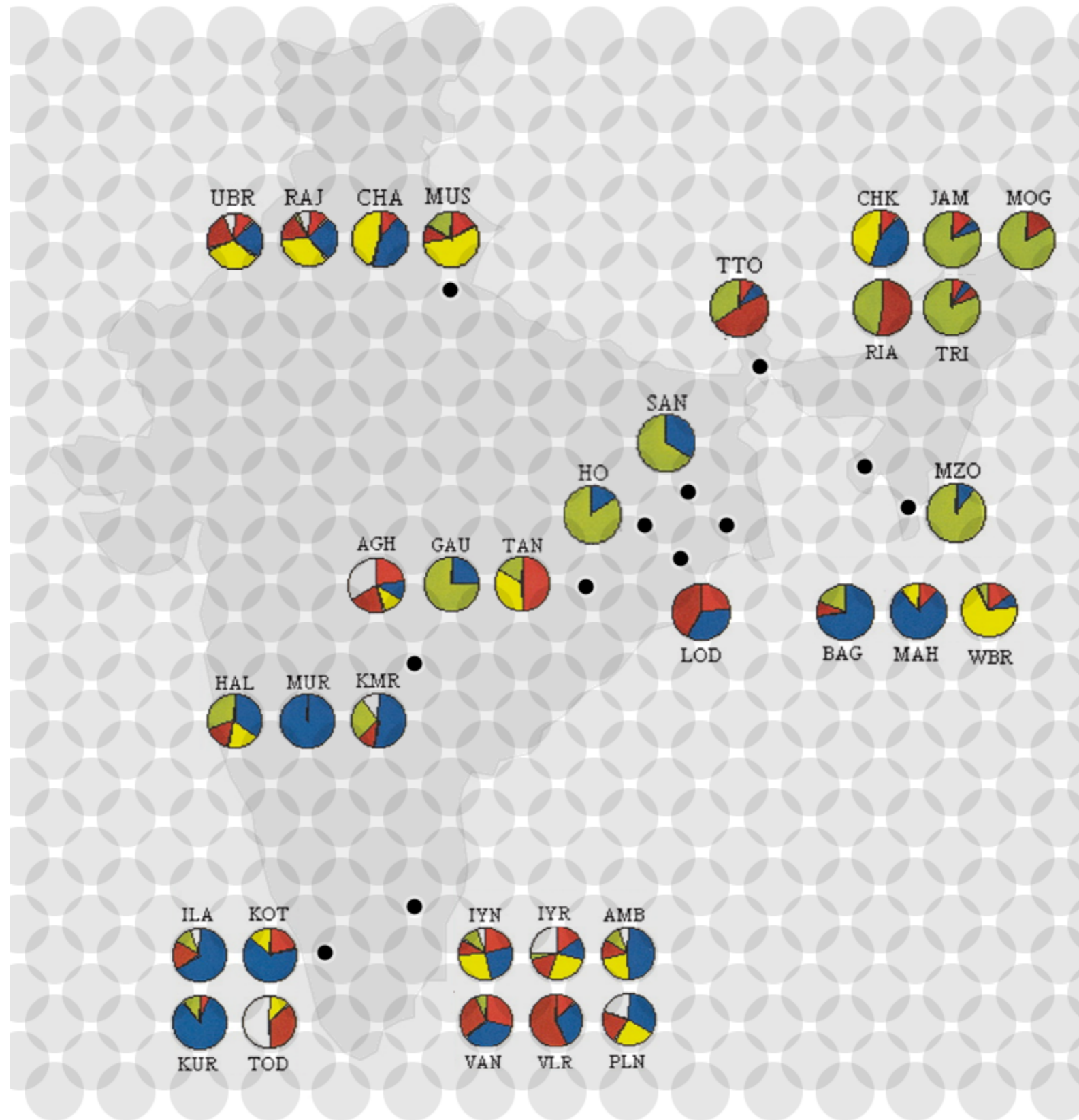


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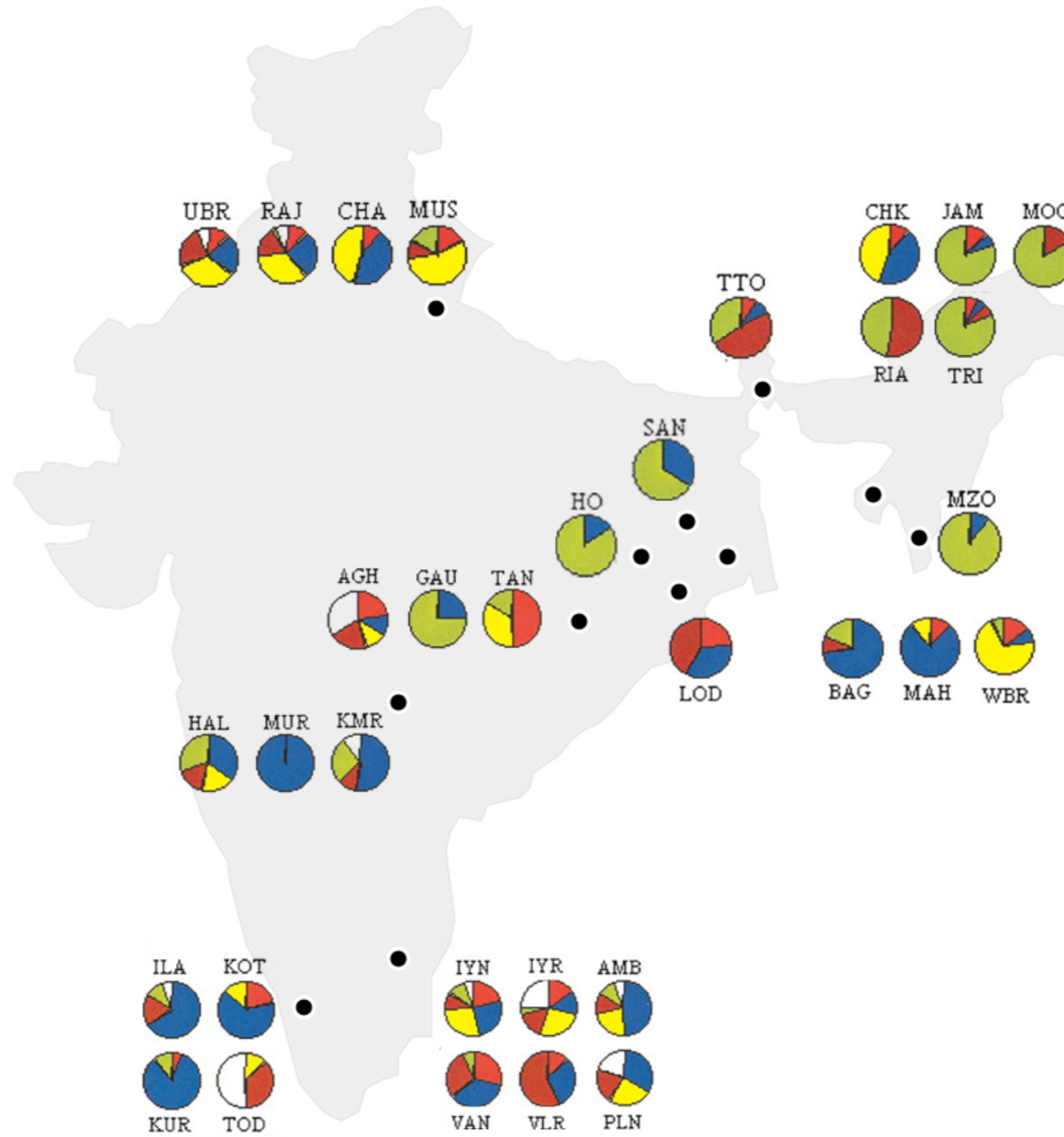




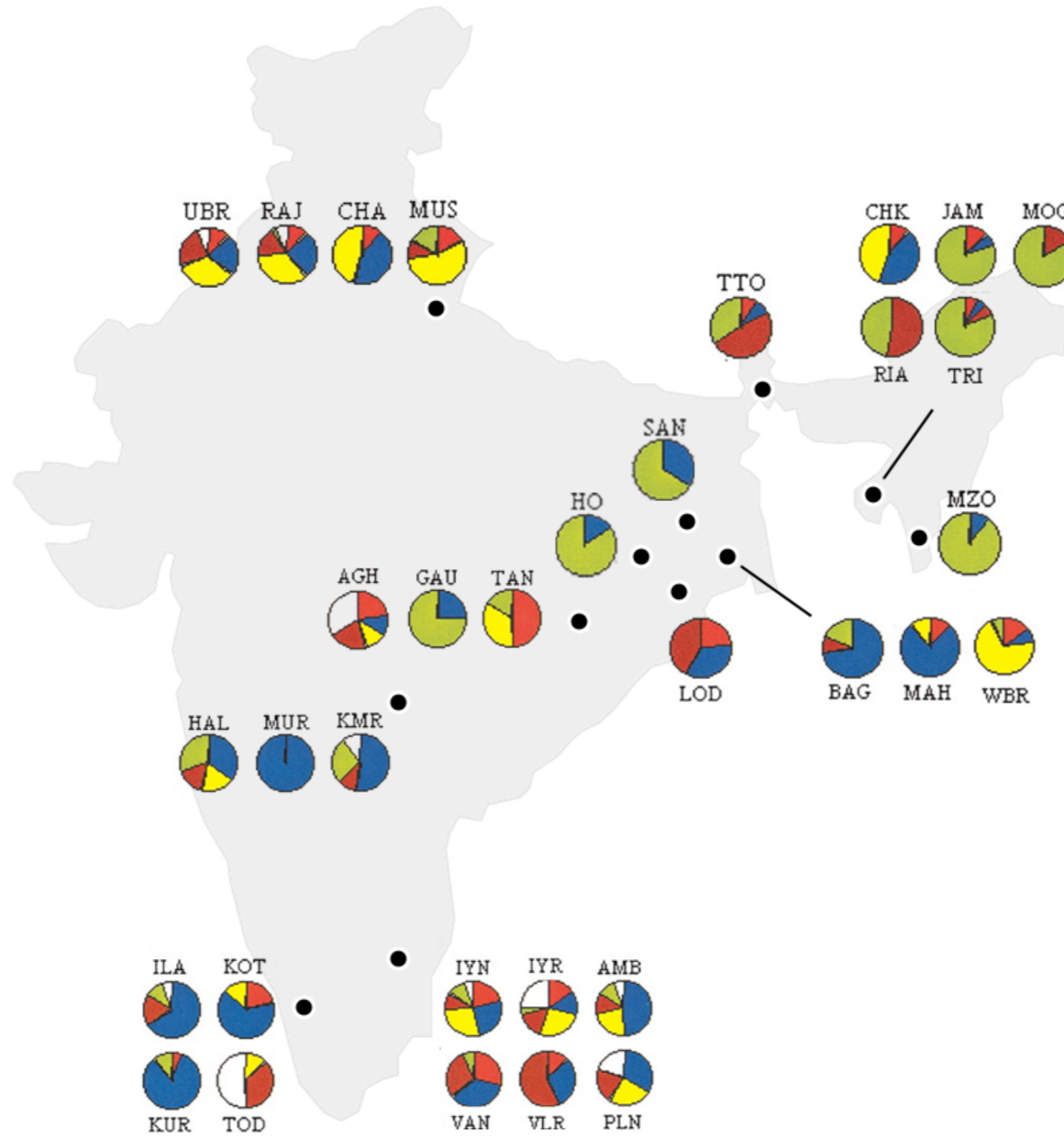
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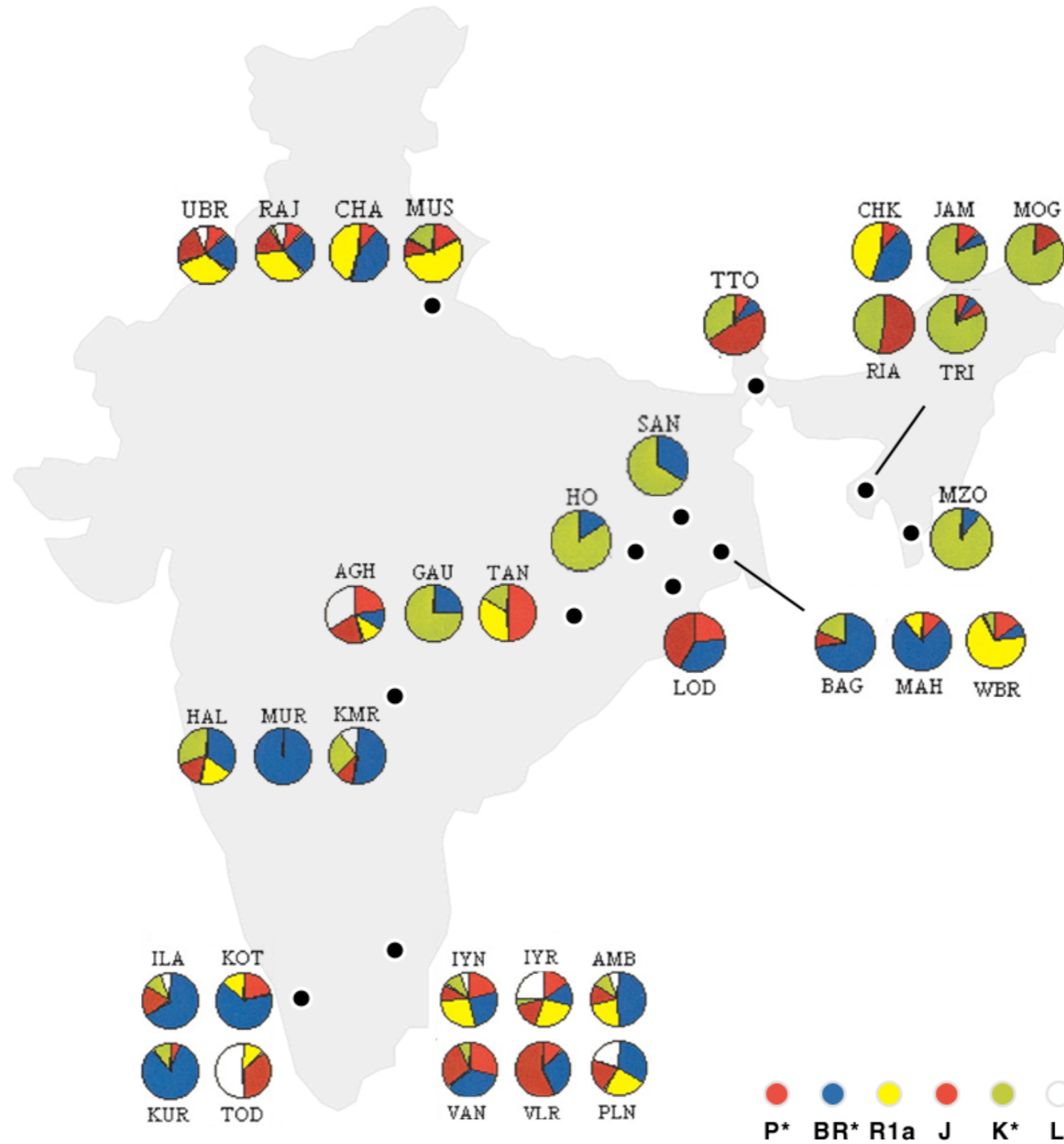
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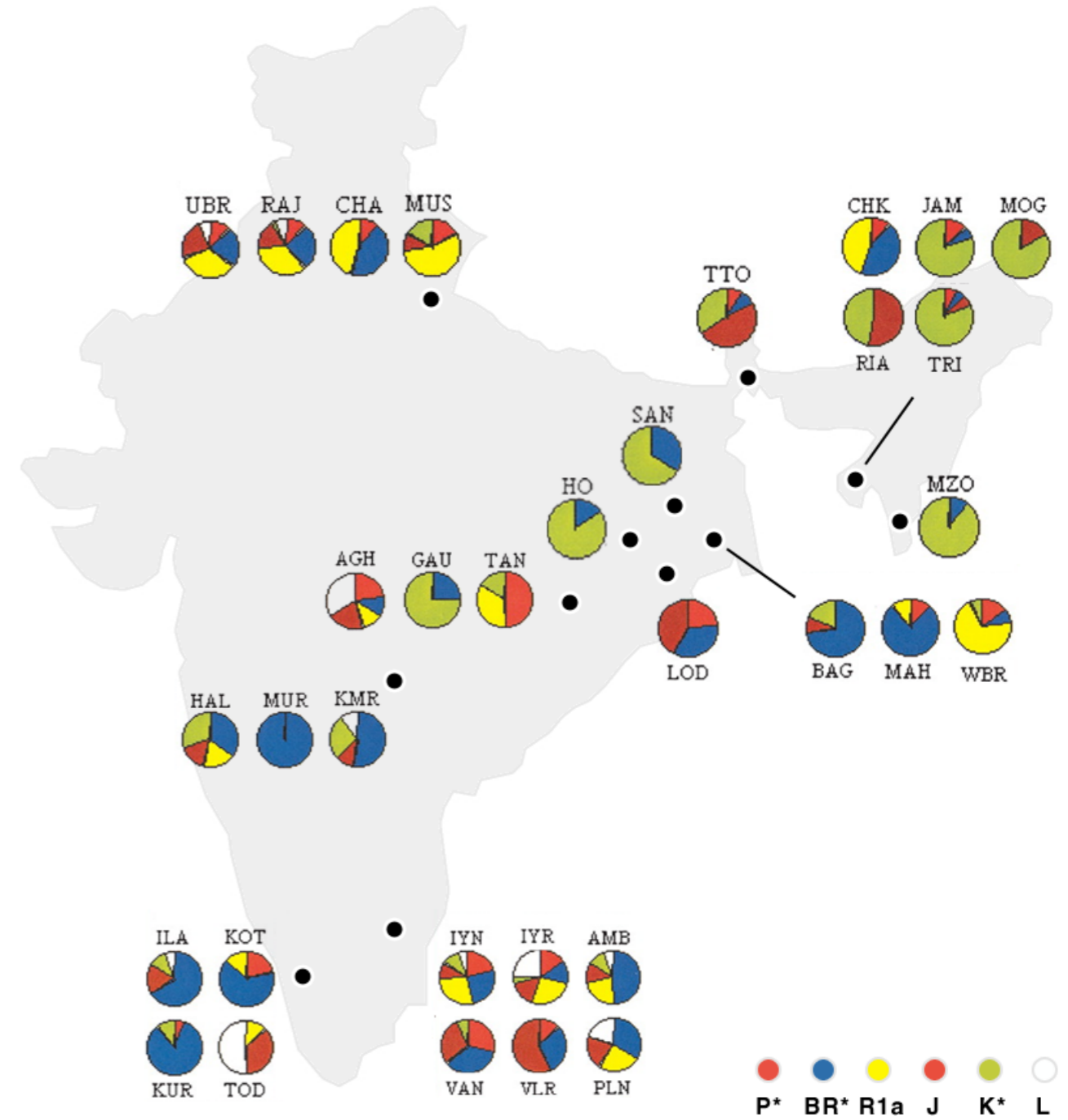
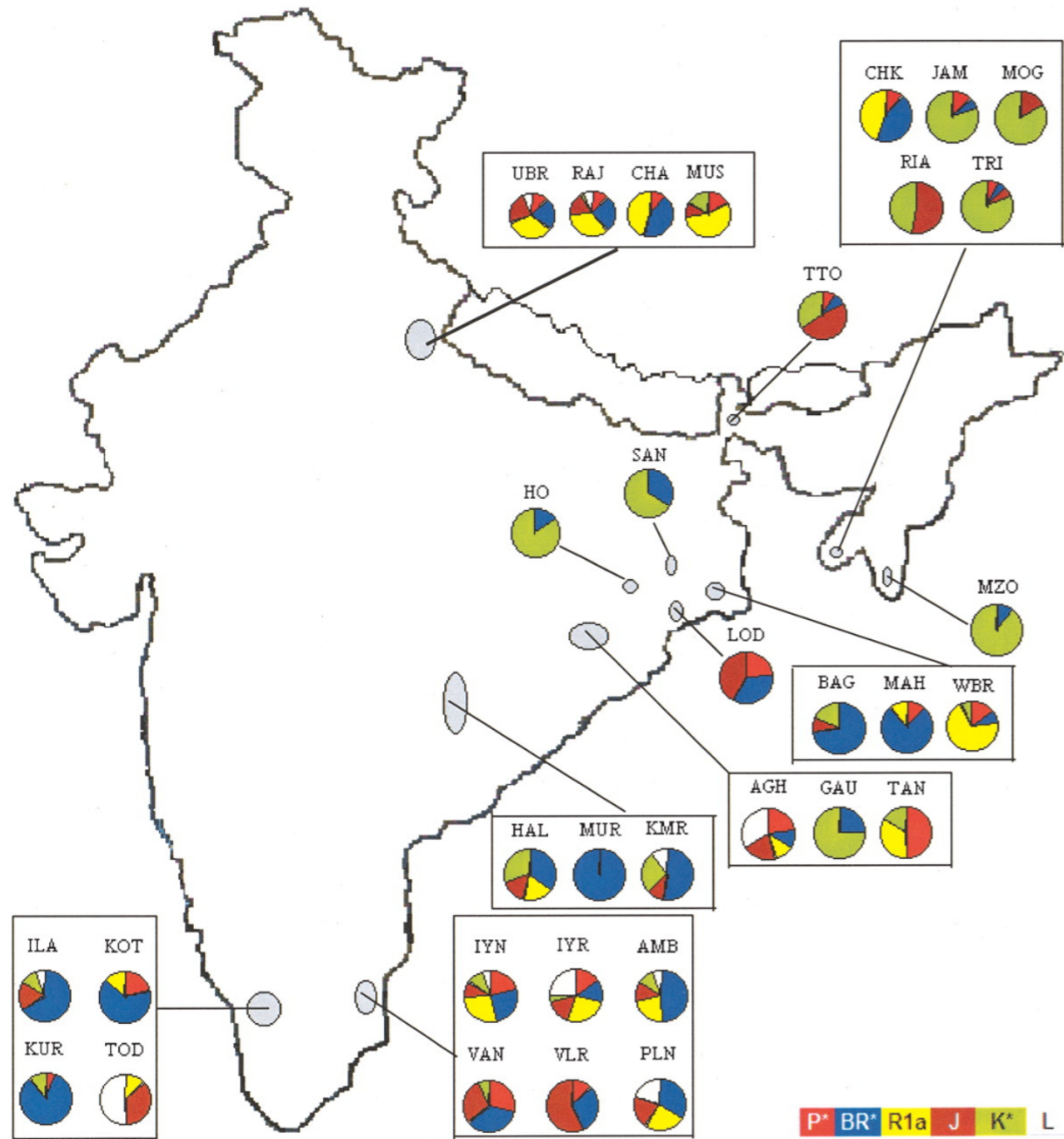
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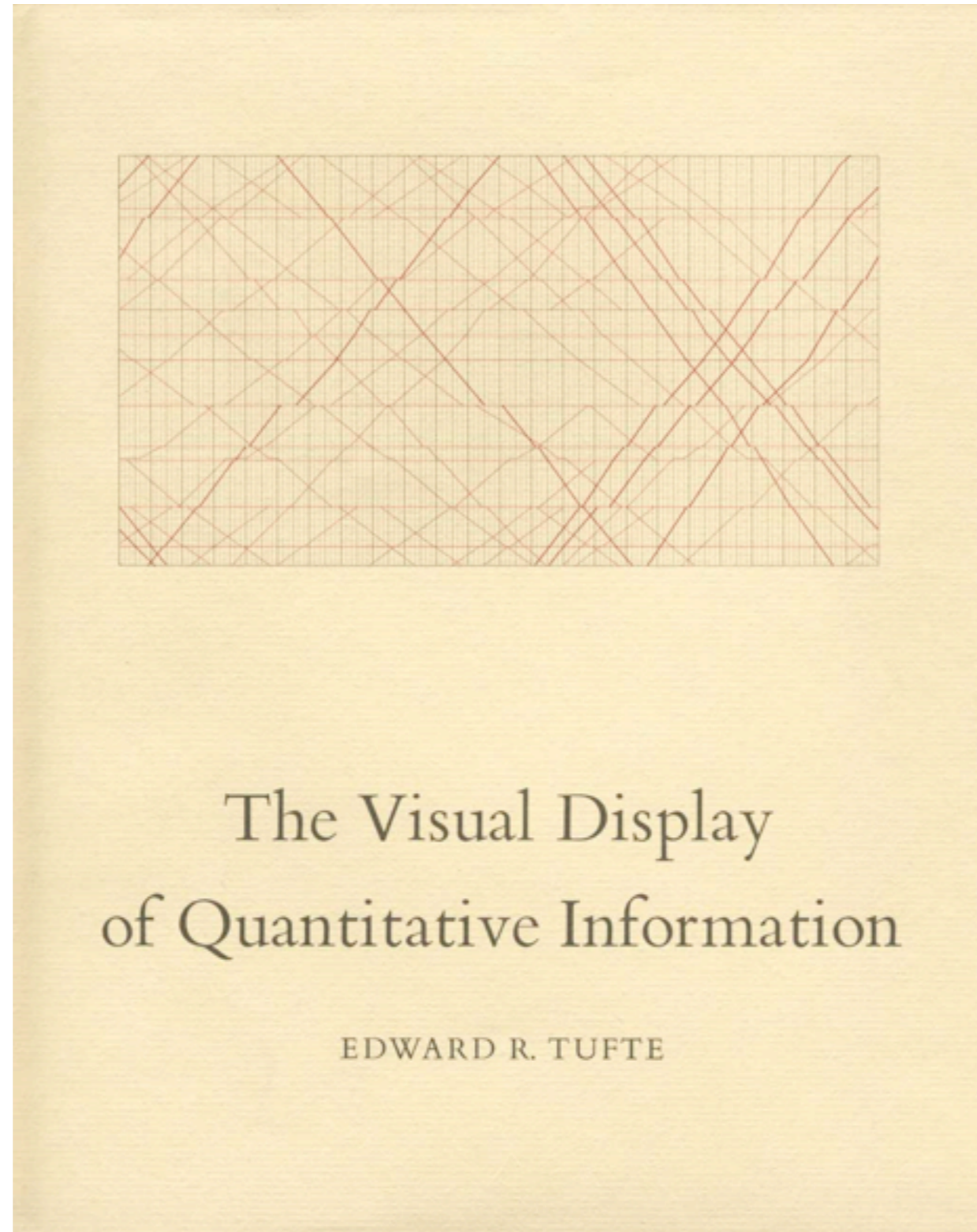


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# REQUIRED READING



TECH DEV

GENOMICS

SEQUENCING

INFORMATICS

COMPUTING



CANADA'S MICHAEL SMITH  
**GENOME  
SCIENCES**  
CENTRE

WWW.BCGSC.CA

