#### R/qtl2

#### high-dimensional data and multi-parent populations

#### Karl Broman

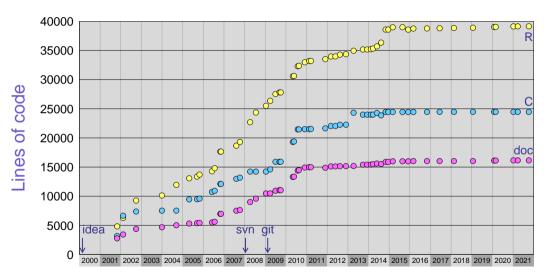
Biostatistics & Medical Informatics, UW-Madison

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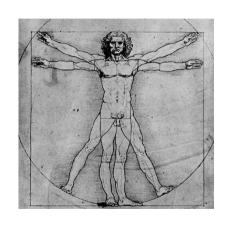
Slides: kbroman.org/Talk\_DOWorkshop2021

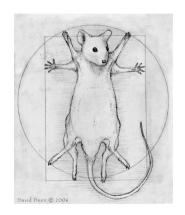


## 21 years of R/qtl



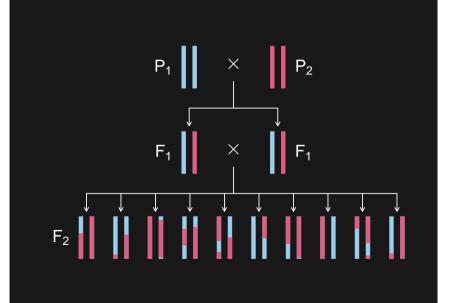




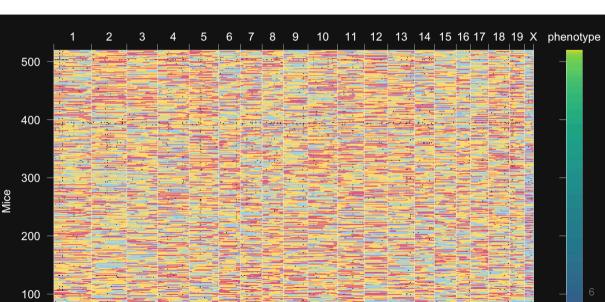


daviddeen.com

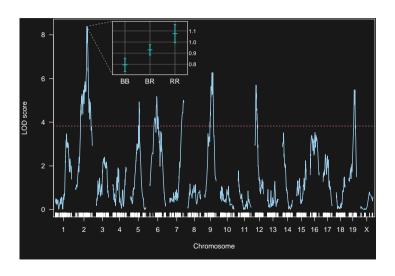
#### Intercross



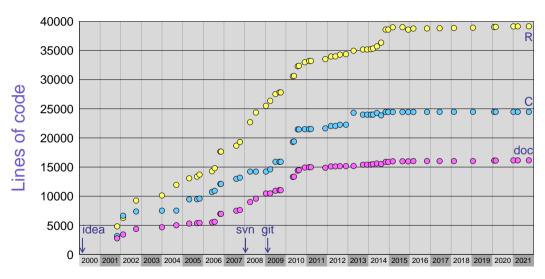
## Data



## QTL mapping



#### 21 years of R/qtl



# Why?

# Good things

#### Good things

- ▶ some of the code
- ▶ basics of the user interface
- ► diagnostics and data visualization
- ► quite comprehensive
- quite flexible

# Bad things

# Input file

	Α	В	С	D	Е	F	G	Н	- 1
1	liver	spleen	sex	pgm	D1Mit18	D1Mit80	D1Mit17	D2Mit379	D2Mit75
2					1	1	1	2	2
3					27.3	51.4	110.4	38.3	48.1
4	61.92	153.16	m	1	BB	SB	SB	SB	SB
5	88.33	178.58	m	1	-	-	-	BB	BB
6	58	131.91	m	1	BB	SB	SB	SB	SB
7	78.06	126.13	m	1	SB	SB	BB	SS	SS
8	65.31	181.05	m	1	-	-	-	SB	SB
9	59.26	191.54	m	1	-	-	-	SS	SS
10	59.47	154.88	m	1	BB	BB	BB	SB	SB
11	65.63	184.12	m	1	-	-	-	SB	SB
12	38.64	133.05	m	1	SB	BB	SB	SB	SB
13	60.94	275.63	m	1	-	-	-	SB	BB
14	51.48	395.25	m	1	-	-	-	SB	BB
15	47.12	260.45	m	1	BB	SB	SB	BB	BB

#### Stupidest code ever

```
n <- ncol(data)
temp <- rep(FALSE,n)
for(i in 1:n) {
  temp[i] <- all(data[2,1:i]=="")
  if(!temp[i]) break
}
if(!any(temp)) stop("...")
n.phe <- max((1:n)[temp])</pre>
```

# Open source means everyone can see my stupid mistakes

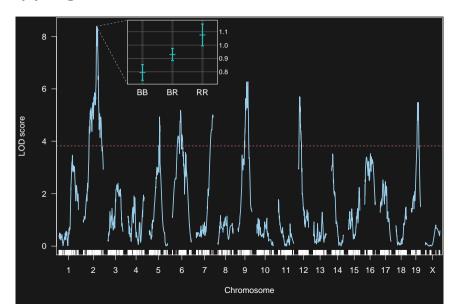
Open source means everyone can see my stupid mistakes

Version control means everyone can see every stupid mistake I've ever made

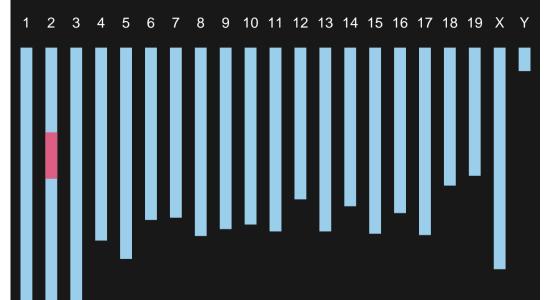
#### **Documentation**

# Support

## QTL mapping



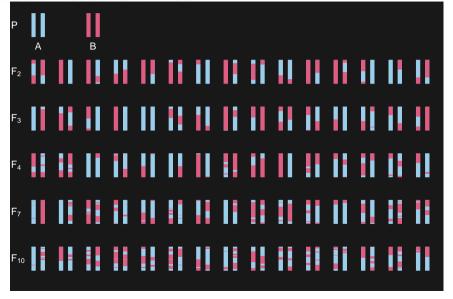
Congenic line



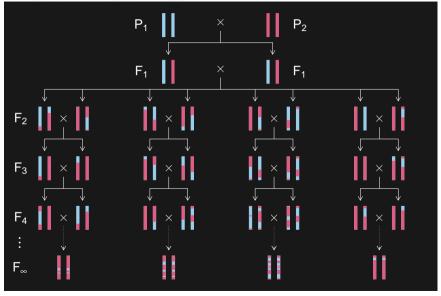
#### Improving precision

- more recombinations
- ▶ more individuals
- ▶ more precise phenotype
- ► lower-level phenotypes
  - transcripts, proteins, metabolites

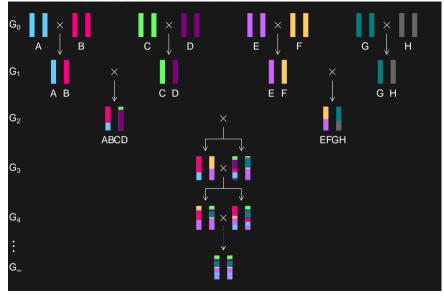
#### Advanced intercross lines



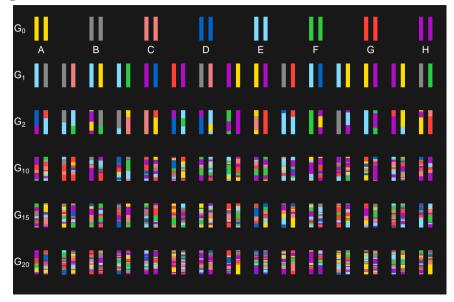
#### Recombinant inbred lines



## **Collaborative Cross**



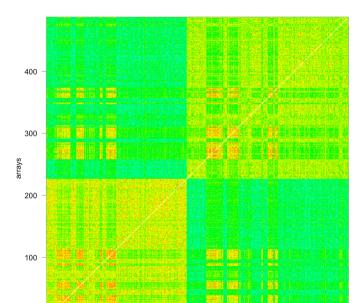
#### Heterogeneous stock



## Genome-scale phenotypes



## Challenges: diagnostics

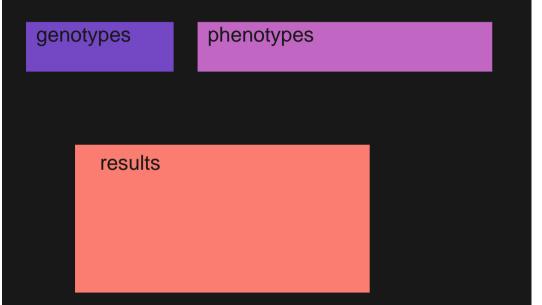


#### Challenges: scale of results

genotypes

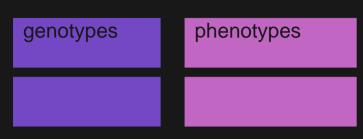
phenotypes

#### Challenges: scale of results

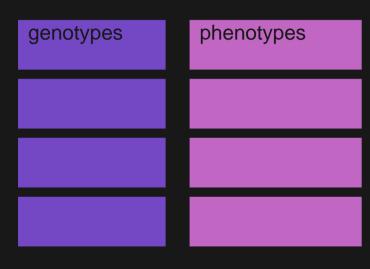


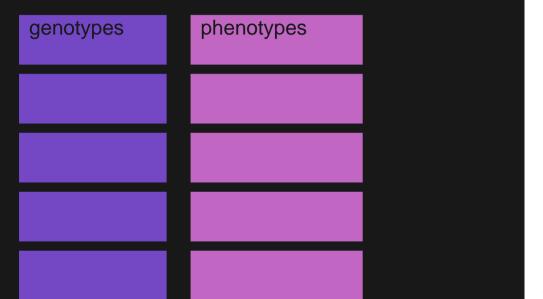
genotypes

phenotypes













## Challenges: metadata

What the heck is "FAD\_NAD SI 8.3\_3.3G"?

What was the question again?



## R/qtl2

- ► High-density genotypes
- ► High-dimensional phenotypes
- ► Multi-parent populations
- ► Linear mixed models

#### R/qtl2: Let's not make the same mistakes

- ► C++ and Rcpp
- ► Roxygen2 for documentation
- Unit tests
- ► A single "switch" for cross type

#### R/qtl2: Let's not make the same mistakes

- ► C++ and Rcpp
- ► Roxygen2 for documentation
- ▶ Unit tests
- ► A single "switch" for cross type
- Yet another data input format
- ► Flatter data structures, but still complex

#### Sustainable academic software

## Acknowledgments

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Quoc Tran

Aaron Wolen

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