R/qtl2

QTL mapping in multi-parent populations

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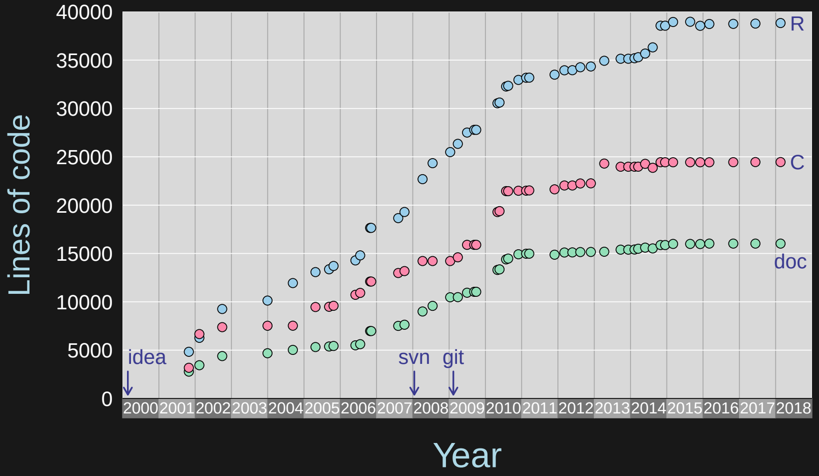
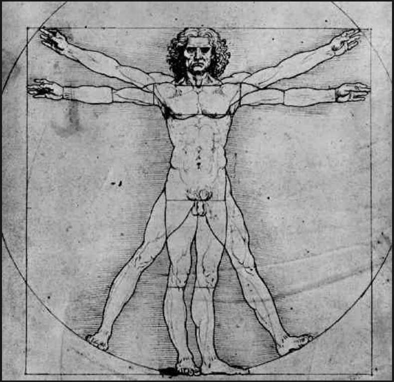
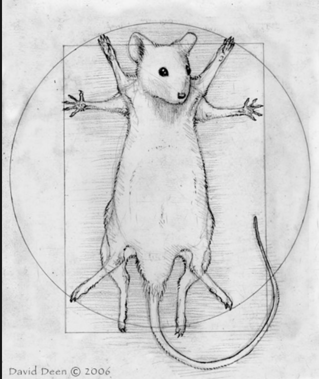
`github.com/kbroman`
`@kwbroman`

Slides: `bit.ly/purdue2018`

18 years of R/qtl

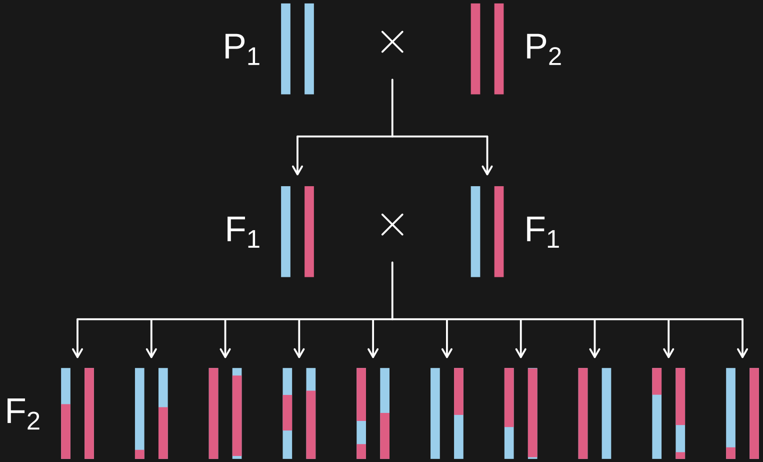




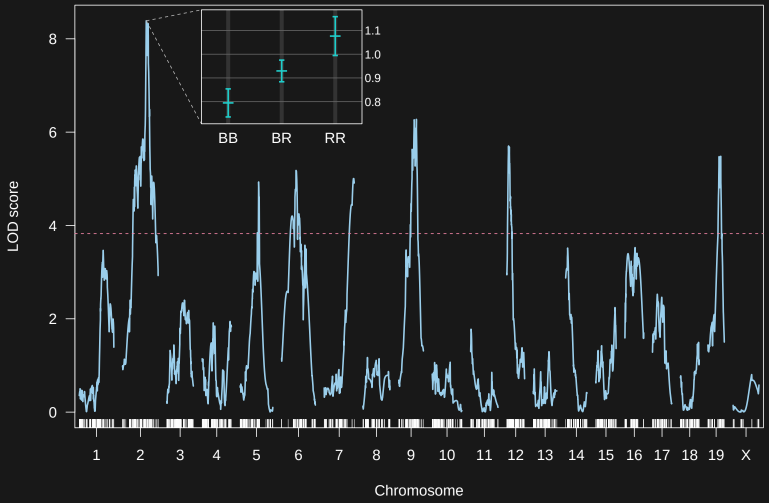


daviddeen.com

Intercross



QTL mapping


1.1
1.0
0.9
0.8
BB
BR
RR
8
6
4
2
0
LOD score
1
2
3
4
5
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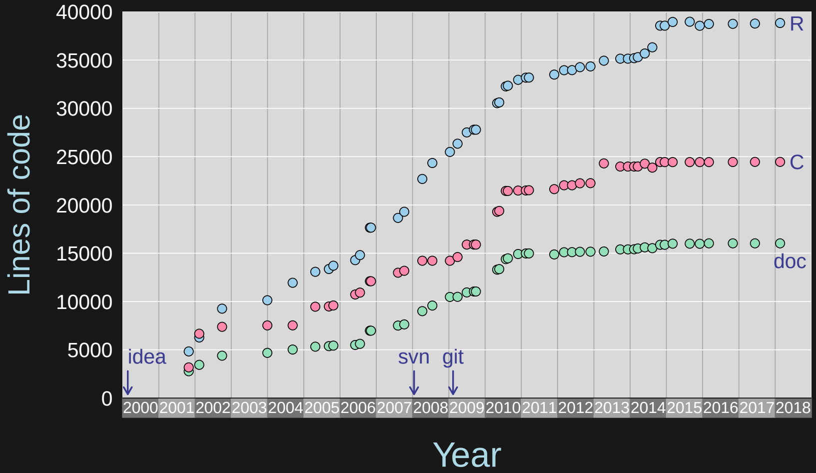
7
8
9
10
11
12
13
14
15
16
17
18
19
X
Chromosome

18 years of R/qtl



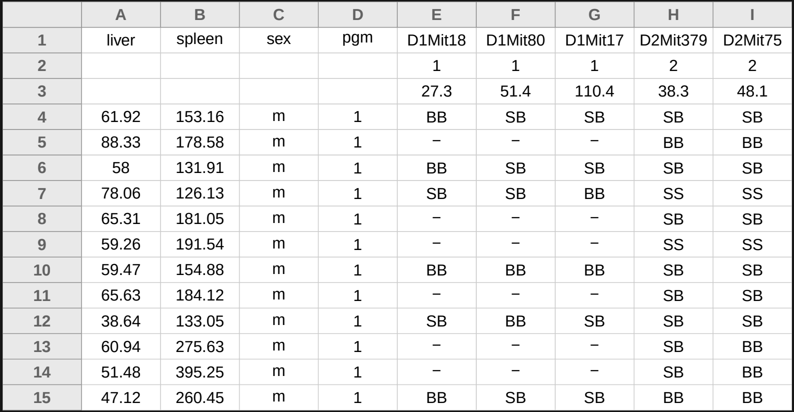
Good things

Good things

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

Bad things

Input file



	A	B	C	D	E	F	G	H	I
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])
```

kbroman.org/blog/2011/08/17/the-stupidest-r-code-ever

Open source means
everyone can see my stupid mistakes

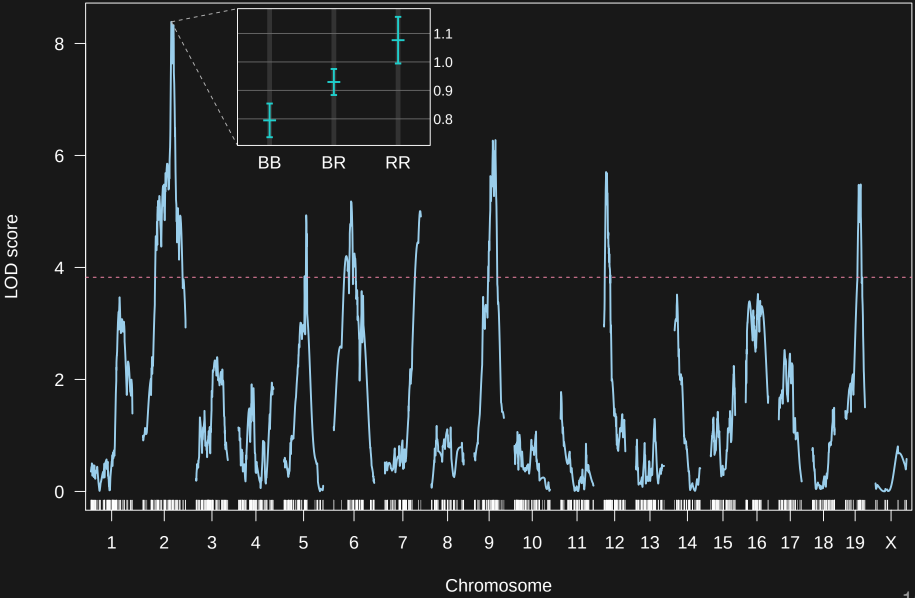
Open source means
everyone can see my stupid mistakes

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

Documentation

Support

QTL mapping


8
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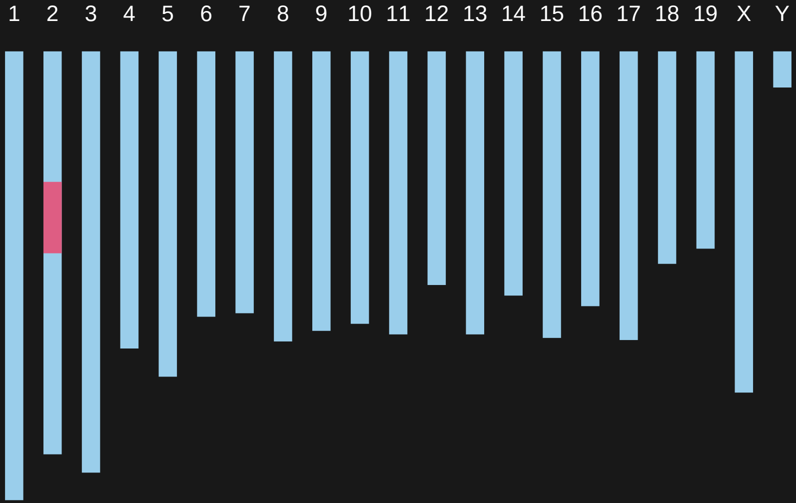
Chromosome

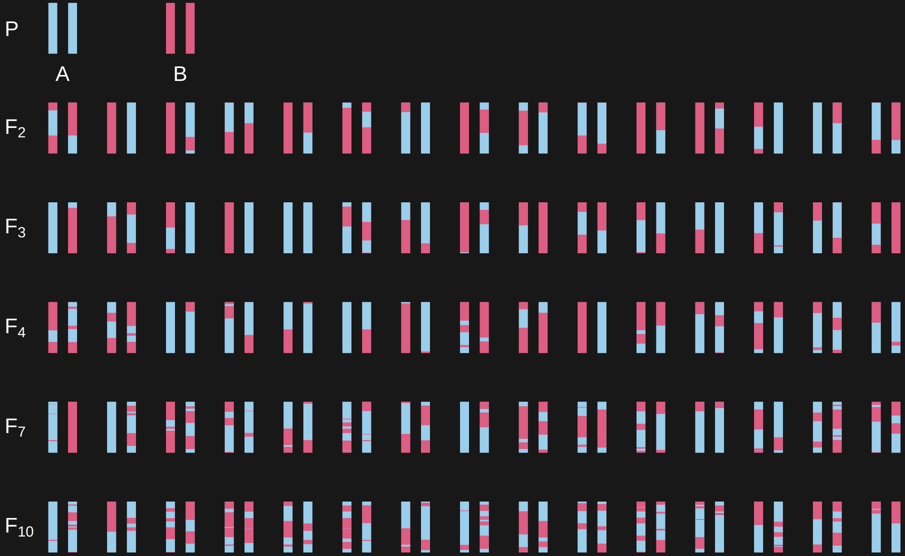
Congenic line



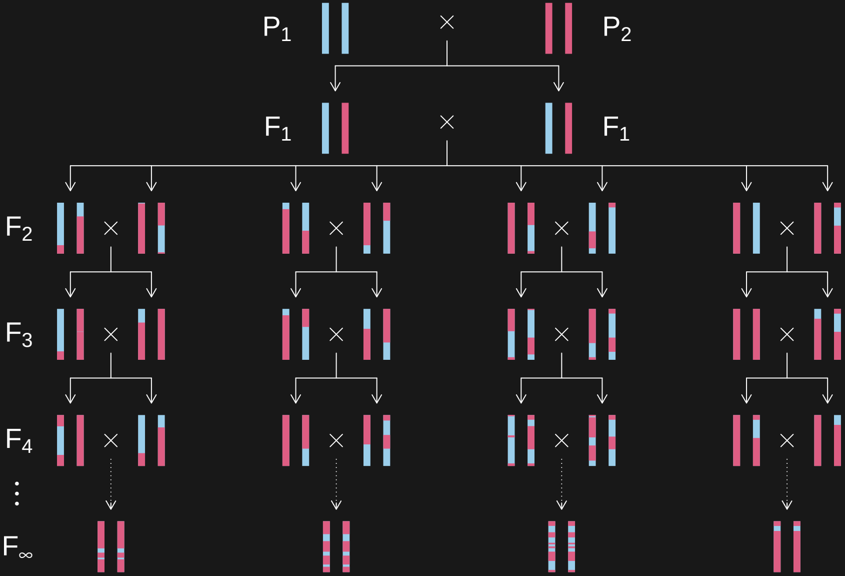
Improving precision

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

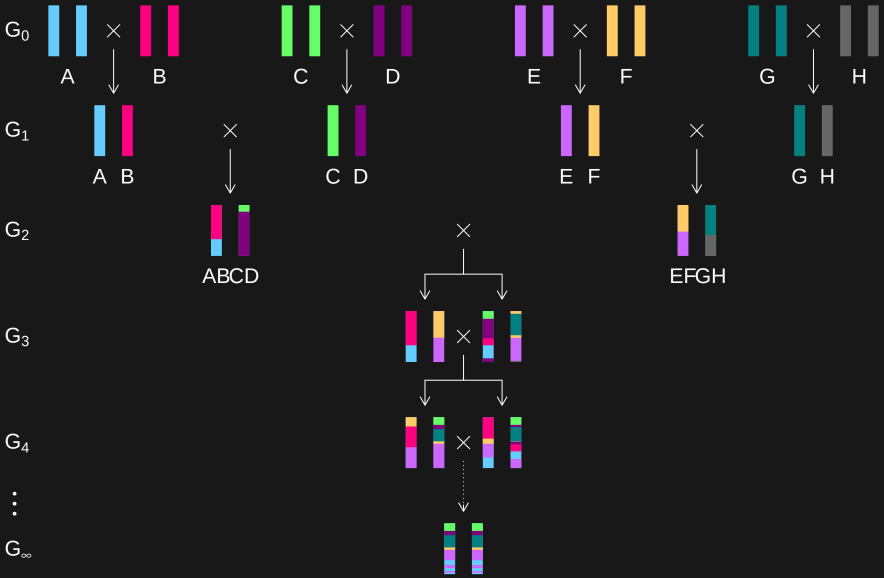
Advanced intercross lines



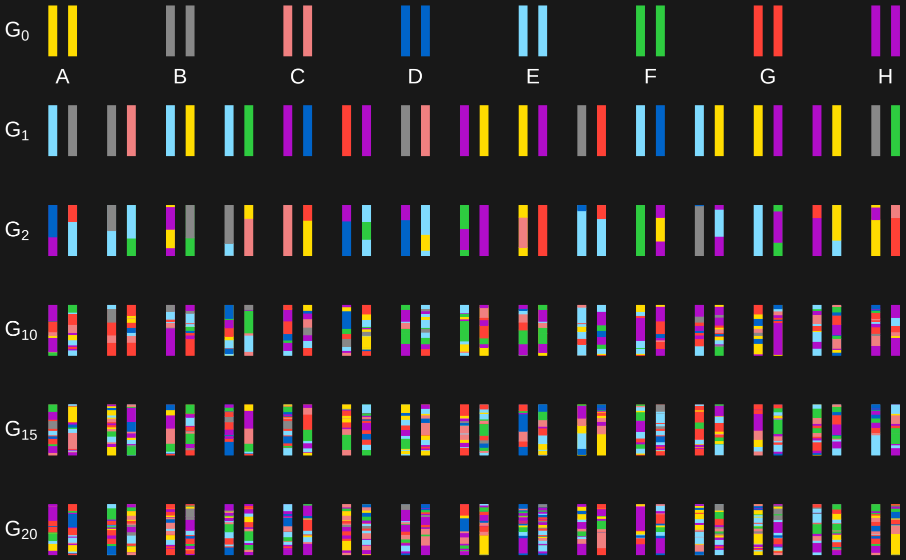
Recombinant inbred lines



Collaborative Cross



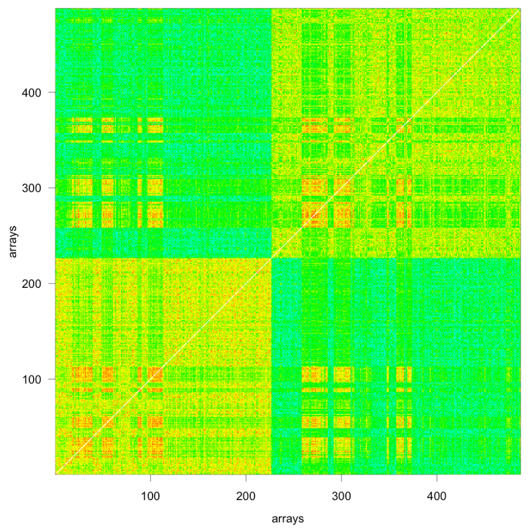
Heterogeneous stock



Genome-scale phenotypes

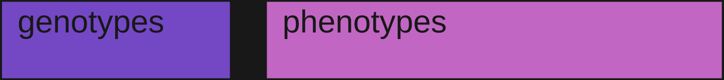
Alan Attie

Challenges: diagnostics



kbroman.org/blog/2012/04/25/microarrays-suck

Challenges: scale of results



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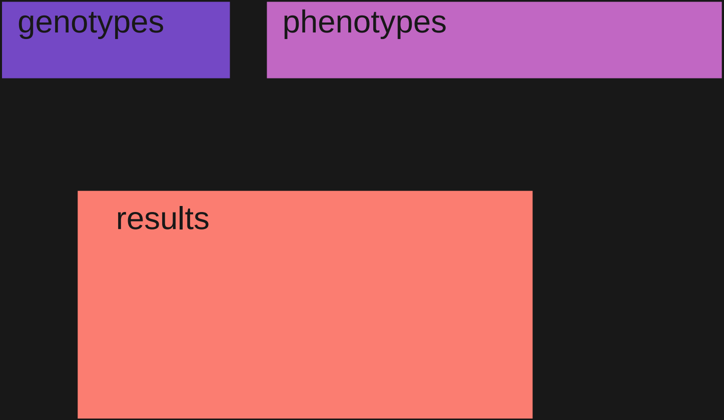

Challenges: organizing, automating

genotypes

phenotypes

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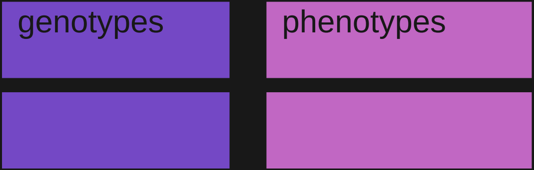

genotypes

phenotypes

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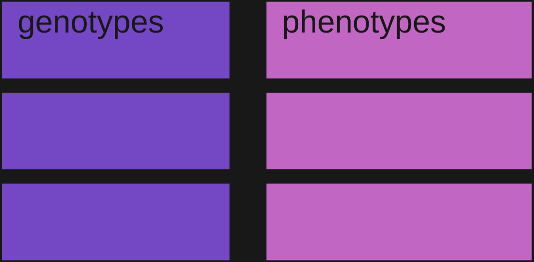
genotypes

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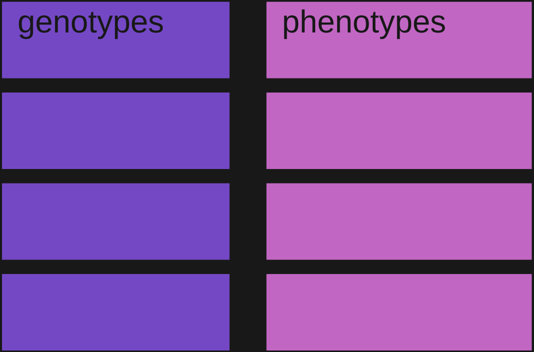

genotypes

phenotypes

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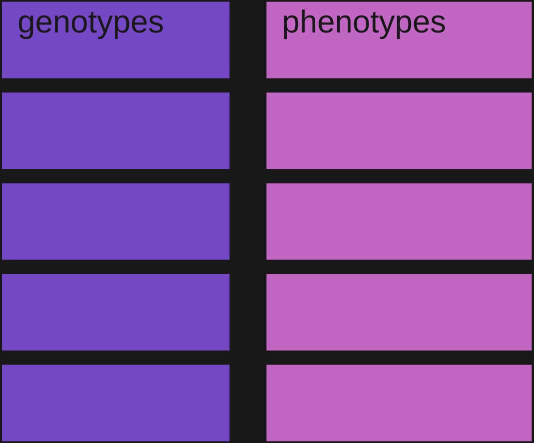

genotypes

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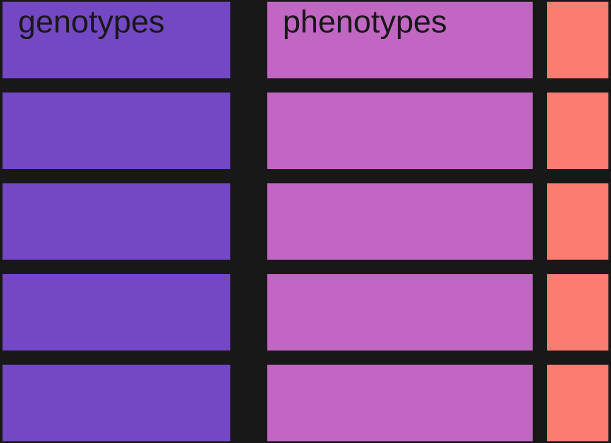
Challenges: organizing, automating



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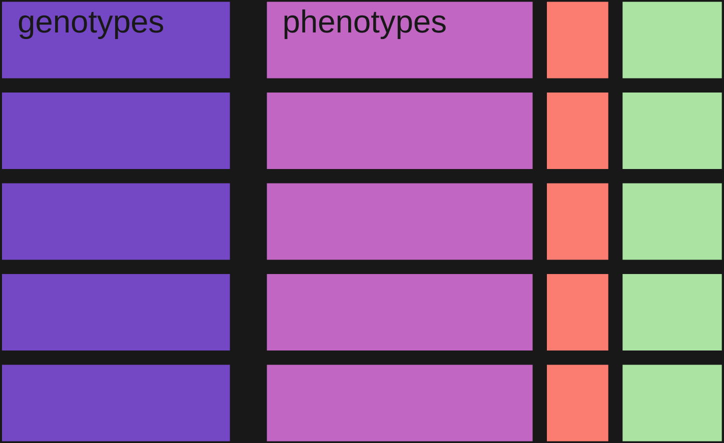

Challenges: metadata

What the heck is `"FAD_NAD SI 8.3_3.3G"`?

What was the question again?

ropenscilabs.github.io/miner_book

R/qtl2

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

`kbroman.org/qtl2`

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

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Laura Shannon
Quoc Tran
Aaron Wolen

NIH/NIGMS

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