

Quantitative Trait Loci (QTL)

Quantitative trait loci

Field

Generation	Procedure
1 Parents	A B
P = G + E	
2 F1	
F2	
F2 : 3	
Alternatives: BC1, RIL, DHL	

3 Laboratory

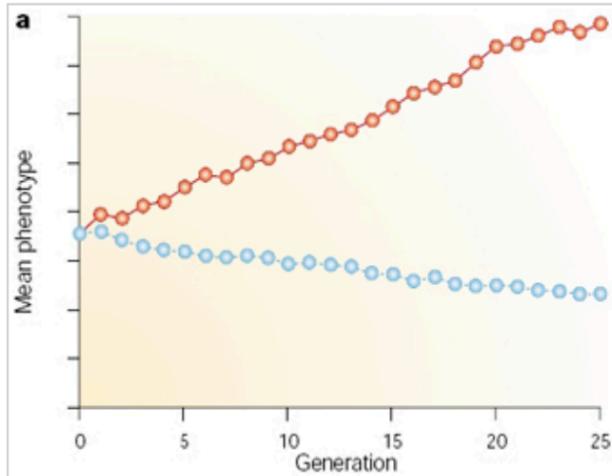
Marker #	Marker						PHT [cm]	
	1	2	3	4	5	.. M		
1	B	B	H	H	A	..	A	210
2	H	A	H	A	A	..	H	190
3	B	B	H	H	H	..	A	203
4	H	H	B	B	B	..	H	159
5	H	B	H	H	A	..	B	206
.
.
N	A	H	H	H	A	..	A	171

4 Office

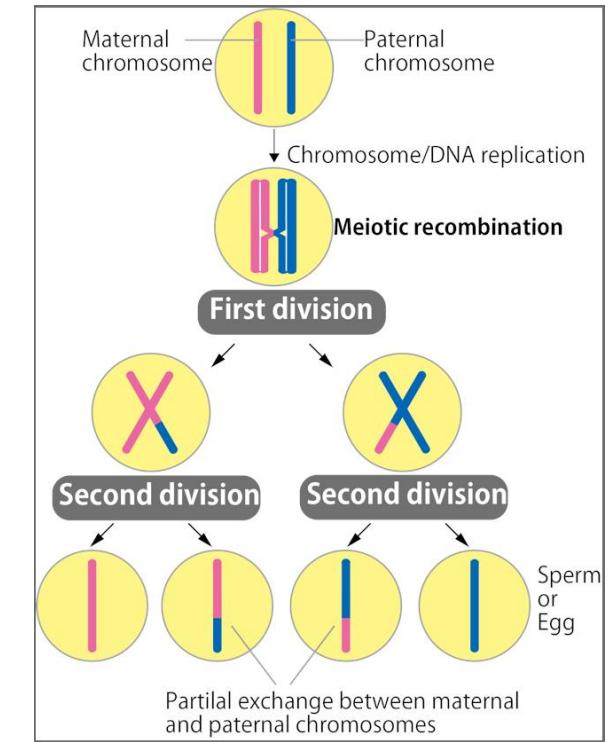
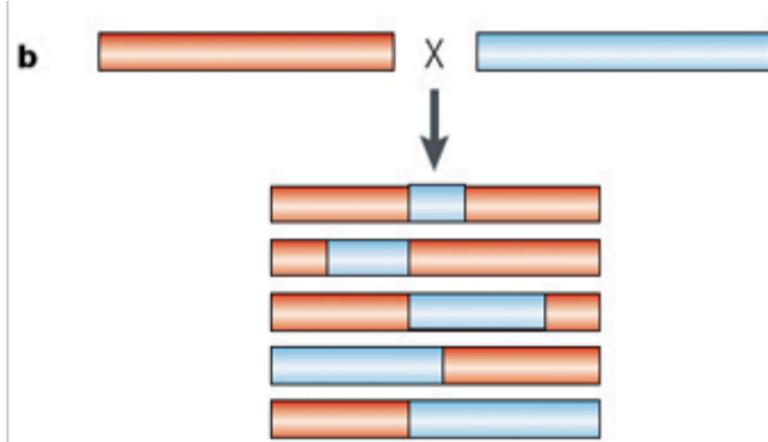
McLain 2014

1 – 2. Parents and Population

Select parents



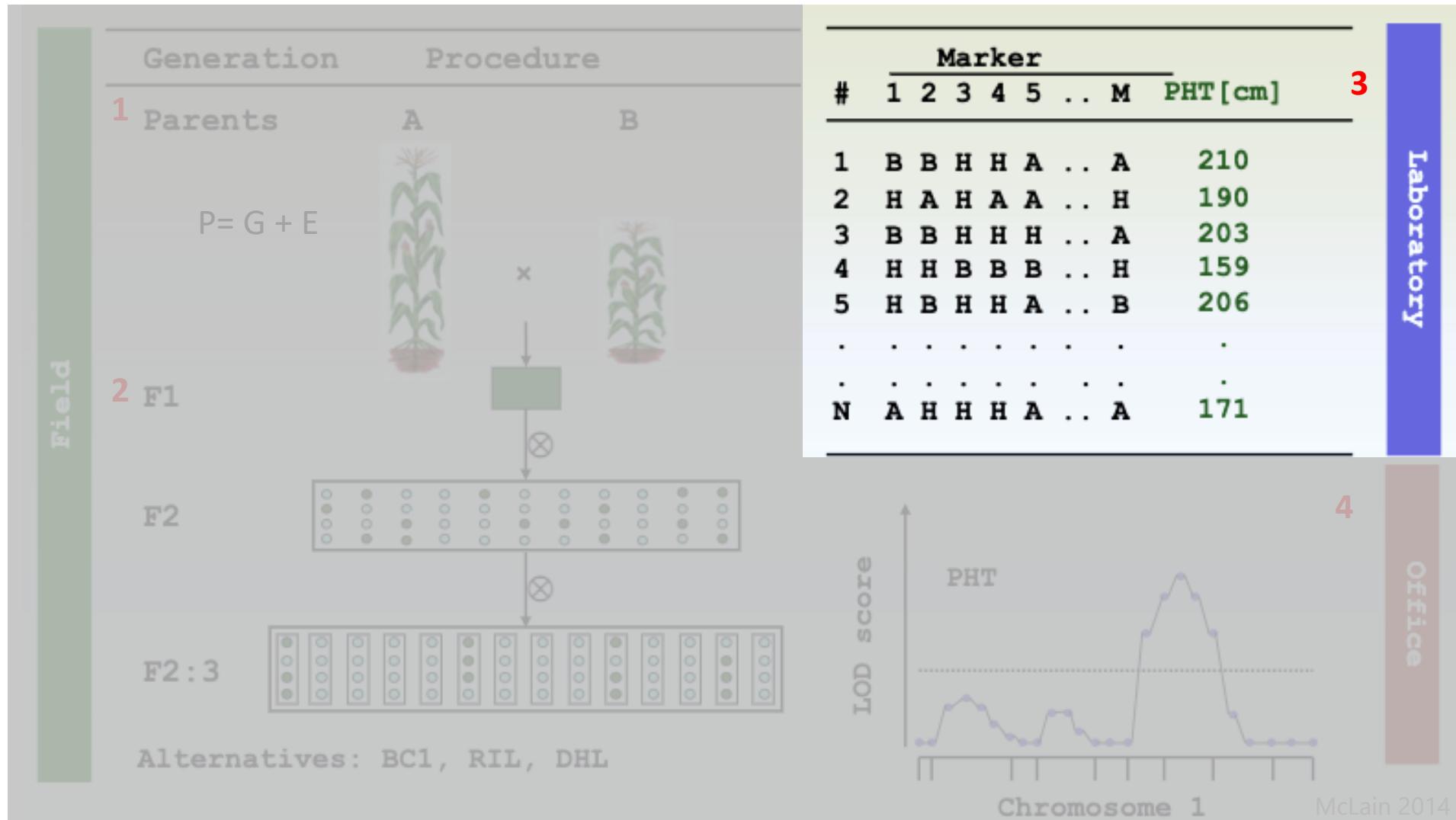
bi-parental populations



It was estimated that amongst nearly 50 eukaryotes belonging to different kingdoms, 80% of chromosome pairs have fewer than 3 crossovers (Fernandes et al., 2018).

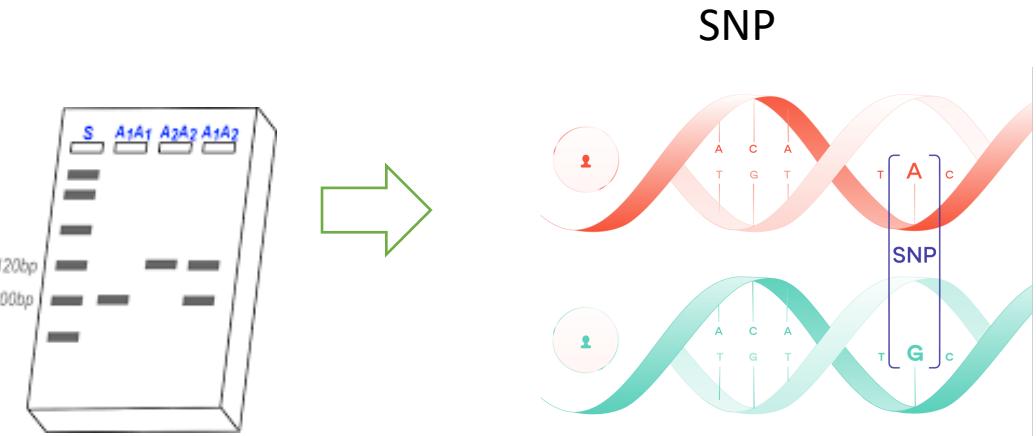
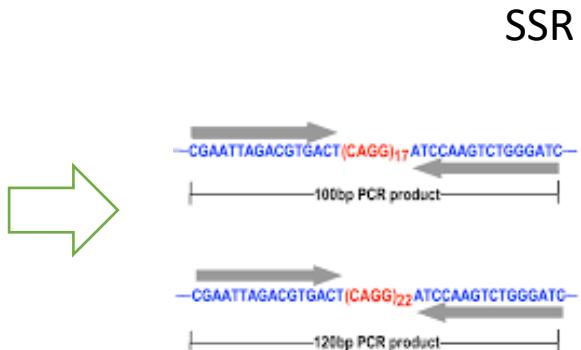
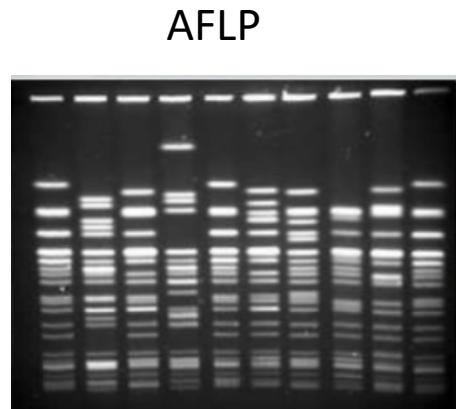
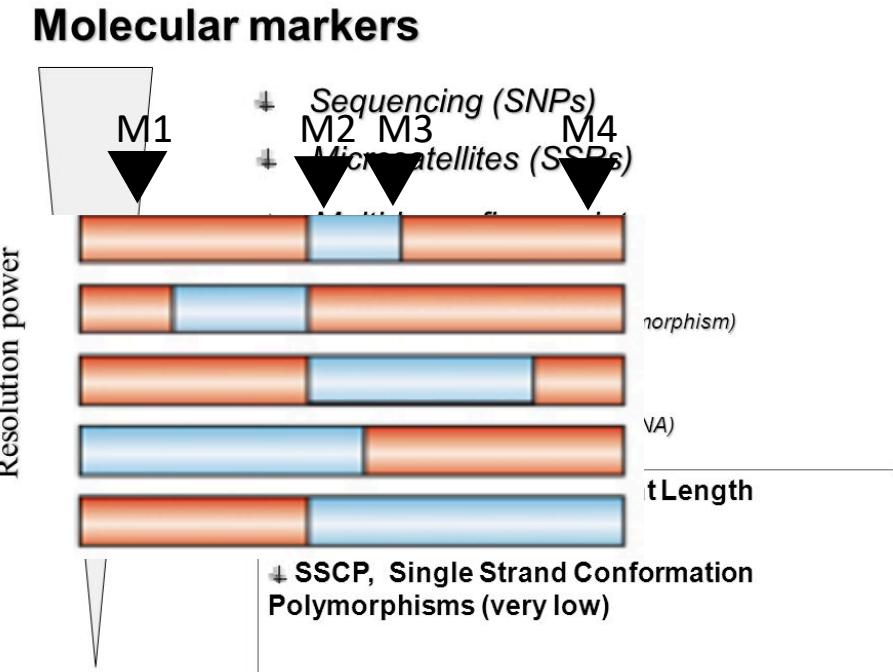


Quantitative trait loci



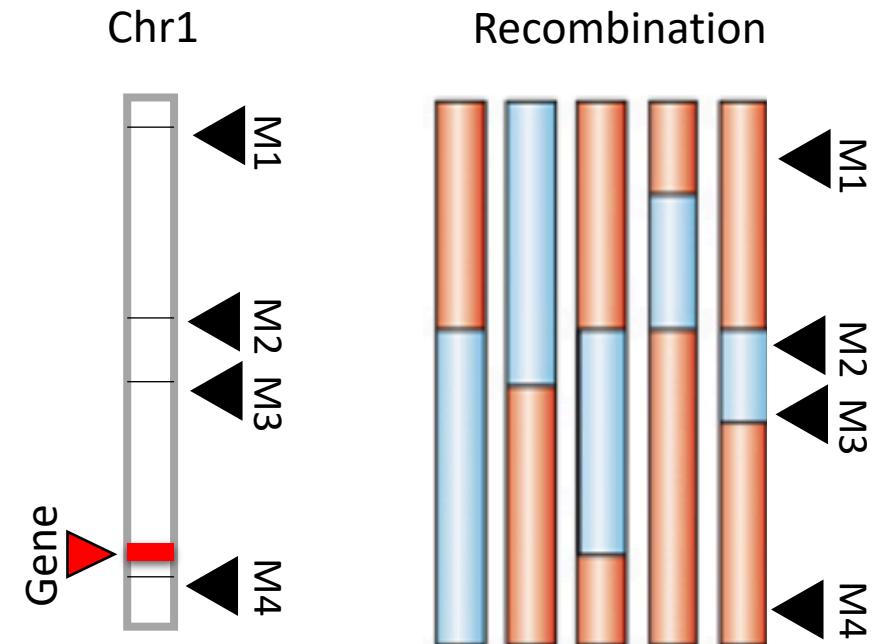
3. Markers and linkage maps

- In genetics, a **molecular marker** is a fragment of DNA that is associated with a certain location within the genome.



3. Markers and linkage maps

- Linkage maps indicate the position and relative genetic distances between markers along chromosomes.
- QTL mapping is based on the principle that genes and markers segregate via chromosome recombination during meiosis.



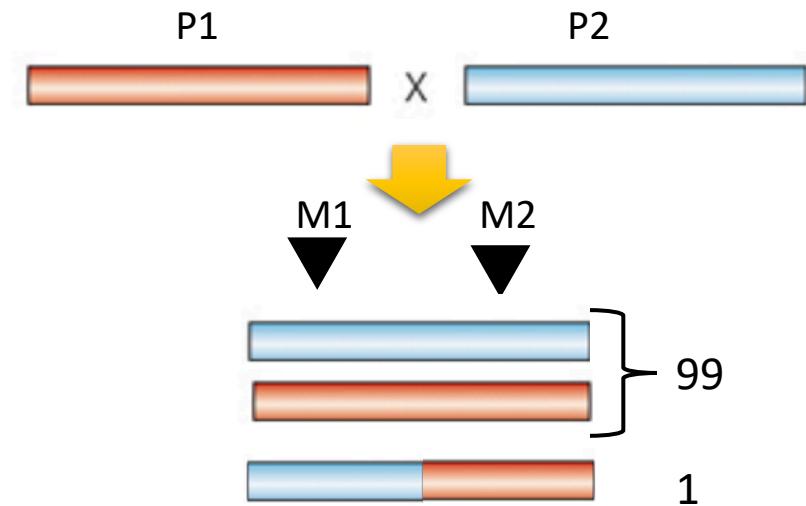
3. Markers and linkage maps

- The **frequency of recombinant genotypes** can be used to calculate the genetic distance between markers and their order in the genome (the lower the recombination between two markers, the closer they are situated on a chromosome).

$$n = 100$$

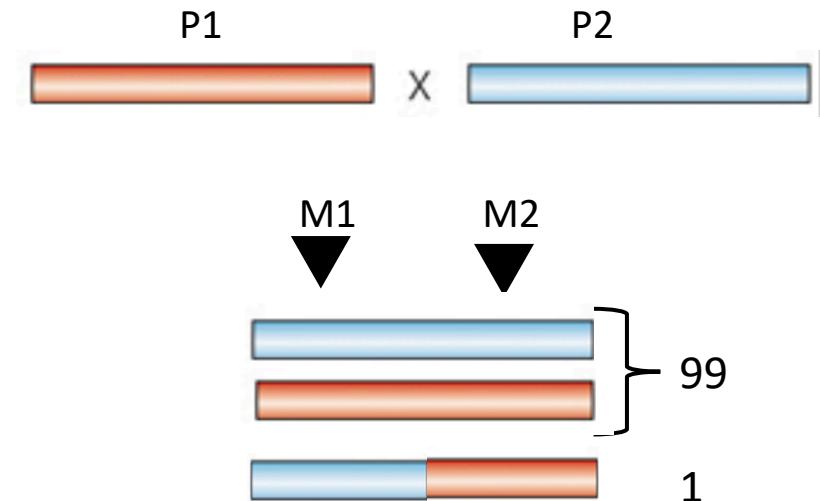
$$\text{Recombination} = 1$$

$$r = 1 / 100 = 0.01$$



3. Markers and linkage maps

- The **frequency of recombinant genotypes** can be used to **calculate the genetic distance** between markers and their order in the genome (the lower the recombination between two markers, the closer they are situated on a chromosome).
- A map unit of 1 centimorgan (cM) corresponds to a recombination frequency of 1%.



$$r = 1 / 100 = 0.01$$

Haldane

Assumes that interference is absent

$$m = -50 \ln(1 - 2r)$$

$$m = -50 \ln(1 - 2 * 0.01) = 1.01 \text{ cM}$$

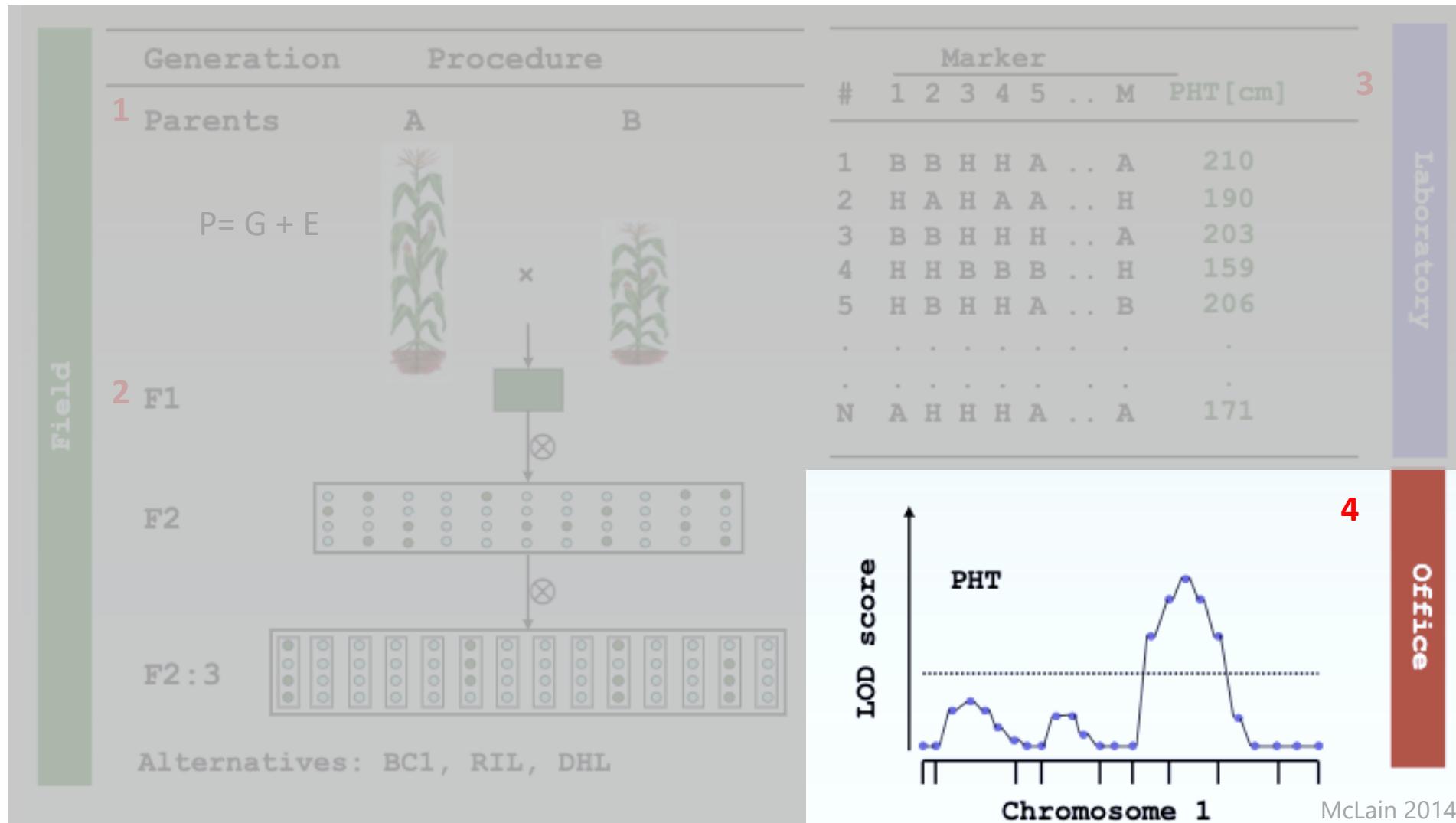
Kosambi

Assumes interference

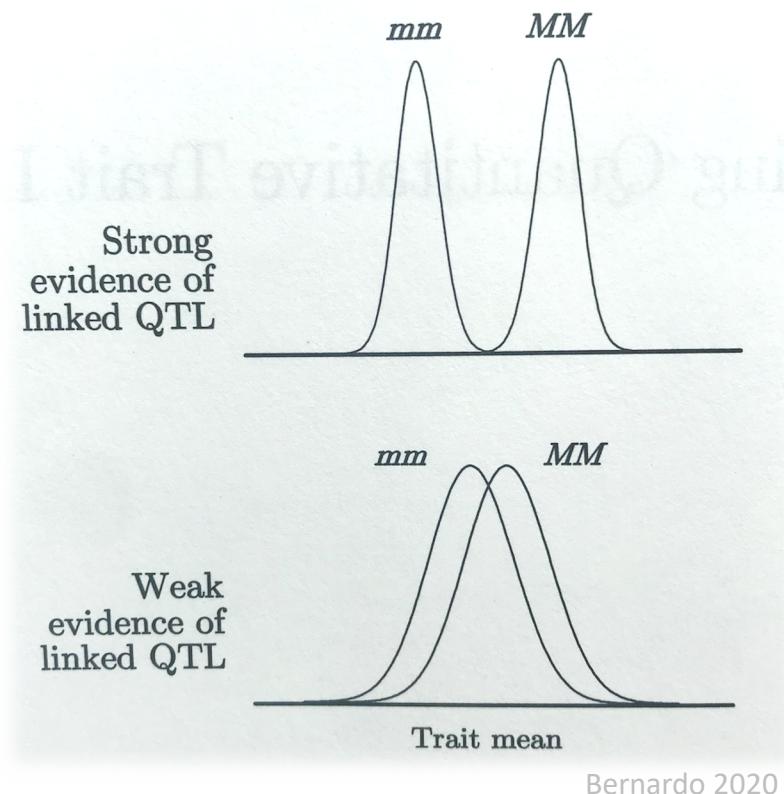
$$m = 25 \ln \left(\frac{1 + 2r}{1 - 2r} \right)$$

$$m = 25 \ln \left(\frac{1 + 2 * 0.01}{1 - 2 * 0.01} \right) = 0.99 \text{ cM}$$

Quantitative trait loci



4. QTL detection



$$P1 = 10 \text{ g}$$

$$P2 = 20 \text{ g}$$

F2:

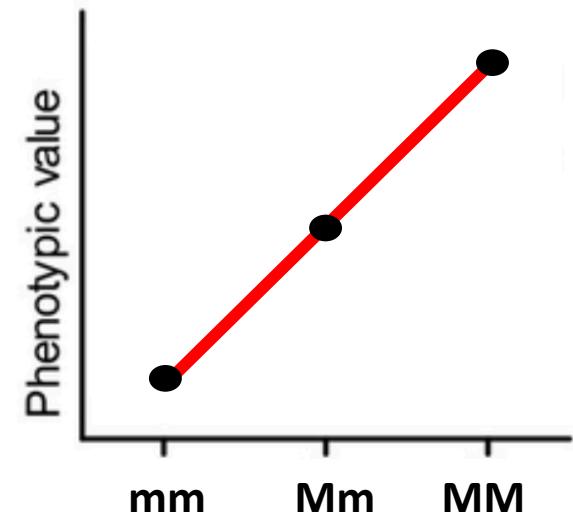
$$mm = 10 \text{ g}$$

$$Mm = 15 \text{ g}$$

$$MM = 20 \text{ g}$$

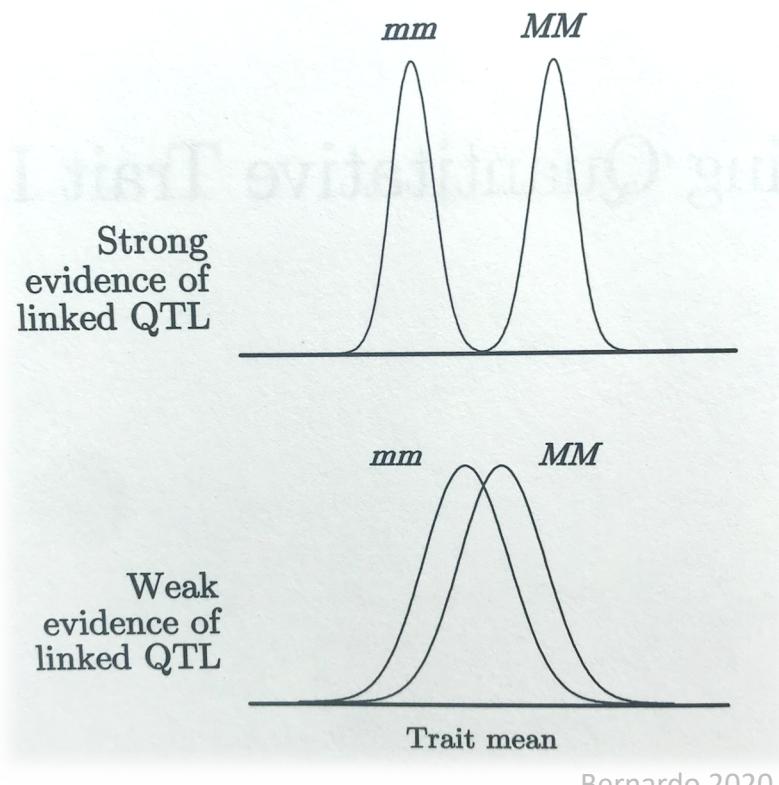
Data suggest
that dominance
is absent

$$\text{Midparent value} = \frac{1}{2} (10+20) = 15$$



$$(\overline{MM} - \overline{mm}) = 2a(1 - 2r)$$

4. QTL detection



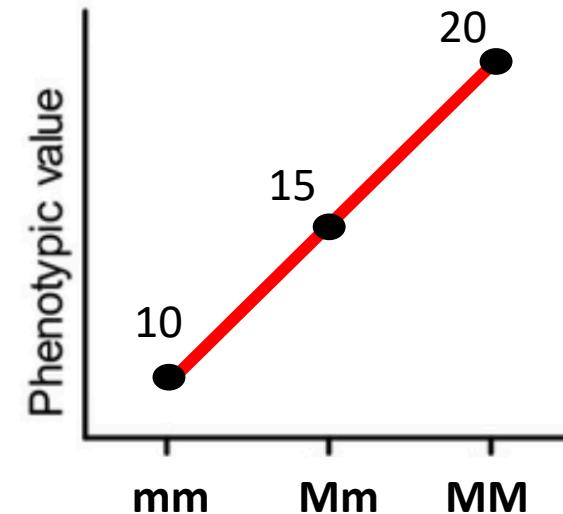
$$P1 = 10 \text{ g}$$
$$P2 = 20 \text{ g}$$

F2:

$$\text{mm} = 10 \text{ g}$$
$$\text{Mm} = 15 \text{ g}$$
$$\text{MM} = 20 \text{ g}$$

$$a = 5$$

$$(\overline{MM} - \overline{mm}) = 2a(1 - 2r)$$



4. QTL detection

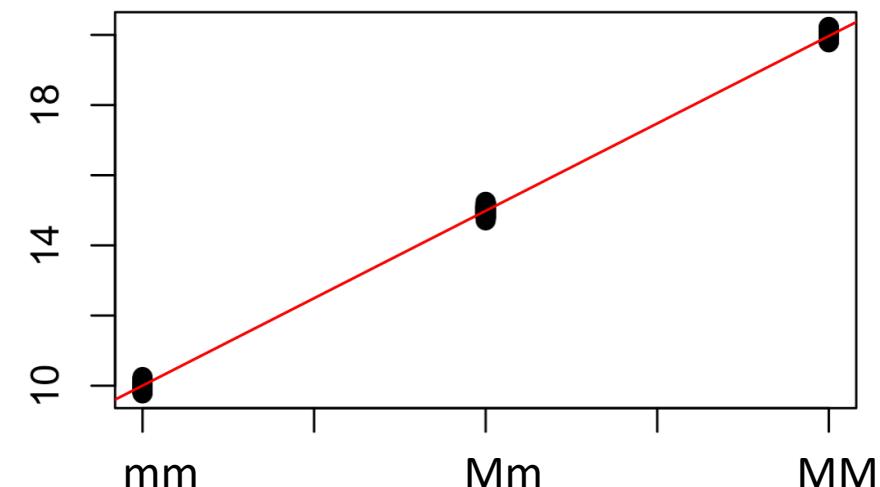
The statistical methods used for single-marker analysis include t-tests, analysis of variance (ANOVA) and linear regression.

$$t = \frac{\overline{MM} - \overline{mm}}{\sqrt{\frac{\hat{V}(MM)}{N} + \frac{\hat{V}(mm)}{N}}}$$

T-test: compare the mean of 2 groups. To compare 3 or more groups, one must use an ANOVA.

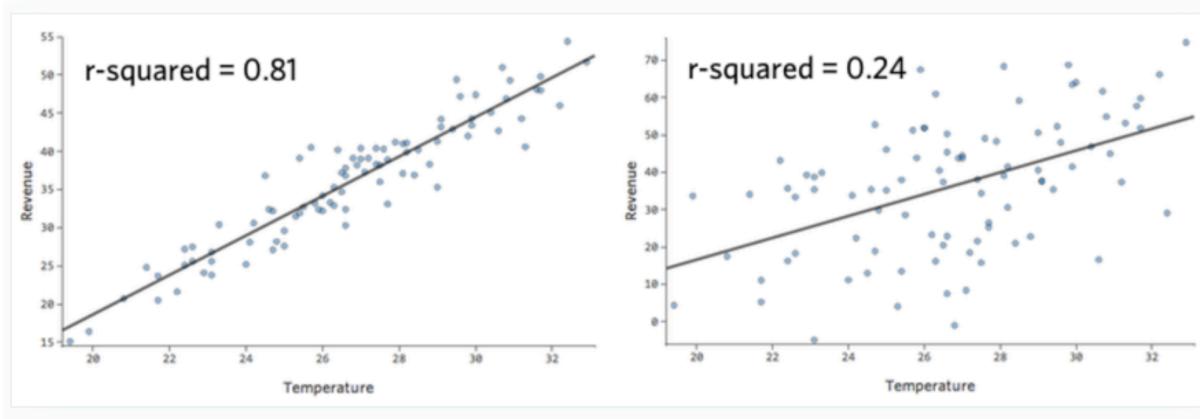
$$y_i = b_0 + b_1 x + e$$

Estimated (or predicted) y value
Estimate of the regression intercept
Estimate of the regression slope
Independent variable
Error term



4. QTL detection

The statistical methods used for single-marker analysis include t-tests, analysis of variance (ANOVA) and linear regression.

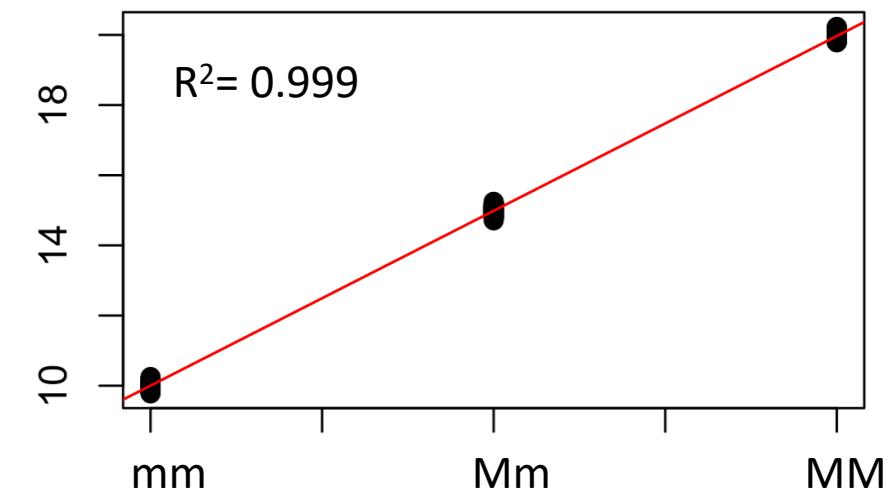
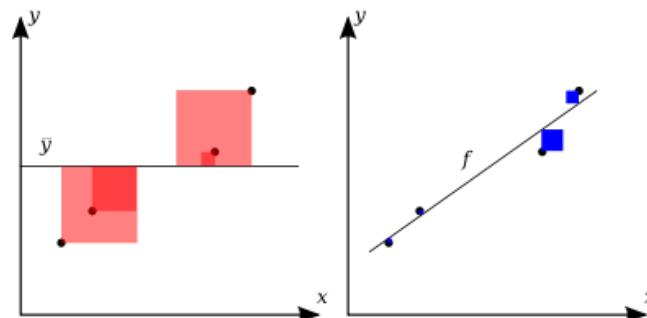


$$y_i = b_0 + b_1 x + e$$

Annotations pointing to parts of the equation:

- Estimated (or predicted) y value
- Estimate of the regression intercept
- Estimate of the regression slope
- Independent variable
- Error term

$$R^2 = 1 - \frac{SS_{\text{res}}}{SS_{\text{tot}}}$$



4. QTL detection

- **Simple Interval Mapping (SIM)**

SIM uses adjacent markers to compensate the recombination between markers.

- **Composite Interval Mapping (CIM)**

CIM uses interval mapping and includes additional genetic markers in the statistical model in addition to an adjacent pair of linked markers for interval mapping.

The main advantage of CIM is that it is more precise and effective at mapping QTLs compared to single-point analysis and interval mapping.

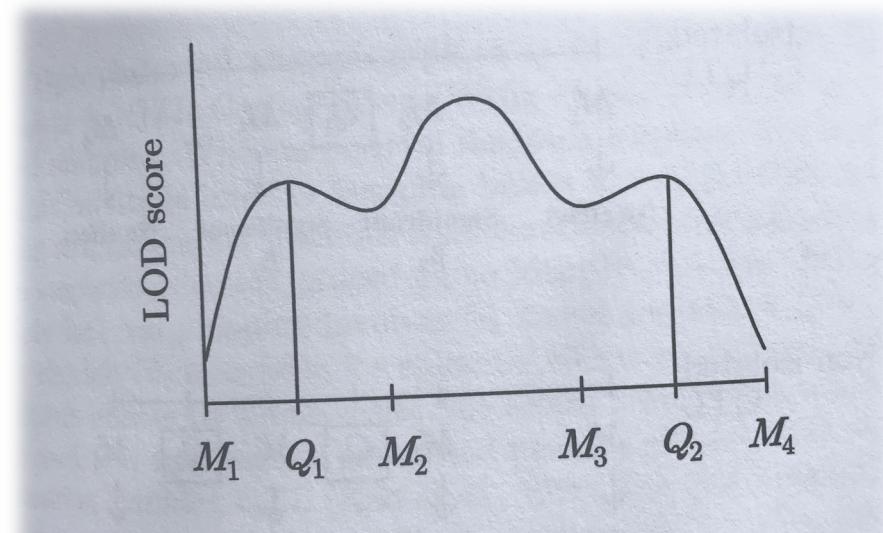


FIGURE 5.3. Ghost QTL from interval mapping.

Bernardo 2020

Real data....

Theoretical and Applied Genetics
<https://doi.org/10.1007/s00122-020-03598-w>

ORIGINAL ARTICLE



QTL analysis of cooking time and quality traits in dry bean (*Phaseolus vulgaris* L.)

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Results

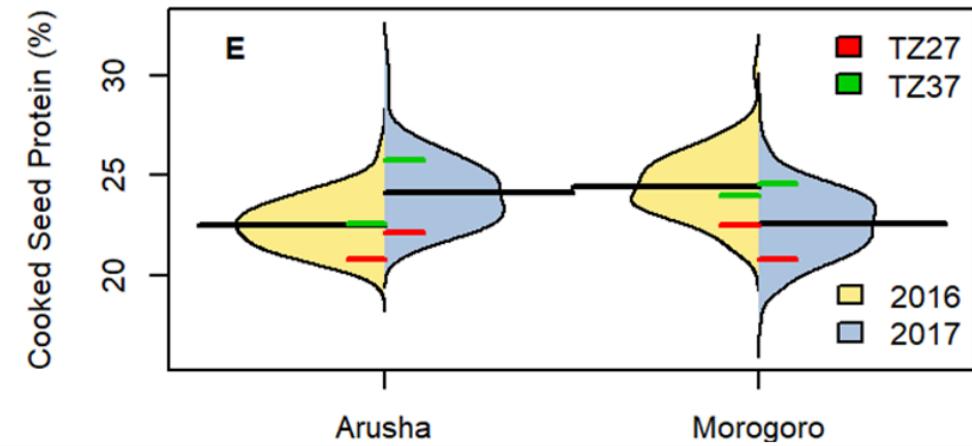
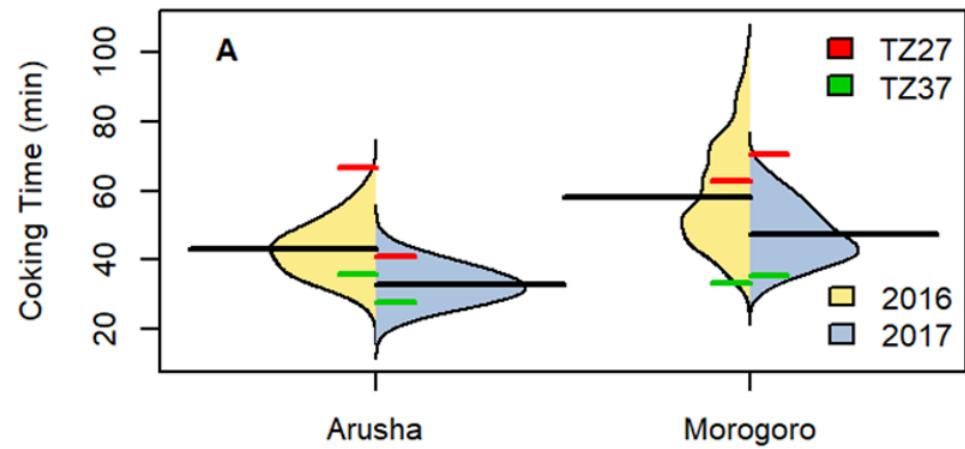


Figure S2. Bean plots represent the distribution of A) cooking time, and E) cooked seed protein concentration for beans grown in Arusha and Morogoro in Tanzania in 2016 and 2017. Frequencies were determined by using the adjusted means calculated with the RCBD with 2 replications. Lines red and green indicate the values for TZ27 and TZ37 respectively.

Results

Composite interval mapping

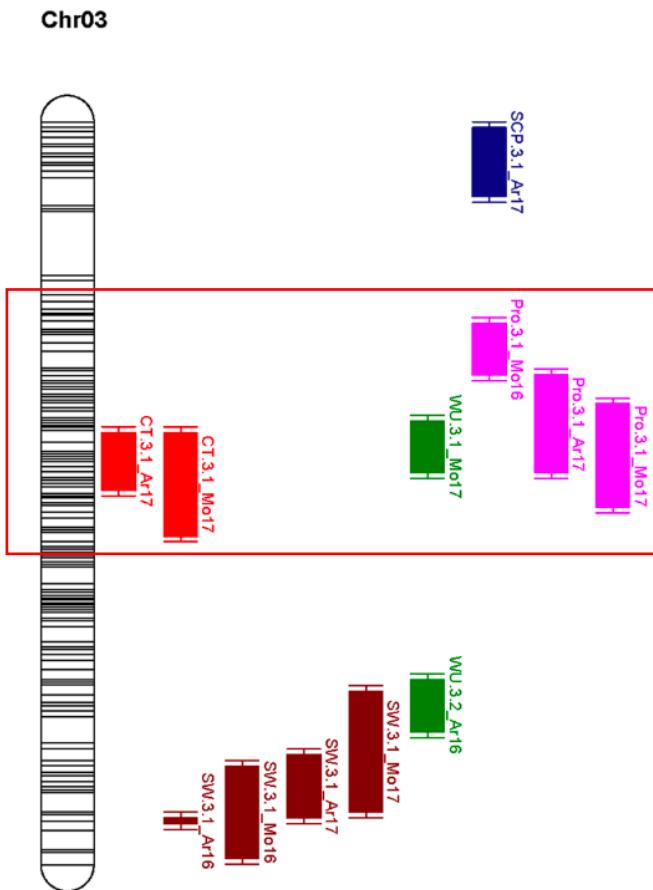


Table 4 Linkage map information of the TT-RIL population of 146 lines

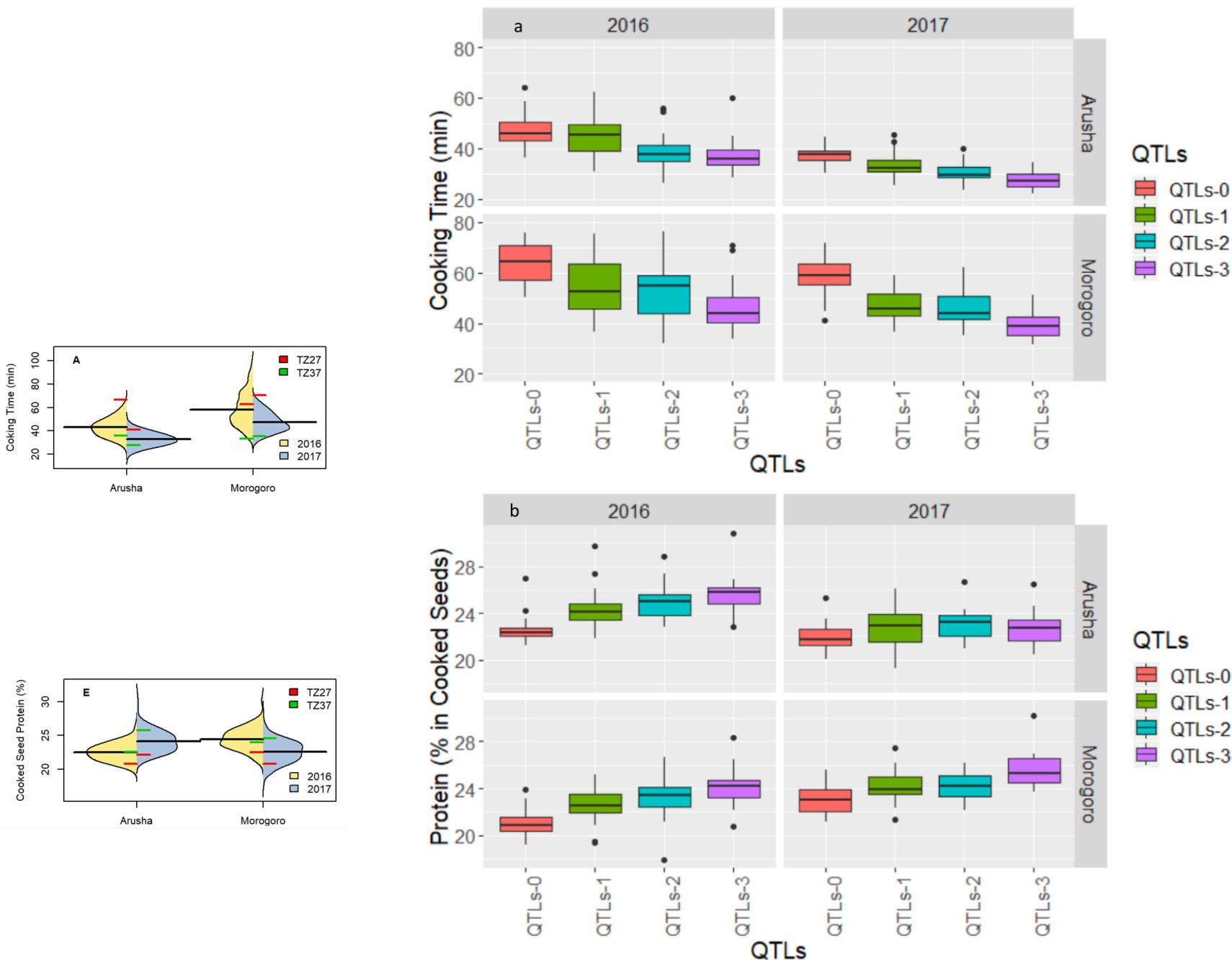
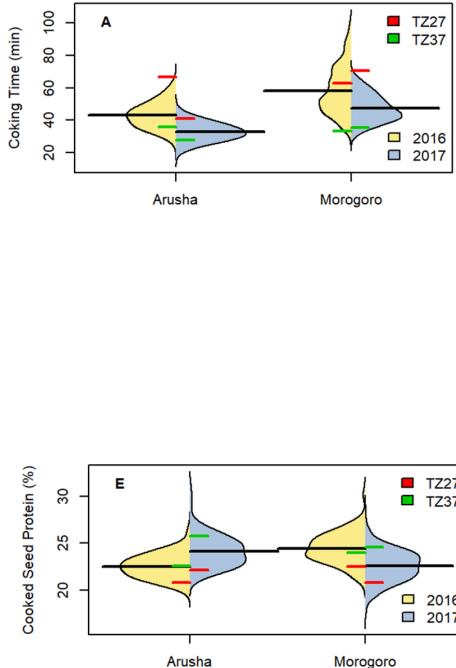
Chromosome #	# of markers	Chromosome size (cM)	Marker density (cM/marker)
1	48	63.7	1.33
2	303	74.4	0.25
3	238	129.1	0.54
4	217	89.5	0.41
5	184	68.7	0.37
6	217	110.6	0.51
7	79	86	1.09
8	200	114.2	0.57
9	115	80.7	0.7
10	117	82.1	0.7
11	233	112.7	0.48
Total	1951	1011.70	0.52

In total, 1951 markers were mapped resulting in an overall genome size of 1011.7 cM. Marker density represented the average number of cM between markers. Marker density varied by linkage group with the average coverage across the entire genome being 1 marker every 0.52 cM

Results

➤ Haplotype Analysis

Arusha				
	2016		2017	
QTLs	*CT-(SD)	Genotypes	*CT-(SD)	Genotypes
QTLs-0	47.6 (5.9)	29	37.5 (3.0)	29
QTLs-1	45.2 (7.6)	40	33.3 (4.6)	44
QTLs-2	38.8 (6.5)	30	30.4 (3.7)	32
QTLs-3	37.1 (6.6)	21	27.4 (3.4)	23
Morogoro				
QTLs	63.4 (8.8)	12	58.6 (6.7)	26
QTLs	56.9 (15.3)	37	47.1 (5.8)	44
QTLs	52.4 (12.0)	27	45.6 (6.6)	31
QTLs	46.7 (10.2)	21	39.6 (5.6)	23
Arusha				
QTLs	*Pro-(SD)	Genotypes	*Pro-(SD)	Genotypes
QTLs-0	22.5 (1)	32	22.0 (1.1)	29
QTLs-1	24.2 (1.4)	49	22.7 (1.6)	42
QTLs-2	25.0 (1.4)	28	23.1 (1.3)	21
QTLs-3	25.6 (1.5)	27	22.7 (1.4)	23
Morogoro				
QTLs	21.0 (1.1)	31	23.1 (1.3)	22
QTLs	22.5 (1.2)	49	24.2 (1.3)	24
QTLs	23.2 (1.7)	27	24.3 (1.3)	14
QTLs	24.1 (1.5)	27	25.6 (1.7)	15



Results

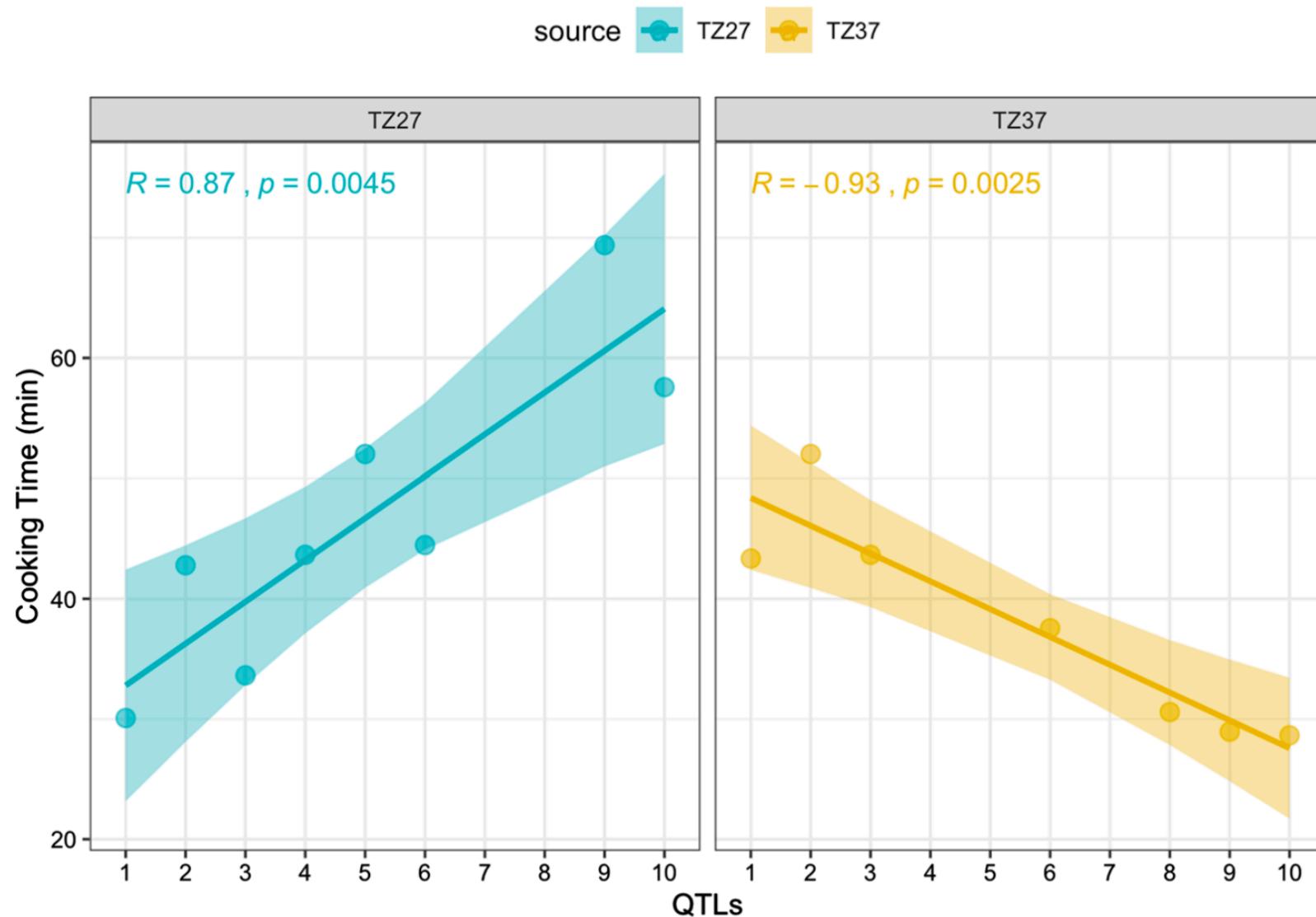
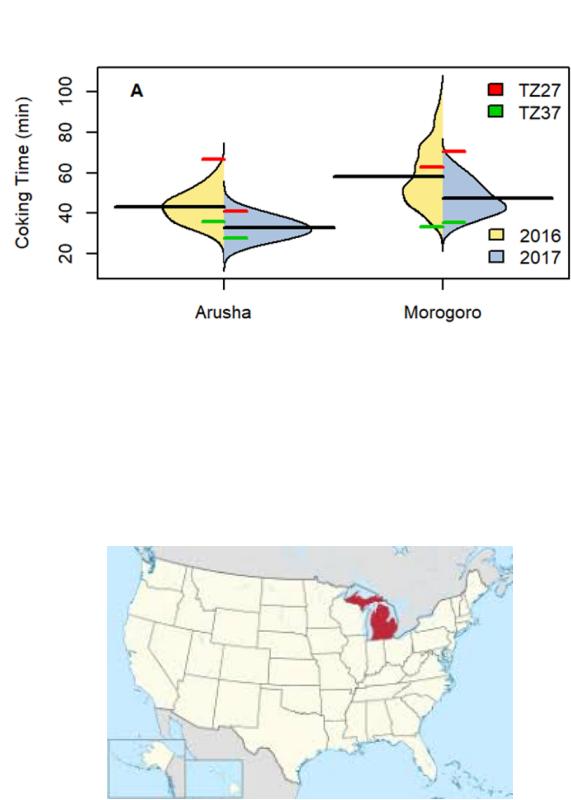


Fig. 4 Phenotypic effect of 10 cooking time QTL in 30 selected RILs grown in Michigan in 2018 on average cooking times of lines carrying 1–10 of the QTL regions from either the fast cooking parental source (TZ-37) or the slow cooking parental source (TZ-27).

Conclusions

- The three most robust QTL for cooking time explained up to 40% of the variation for the trait, and genotypes with all three QTL cooked 11–26 min faster than genotypes without any fast cooking QTL. In addition, the three most robust QTL for cooking time co-localized with QTL for increased cooked bean protein concentration, suggesting an added benefit to using these QTL in breeding. The validation data also suggested these QTL would function in beans grown in Michigan.