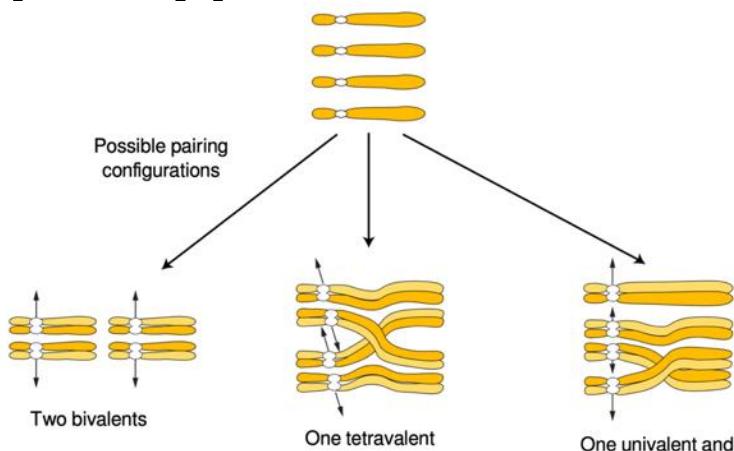
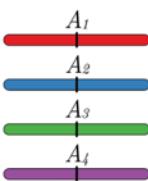


Gamete formation in autopolyploids*

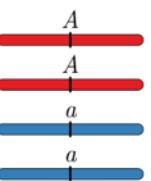


Griffiths et al. (2004)

Multiallelic



Biallelic



A_1A_2

A_1A_3

A_1A_4

A_2A_3

A_2A_4

A_3A_4

$A A$

$A a$

$A a$

$A a$

$A a$

$a a$

Number of possible gamtes considering one locus with no double-reduction in one and two parents

Ploidy	$(\frac{p}{2})$	$(\frac{p}{2})^2$
4	6	36
6	20	400
8	70	4900
10	252	63504
12	924	853776
14	3432	11778624
16	12870	165636900

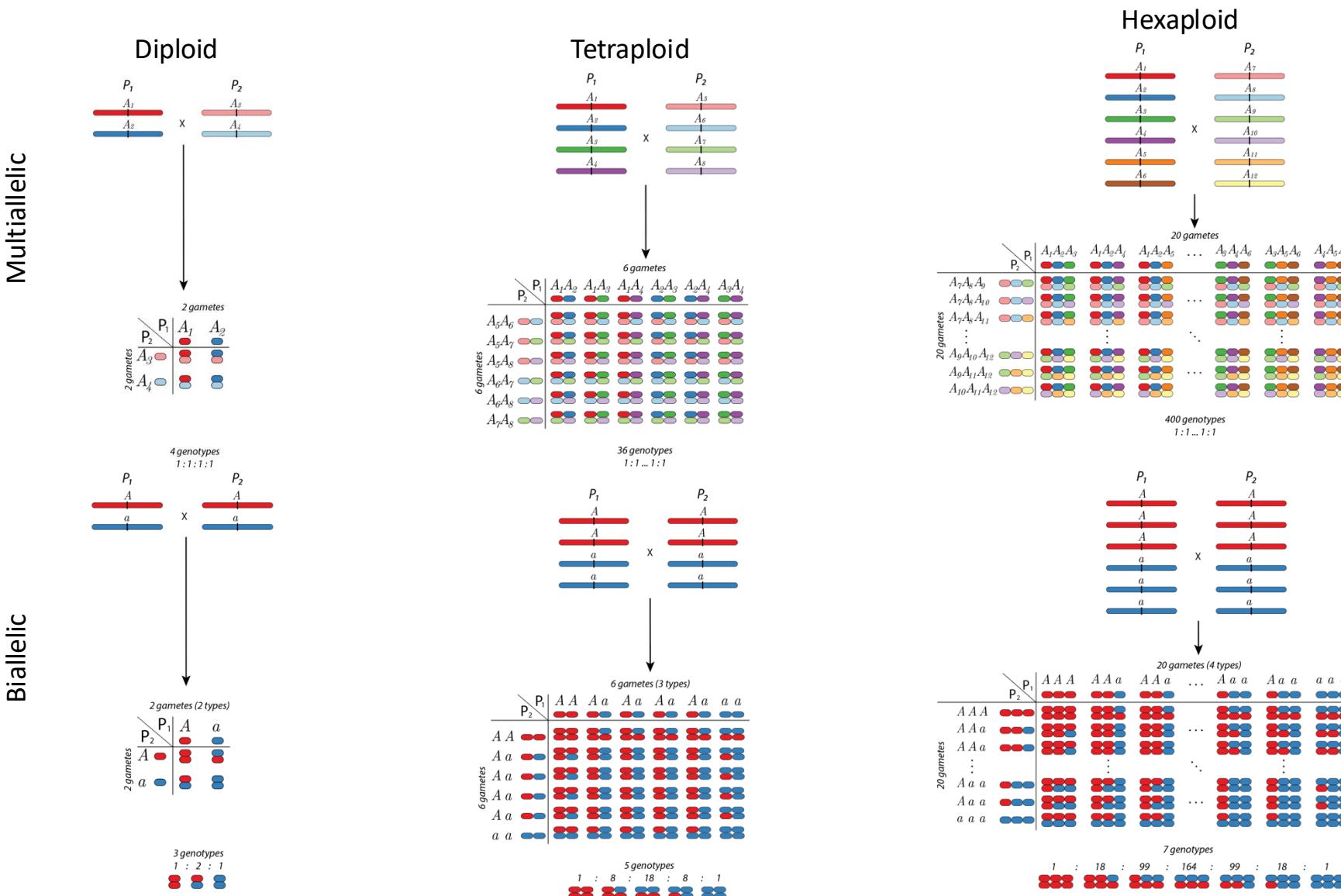
```
> ploidy <- c(2,4,6,8)
> choose(ploidy, ploidy/2)
[1] 2 6 20 70
```

If chromosome arms pair at random, bivalents occur when there is pairing in both arms of two chromosomes instead of a switch in partners between one of these chromosomes and a third chromosome. Studies on induced autotetraploids in different plant species indicate that there are more bivalents and fewer quadrivalents at metaphase I than would be expected if partner exchange occurs without restrictions. There also is evidence that the proportion of bivalents to multivalents increases over a span of generations. The suggested explanations for the reduction in quadrivalents focus on the pairing mechanism during the zygotene and pachytene stages of meiosis and the locations of chiasmata.

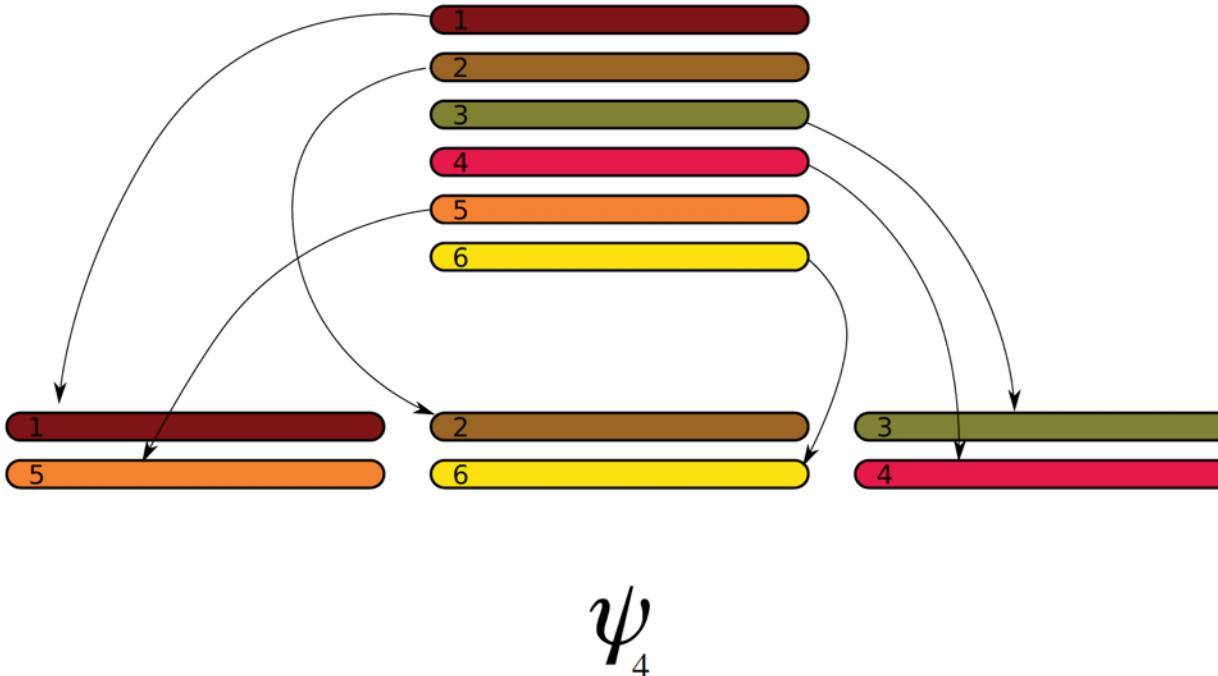
Apples et al. (1998)

*random pairing and no double reduction

How genetic information is transmitted?



Gamete formation in polyploids*

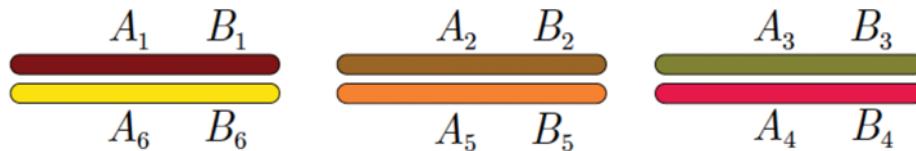


In this case: 15 possible configurations. For any ploidy level p

$$\frac{1}{\frac{p}{2}!} \prod_{i=1}^{\frac{p}{2}} \binom{2i}{2}$$

<https://doi.org/10.1534/g3.119.40037>

Expected gametic frequency given a bivalent configuration



$$\begin{bmatrix} \frac{1-r}{2} & \frac{r}{2} \\ \frac{r}{2} & \frac{1-r}{2} \end{bmatrix} \otimes \begin{bmatrix} \frac{1-r}{2} & \frac{r}{2} \\ \frac{r}{2} & \frac{1-r}{2} \end{bmatrix} \otimes \begin{bmatrix} \frac{1-r}{2} & \frac{r}{2} \\ \frac{r}{2} & \frac{1-r}{2} \end{bmatrix}$$

In general:

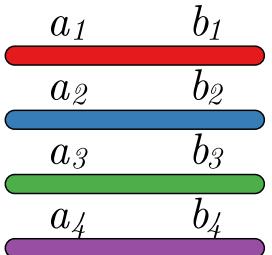
$$\mathbf{V}_1 \otimes \mathbf{V}_2 \otimes \cdots \otimes \mathbf{V}_{\frac{p}{2}}$$

All elements of this product are of the form

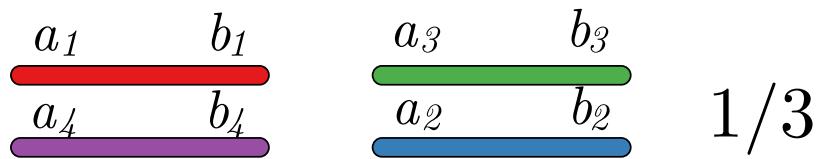
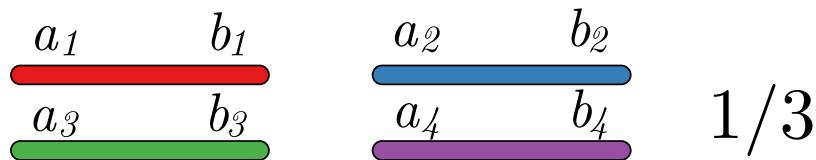
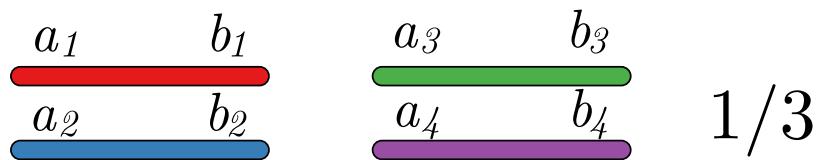
$$\Pr(G_A, G_B \mid \psi_j, r) = \frac{(1-r)^{(\frac{p}{2}-l)} p^l}{2^{\frac{p}{2}}}$$

l : known number of recombinant bivalents between loci A and B

Recombination Fraction - autotetraploid



Bivalent pairing at meiosis



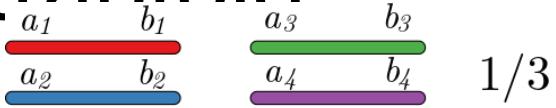
Recombination Fraction - autotetraploid

First bivalent pairing at meiosis

$$\begin{bmatrix}
 \Pr(a_1, b_1) & \Pr(a_2, b_1) \\
 \Pr(a_1, b_2) & \Pr(a_2, b_2)
 \end{bmatrix} \otimes \begin{bmatrix}
 \Pr(a_3, b_3) & \Pr(a_4, b_3) \\
 \Pr(a_3, b_4) & \Pr(a_4, b_4)
 \end{bmatrix} = \begin{bmatrix}
 \Pr(a_1, b_1, a_3, b_3) & \Pr(a_1, b_1, a_4, b_3) & \Pr(a_2, b_2, a_3, b_3) & \Pr(a_2, b_1, a_4, b_3) \\
 \Pr(a_1, b_1, a_3, b_4) & \Pr(a_1, b_1, a_4, b_4) & \Pr(a_2, b_2, a_3, b_4) & \Pr(a_2, b_1, a_4, b_4) \\
 \Pr(a_1, b_2, a_3, b_3) & \Pr(a_1, b_2, a_4, b_3) & \Pr(a_2, b_2, a_3, b_3) & \Pr(a_2, b_2, a_4, b_3) \\
 \Pr(a_1, b_2, a_3, b_4) & \Pr(a_1, b_2, a_4, b_4) & \Pr(a_2, b_2, a_3, b_4) & \Pr(a_2, b_2, a_4, b_4)
 \end{bmatrix}$$

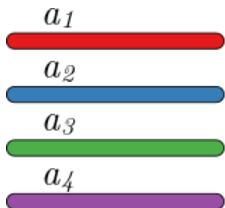
$$\begin{bmatrix}
 \frac{1-r}{2} & \frac{r}{2} \\
 \frac{r}{2} & \frac{1-r}{2}
 \end{bmatrix} \otimes \begin{bmatrix}
 \frac{1-r}{2} & \frac{r}{2} \\
 \frac{r}{2} & \frac{1-r}{2}
 \end{bmatrix} = \begin{bmatrix}
 \frac{(1-r)^2}{4} & \frac{(1-r)r}{4} & \frac{(1-r)r}{4} & \frac{r^2}{4} \\
 \frac{(1-r)r}{4} & \frac{(1-r)^2}{4} & \frac{r^2}{4} & \frac{(1-r)r}{4} \\
 \frac{(1-r)r}{4} & \frac{r^2}{4} & \frac{(1-r)^2}{4} & \frac{(1-r)r}{4} \\
 \frac{r^2}{4} & \frac{(1-r)r}{4} & \frac{(1-r)r}{4} & \frac{(1-r)^2}{4}
 \end{bmatrix}$$

Recombination Fraction - autotetraploid



1/3

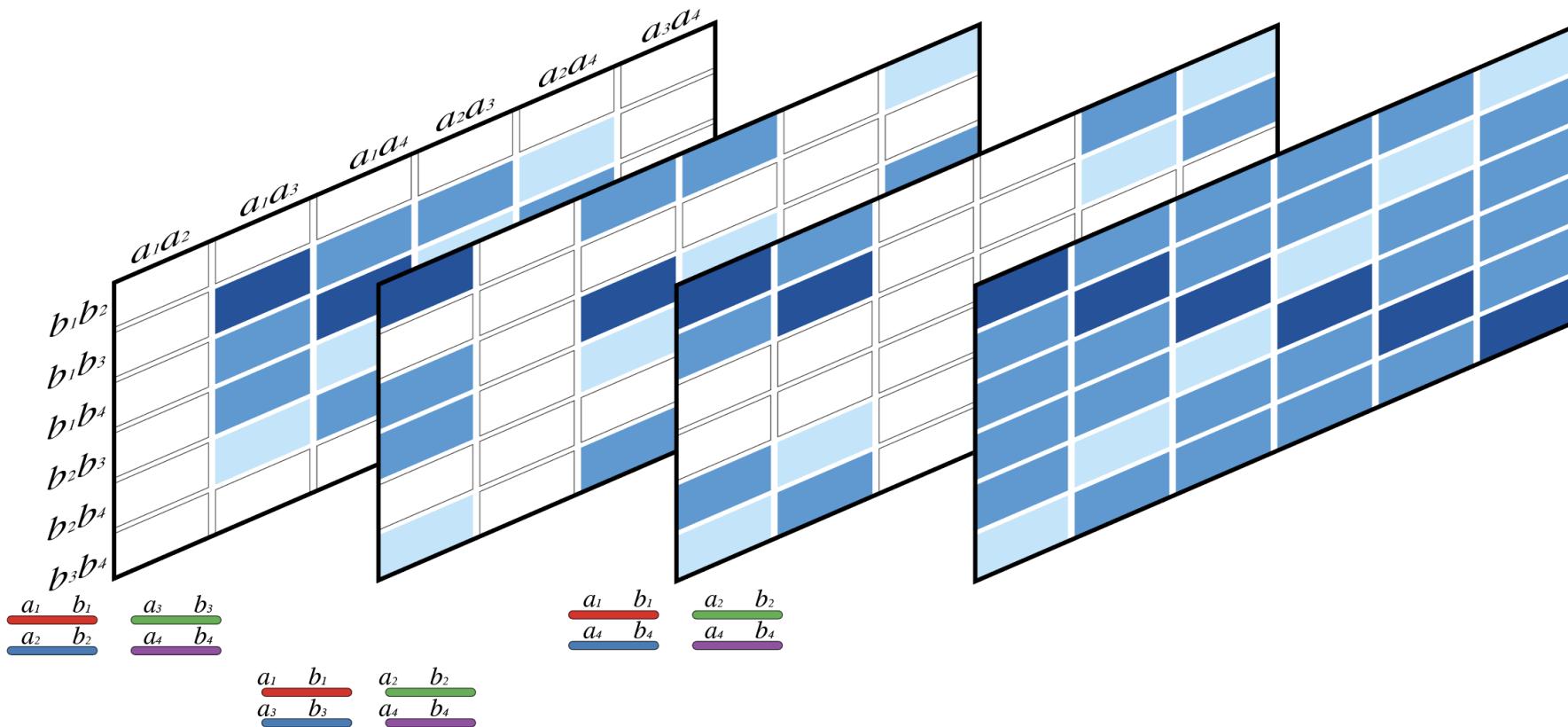
	$a_1 a_3$	$a_1 a_4$	$a_2 a_3$	$a_2 a_4$
$b_1 b_3$	$\frac{(1-r)^2}{4}$	$\frac{(1-r)r}{4}$	$\frac{(1-r)r}{4}$	$\frac{r^2}{4}$
$b_1 b_4$	$\frac{(1-r)r}{4}$	$\frac{(1-r)^2}{4}$	$\frac{r^2}{4}$	$\frac{(1-r)r}{4}$
$b_2 b_3$	$\frac{(1-r)r}{4}$	$\frac{r^2}{4}$	$\frac{(1-r)^2}{4}$	$\frac{(1-r)r}{4}$
$b_2 b_4$	$\frac{r^2}{4}$	$\frac{(1-r)r}{4}$	$\frac{(1-r)r}{4}$	$\frac{(1-r)^2}{4}$



$a_1 a_2$
 $a_1 a_3$
 $a_1 a_4$
 $a_2 a_3$
 $a_2 a_4$
 $a_3 a_4$



Recombination Fraction - autotetraploid



$$\Pr(G_A, G_B \mid r) = \sum_j \Pr(G_A, G_B \mid \psi_j, r) \Pr(\psi_j)$$

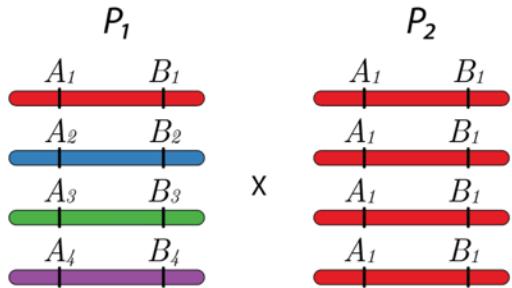
Recombination Fraction - autotetraploid

	a_1a_2	a_1a_3	a_1a_4	a_2a_3	a_2a_4	a_3a_4
b_1b_2	$\frac{(1-r)^2}{6}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{r^2}{6}$
b_1b_3	$\frac{(1-r)r}{12}$	$\frac{(1-r)^2}{6}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{r^2}{6}$	$\frac{(1-r)r}{12}$
b_1b_4	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)^2}{6}$	$\frac{r^2}{6}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$
b_2b_3	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{r^2}{6}$	$\frac{(1-r)^2}{6}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$
b_2b_4	$\frac{(1-r)r}{12}$	$\frac{r^2}{6}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)^2}{6}$	$\frac{(1-r)r}{12}$
b_3b_4	$\frac{r^2}{6}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)^2}{6}$

$$\begin{aligned}
 L &= \Pr(\text{model} | \text{data}) = \prod_n \Pr(\text{loc}_B, \text{loc}_A) \\
 &= \left[\frac{(1-r)^2}{6} \right]^{n_1} \left[\frac{(1-r)r}{12} \right]^{n_2} \left[\frac{r^2}{6} \right]^{n_3}
 \end{aligned}$$

Recombination Fraction – autotetraploid

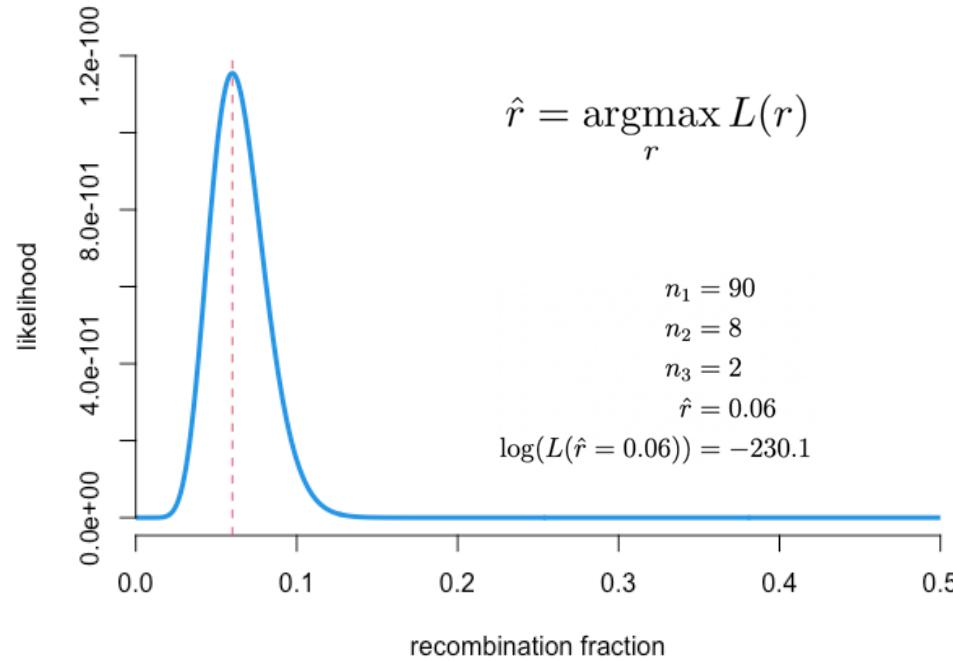
Fully informative marker



$$L(r) = \left[\frac{(1-r)^2}{6} \right]^{n_1} \left[\frac{(1-r)r}{12} \right]^{n_2} \left[\frac{r^2}{6} \right]^{n_3}$$

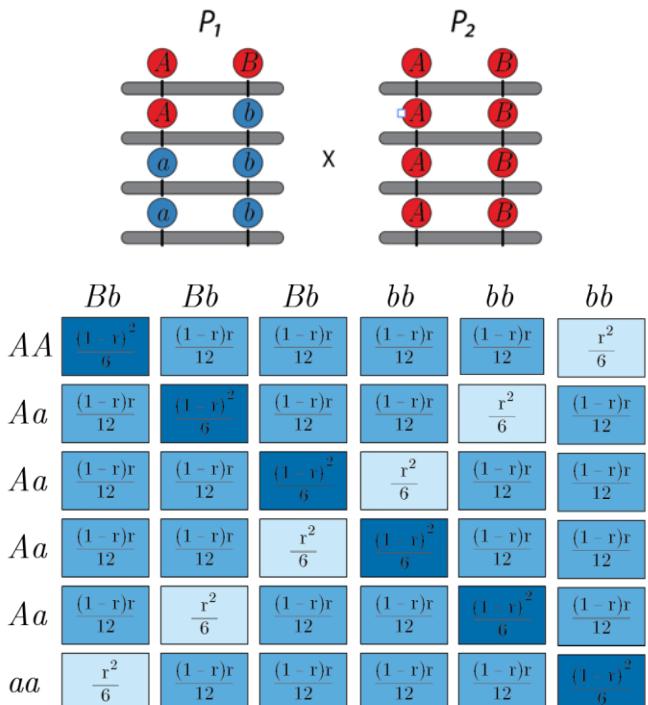
$\Pr(G_A, G_B \mid r)$

	B_1B_2	B_1B_3	B_1B_4	B_2B_3	B_2B_4	B_3B_4
A_1A_2	$\frac{(1-r)^2}{6}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{r^2}{6}$
A_1A_3	$\frac{(1-r)r}{12}$	$\frac{(1-r)^2}{6}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{r^2}{6}$	$\frac{(1-r)r}{12}$
A_1A_4	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)^2}{6}$	$\frac{r^2}{6}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$
A_2A_3	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{r^2}{6}$	$\frac{(1-r)^2}{6}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$
A_2A_4	$\frac{(1-r)r}{12}$	$\frac{r^2}{6}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)^2}{6}$	$\frac{(1-r)r}{12}$
A_3A_4	$\frac{r^2}{6}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)r}{12}$	$\frac{(1-r)^2}{6}$



Recombination Fraction – autotetraploid

Partially informative marker – Duplex/simplex – Association

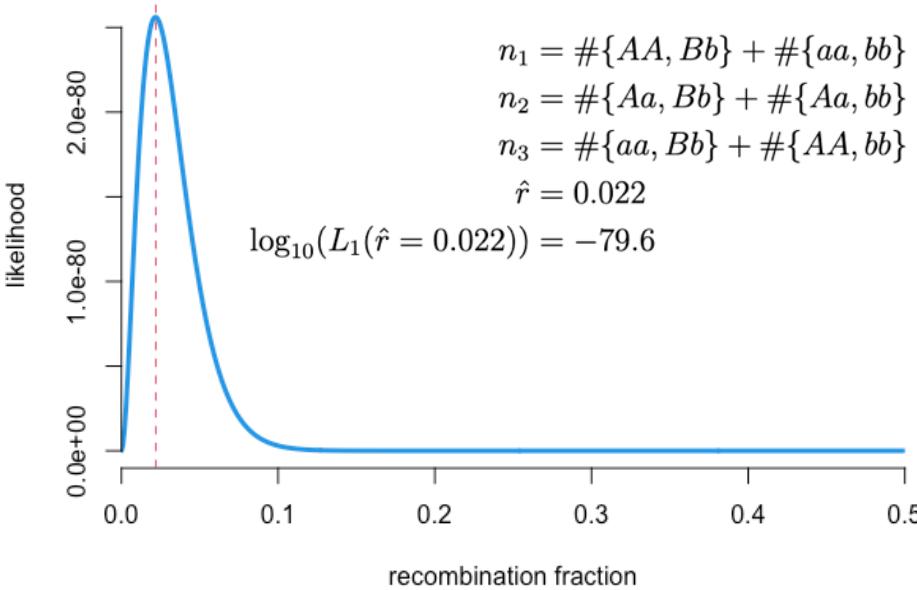


	Bb	bb
AA	$\frac{(1-r)}{6}$	$\frac{r}{6}$
Aa	$\frac{1}{3}$	$\frac{1}{3}$
$a\ a$	$\frac{r}{6}$	$\frac{(1-r)}{6}$

$$L_1(r) = \left[\frac{(1-r)}{6} \right]^{n_1} \left[\frac{1}{3} \right]^{n_2} \left[\frac{r}{6} \right]^{n_3}$$

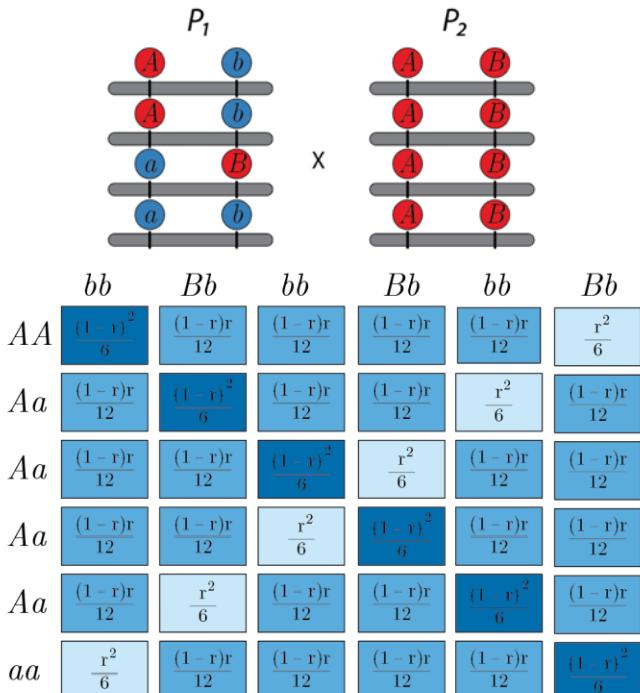
$$\begin{aligned} n_1 &= \#\{AA, Bb\} + \#\{aa, bb\} = 90 \\ n_2 &= \#\{Aa, Bb\} + \#\{Aa, bb\} = 8 \\ n_3 &= \#\{aa, Bb\} + \#\{AA, bb\} = 2 \\ \hat{r} &= 0.022 \end{aligned}$$

$$\log_{10}(L_1(\hat{r} = 0.022)) = -79.6$$



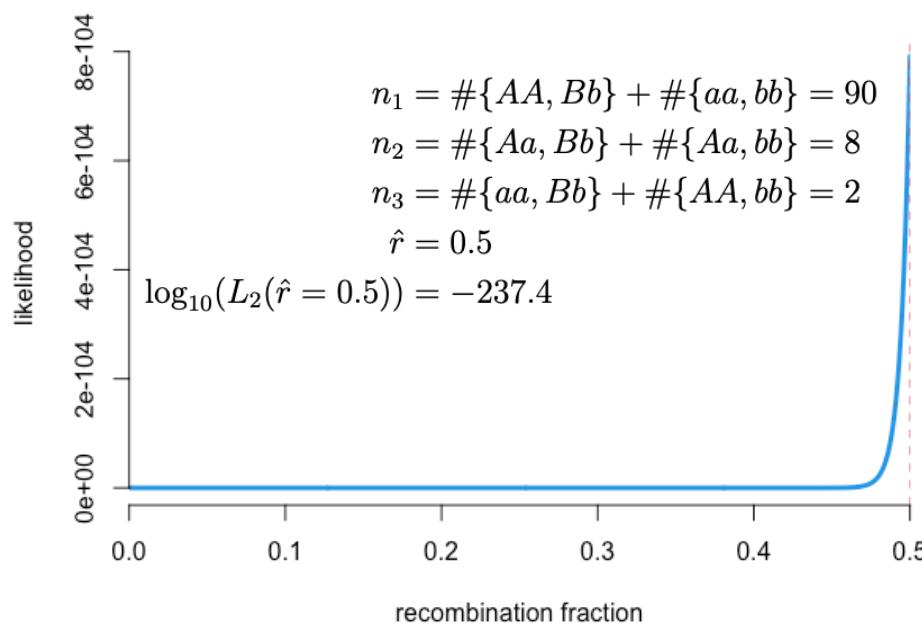
Recombination Fraction – autotetraploid

Partially informative marker – Duplex/simplex –
Repulsion

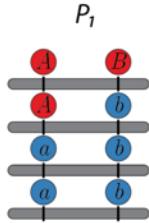


	Bb	bb
AA	$\frac{r}{6}$	$\frac{(1-r)}{6}$
Aa	$\frac{1}{3}$	$\frac{1}{3}$
$a\ a$	$\frac{(1-r)}{6}$	$\frac{r}{6}$

$$L_2(r) = \left[\frac{r}{6} \right]^{n_1} \left[\frac{1}{3} \right]^{n_2} \left[\frac{(1-r)}{6} \right]^{n_3}$$

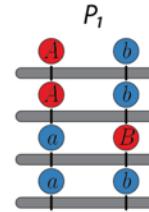


Recombination Fraction – assessing linkage phases



$$\hat{r}_1 = \arg \max_r L_1(r) \implies L_1(0.022) = -79.6$$

Compare likelihoods choosing the **most likely** configuration



$$\hat{r}_2 = \arg \max_r L_2(r) \implies L_2(0.5) = -237.4$$

logarithm-of-odds (LOD-score)

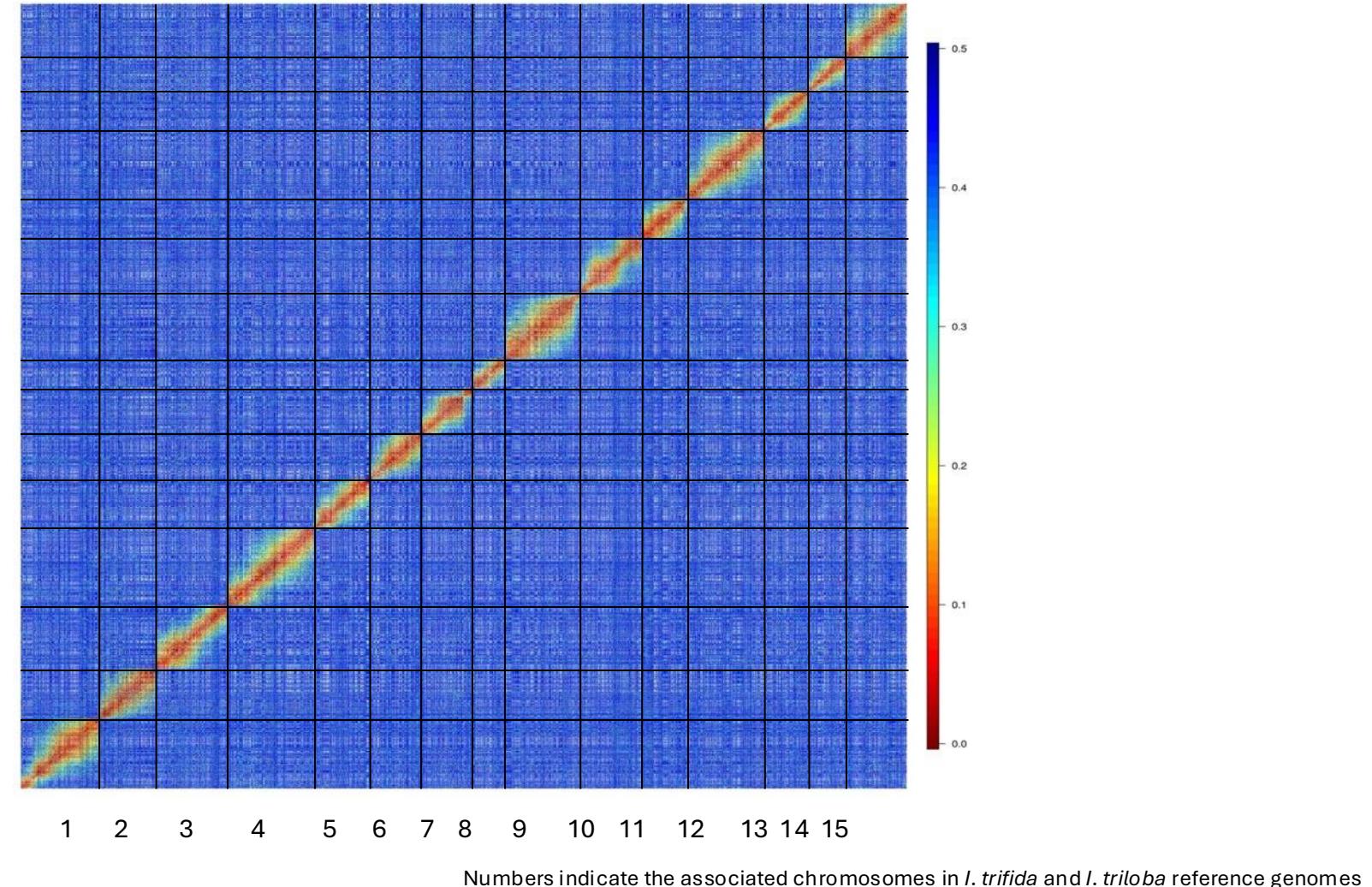
$$\begin{aligned} LOD &= \log_{10} \left(\frac{L_1}{L_2} \right) \\ &= \log_{10}(L_1) - \log_{10}(L_2) \\ &= -79.6 - (-237.4) = 157.8 \end{aligned}$$

Markers A and B are most likely in **association**

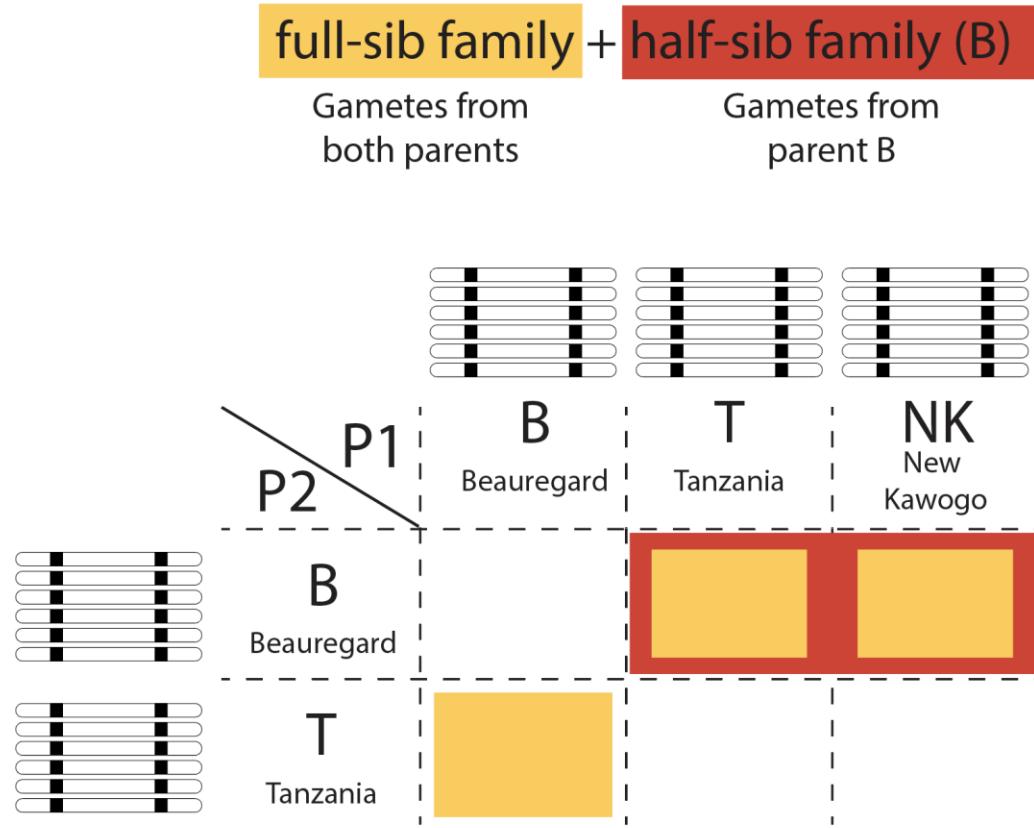
- Pairwise MLEs of r are used to group markers into linkage groups and order markers within each linkage group using optimization algorithms such as MDS.
- Given a sequence of ordered markers, it is possible to extend the idea of comparing the likelihoods of competing linkage phases throughout multiple markers.

```
> x <- make_seq_mappoly(tetra.solcap, 1:10)
> plot_mrk_info(tetra.solcap,4)
> plot_mrk_info(tetra.solcap,6)
> y <- est_pairwise_rf(x, verbose = FALSE)
> y$pairwise`^4-6`  
          LOD_ph           rf      LOD_rf
3-2    0.00000 0.002770179 7.034134e+01
2-2   -64.47357 0.217875201 5.867772e+00
3-0   -69.52225 0.396840480 8.190949e-01
2-0   -70.34559 0.499954162 4.246663e-03
3-1   -74.18865 0.283120852 6.698376e+00
2-1   -80.88787 0.499954162 8.493239e-04
```

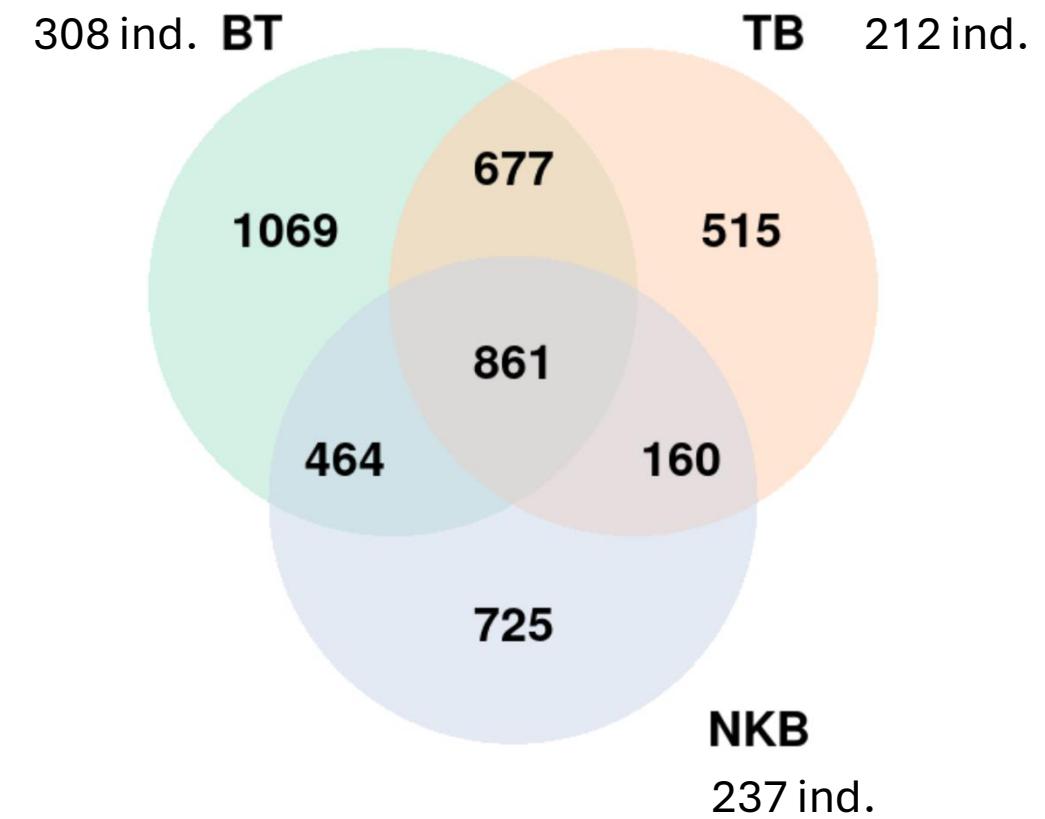
Ordering with MDS – 15 linkage groups, 30684 SNPs



BT, TB and NKB



For chromosome 9 - number of markers and individuals (after filtering)



Two-point analysis

Biparental

BT
9 phases

	LOD_ph	rf	LOD_rf
2-2	0.00000	6.303100e-02	40.134508756
2-1	-14.46833	4.584503e-05	28.229669247
1-2	-18.77941	4.311770e-02	22.925996282
0-2	-37.54139	2.727289e-01	2.593118364
2-0	-38.58133	1.623299e-01	3.219101398
1-1	-40.46726	1.674657e-01	3.772509586
0-0	-41.80496	4.999542e-01	0.004529388
0-1	-42.69973	4.999542e-01	0.001729664
1-0	-43.34615	4.999542e-01	0.001901203

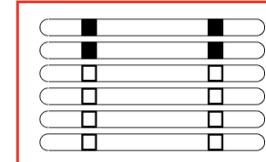
NKB
12 phases

	LOD_ph	rf	LOD_rf
2-3	0.0000000	0.20035876	1.407721e+01
1-3	-0.4124350	0.05582791	1.409992e+01
2-2	-0.5311166	0.05290159	1.475879e+01
0-3	-10.4426208	0.17700292	3.634587e+00
2-1	-11.1272382	0.11745519	4.162664e+00
1-2	-11.5603771	0.11079442	4.417476e+00
2-0	-14.0780042	0.49995416	7.967426e-04
0-0	-14.0810455	0.49995416	3.838075e-03
1-0	-14.5147546	0.49995416	2.395093e-03
0-2	-15.2907315	0.49995416	8.294299e-04
0-1	-15.2923779	0.49995416	2.475841e-03
1-1	-15.9787141	0.49995416	8.606913e-04

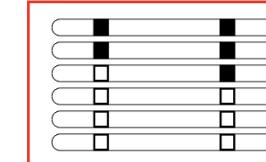
"S9_1149652"

"S9_1150612"

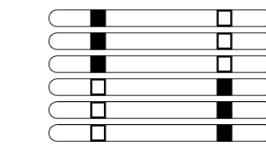
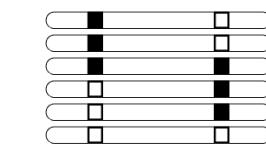
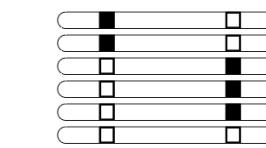
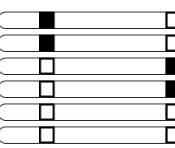
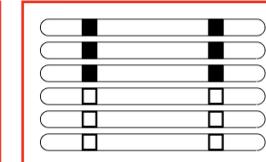
T
Tanzania



B
Beauregard

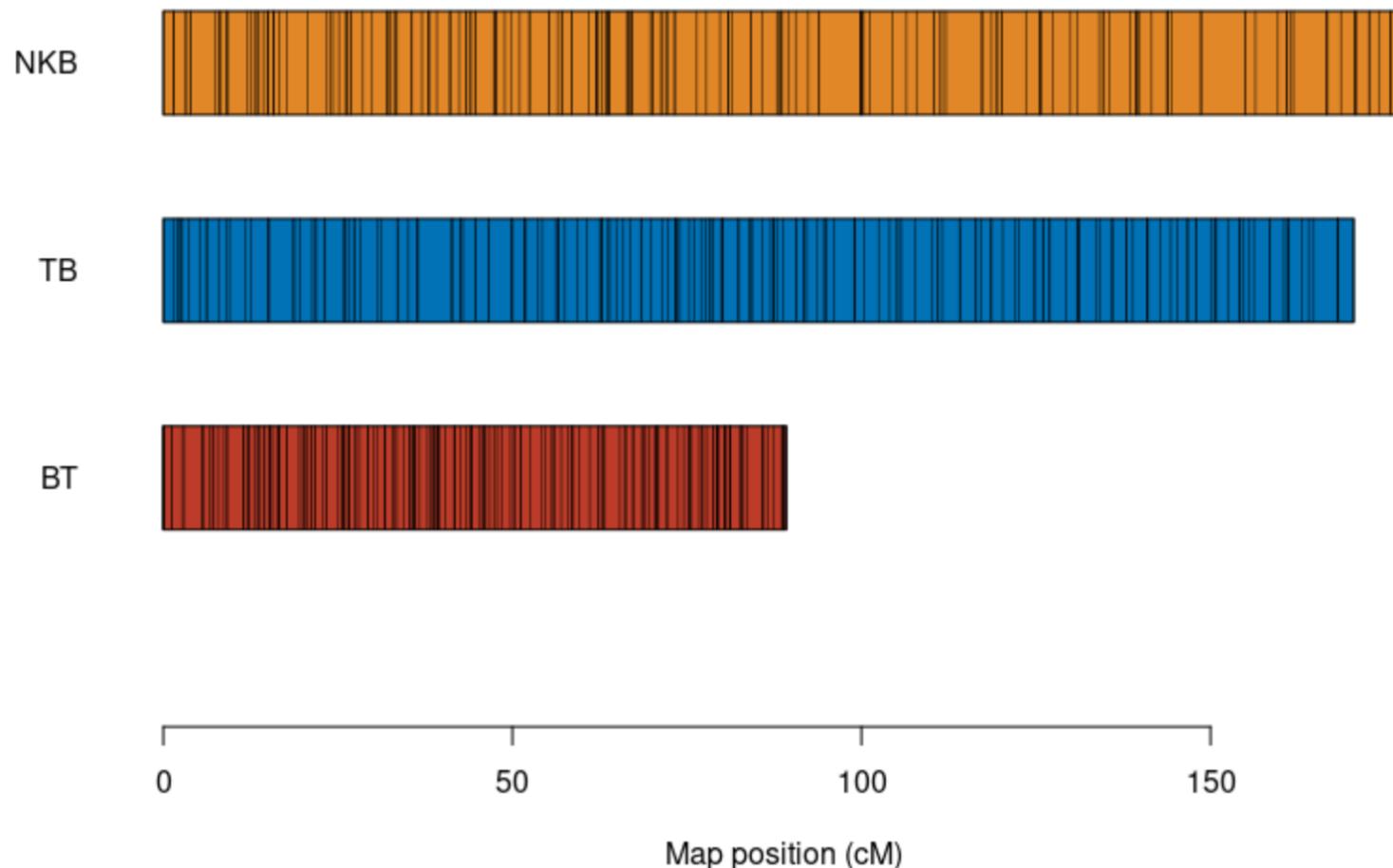


NK
New
Kawogo



Example using BT, TB and NKB

MAPpoly results: first 200 markers, chromosome 9

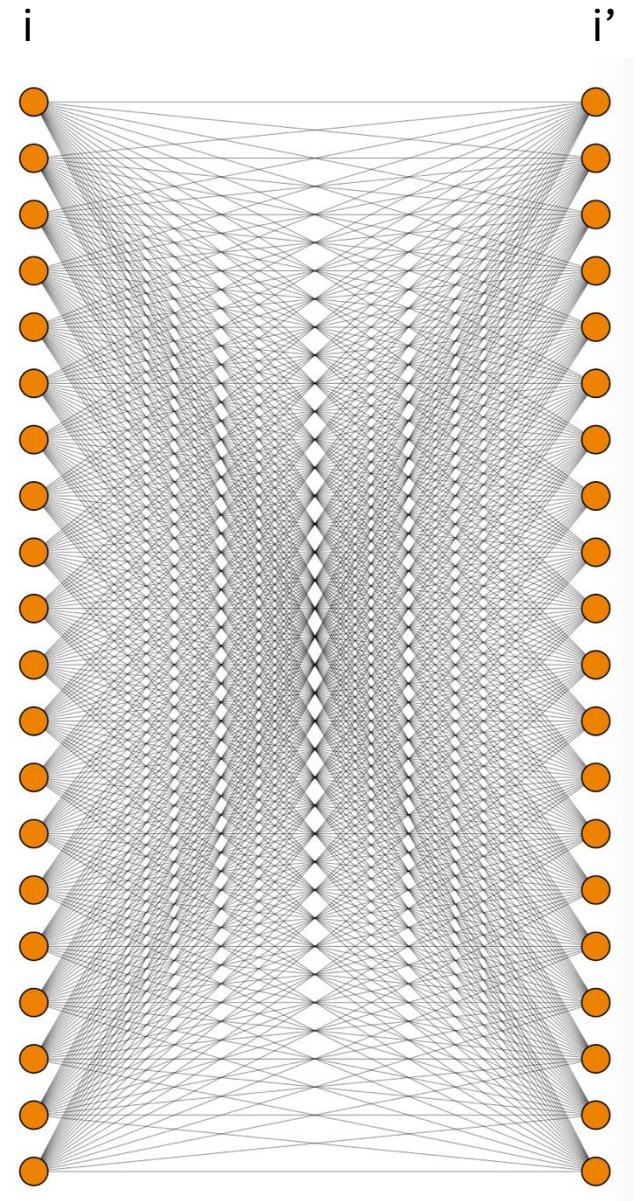


Transition probability

1 Gametic transition probability

$$\Pr(g_{i'}|g_i) = \frac{(1-r)^{\frac{m}{2}-l}(r)^l}{\binom{\frac{m}{2}}{l}}, \quad i, i' \in (1, \dots, 20). \quad (1)$$

where l denotes the number of total recombinant bivalents between two adjacent loci, r is the recombination fraction and m is the even ploidy level

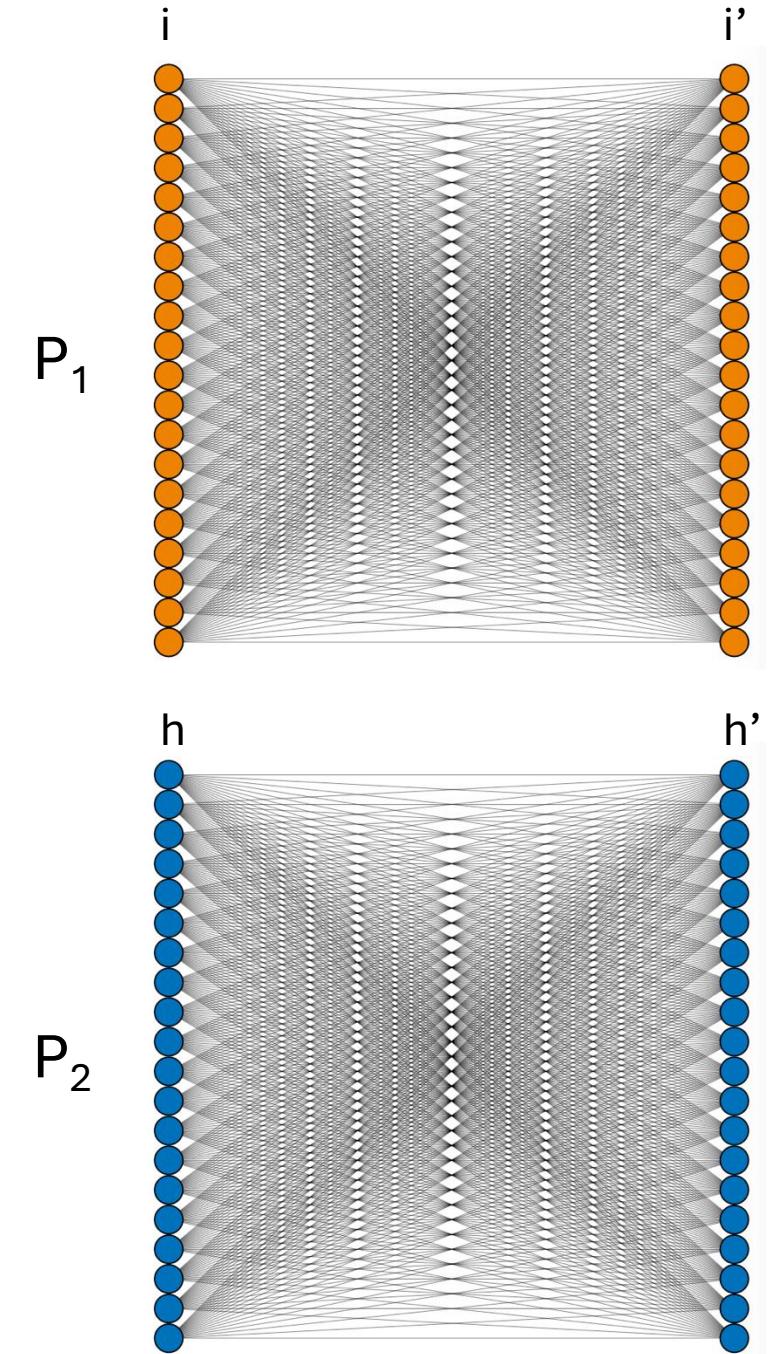


Transition probability

2 Transition probability for any pair of parents

$$\begin{aligned}\Pr(\mathcal{G}_{j'}|\mathcal{G}_j) &= \Pr(g_{i'}^{P^1}|g_i^{P^1})\Pr(g_{h'}^{P^2}|g_h^{P^2}) \\ &= \frac{(1-r)^{\frac{m_1}{2} + \frac{m_2}{2} - l_{P^1} - l_{P^2}} r^{l_{P^1} + l_{P^2}}}{\binom{\frac{m_1}{2}}{l_{P^1}} \binom{\frac{m_2}{2}}{l_{P^2}}} \quad (2)\end{aligned}$$

where \mathcal{G}_j denotes the genotype of an individual derived from the union of gametes from parents P^1 and P^2 with even ploidy levels m_1 and m_2 respectively, $i, i' \in \{1, \dots, \binom{m_1}{2}\}$, $h, h' \in \{1, \dots, \binom{m_2}{2}\}$, $j = (i-1)\binom{m_1}{2} + h$ and $j' = (i'-1)\binom{m_1}{2} + h'$. l_{P^1} and l_{P^2} denote the number of recombinant bivalents between two adjacent loci in P^1 and P^2 .

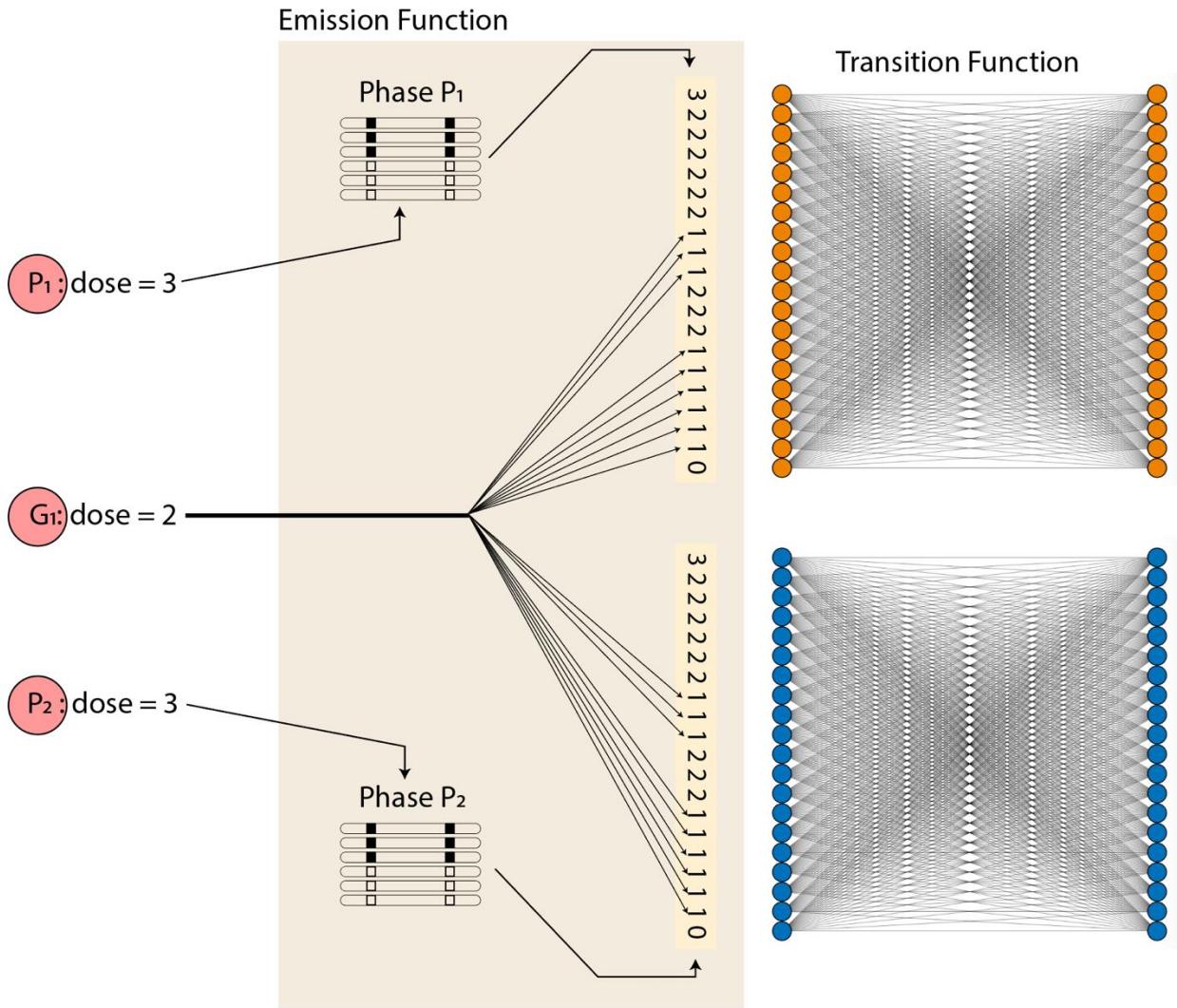


Emission and transition for a trio

3 Emission function

$$b_j(O) = \Pr(O|\mathcal{G}_j, \varphi^{P^1}, \varphi^{P^2}) = \begin{cases} 1 - \epsilon & \text{if } O = \delta(k, j) \\ \frac{\epsilon}{m} & \text{otherwise} \end{cases} \quad (3)$$

where $\delta(j) = |(\varphi^{P^1} \cup \varphi^{P^2}) \cap \mathcal{G}_j|$ and ϵ denotes the global genotype error rate.



Two-point analysis

Biparental

BT
9 phases

	LOD_ph	rf	LOD_rf
2-2	0.00000	6.303100e-02	40.134508756
2-1	-14.46833	4.584503e-05	28.229669247
1-2	-18.77941	4.311770e-02	22.925996282
0-2	-37.54139	2.727289e-01	2.593118364
2-0	-38.58133	1.623299e-01	3.219101398
1-1	-40.46726	1.674657e-01	3.772509586
0-0	-41.80496	4.999542e-01	0.004529388
0-1	-42.69973	4.999542e-01	0.001729664
1-0	-43.34615	4.999542e-01	0.001901203

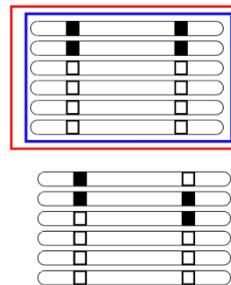
NKB
12 phases

	LOD_ph	rf	LOD_rf
2-3	0.0000000	0.20035876	1.407721e+01
1-3	-0.4124350	0.05582791	1.409992e+01
2-2	-0.5311166	0.05290159	1.475879e+01
0-3	-10.4426208	0.17700292	3.634587e+00
2-1	-11.1272382	0.11745519	4.162664e+00
1-2	-11.5603771	0.11079442	4.417476e+00
2-0	-14.0780042	0.49995416	7.967426e-04
0-0	-14.0810455	0.49995416	3.838075e-03
1-0	-14.5147546	0.49995416	2.395093e-03
0-2	-15.2907315	0.49995416	8.294299e-04
0-1	-15.2923779	0.49995416	2.475841e-03
1-1	-15.9787141	0.49995416	8.606913e-04

"S9_1149652"

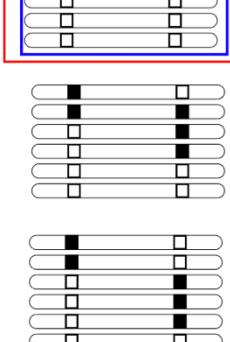
T

Tanzania



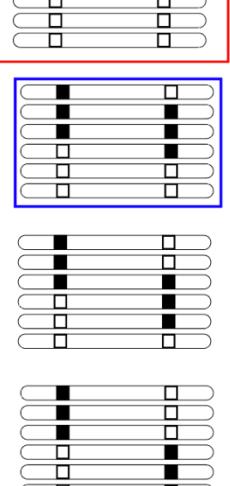
B

Beauregard



NK

New
Kawogo



"S9_1150612"

\$Br	[,1]	[,2]	[,3]	[,4]	[,5]	[,6]
[1,]	1	1	0	0	0	0
[2,]	1	1	1	0	0	0

\$Tz	[,1]	[,2]	[,3]	[,4]	[,5]	[,6]
[1,]	1	1	0	0	0	0
[2,]	1	1	0	0	0	0

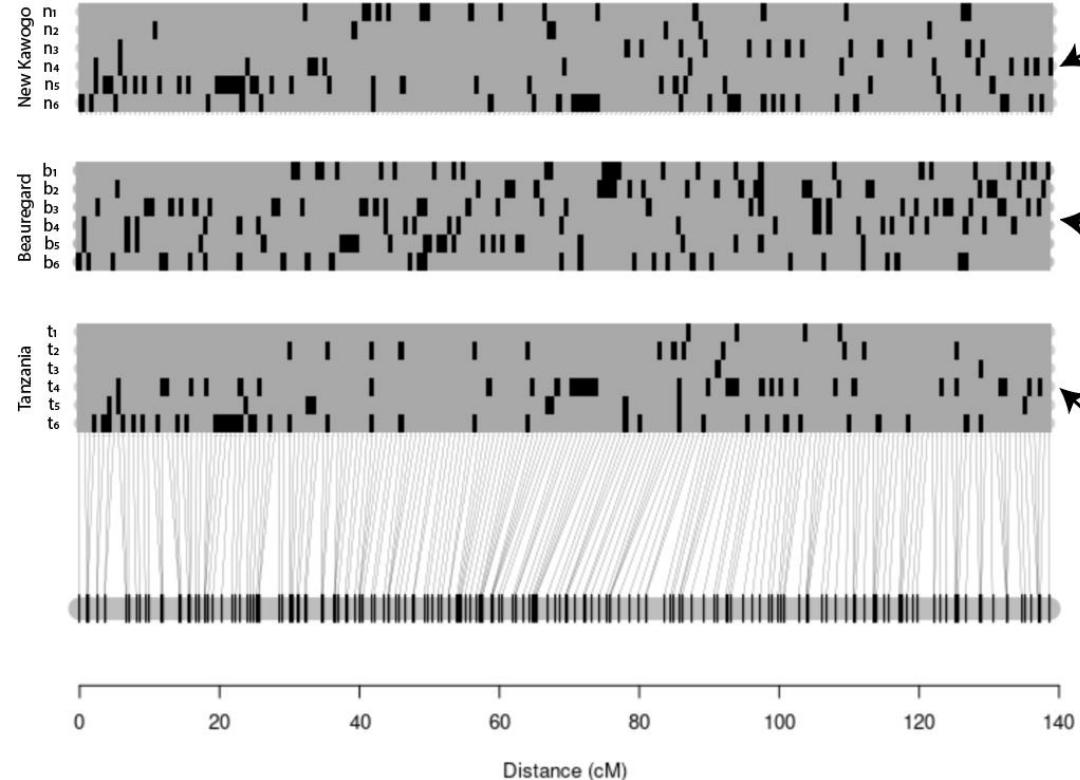
\$NK	[,1]	[,2]	[,3]	[,4]	[,5]	[,6]
[1,]	1	1	1	0	0	0
[2,]	0	1	1	1	0	0

Multiparental

36 phases

phase: 1	--->	1347.491	0.05568119
phase: 2	--->	1340.776	0.01140695
phase: 3	--->	1280.699	0.01653743
phase: 4	--->	1317.229	0.0297278
phase: 5	--->	1316.649	0.01156901
phase: 6	--->	1246.391	0.0168745
phase: 7	--->	1300.952	0.04167072
phase: 8	--->	1261.465	0.01637306
phase: 9	--->	1294.933	0.3891418
phase: 10	--->	1352.602	0.01418067
phase: 11	--->	1328.605	0.01147952
phase: 12	--->	1261.19	0.0299921
phase: 13	--->	1345.915	0.0105054
phase: 14	--->	1304.478	0.01164158
phase: 15	--->	1259.206	0.09527872
phase: 16	--->	1318.737	0.011544
phase: 17	--->	1249.294	0.01644563
phase: 18	--->	1304.026	0.4999
phase: 19	--->	1340.757	0.01427433
phase: 20	--->	1290.181	0.01545397
phase: 21	--->	1287.129	0.2338434
phase: 22	--->	1334.07	0.01059906
phase: 23	--->	1266.054	0.01561603
phase: 24	--->	1295.068	0.4186853
phase: 25	--->	1306.892	0.01163767
phase: 26	--->	1253.631	0.08398184
phase: 27	--->	1303.717	0.4999
phase: 28	--->	1303.747	0.01831786
phase: 29	--->	1288.718	0.1308722
phase: 30	--->	1298.22	0.3941346
phase: 31	--->	1297.059	0.01464259
phase: 32	--->	1279.656	0.1887354
phase: 33	--->	1305.097	0.4999
phase: 34	--->	1269.881	0.0156812
phase: 35	--->	1296.183	0.4283121
phase: 36	--->	1306.915	0.4999

Joint analysis



BT

