

Linkage and Chromosome Mapping

I.

1st year, 2nd semester, week 11

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Terminology, definitions

- The term **recombination ratio (fraction)**, Θ (Greek letter theta), is used for expression of linkage intensity (strength).
- The unit of **cM** (centimorgan) is, in contemporary textbooks, used for **map distance**.
- These two variables are identical only for small values – namely, maximum possible value of Θ is 0.5, *i.e.* 50 %, whereas the length of a chromosome after counting up of individual segments at genetic mapping can be even 120 to 150 cM.
- Roughly, we may declare that two loci are separated by genetic distance 1 cM (one centimorgan), if the recombination fraction make 1 % or $\Theta = 0.01$.
- In the text of the book the older terminology is used (linkage intensity is marked as ***p***).

Intrachromosomal recombination

P (homozygous parents)

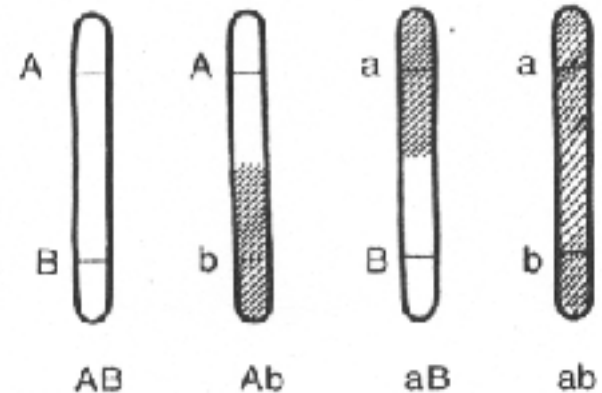
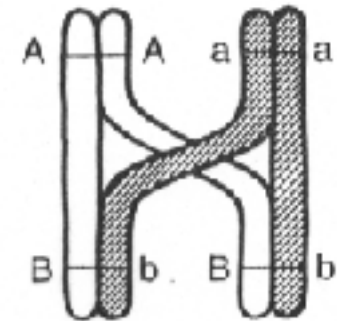
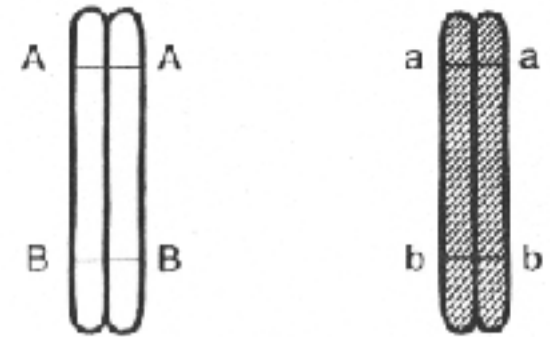
F₁ (heterozygote)

crossing-over in prophase I

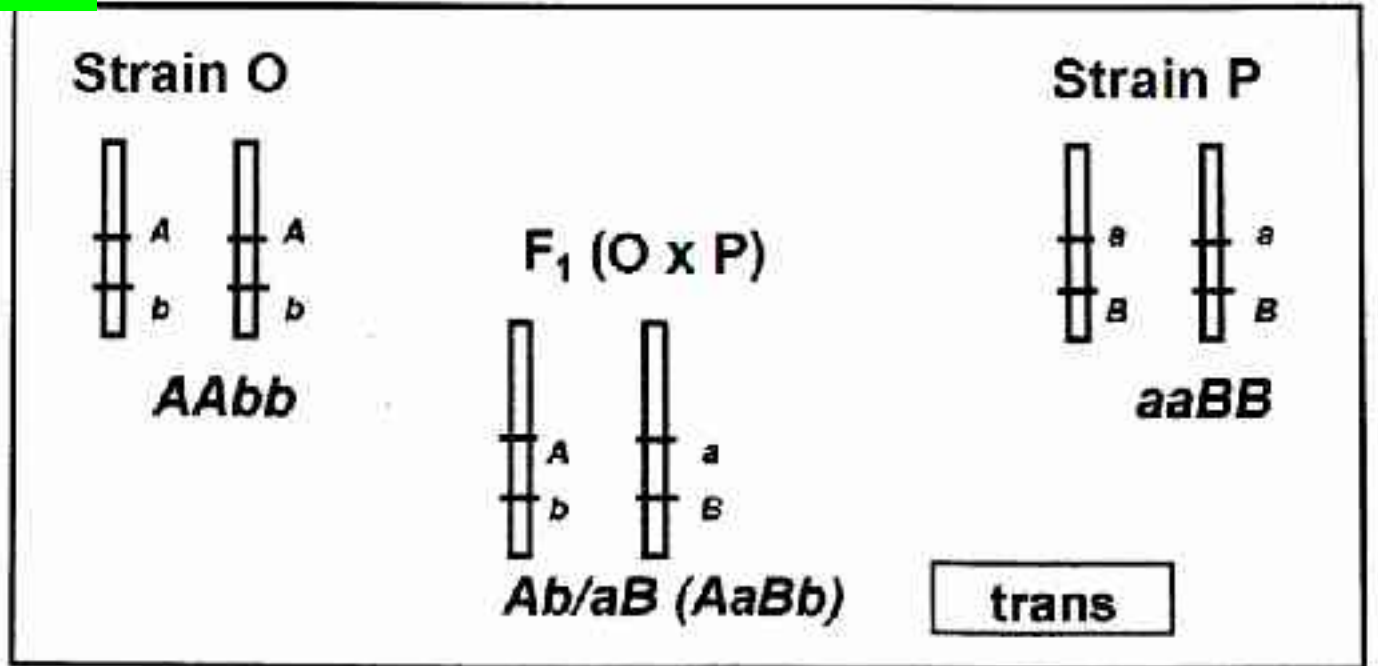
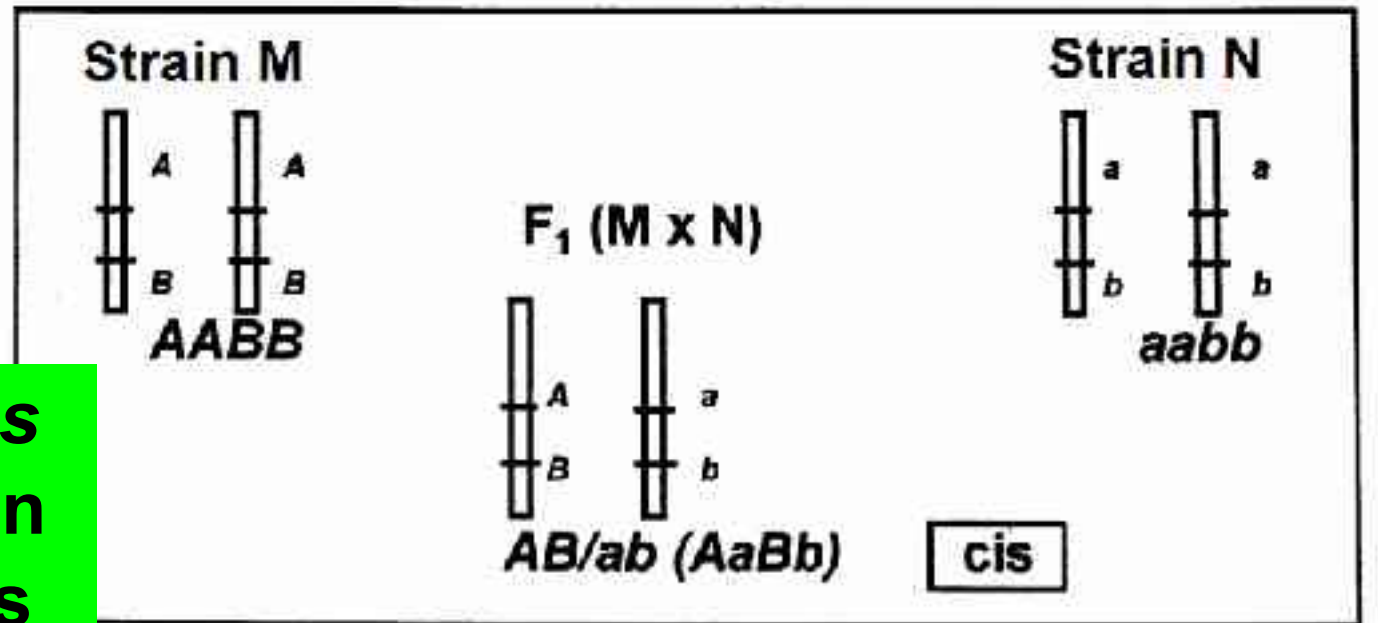
disjunction and distribution of chromatids in anaphase II

chromosomes in gametes of F₁ generation

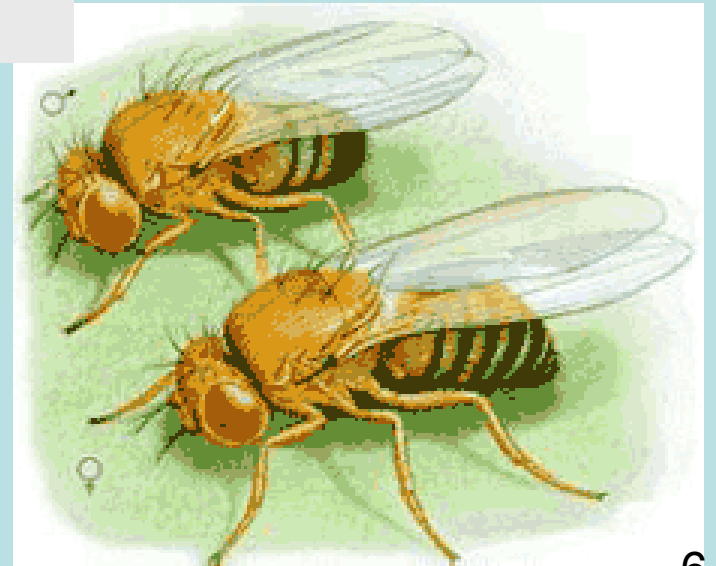
genotypes of gametes



***cis* and *trans*
configuration
in F₁ hybrids**



Drosophila melanogaster



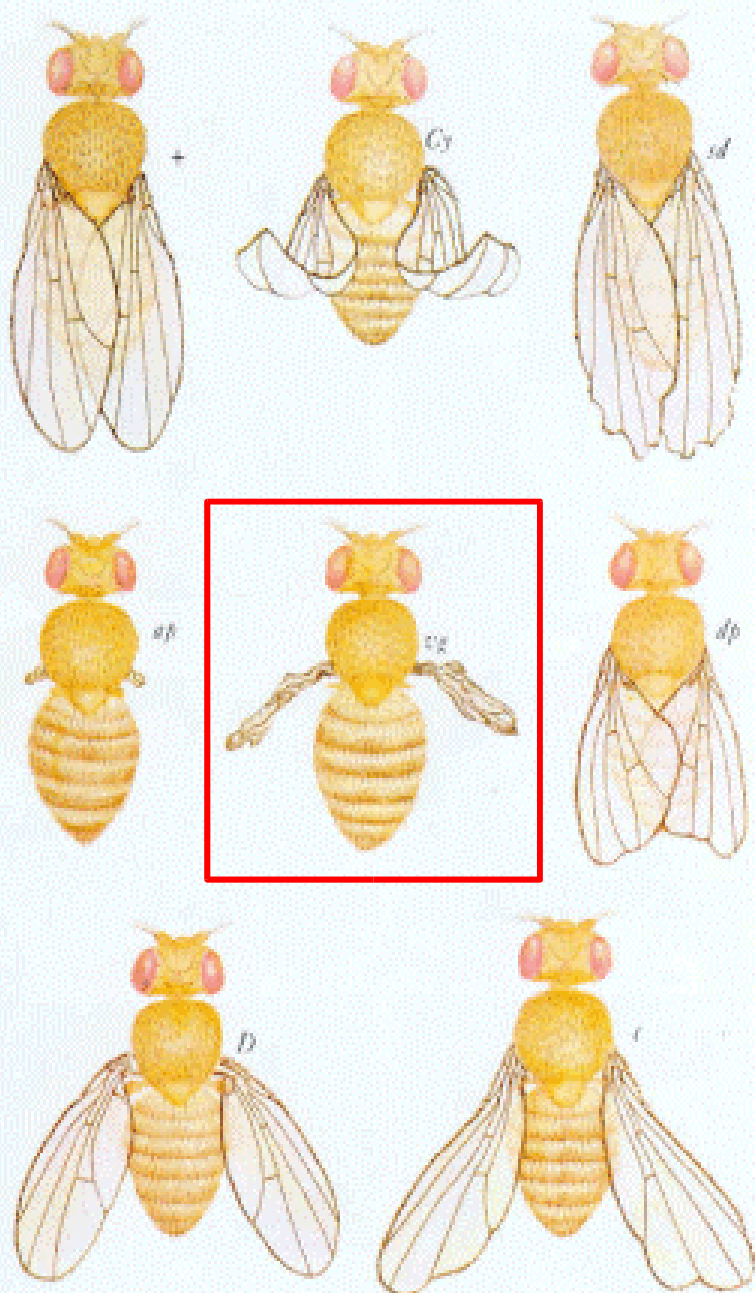
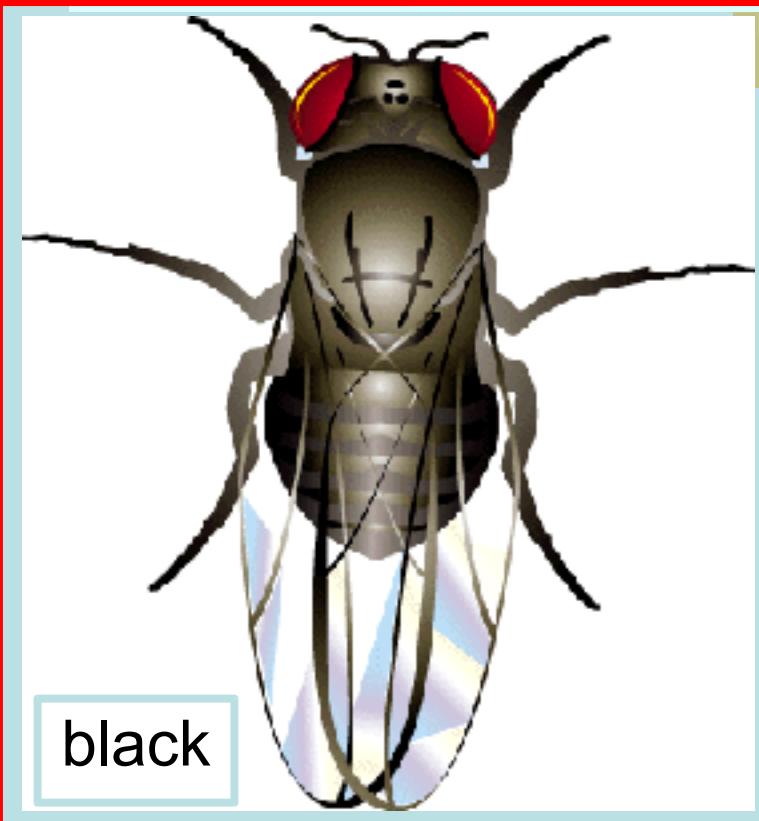
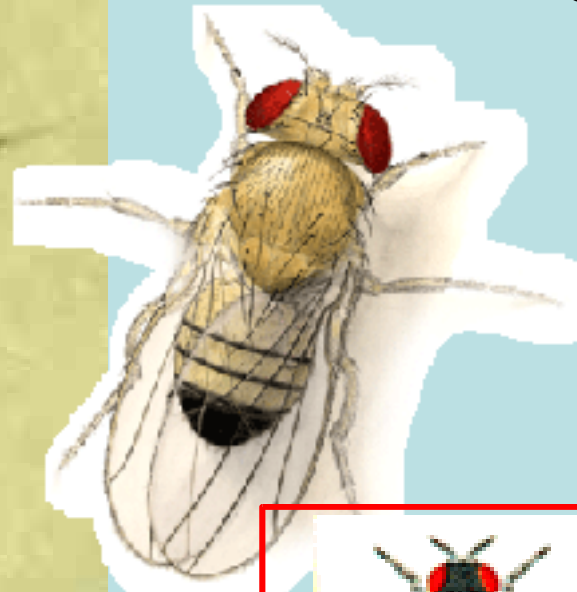


Figure 24 Wing mutations: +, wild; Cy, curly; sd, scalloped; ap, apterous; vg, vestigial; dp, dumpy; D, Dichaeete; c, curved.

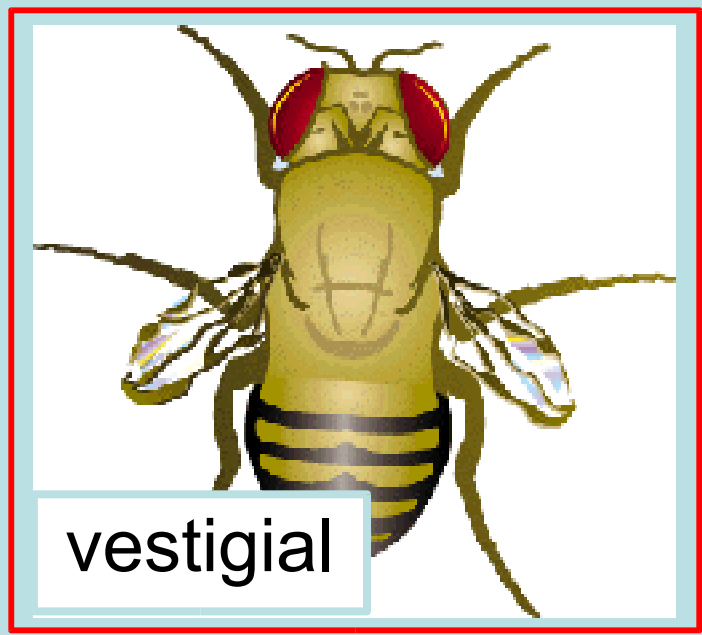


vestigial

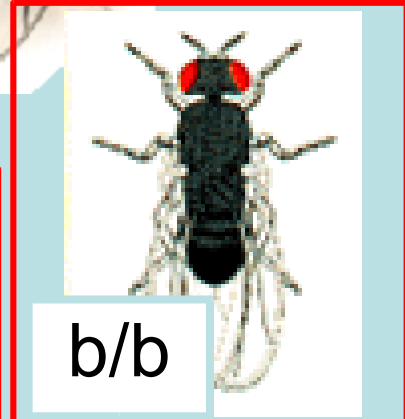




black



vestigial



b/b



vg/vg

Linkage in Drosophila - Task 5/p, 100KrOt

Phenotypes



x

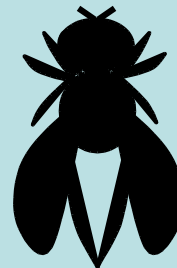


Genotypes

+ b
+ vg

b b
vg vg

Phenotypes
of Bc
population



Number

91

411

413

84

Genotypes

+ + / b vg



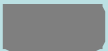
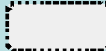
b + / b

+ vg / b vg

b vg / b vg

Configuration:
trans

$$\Theta = \frac{91 + 84}{91 + 411 + 413 + 84} = \frac{175}{999} = 0.175$$

Genes: A Traits: "shape"  (smooth)  (wrinkled)
 B "colour"  (deep)  (pale)

Back - cross (Bc) - cis configuration (coupling)

map distance $p = 0$ cM (recombination ratio $\theta = 0.0$)



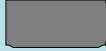
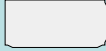
double heterozygote (F1 hybrid) x recessive homozygote



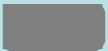
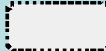
Phenotype AB  ab 

Genotype *AB/ab* *ab/ab*

Gametes AB, ab (original) Ab, aB (recombinants) ab

frequency by 50 % (0.5) 0% (0.0) 100 % (1.0)

Distance $p =$	Genotype	<i>AB/ab</i>	<i>Ab/ab</i>	<i>aB/ab</i>	<i>ab/ab</i>
	Phenotype				
0 cM	frequency	0.5	0	0	0.5

Genes: A Traits: "shape"  (smooth)  (wrinkled)
 B "colour"  (deep)  (pale)

Back - cross (Bc) - cis configuration (coupling)

map distance $p = 20 \text{ cM}$ ($\theta = 0.2$)



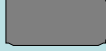
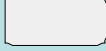
double heterozygote (F1 hybrid) x recessive homozygote





Phenotype AB  ab 

Genotype *AB/ab* *ab/ab*

Gametes AB, ab (original) Ab, aB (recombinants) ab

frequency by 40 % (0.4) by 10% (0.1) 100 % (1.0)

Distance $p =$	Genotype	<i>AB/ab</i>	<i>Ab/ab</i>	<i>aB/ab</i>	<i>ab/ab</i>
	Phenotype				
20 cM	frequency	0.4	0.1	0.1	0.4

Genes: A Traits: "shape"  (smooth)  (wrinkled)
 B "colour"  (deep)  (pale)

Back - cross (Bc) - cis configuration (coupling)

map distance, „linkage“ $p = 50 \text{ cM}$ ($\theta = 0.5$)





double heterozygote (F_1 hybrid) x recessive homozygote





Phenotype AB  ab 

Genotype AB/ab ab/ab

Gametes AB, ab (original) Ab, aB (recombinants) ab

frequency by 25 % (0.25) by 25% (0.25) 100 % (1.0)

Distance $p =$	Genotype	AB/ab	Ab/ab	aB/ab	ab/ab
	Phenotype				
50 cM	frequency	0.25	0.25	0.25	0.25

Genes: A Traits: "shape"  (smooth)  (wrinkled)
 B "colour"  (deep)  (pale)





Back - cross (Bc) - cis configuration (coupling)





double heterozygote (F1 hybrid) x recessive homozygote

Phenotype AB  ab 

Genotype *AB/ab* *ab/ab*

Gametes AB, ab (original) Ab, aB (recombinants) ab

	Distance p =	Genotype Phenotype	<i>AB/ab</i> 	<i>Ab/ab</i> 	<i>aB/ab</i> 	<i>ab/ab</i> 
a)	0 cM	frequency	0.5	0	0	0.5
b)	20 cM	frequency	0.4	0.1	0.1	0.4
c)	50 cM	frequency	0.25	0.25	0.25	0.25

Genes: A Traits: "shape"  (smooth)  (wrinkled)
 B "colour"  (deep)  (pale)





Back - cross (Bc) - trans configuration (repulsion)

double heterozygote (F1 hybrid) x recessive homozygote

Phenotype AB  ab 

Genotype *Ab/aB* *ab/ab*

Gametes *Ab, aB* (original) *AB, ab* (recombinants) *ab*

	Distance p =	Genotype	<i>AB/ab</i>	<i>Ab/ab</i>	<i>aB/ab</i>	<i>ab/ab</i>
		Phenotype				
a)	0 cM	frequency	0	0.5	0.5	0
b)	20 cM	frequency	0.1	0.4	0.4	0.1
c)	50 cM	frequency	0.25	0.25	0.25	0.25

P **Ab/Ab** **x** **aB/aB**

gametes

Ab

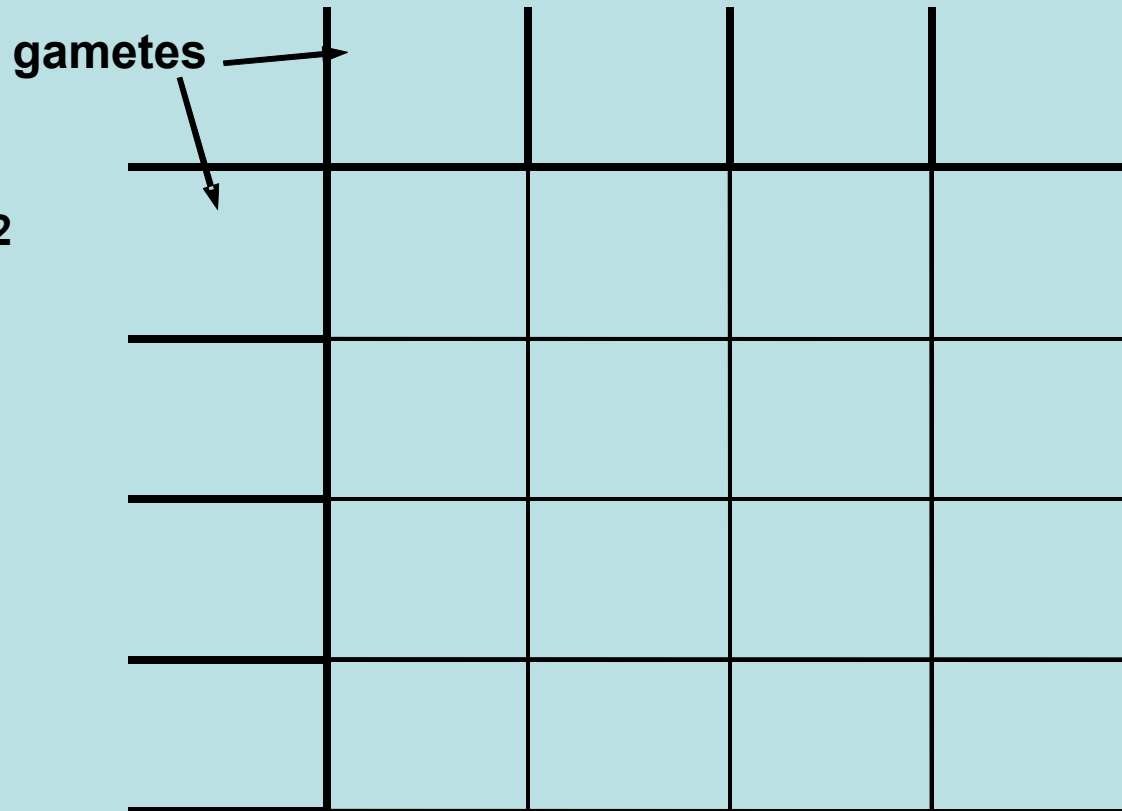
aB

F₁

Ab/aB

($\theta = 0.2$)

F₂



P **Ab/Ab** x **aB/aB**

gametes **Ab** **aB**

F₁ **Ab/aB** ($\theta = 0.2$)

		gametes			
		<i>AB</i>	<i>Ab</i>	<i>aB</i>	<i>ab</i>
F₂	<i>AB</i>				
	<i>Ab</i>				
	<i>aB</i>				
	<i>ab</i>				

P **Ab/Ab** x **aB/aB**
 gametes **Ab** **aB**
F₁ **Ab/aB** ($\theta = 0.2$)

gametes		<i>AB</i>	<i>Ab</i>	<i>aB</i>	<i>ab</i>
frequency		0.1	0.4	0.4	0.1
F₂	<i>AB</i> 0.1				
	<i>Ab</i> 0.4				
	<i>aB</i> 0.4				
	<i>ab</i> 0.1				

P **Ab/Ab** x **aB/aB**
 gametes **Ab** **aB**
F₁ **Ab/aB** ($\theta = 0.2$)

gametes		AB	Ab	aB	ab
		0.1	0.4	0.4	0.1
F ₂	AB 0.1	AB	AB	AB	AB
	Ab 0.4	AB	Ab	AB	Ab
	aB 0.4	AB	AB	aB	aB
	ab 0.1	AB	Ab	aB	ab
		phenotypes			

P **Ab/Ab** x **aB/aB**

gametes **Ab** **aB**

F₁ **Ab/aB** ($\theta = 0.2$)

gametes		AB	Ab	aB	ab
		frequency	0.1	0.4	0.4
F₂	AB	AB	AB	AB	AB
	0.1	0.01	0.04	0.04	0.01
	Ab	AB	Ab	AB	Ab
	0.4	0.04	0.16	0.16	0.04
	aB	AB	AB	aB	aB
0.4	0.04	0.16	0.16	0.04	
ab	AB	Ab	aB	ab	
0.1	0.01	0.04	0.04	0.01	
		frequency			

P **Ab/Ab** x **aB/aB**
 gametes **Ab** **aB**
F₁ **Ab/aB** ($\theta = 0.2$)

gametes		AB	Ab	aB	ab
frequency		0.1	0.4	0.4	0.1
F₂	AB	AB 0.01	AB 0.04	AB 0.04	AB 0.01
	Ab	AB 0.04	Ab 0.16	AB 0.16	Ab 0.04
	aB	AB 0.04	AB 0.16	aB 0.16	aB 0.04
	ab	AB 0.01	Ab 0.04	aB 0.04	ab 0.01

		<i>AB</i>	<i>Ab</i>	<i>aB</i>	<i>ab</i>
		0.1	0.4	0.4	0.1
F₂	<i>AB</i>	AB 0.1	AB 0.04	AB 0.04	AB 0.01
	<i>Ab</i>	AB 0.04	Ab 0.16	AB 0.16	Ab 0.04
	<i>aB</i>	AB 0.04	AB 0.16	aB 0.16	aB 0.04
	<i>ab</i>	AB 0.01	Ab 0.04	aB 0.04	ab 0.01

phenotype frequency

Efficiency of methods of linkage estimation

Homework: Task 4/str. 100*KrOt*.