Linkage
and Chromosome Mapping

II.

1st year, 2nd semester, week 11

5 and 6/5/2008

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 Θ = 0.01.
- In the text of the book the older terminology is used (linkage intensity is marked as ***p***).

Efficiency of methods of linkage

Task 5/p. 100*KrOt*

If Θ = 0.2 and phase is trans then expected phenotypic ratio in Bc generation is

AB 0.1 : Ab 0.4 : aB 0.4 : ab 0.1

If the frequency 0.1 is to be represented by 10 individuals then at least 100 individuals should be tested.

If Θ = 0.2 and phase is trans then expected phenotypic ratio in F_2 generation is

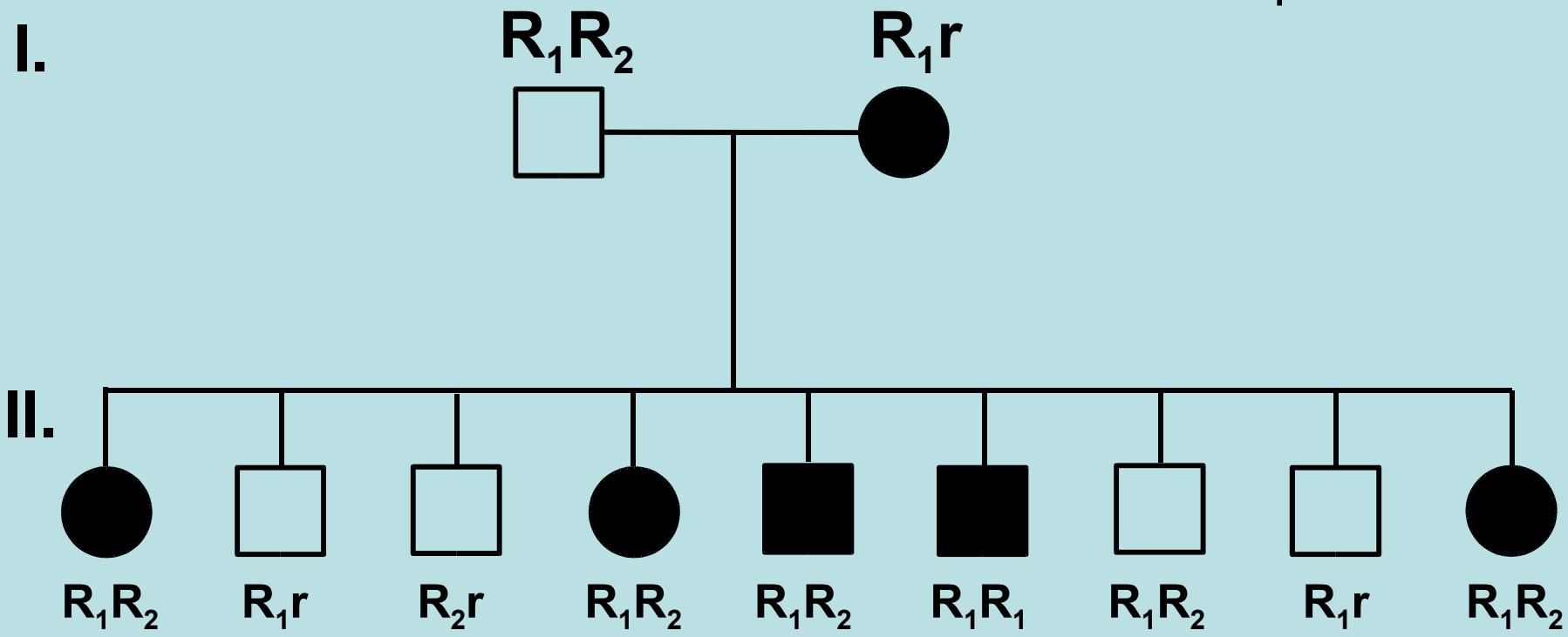
AB 0.51 : Ab 0.24 : aB 0.24 : ab 0.01

If the frequency 0.01 is to be represented by 10 individuals then at least 1000 individuals should be tested in total,

***i.e.* 10x more individuals than in Bc are needed.**

Linkage: Rh system and elliptocytosis

Task 8/p. 102*KrOt*



r…….dce
R1…….DCe
R2…….DcE

■ ● – elliptocytosis (heterozygotes *El/el*)

Rh system and elliptocytosis – answers

Task 8/p. 102*KrOt*

a) Do these traits cosegregate?	**yes**
b) Linkage configuration of the genes in	**father: R1 el / R2 el** **mother: R1 El / r el**
c) Can you find any recombination?	**yes**
d) frequency of recombination **map distance between loci**	**1/9 (11.1 %)** **11.1 cM**

This type of elliptocytosis (autosomal dominant) is due to the defect of a protein of RBC membrane (protein 4.1). Related gene maps to 1p33-p32, the Rh system (antigen D) maps to 1p36.2-p34 (1p36.11), the map distance between genes is reported in the range 5 to 22 cM.

Somatic cell hybridisation

Úkol č. 24/str. 63 *Kot* - **zadání**

linie	znak H	**Lidský chromosom**																						
		1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	Y	X
1	+	+	-	+	+	+	+	-	+	+	+	-	-	-	-	-	-	-	+	+	+	-	-	-	+
2	-	-	-	+	-	-	-	+	-	+	-	+	-	+	+	+	-	-	-	-	-	-	-	-	+
3	+	-	-	+	+	-	-	-	-	-	-	-	-	-	-	-	+	-	-	+	-	+	-	-	+
4	+	+	-	+	+	-	+	-	+	-	+	+	+	+	+	-	-	-	-	+	+	+	+	+	+
5	+	-	+	+	+	-	-	+	+	-	+	+	+	-	+	+	+	-	-	-	+	-	-	-	+
6	+	-	-	+	-	-	+	+	-	-	+	+	-	+	-	-	-	-	-	-	+	-	+	-	+
7	+	+	+	+	-	-	-	+	-	+	+	+	-	+	-	-	+	-	+	-	-	+	+	-	+
8	-	-	-	-	-	-	-	-	-	-	-	+	-	+	-	-	-	-	+	-	-	-	+	-	+
9	+	-	-	+	+	+	+	-	-	-	-	+	+	-	+	+	+	+	+	-	-	+	+	-	+
10	+	-	+	+	+	+	+	-	-	-	-	+	-	-	-	-	+	-	+	+	-	+	-	+	+

Hybridomovová technika

Úkol č. 24/str. 63 *Kot* - **řešení**

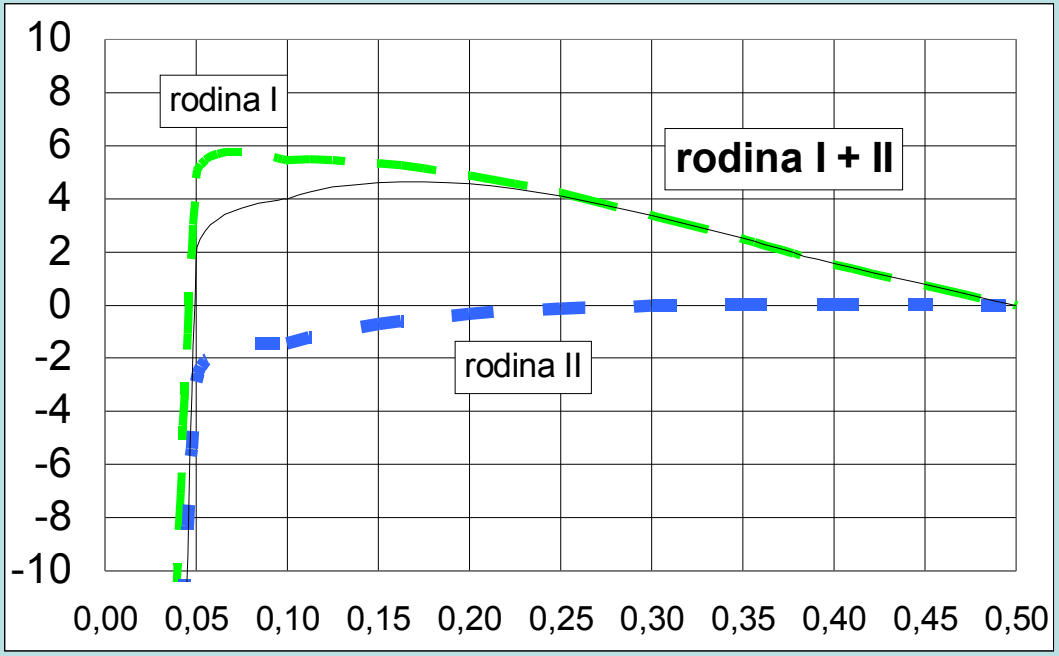
linie	znak H	Lidský chromosom																							
		1	2	**3**	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	Y	X
1	+	+	-	+	+	+	+	-	+	+	+	-	-	-	-	-	-	-	+	+	+	-	-	-	+
2	-	-	-	+	-	-	-	+	-	+	-	+	-	+	+	+	-	-	-	-	-	-	-	-	+
3	+	-	-	+	+	-	-	-	-	-	-	-	-	-	-	-	+	-	-	+	-	+	-	-	+
4	+	+	-	+	+	-	+	-	+	-	+	+	+	+	+	-	-	-	-	+	+	+	+	+	+
5	+	-	+	+	+	-	-	+	+	-	+	+	+	-	+	+	+	-	-	-	+	-	-	-	+
6	+	-	-	+	-	-	+	+	-	-	+	+	-	+	-	-	-	-	-	-	+	-	+	-	+
7	+	+	+	+	-	-	-	+	-	+	+	+	-	+	-	-	+	-	+	-	-	+	+	-	+
8	-	-	-	-	-	-	-	-	-	-	-	+	-	+	-	-	-	-	+	-	-	-	+	-	+
9	+	-	-	+	+	+	+	-	-	-	-	+	+	-	+	+	+	+	+	-	-	+	+	-	+
10	+	-	+	+	+	+	+	-	-	-	-	+	-	-	-	-	+	-	+	+	-	+	-	+	+
počet asociací		5	5	**9**	8	5	7	4	5	3	7	6	5	3	4	3	7	3	5	6	6	7	5	4	8

Vazba Rh a eliptocytózy

Úkol č. 28/str. 64*Kot*

Maximální hodnota (zarámovaná) odpovídá Θ = 0,15.

Θ	Rodina I lod	Rodina II lod	Celkem
0,00	- ∞	- ∞	- ∞
0,05	4,74	-2,90	1,84
0,10	5,44	-1,44	4,00
0,15	5,35	-0,73	4,62
0,20	4,89	-0,34	4,55
0,25	4,22	-0,12	4,10
0,30	3,39	-0,01	3,38
0,35	2,49	0,03	2,52
0,40	1,54	0,02	1,56
0,50	0,00	0,00	0,00



An example of test question

- In a broad back-cross population, the phenotypic representation has been reported as follows:
- Both A and B traits have been observed in 18 offspring of this cross of double heterozygote *AaBb* with double recessive homozygote *aabb*.
- 20 individuals expressed neither trait A nor B.
- 378 individuals had the A trait but not the B, and
- 384 individuals had the B trait but not the A.

a) What are the genotypes of offsprings? And what are their frequencies in %?

b) Calculate the recombination fraction (ratio).

c) Determine the linkage configuration (phase) in F_1 hybrid.

An example of test question - answers

a)

number	**18**	**378**	**384**	**20**
phenotype	**AB**	**Ab**	**aB**	**ab**
genotype	**AB/ab**	**Ab/ab**	**aB/ab**	**ab/ab**

b)

$$\Theta = \frac{18 + 20}{18 + 378 + 384 + 20} = \frac{38}{800} = 0.0475$$

c) Configuration: **trans**