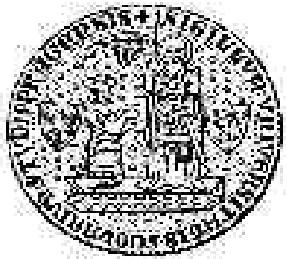
Molecular genetics V

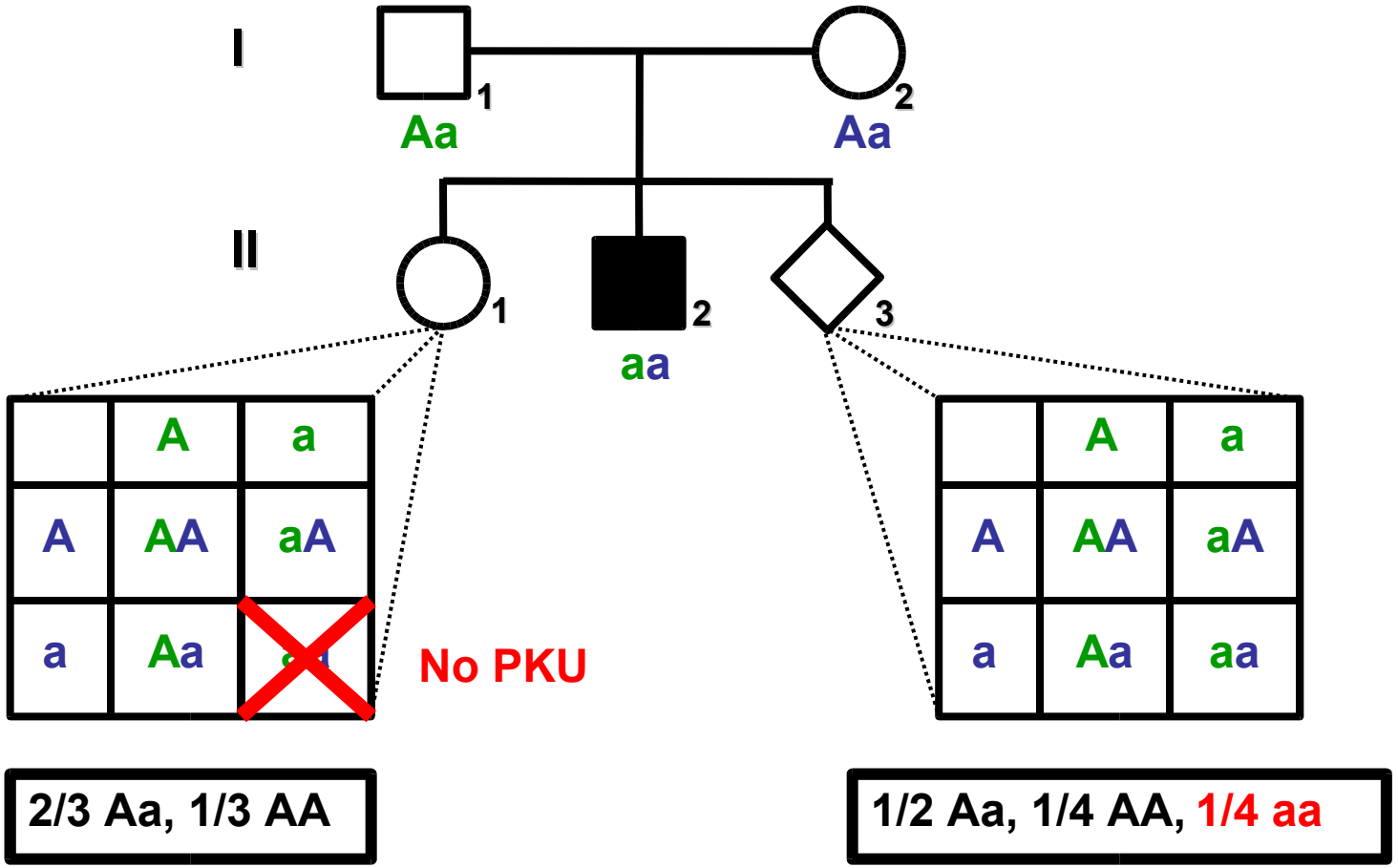
winter semester

(Nov. 18th – 21th, 2008)



Inst. of Biology and Medical Genetics 1.LF UK a VFN, Praha

Task 3, p. 134, PKU

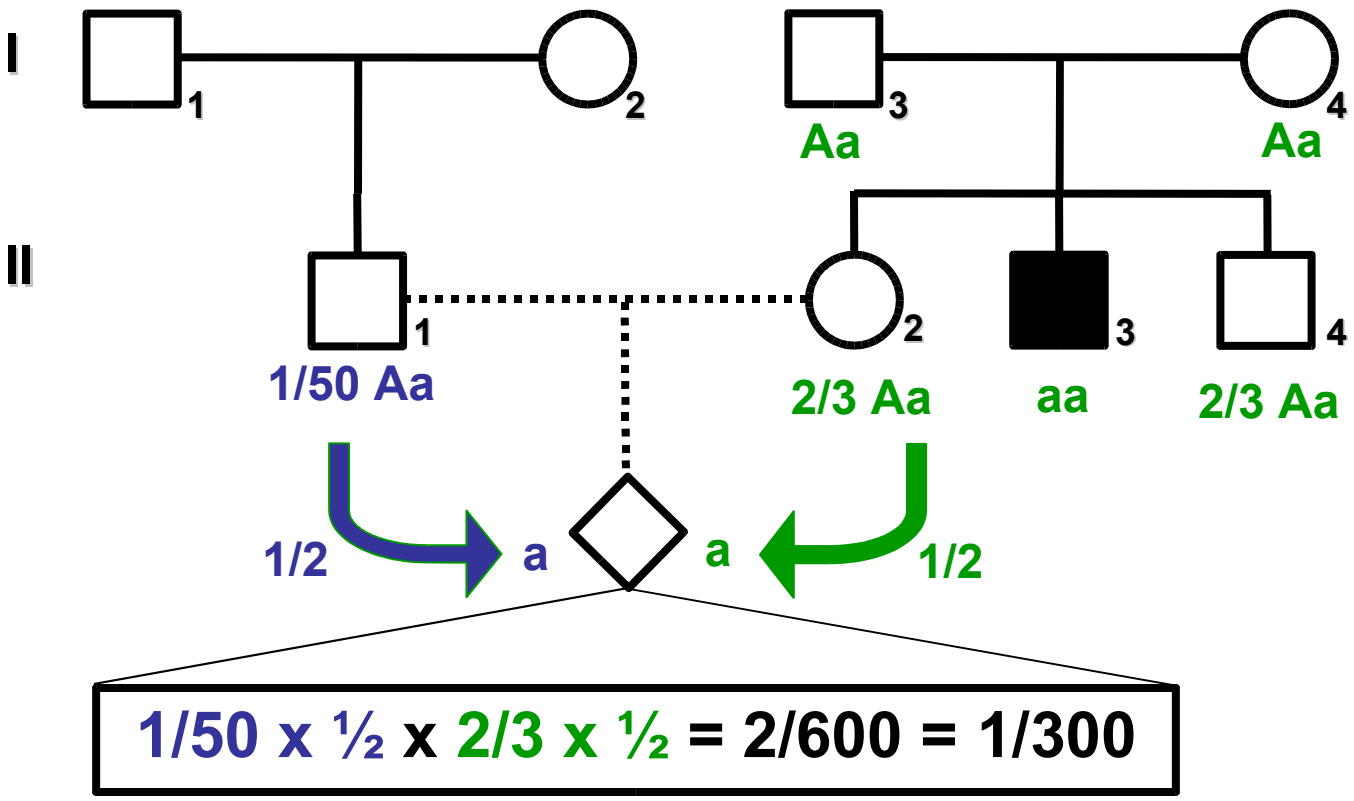


Prenatal dg. was not possible in late 1970s.

Risk 25% (i.e. > 10%) - possibility to terminate the pregnancy upon mother´s request.

In case the pregnancy continues: after the child is born, PKU screening will be performed, and put on special diet if tested positive (i.e. affected).

Task 4a, p. 134, PKU – incidence 1/10,000



$q^2 = 1/10000$

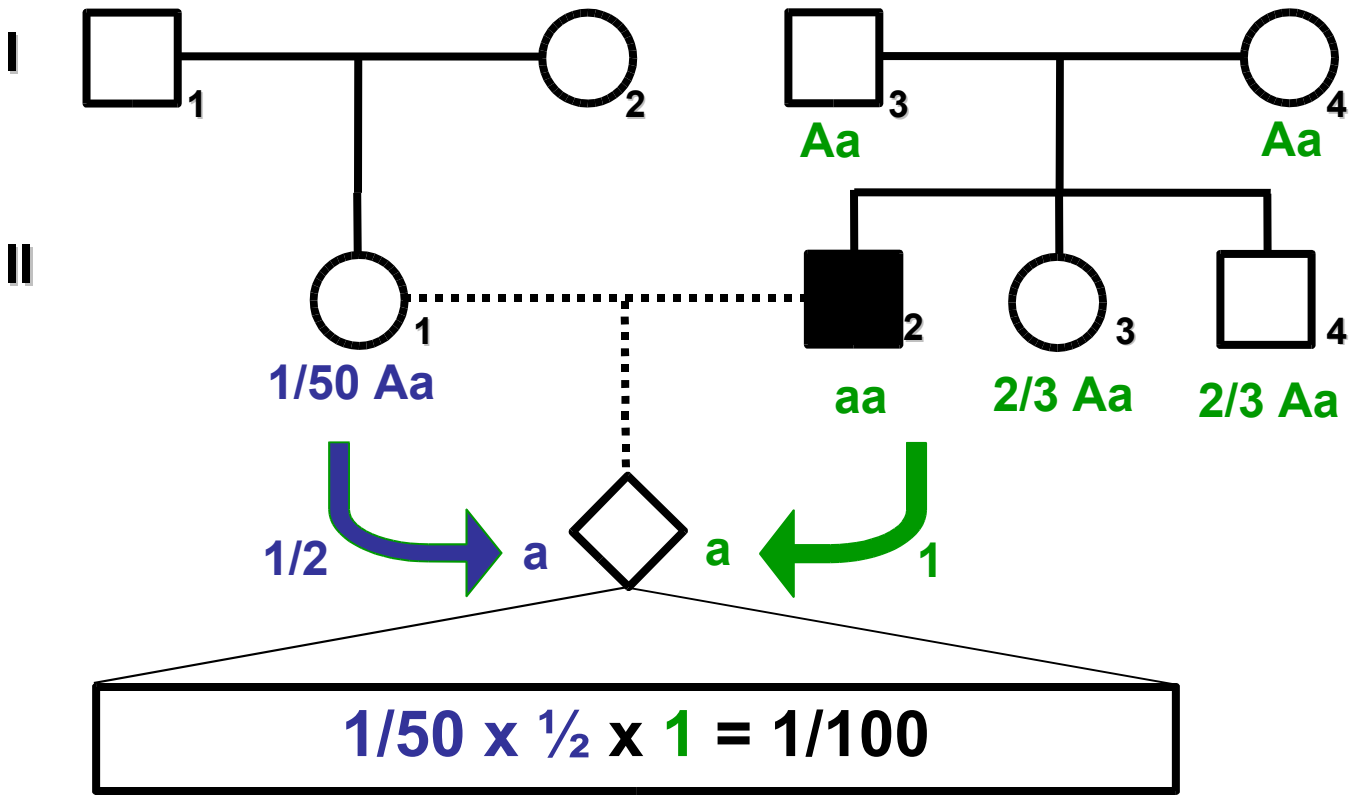
$q = 1/100$

$2pq = 2 \times 99/100 \times 1/100 = 1/50$

Low risk, DNA analysis recommended,

Importance of TIME factor….

Task 4b, p. 134, PKU – incidence 1/10,000



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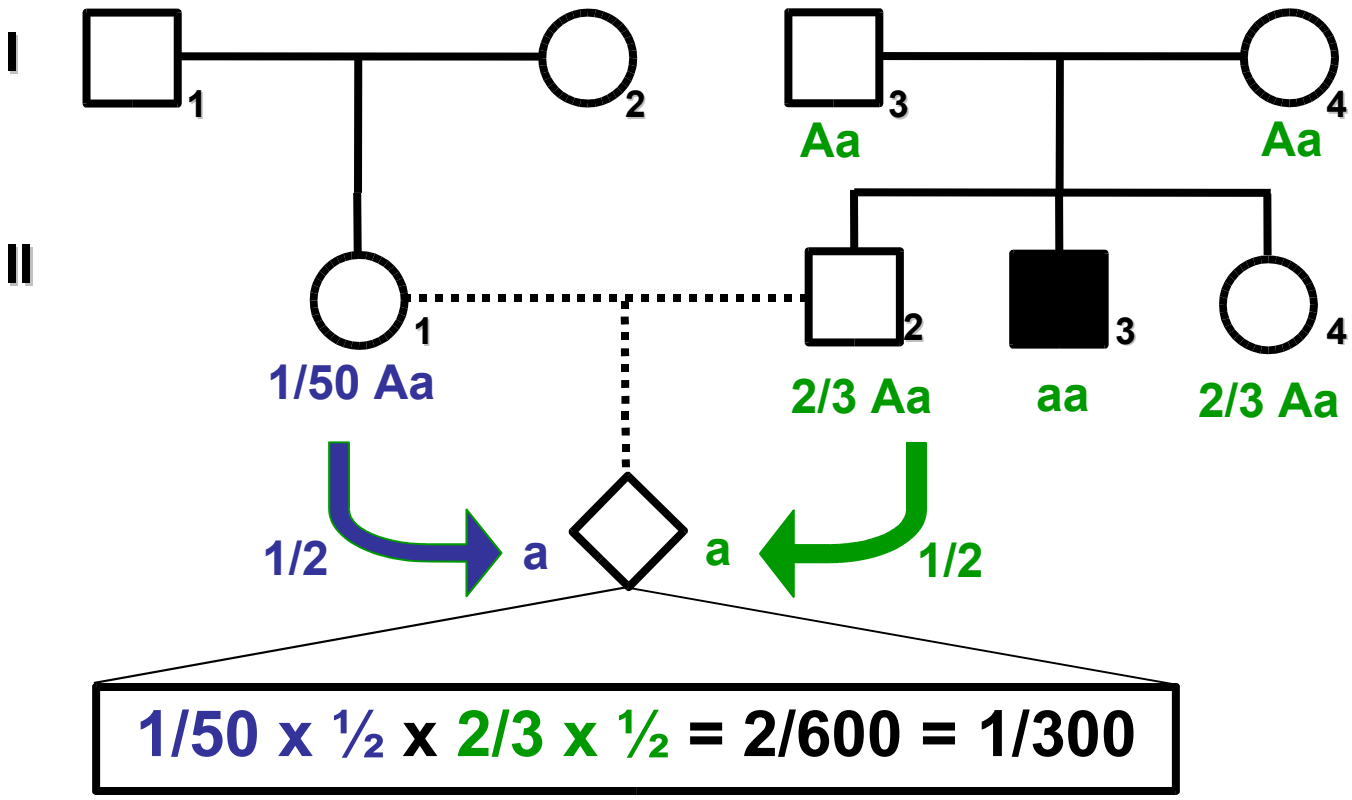
Task 4b, p. 134, PKU – incidence 1/10,000



$q^2 = 1/10000$

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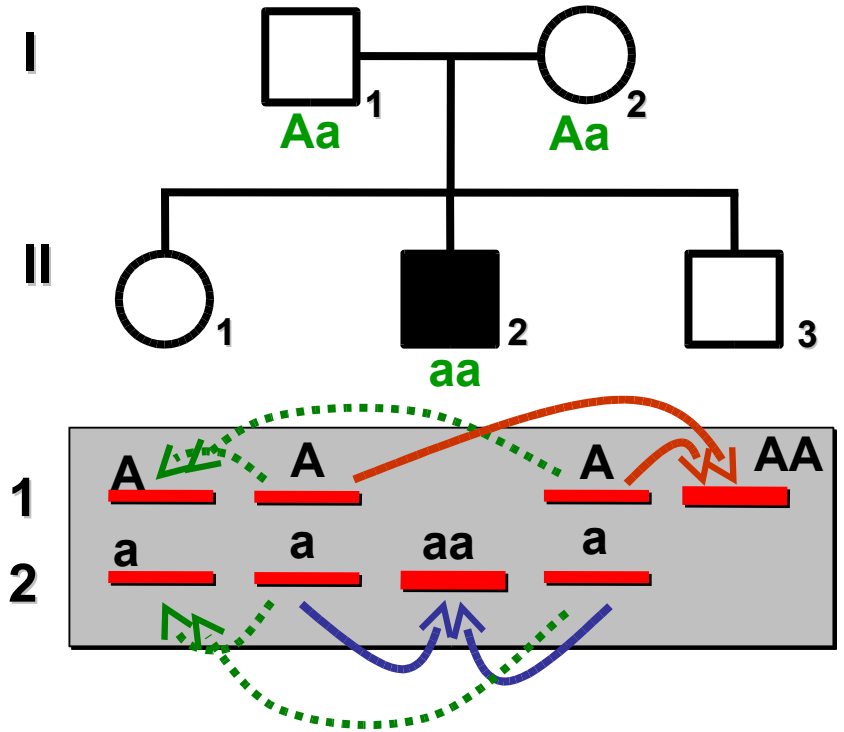
Task 6, p. 135



a) Yes, the familiy IS INFORMATIVE concerning the genotypes of II/1-3.

b) Intragenic probe, i.e. the daughter II/1 IS HETEROZYGOUS.

c) Intragenic probe, i.e. the son II/3 IS a DOMINANT HOMOZYGOTE.

d) NO, indirect diagnostics cannot be used outside the context of the family.

Task 7, p. 135

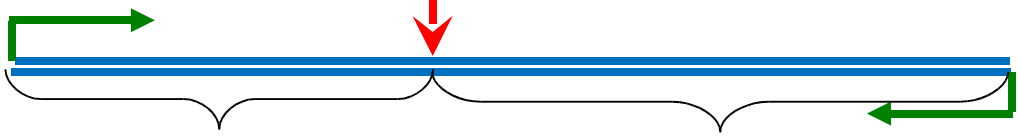
R408W
(Sty I)

StyI
5′. . . C▼C W W G G . . . 3′
3′. . . G G W W C▲C . . . 5′

Mutated allele

R408W

ProTrp
cctTGG
*



108 bp

137 bp

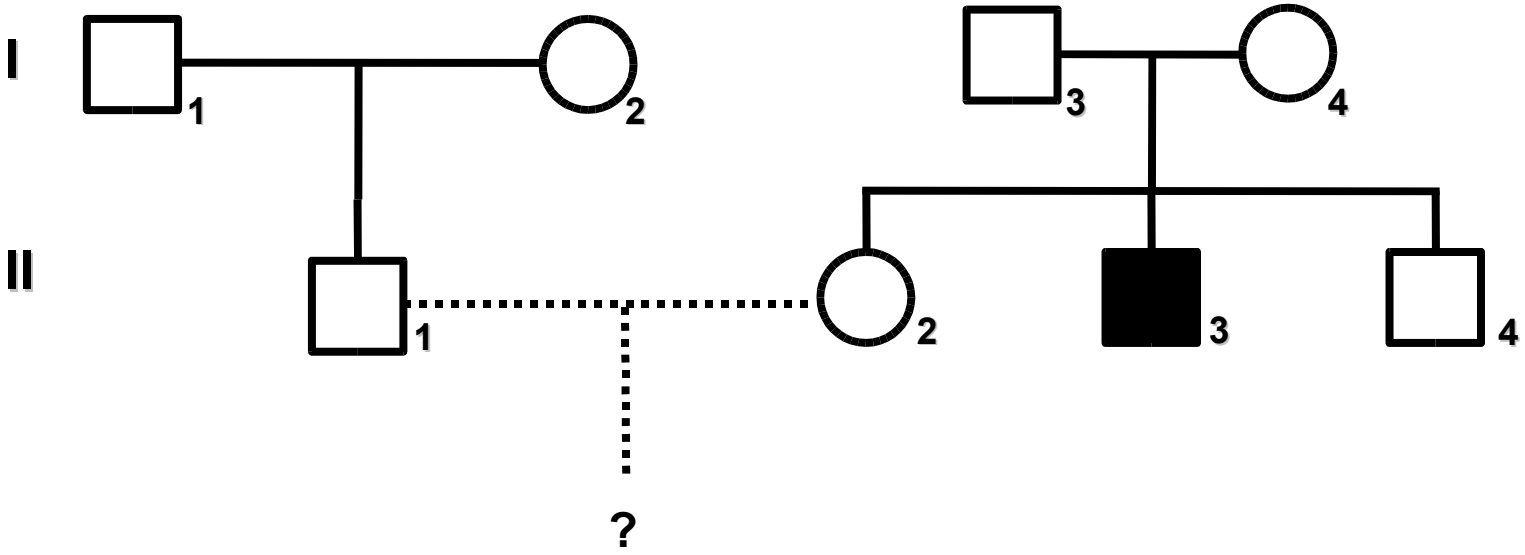
Normal allele

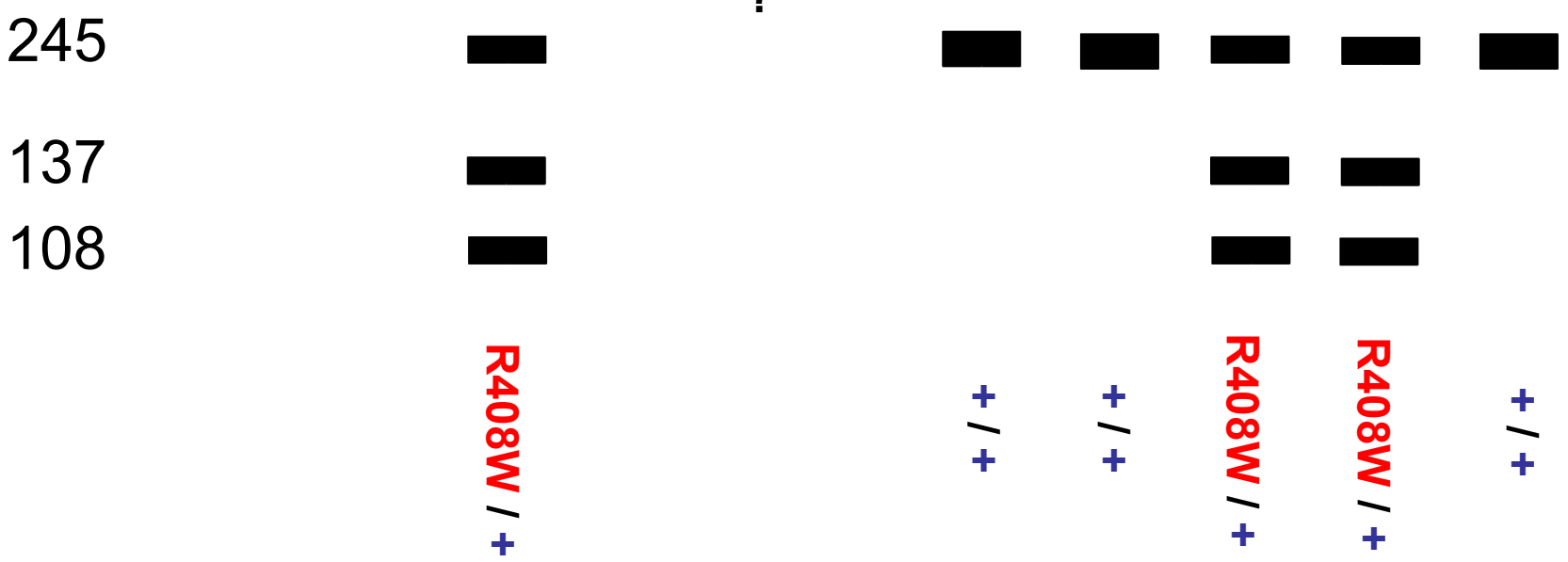
+

ProArg
cctCGG

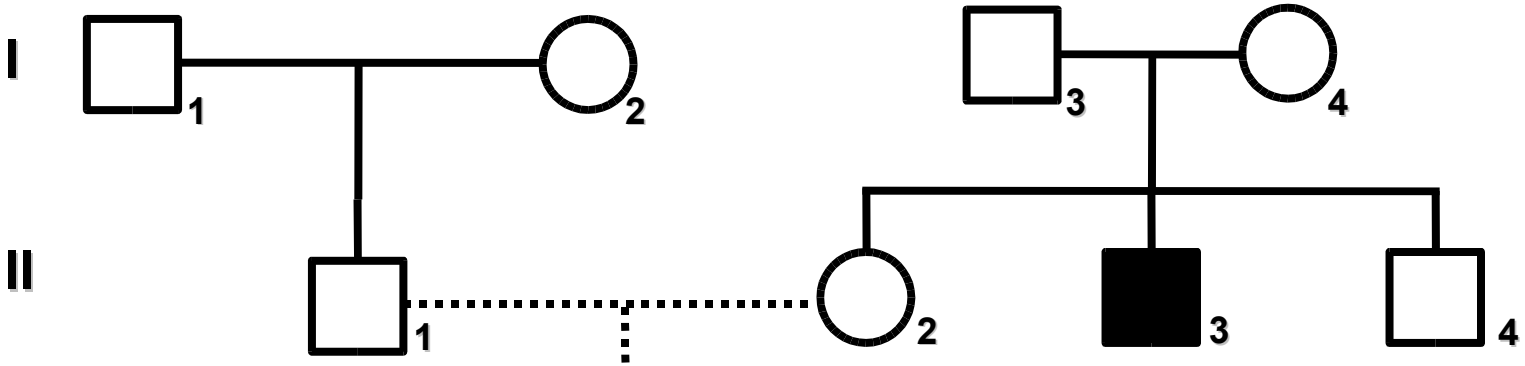
245 bp

Task 7a, p. 135





Task 7b, p. 135



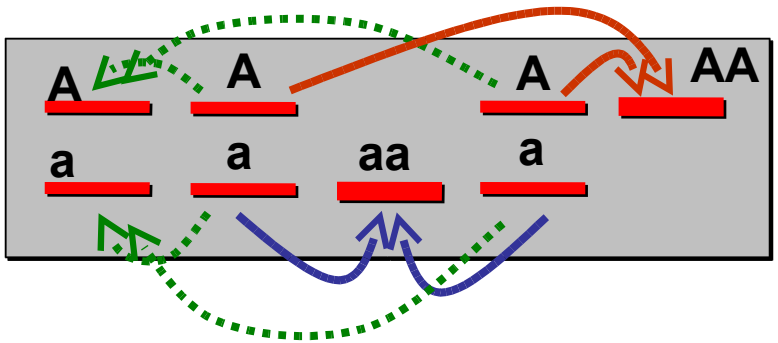
Aa

R408W / +

Aa

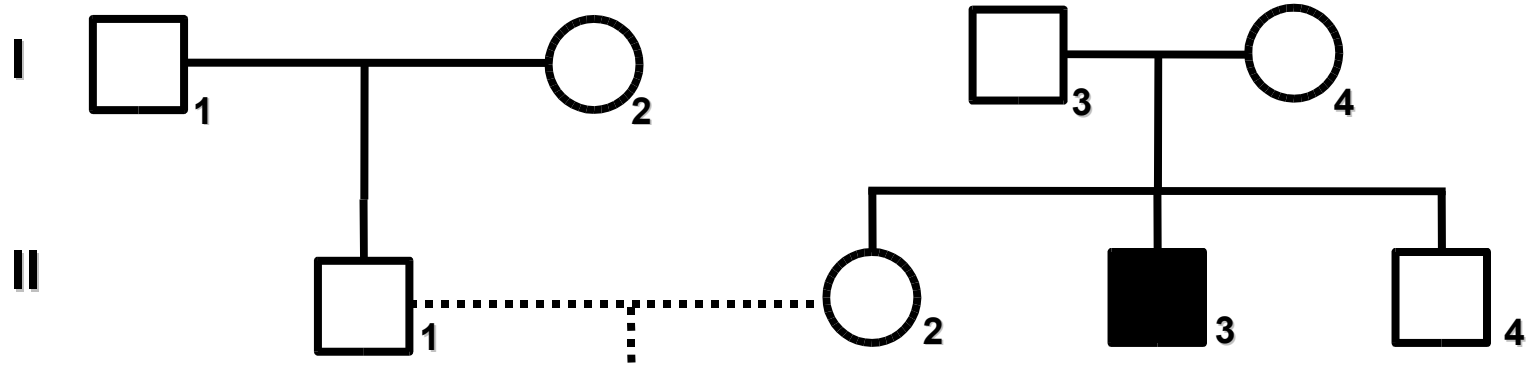
mut / +

	+	R408W
+	+/+	+/R408W
mut	+/mut	mut/R408W



1/2 Aa, 1/4 AA, 1/4 aa

Task 8, p. 136 SSCP for EXON 6.



b) CVS (10 – 12 week of pregnancy),

AMC (15th – 20th week)

PUBS - later

GENOTYPE

6mut / +

R408W / +