----------**Chapter 1**----------

(each task of this chapter is dedicated as 1.x (x meaning the exact task. Solutions should be for both editions 2005 and 2009, if answers do not match in between these two editions it is clearly stated)

1.1

Phenotypic ratios: 1:1, all alike, 3:1

1.2

Yellow – A, Green – a, complete dominance of A over a

1.3

P1 – aa, P2 – AA, F1 – Aa, F2 – AA,Aa,aa

1.4

1.4.1(Question should be rather: Another sibling of these two brothers) – 25%
1.4.2 – 50%
1.4.3 – 0% (population risk of heterozygot*2/3*1/4)

1.5

Polydactyly with luxation – recessive trait, monogenic
Normodactylic rats +/+ or Lx/+
Polydactylic rats Lx/Lx

	normodactylic	**polydactylic**	**Total**
(Lx/Lx) x (+/+)	87	0	87
(+/Lx) x (Lx/Lx)	160	160	320

1.6

Type of inheritance: monogenic
M and N are CODOMINANT

1.7

ABO test: serologic test of agglutation

Phenotype	A	B	AB	O
Genotype	AA or AO	BB or BO	AB	OO

1.7.a – 4
1.7.b – 6

1.8

Mother	**Child**	**possible fathers**	**impossible fathers**
A	A	A,B,AB,O	-
	B	B,AB	A,O
	AB	B,AB	A,O
	O	A,B,O	AB
B	A	A,AB	B,O
	B	A,B,AB,O	-
	AB	A,AB	B,O
	O	A,B,O	AB
AB	A	A,B,AB,O	-
	B	A,B,AB,O	-
	AB	A,B,AB	O
	O	impossible combination	
O	A	A,AB	B,O
	B	B,AB	A,O
	AB	impossible combination	

Mother	Child	possible fathers	impossible fathers
	O	A,B,O	AB

1.9

Genotypes: II/2 (BO), II/3 (BB), II/4 (BO), all other O bloodgroups having OO genotype
Missing genotypes: II/1 – AO, III/2 – AO, III/3 – BO, III/4 – BO, IV/2 – AO, IV/3 – BO

1.10

1.10.a – AABB (all gametes AB) crossed with aabb (all gametes ab)
1.10.b – all AaBb (gametes: AB, Ab,aB,ab)
1.10.c – 1 AABB, 2 AABb, 1AAbb, 2AaBB, 4AaBb, 2 Aabb, 1 aaBB, 2 aaBb, 1 aabb
Phenotypic ratio in F2: 9:3:3:1

1.11

Question marks in the first row: AaBb x AaBb (double heterozygotes for polydactyly $^{+/Lx}$ (Aa) and ikterism $^{+/j}$(Bb))
Question marks in the second row: AbBb x aabb, or Abbb x aaBb
Genotypes of offspring in the first row: AaBb-Aabb-aaBb-aabb
Genotypes of offspring in the second row: AaBb-Aabb-aaBb-aabb

1.12

Dihybridism of two monogenic traits
305 black, short fur (AaBb)
100 black, Angora fur (aaBb)
108 white, short fur (Aabb)
32 white, Angora fur (aabb)

Their parents were double heterozygotes: AaBb x AaBb, both black with short fur

1.13

1.13.a – genotypes of puppies: 50% BbSs and 50%Bbss, parent female – bbss, parent male – BBSs (if there was just few puppies then the male parent could have also been BbSs)

1.13.b.1 – yes (BbSs x BbSS parents)

1.13.b.2 – yes (bbss x BbSs parents)

1.14

Mother	Child	possible Father	excluded Father
O,M	O,MN	A, B, O, N, MN	AB, M
O,Rh^+	O,Rh^-	A, B, O, Rh^+, Rh^-	AB
O,Rh^-	A,Rh^+	A, AB, Rh^+	O, B, Rh^-
O,MN	B,MN	B, AB, N, M, MN	O, A
A,N	O,MN	A, B, O, M, MN	AB, N
A,MN	A,N	A, B, O, AB, MN, N	M
A,Rh^+	B,Rh^-	B, AB, Rh^+, Rh^-	A, O
A,Rh^-	A,Rh^+	A, B, AB, O, Rh^+	Rh^-
A,N	AB,MN	B, AB, MN, M	A, O, N
B,MN	O,N	A, B, O, MN, N	AB, M
B,Rh^+	B,Rh^-	A, B, O, AB, Rh^+, Rh^-	None
B,Rh^-	B,Rh^-	A, B, AB, O, Rh^+, Rh^-	None
B,M	O,M	A, B, O, MN, M	AB, N
AB,N	A,N	A, B, AB, O, MN, N	M
AB,Rh^+	B,Rh^-	A, B, AB, O, Rh^+, Rh^-	None
AB,Rh^-	AB,Rh^+	A, B, AB, Rh^+	O, Rh^-

Mother	Child	possible Father	excluded Father
AB,MN	AB,M	A, B, AB, MN, M	O, N

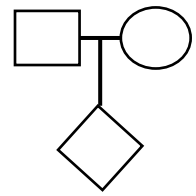
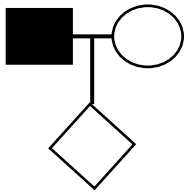
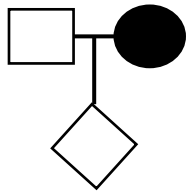
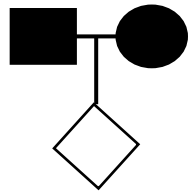
----------**Chapter 2** ----------

(each task of this chapter is dedicated as 1.x (x meaning the exact task. Solutions should be for both editions 2005 and 2009, if answers do not match in between these two editions it is clearly stated)

2. 1

Your own family pedigree.

2.2

	Autosomal dominant inheritance	**Autosomal recessive inheritance**	**Gonosomal recessive inheritance**
	aa x aa, risk for offspring close to 0	AA x AA –risk for offspring close to 0 Aa x AA – risk for offspring close to 0 Aa x Aa – 25 % risk for offspring having aa genotype	X^+Y x X^+X^+ - risk for offspring close to 0 X^+Y x X^-X^+- risk only for boys – 25% having X^-Y genotype
	Aa x aa, risk for offspring having Aa genotype 50%	aa x AA – risk close to 0, aa x Aa – 50% risk for offspring having aa genotype	X^-Y x X^+X^+ - risk close to 0, all girl offspring will be „only“ carriers X^-X^+
	aa x Aa, risk for offspring having Aa genotype 50%	AA x aa – risk close to 0, Aa x aa – 50% risk for offspring having aa genotype	X^+Y x X^-X^- - risk for offspring depends on sex, all girls will be carriers, all boys will be affected
	Aa x Aa, risk for offspring 75% if AA genotype is compatible with life, or 66% for all viable fetuses, AA x AA (rare combination) – 100% risk AA x Aa (rare combination) – 100% risk	ˇ aa x aa – 100% risk for offspring having aa genotype	X^-Y x X^-X^- - rare combination, risk for offspring 100%

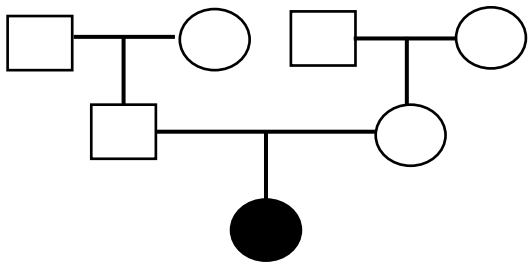
2.3

Types of matings		Genotypes of the progeny (%)		
		AA	**Aa**	**aa**
1.	AA x AA	100		
2.	AA x Aa	50	50	
3.	AA x aa		100	
4.	Aa x Aa	25	50	25
5.	Aa x aa		50	50
6.	aa x aa			100

Autosomal dominant traits:
- Most common type Aa x aa,
- Aa x Aa mating appears in assortative matings.
- AA genotype is not very common in medical counseling.

Autosomal recessive traits:
- Most common type Aa x Aa (two heterozygotes come to clinical genetics department because their child has AR disease.)
- Aa x aa is also possible.
- aa x aa is possible, but very rare.

2.4



1. It could be AR inheritance due to Aa x Aa mating
2. It could be AD with „de novo“ mutation
3. It could be XD with „de novo“ mutation
4. It could be mtDNA inheritance, heteroplasmy
5. It does not necesserily need to be genetic cause, phenocopy or other

2.5

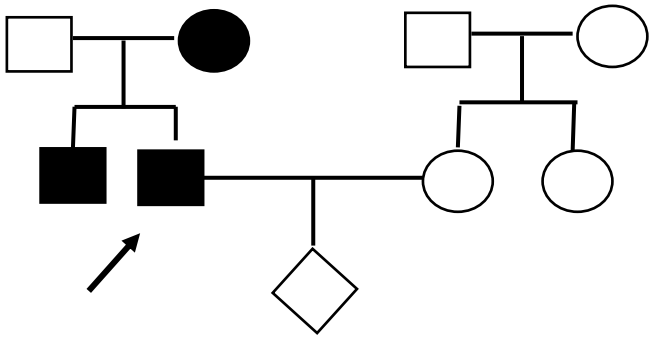
2.5.b – A autosomal dominant, III/6 50%, IV/1 50%, IV/2 0%

2.5.b – B autosomal recessive, III/6 25%,

IV/1 depends on the partner: AA – 0%
Aa – 50%
20% aa – 100%

IV/2 again it depends on the partner: AA – 0%
Aa – 2/3*1/4
aa – 2/3*1/2

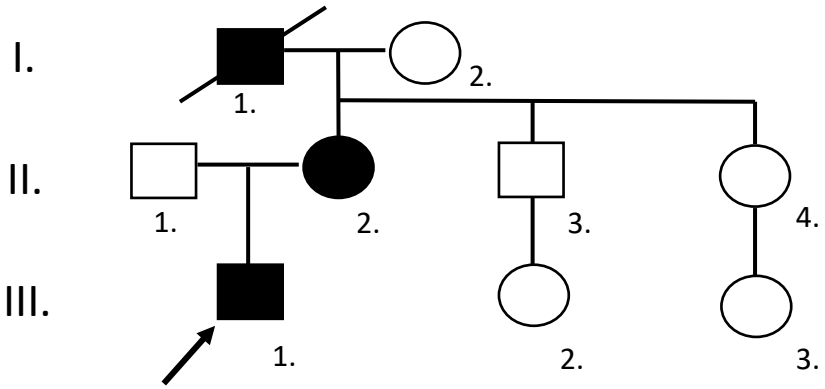
2.6



2.6.a – Aa, aa
2.6.b – 50%
2.6.c – 50%
2.6.d – 25%

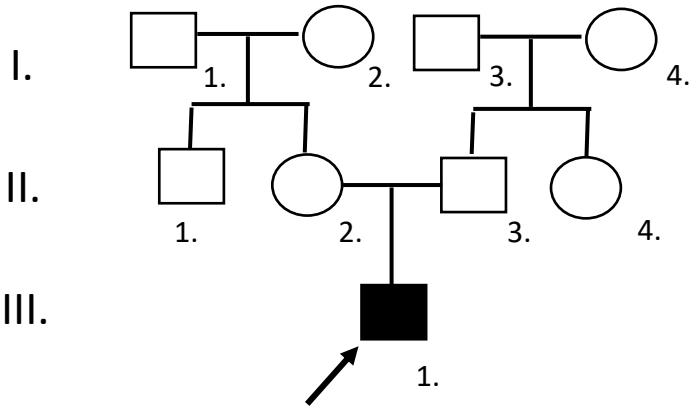
2.7

Cumulative risk for a family with defect in Hungtingtin gene is 100%.



I/2 – 0%
II/1 – 0%
II/3 – 80% if a carrier of Aa genotype
II/4 – 50% if a carrier of Aa genotype
III/2 – 20% if a carrier of Aa genotype
III/3 – very low risk at the beginning, but if a carrier of Aa it will steeply increase

2.8



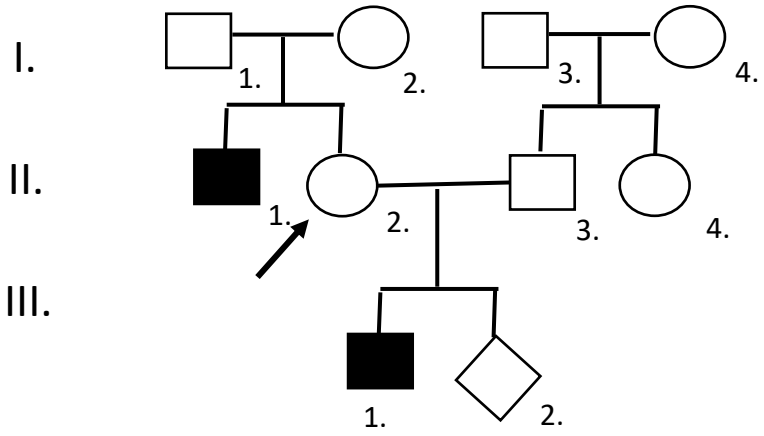
2.8.a – parents Aa, Aa
2.8.b – 25%
2.8.c – 2/3, 66,6%
2.8.d – Aa x Aa, Aa x aa, aa x aa

2.9

Haemophilia A or B:

1. $X^+X^+ \times X^+Y$ – close to 0% of affected offspring
2. $X^hX^+ \times X^+Y$ – 50% of sons and in total 25% of children affected
3. $X^+X^+ \times X^hY$ – all daughters are carriers but none of the children is affected
4. $X^hX^+ \times X^hY$ – 50% of sons affected, 50% of daughters affected
5. $X^hX^h \times X^+Y$ – all sons affected, all daughters carriers

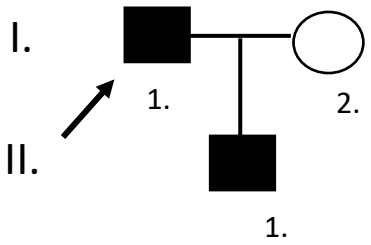
2.10



2.10.a – 25%
2.10.b – 50%
2.10.c – 0%

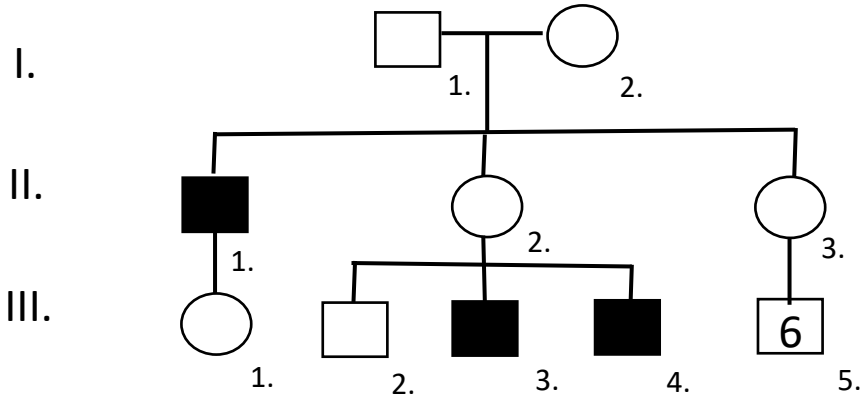
2.11

I/1. – $X^{d-}Y$
I/2. – $X^{d-}X^+$
II/1. – $X^{d-}Y$



2.11.a – 50%
2.11.b – 50%

2.12



2.12.b – X linked recessive inheritance

2.12.c – I/1 $X^{+}Y$

I/2 $X^{d}X^{+}$

II/1 $X^{d}Y$

II/2 $X^{d}X^{+}$

II/3 $X^{+}X^{+}$

III/1 $X^{d}X^{+}$

III/2 $X^{+}Y$

III/3 a III/4 $X^{d}Y$

III/5 $X^{+}Y$

2.13 – The probability is 0. Fathers give Y chromosomes to their sons

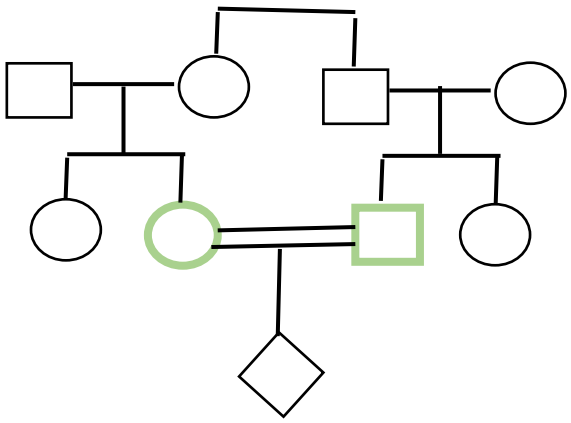
2.14 – In this large family we can see that Duchenne muscular dystrophy affected only sons, which is compatible with X- linked recessive disorders. Surprisingly (maybe a mistake of the authors), all male affected individuals from the second and third generation are still alive. The average life span of people affected with this disease is 30 – 40 years of age. We can also see that none of the affected individuals was not capable of having offspring.

2.12.b – women 100% carriers: I/2, II/3, II/7, III/5, III/8.

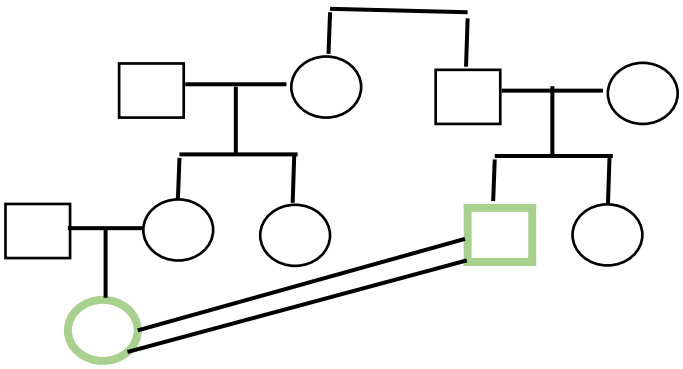
2.15 – Disease „hypophosphatemic rickets“ is and XD disease as well presented in both of the pedigrees. The only problems arises in the first pedigree in Fig. No 2/9 where I/1 or I/2 are not affected. The mutant allele was most probably present in the father I/1 as a mosaic, surely present in gonads. He was not affected but trasmitted the disease to all of his three daughters. Both pedigrees show typical characteristics thereafter of the XD inheritance. More females than males are affected, in every generation we see somebody effected, it never passes from an affected male to his male offspring.

2.16

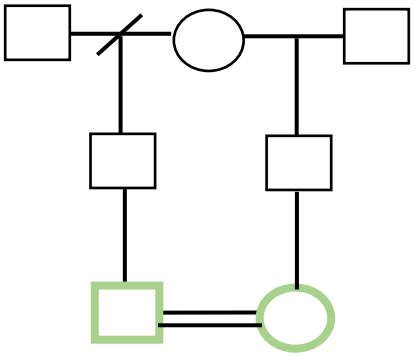
2.16.a - r = 1/8



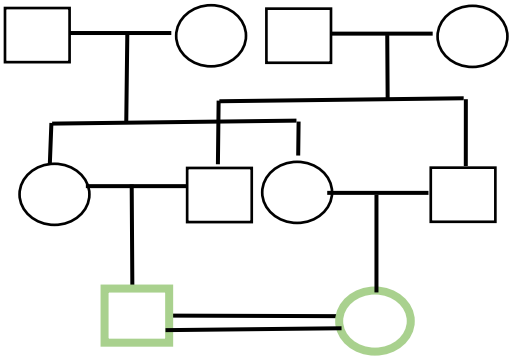
2.16.b – r = 1/16



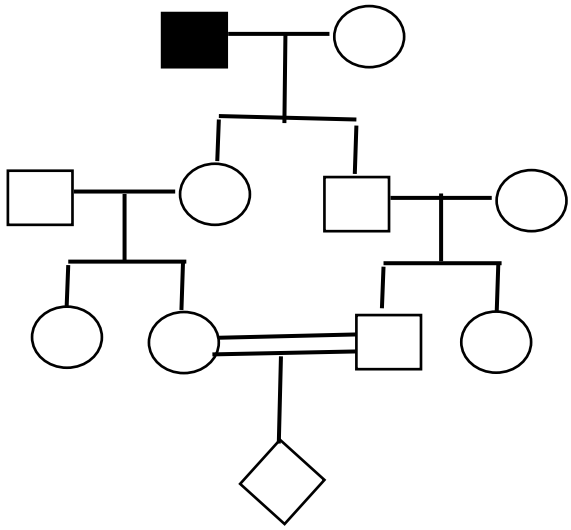
2.16.c – r = 1/16



2.16.d – r = 1/8 (only in 2005 edition)

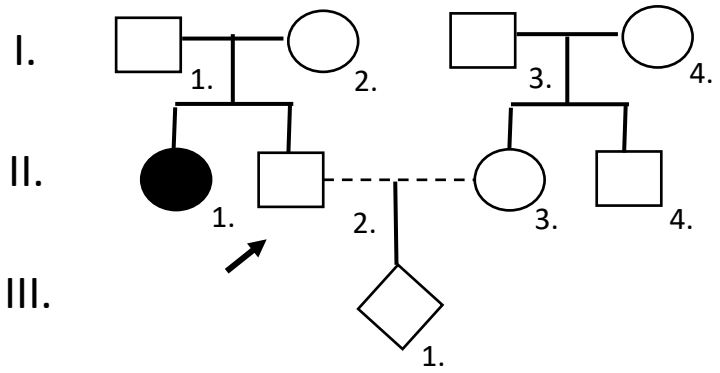


2.17



2.17.a – PKU for IV/1 – 1/16
2.17.b – Brachydactyly for IV/1 – close to 0%
2.17.c – Haemophilia A for IV/1 – 25% for boys, 0% for girls

2.18



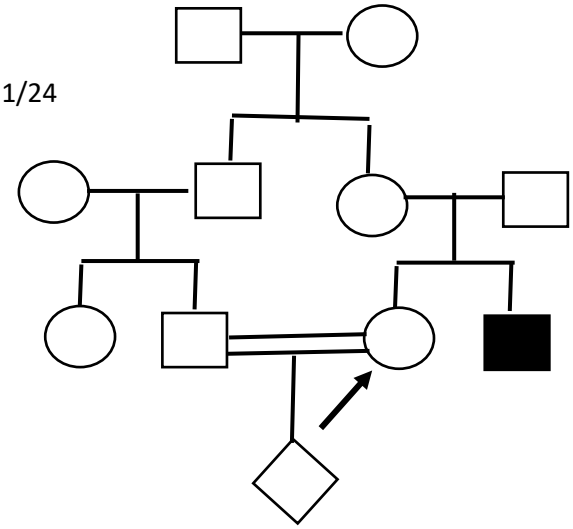
2.18.a – I/1 Aa 100%, I/2 Aa 100%, I/3 unknown AA most probable, I/4 unknown AA most probable
– II/1 aa 100%, II/2 AA for 33,3% and Aa for 66,6%, II/3 AA most probable (Aa depends on the incidence of PKU in the population), II/4 AA most probable
- III/1 will be more probably AA (2/3); Aa genotype less probable (1/3), aa possibility close to 0%.

2.18.b – very low (2/3 * (2*0.01*0.99)*1/4) [0.0033]

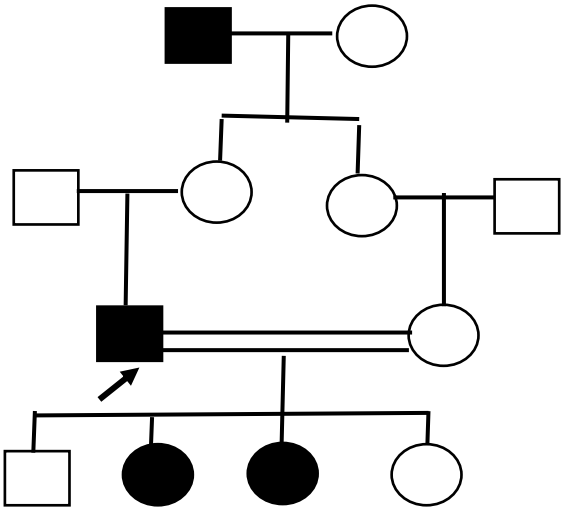
2.18.c – the use of direct or indirect DNA diagnostic methods could help to determine ones genotype

2.19

Risk = 1/2*1/2*2/3*1/4 = 1/24

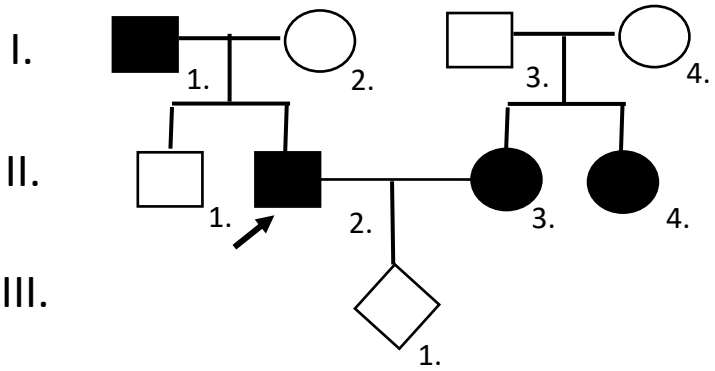


2.20



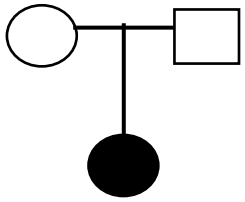
2.20.b – because mother of IV/2 and IV/3 is a carrier X^hX^+ . Both of these daughters recieved X^h allele from both of the parents (father gave one X^h and mother gave the other one X^h).

2.21



Hearing disability or deafness are very heterogenious disorders and not always there is a real genetic cause and if there is one, there are different modes of inheritance that can apply. From our pedigree it seems that family on the left side represents AD inheritance (defect in one allele of gene 1) whereas the family on the right side represents AR inheritance (defect in both alleles of gene 2). The risk for the child III/1 is thus 50% (because of the AD inheritance of deafness in family on the left side).

2.22 (only in 2005 edition)



AGS
Healthy homozygote: A10A26B12B18
Heterozygote: A3A10B17B18 or A26B12B40
Affected homozygote: A3A26B17B40

----------**Chapter 3**----------

All tasks concern observation of mitotic samples on microscope