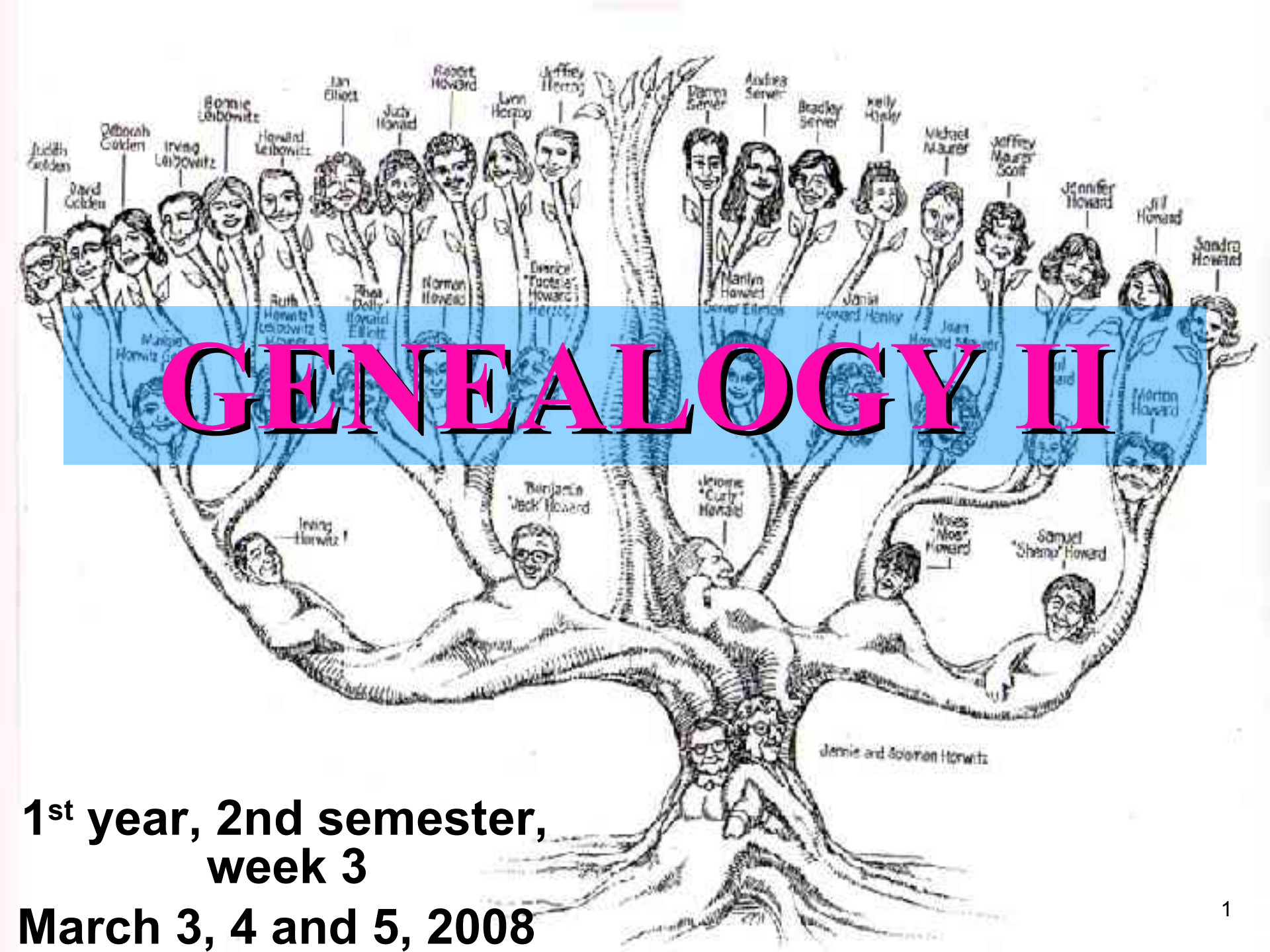


GENEALOGY II

1st year, 2nd semester, week 3

March 3, 4 and 5, 2008

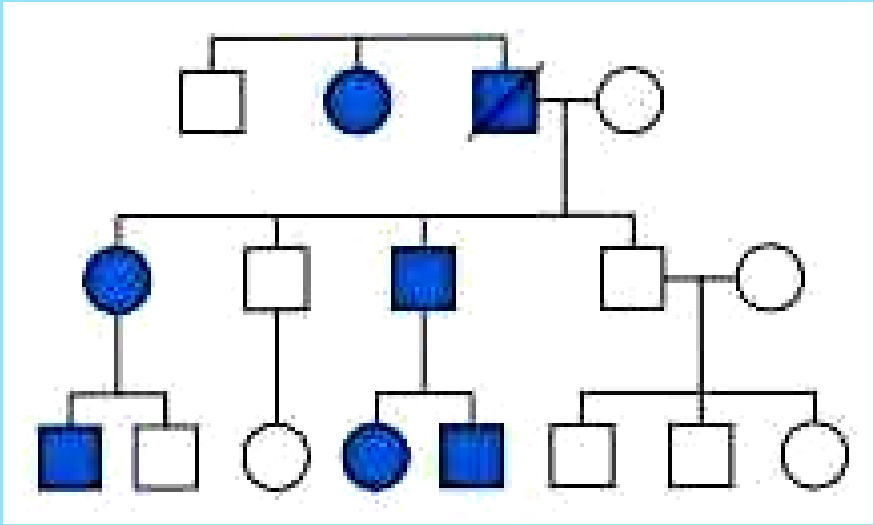
AUTOSOMAL DOMINANT INHERITANCE - AD

Characteristics:

- direct transmission from an affected parent to an affected child (does not skip generations)
- males and females are equally likely to be affected
- transmission from father to son

Examples:

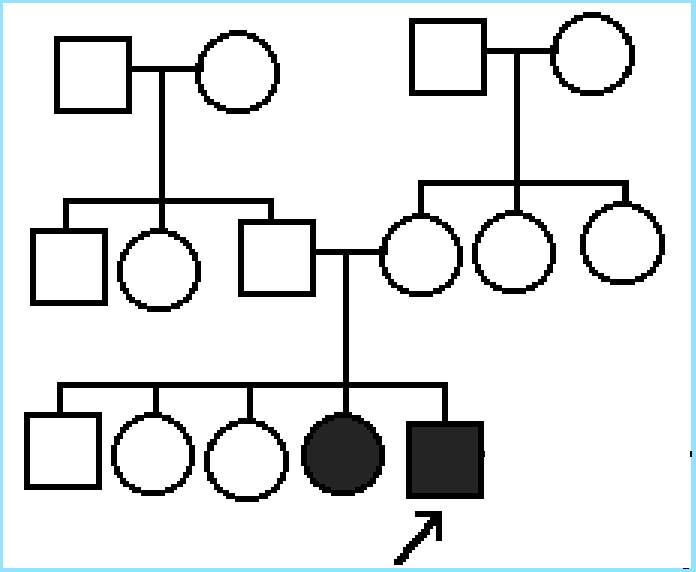
- achondroplasia, brachydactyly
- polycystic kidney disease
- familial hypercholesterolemia
- dentinogenesis imperfecta, osteogenesis imperfecta
- dysostosis cleidocranialis



AUTOSOMAL RECESSIVE INHERITANCE - AR

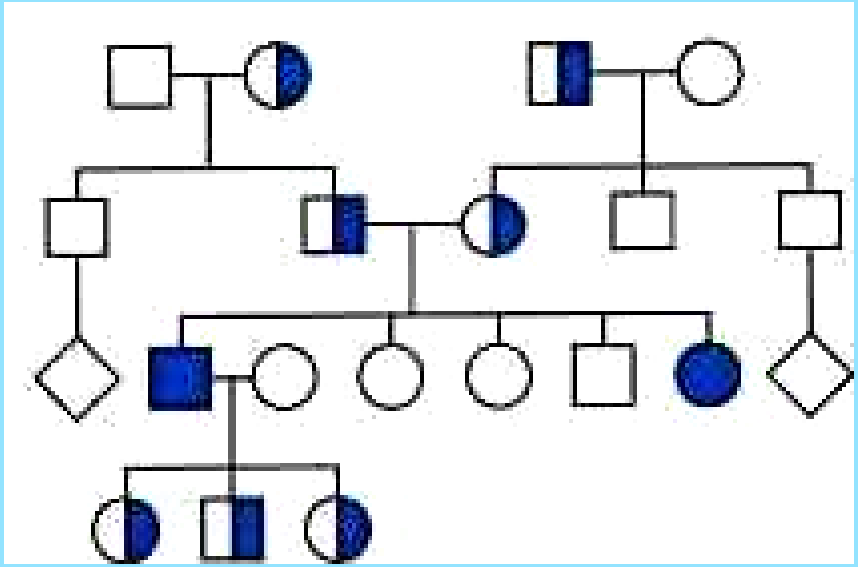
Characteristics:

- the trait is often found in clusters of siblings but not in their parents and offspring
- males and females are equally affected
- parents could be relatives



Examples:

- cystic fibrosis
- phenylketonuria
- sickle cell anemia
- albinism



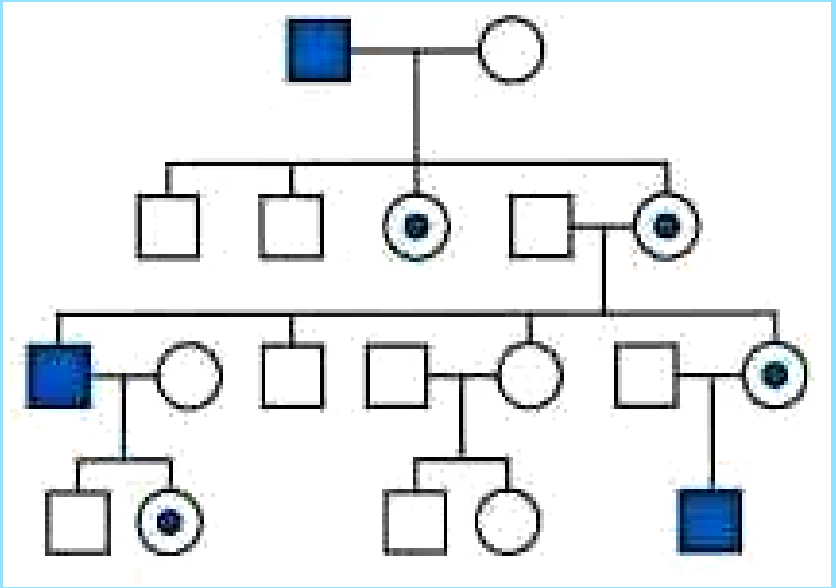
X-LINKED RECESSIVE INHERITANCE (GONOSOMAL RECESSIVE – GR)

Characteristics:

- males are much more likely to be affected
- affected males get the disease from their mothers – healthy carriers
- no transmission from father to son
- transmission from an affected grandfather to his grandsons

Examples:

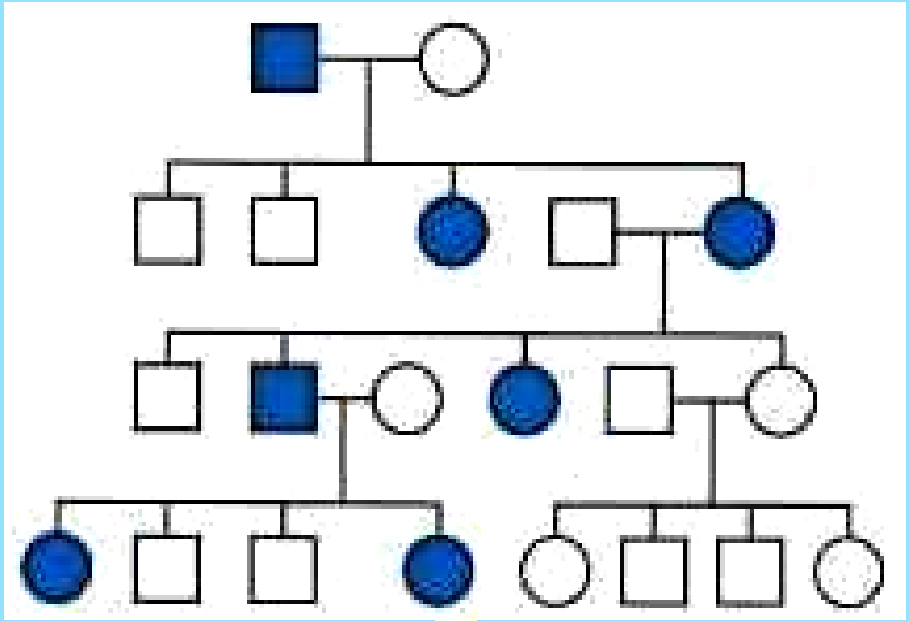
- haemophilia A, haemophilia B
- Duchenne muscular dystrophy
- color blindness
- anhidrotic ectodermal dysplasia



X-LINKED DOMINANT INHERITANCE (GONOSOMAL DOMINANT) - GD

Characteristics:

- direct transmission from generation to generation
- females are more likely to be affected (two X chromosomes)
- males are usually more severely affected than females
- no transmission from father to son



Examples:

- vitamin D-resistant rickets
- incontinentia pigmenti
- amelogenesis imperfecta

(X-linked)

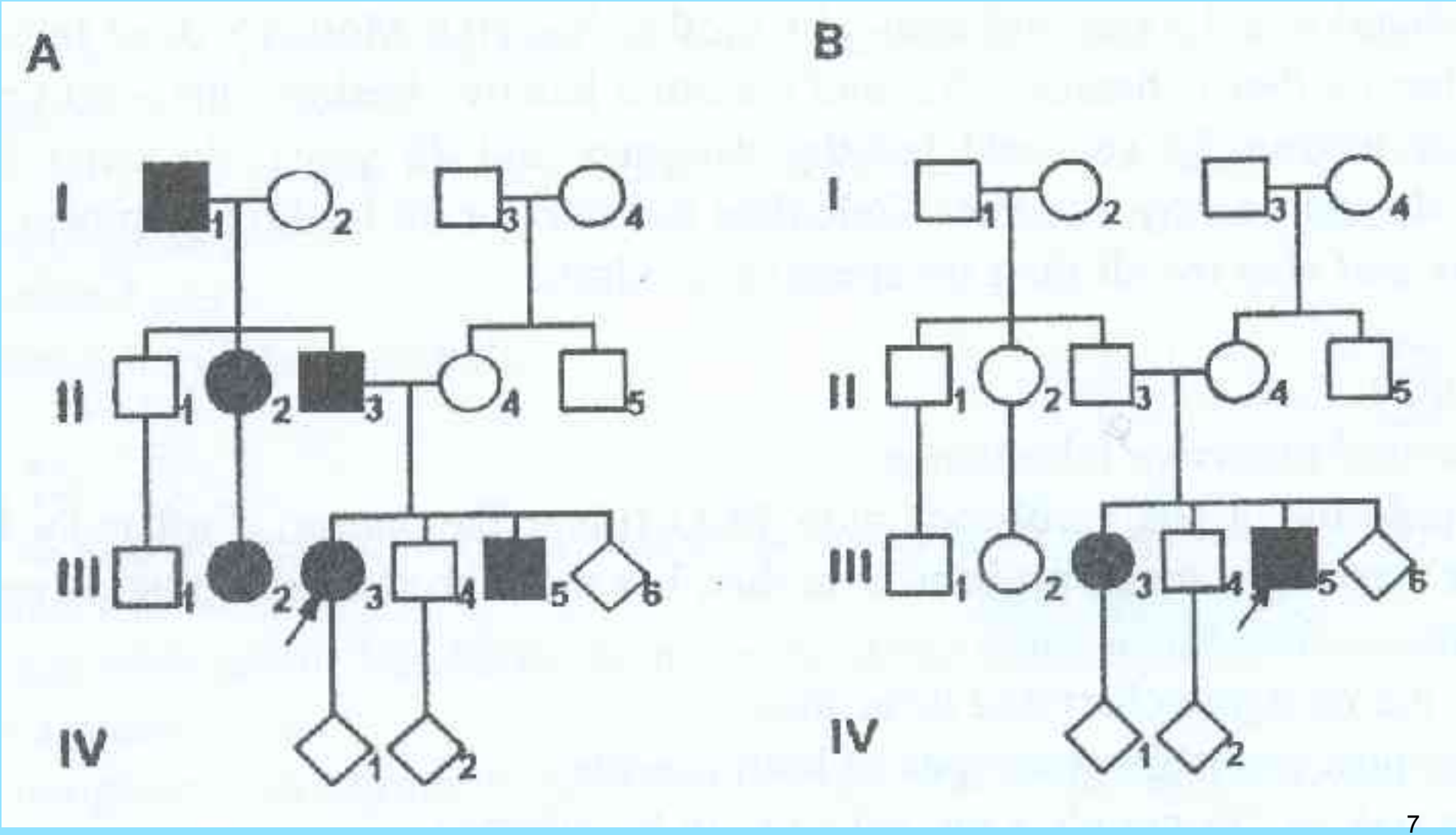
Types of mating - Autosomal inheritance (*KrOt* p. 16/Task 3)

Genotypes of the couple	Genotypes of the progeny (%)		
	AA	*Aa*	*aa*
AA × *AA*	100		
AA × *Aa*	50	50	
AA × *aa*		100	
Aa × *Aa*	25	50	25
Aa × *aa*		50	50
aa × *aa*			100

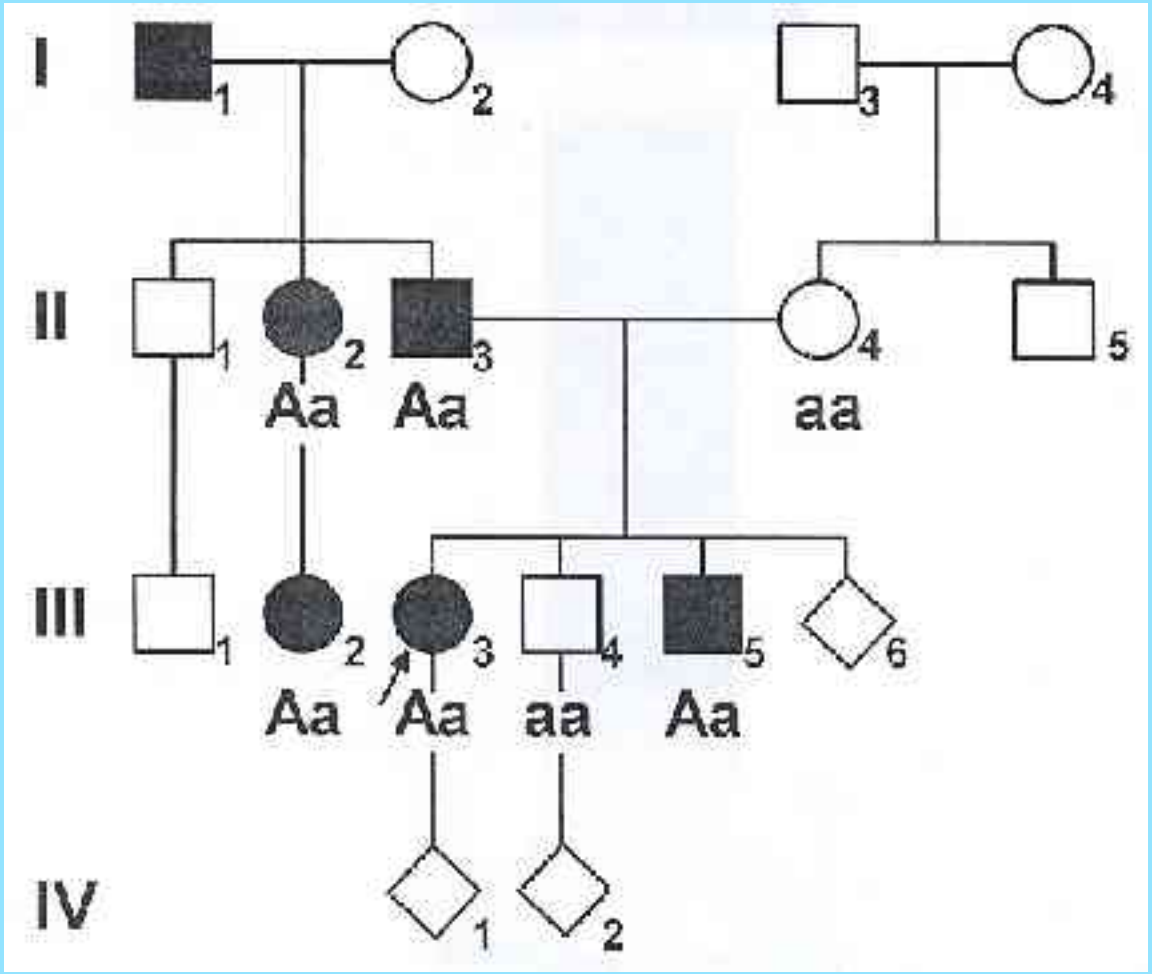
Phenotype	dominant	recessive

Most important in
AR
AD

Typical Autosomal Pedigrees (*KrOt* p. 17/Task 5b)



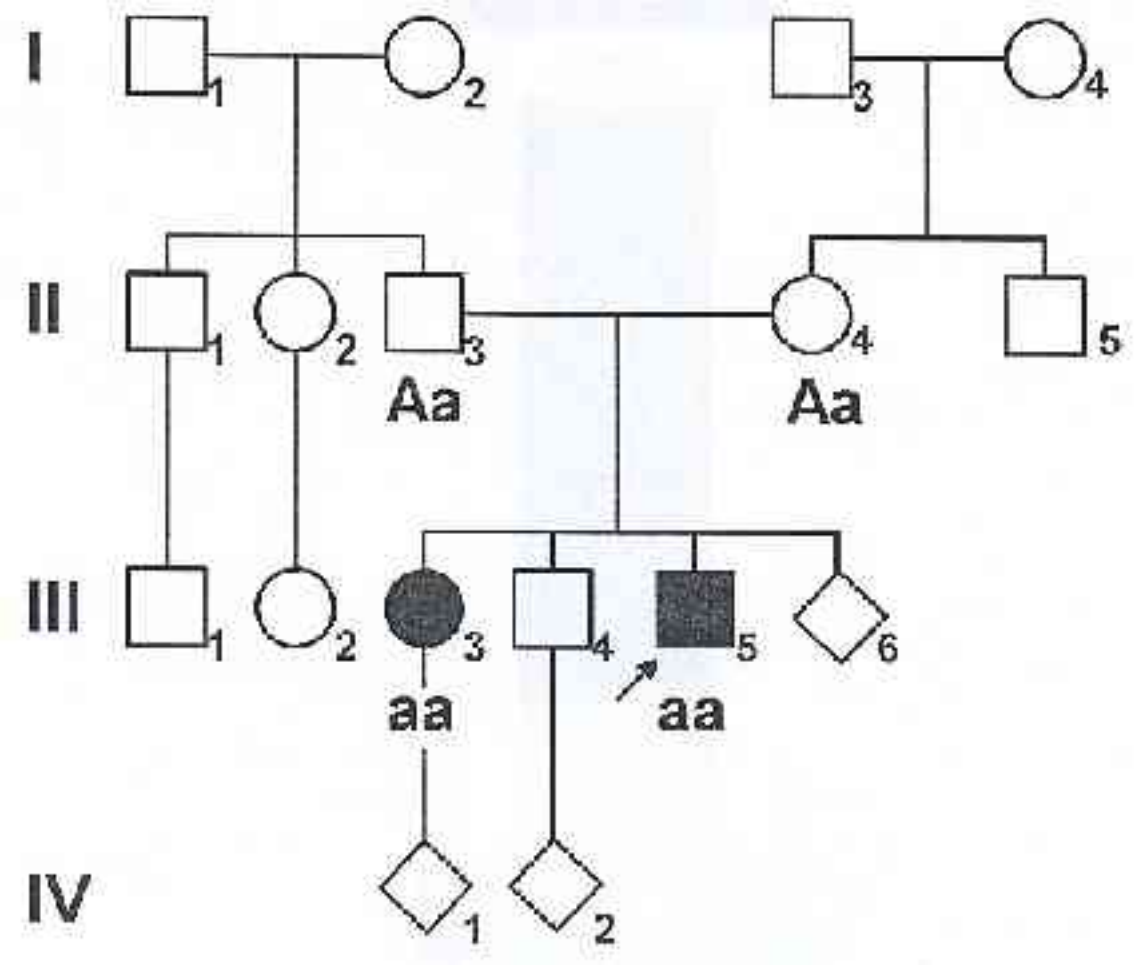
Typical Autosomal Pedigrees (*KrOt* p. 17/Task 5b)



Pedigree A
is typical for
(compatible with)
AD

Risk for:
III/6 is 50 %
IV/1 is 50 %
IV/2 is 0 %

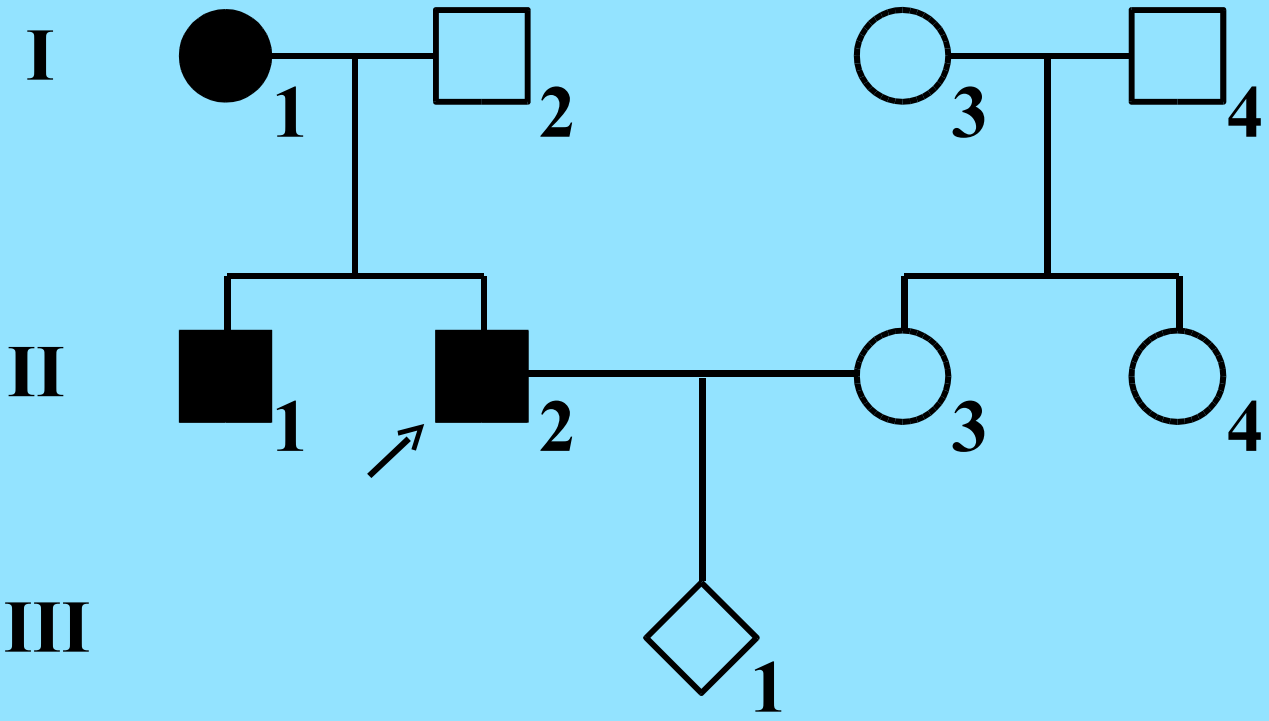
Typical Autosomal Pedigrees (*KrOt* p. 17/Task 5b)



Pedigree B
is typical for
(compatible with)
AR

Risk for:
III/6 is 25 %
IV/1 is ≅ 0 %
IV/2 is ≅ 0 %

Brachydactyly - Autosomal Dominant Inheritance in Pedigree (*KrOt* p. 17/Task 6)



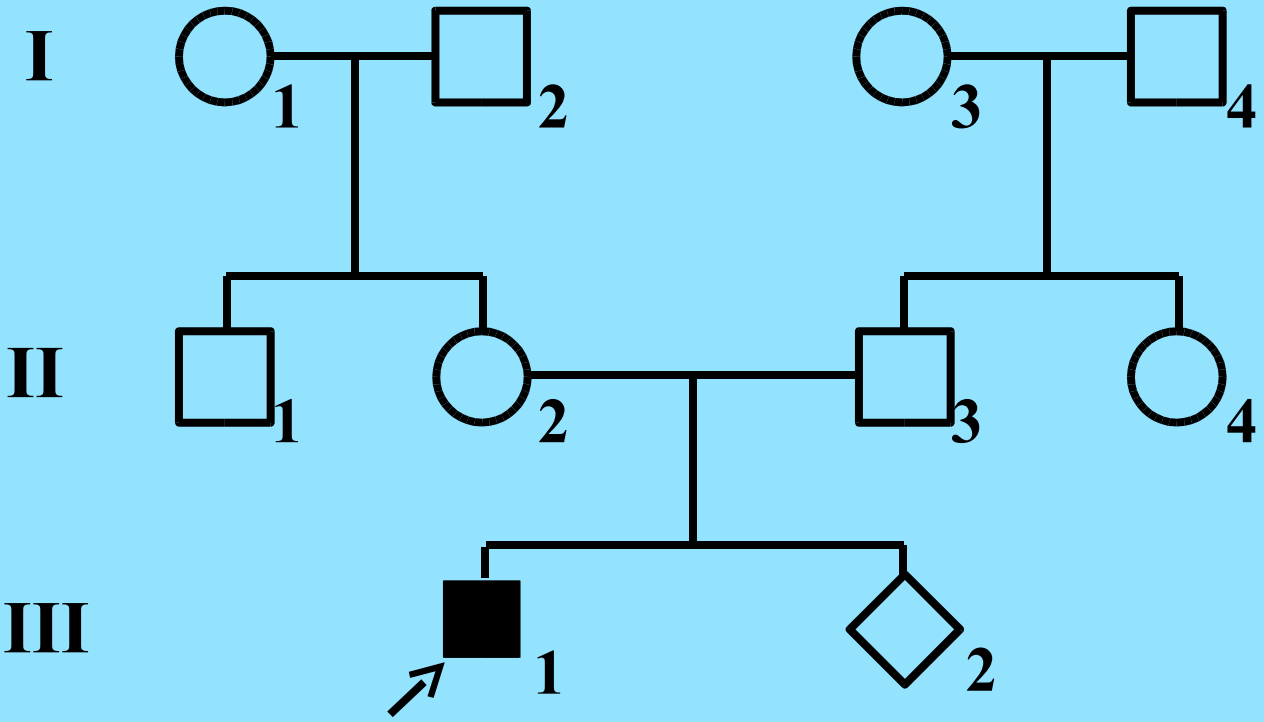
a) II/2 – *Aa*, II/3 - *aa*

b) 50%

c) 50%

d) 25%

PKU - Autosomal Recessive Inheritance in Pedigree (*KrOt* p. 18/Task 8)



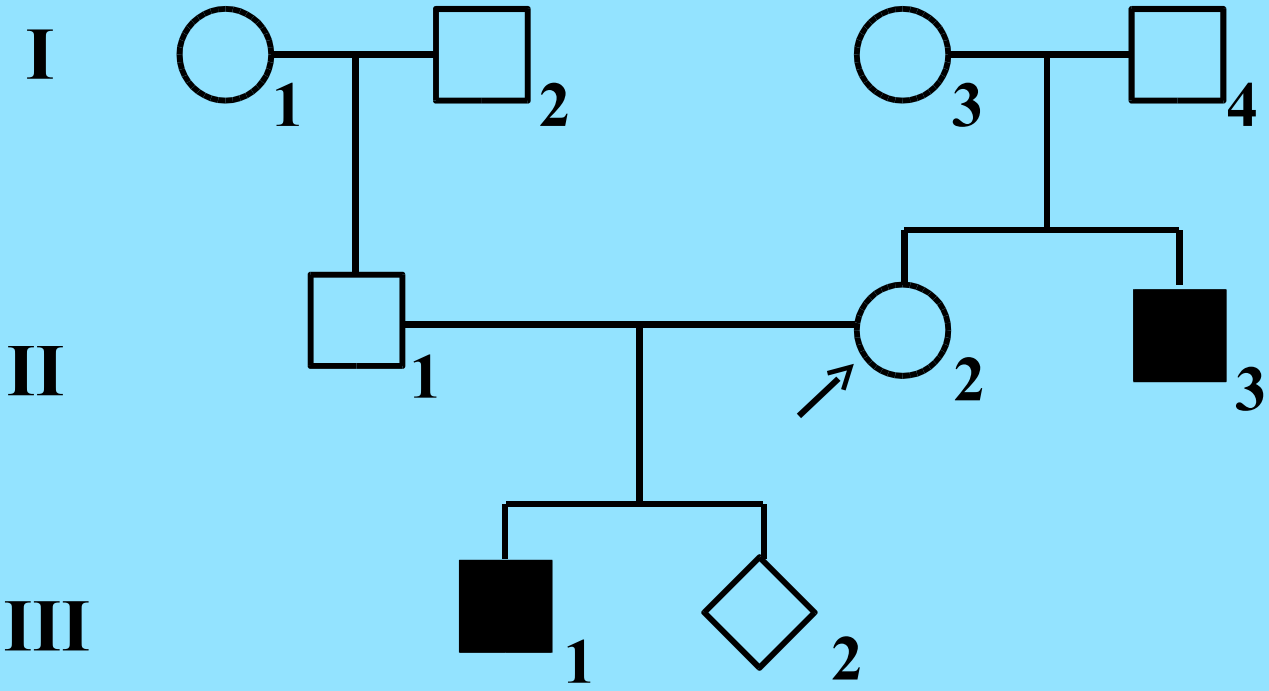
a) II/2, II/3 – both *Aa*

b) 25%

c) 2/3

d) *Aa* × *Aa*, *Aa* × *aa*, *aa* × *aa*

Haemophilia A – Gonosomal (X-linked) Recessive Inheritance in Pedigree (*KrOt* p. 18/Task 10)



Genotypes:

II/1 – $X^{+}Y$

II/2 – $X^{+}X^{h}$

III/1 - $X^{h}Y$

Answers:

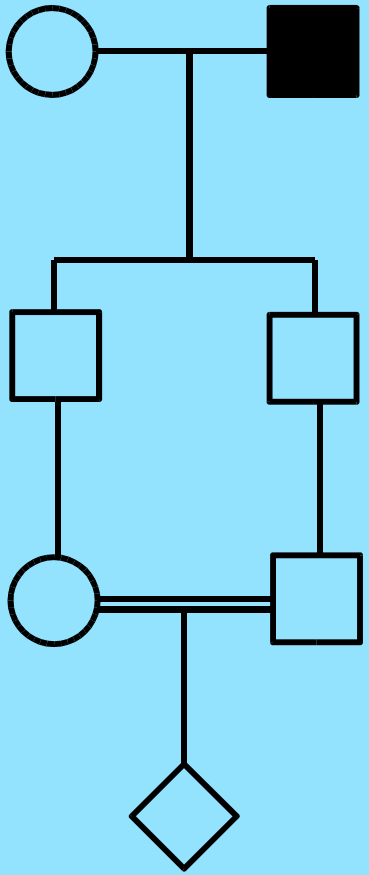
a) 25%

b) 50%

c) ≅ 0%

Risk of consanguineous marriage

(*KrOt* p. 21/Task 17)

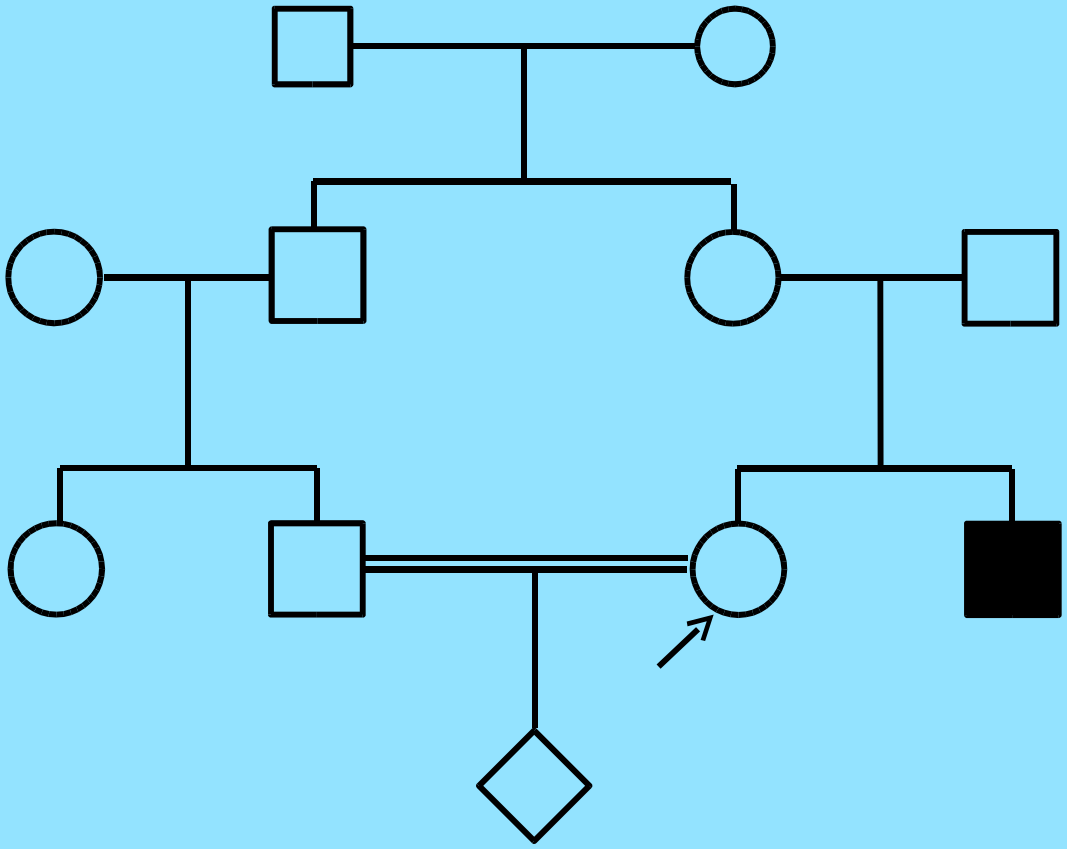


a) PKU (AR) - risk: 1/2 × 1/2 × 1/4 = 1/16

b) brachydactyly (AD) - risk ≅ 0

c) haemophilia A (GR) - risk ≅ 0

Consanguineous marriage of cousins - HPA (KrOt p. 21/Task 19)



Probability: 2/3 × 1 × 1/2 × 1/2 × 1/4 = 1/24 (cca 4 %)