GENEALOGY

I.

II.

III.

IV.

V.

Symbol	Description	Symbol	Description
☐	Male	■	Affected male
○	Female	●	Affected female
◇	Sex unspecified	◆	Affected individual of unspecified sex
3 (in square)	Three males	Male – heterozygote	Male – heterozygote
2 (in circle)	Two females	Female - heterozygote	Female - heterozygote
Monozygotic twins	Monozygotic twins	Carrier (X-linked)	Carrier (X-linked)
Dizygotic twins	Dizygotic twins	Proband	Proband
Adopted into a family	Adopted into a family	Deceased male	Deceased male
		Prenatal death	Prenatal death
Adopted out of a family	Adopted out of a family	Miscarriage	Miscarriage
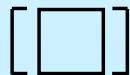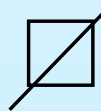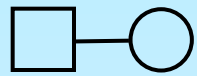
Marriage
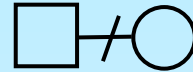
Divorce
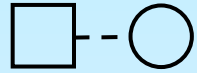
Extramarital mating
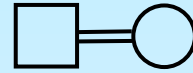
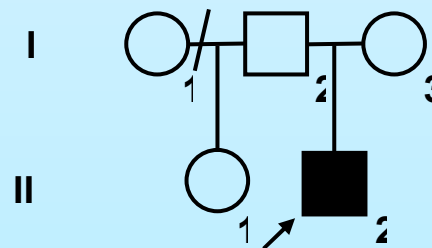**Consanguineous mating**
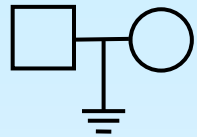
Sterility
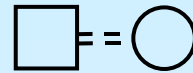
Consanguineous extramarital mating

Numbering of individuals in pedigrees

Sibs
II/2 is proband

Half-sibs

I

1   2

II

1   2   3   4   5   6

III

1   2   3   4   5   6

IV

1   2
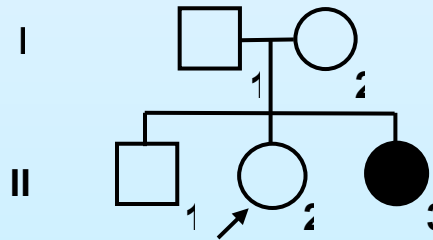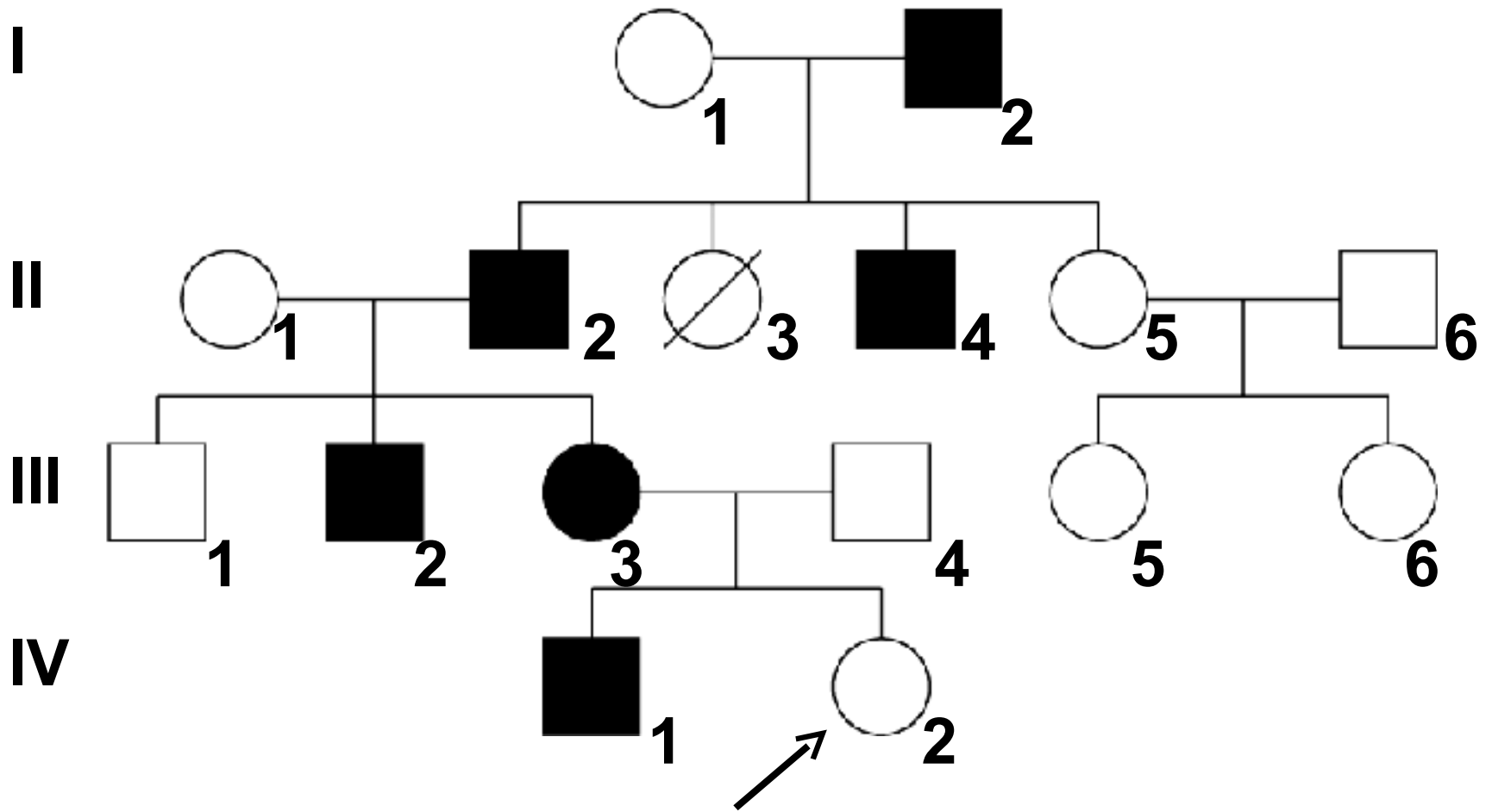
LEGEND:

ROYAL HAEMOPHILIA

Albert — Victoria

Elizabeth II

Charles

Alexei

■ = hemophilia
= age at death
⊙ = carrier for hemophilia

Our Family Tree

Grandma

Stacey

My Mum

Grandma

My Dad

Alien family

My sister

My sister

James

My sister

Ben

My brother

Stephen

My sister

My sister

My baby nephew

My baby Cousin
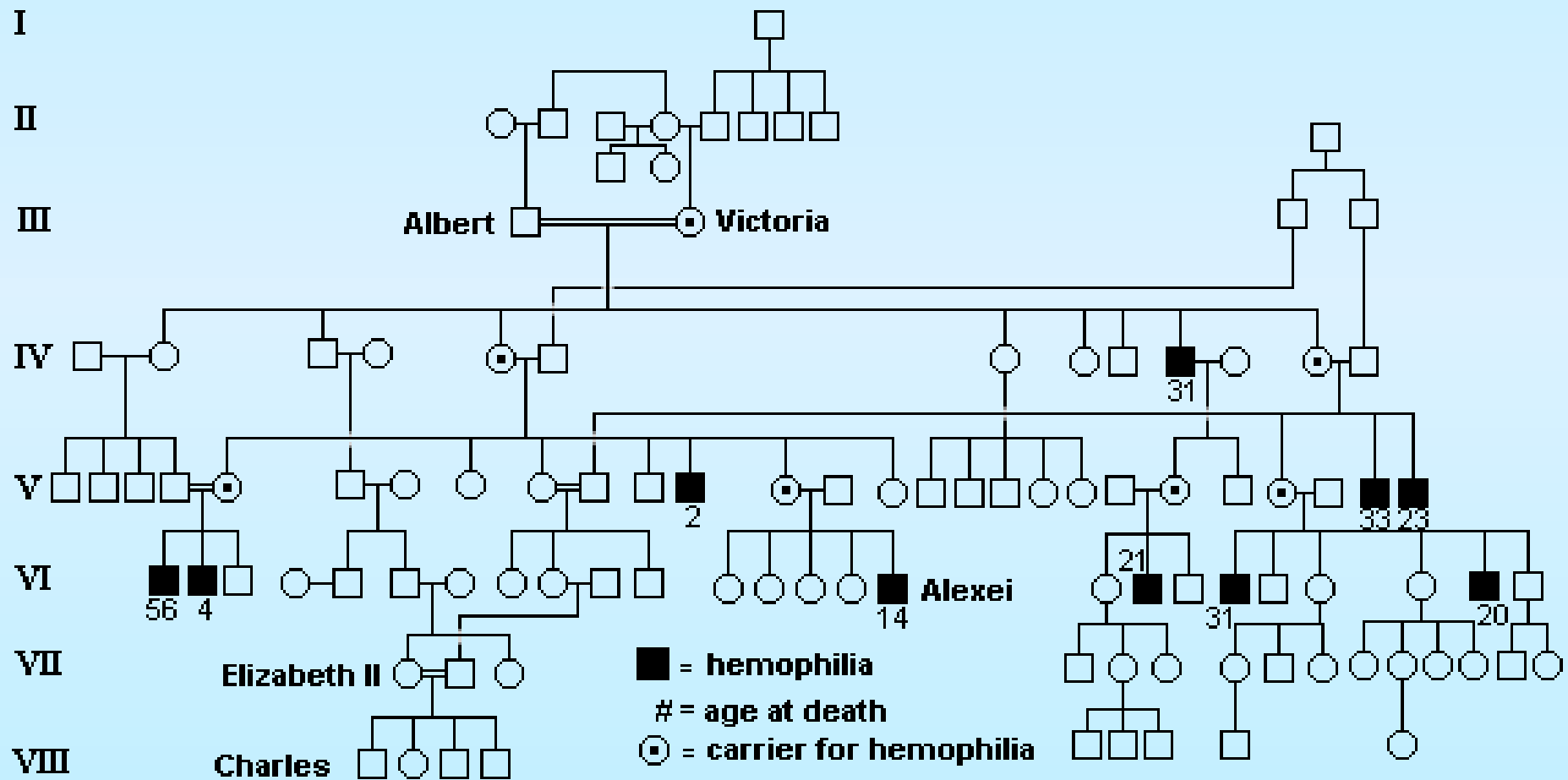
AUTOSOMAL DOMINANT INHERITANCE - AD

Characteristics:

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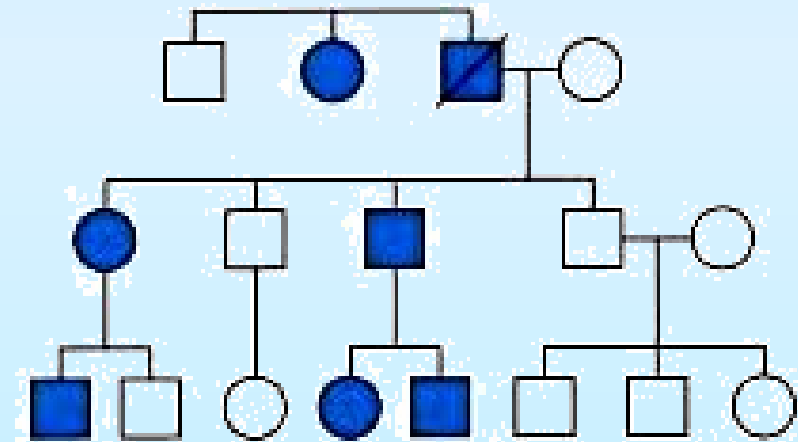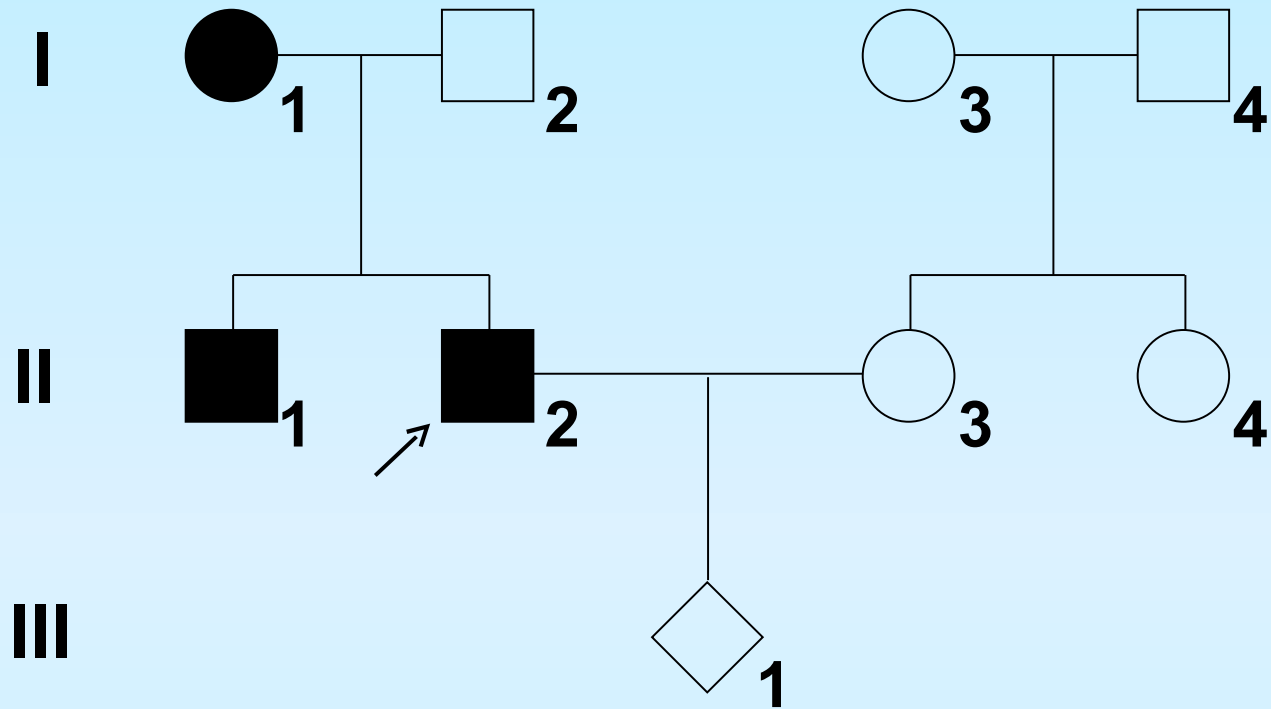- males and females are equally likely to be affected
- transmission from father to son

Examples:

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

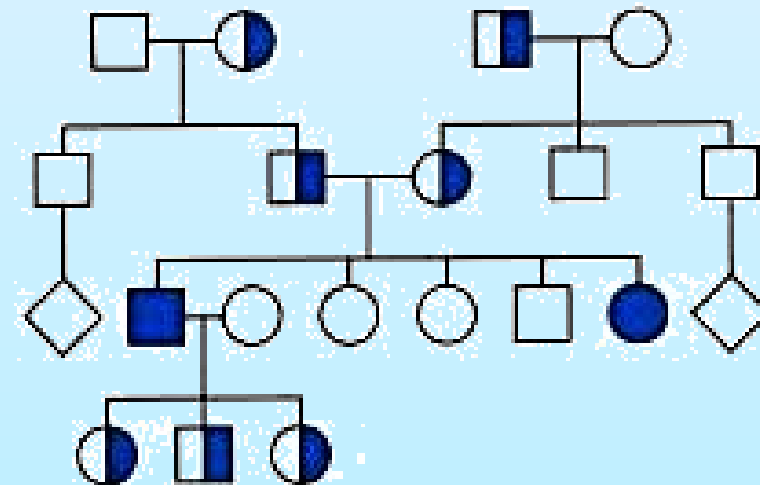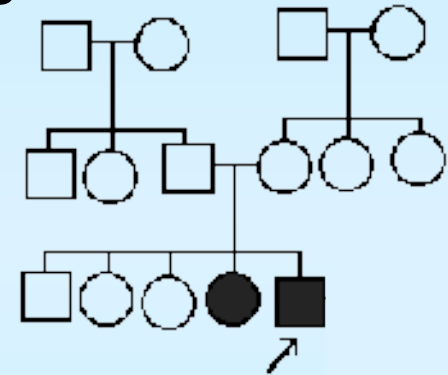a) II/2 – Aa, II/3 - aa

b) 50%

c) 50%

d) 25%

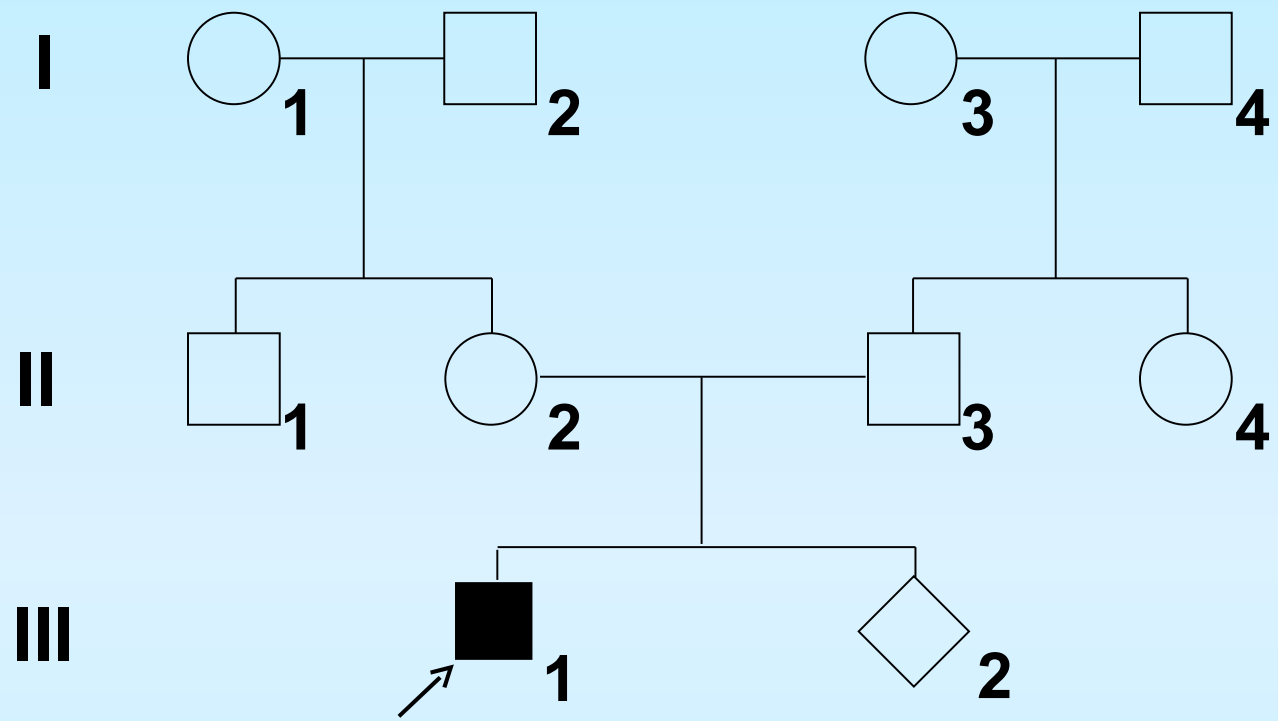# AUTOSOMAL RECESSIVE INHERITANCE - AR

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
- xxx

a) II/2, II/3 – Aa

b) 25%

c) 2/3

d) Aa x Aa, Aa x aa, aa x aa

X-LINKED RECESSIVE INHERITANCE (GONOSOMAL RECESSIVE – GR)
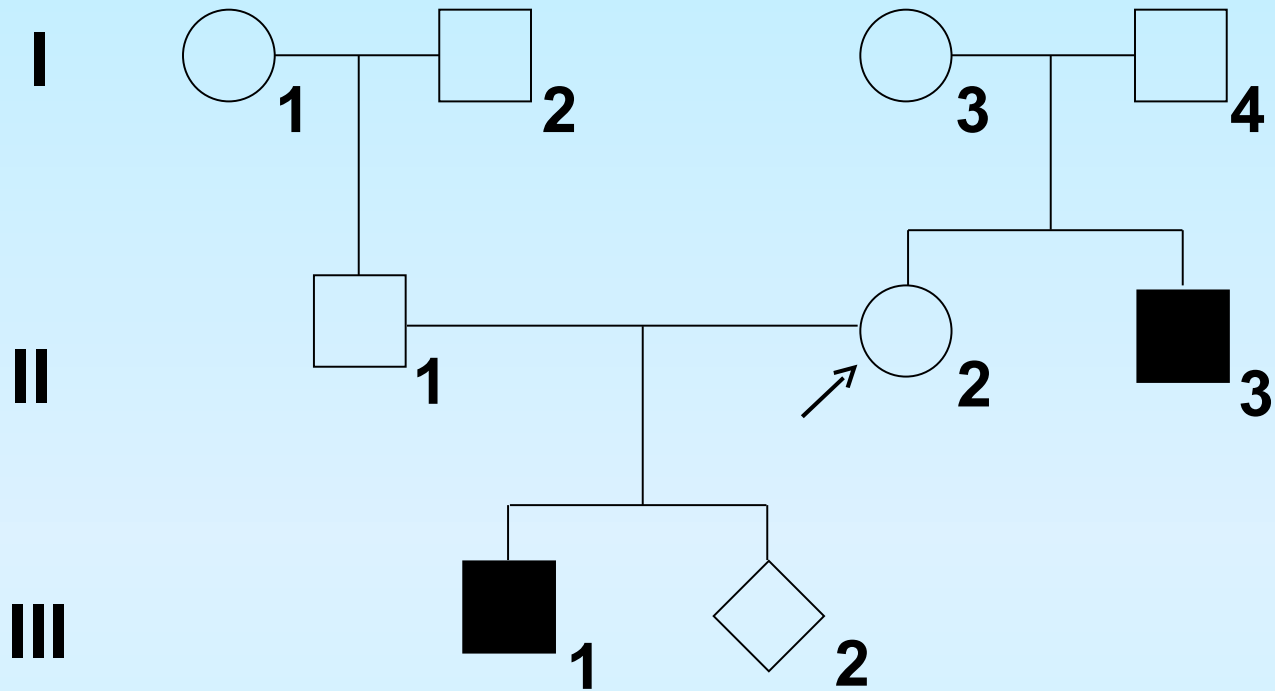
Characteristics:

- 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
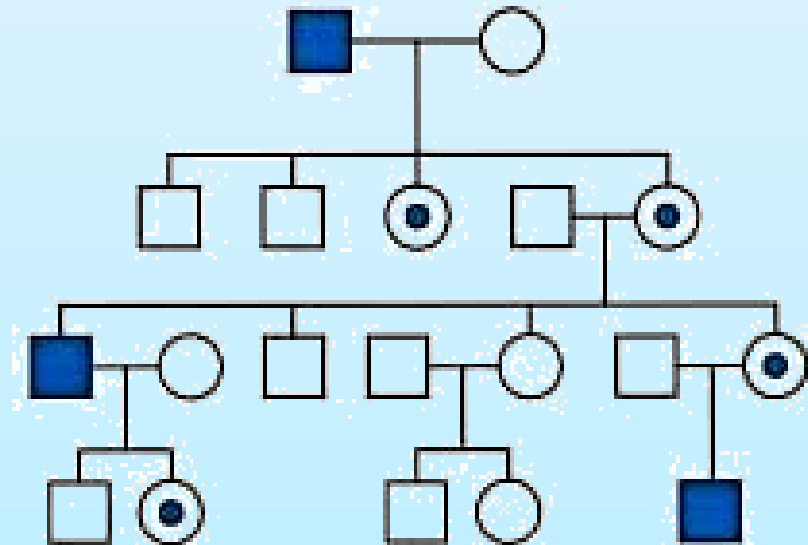
p. 18, task 10 – haemophilia A

I
1 2 3 4

II
1 2 3

III
1 2

a) 25%
b) 50%
c) ≅ 0%

# 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)
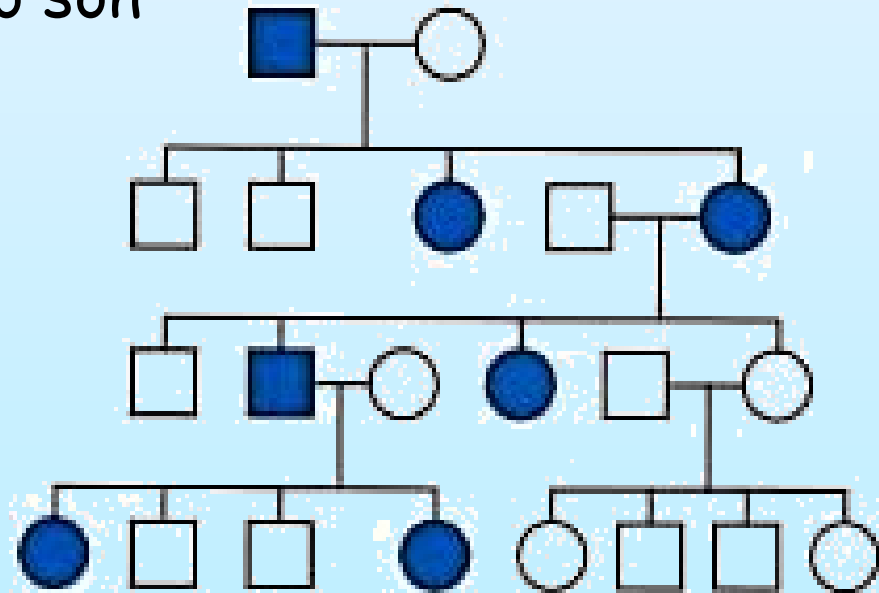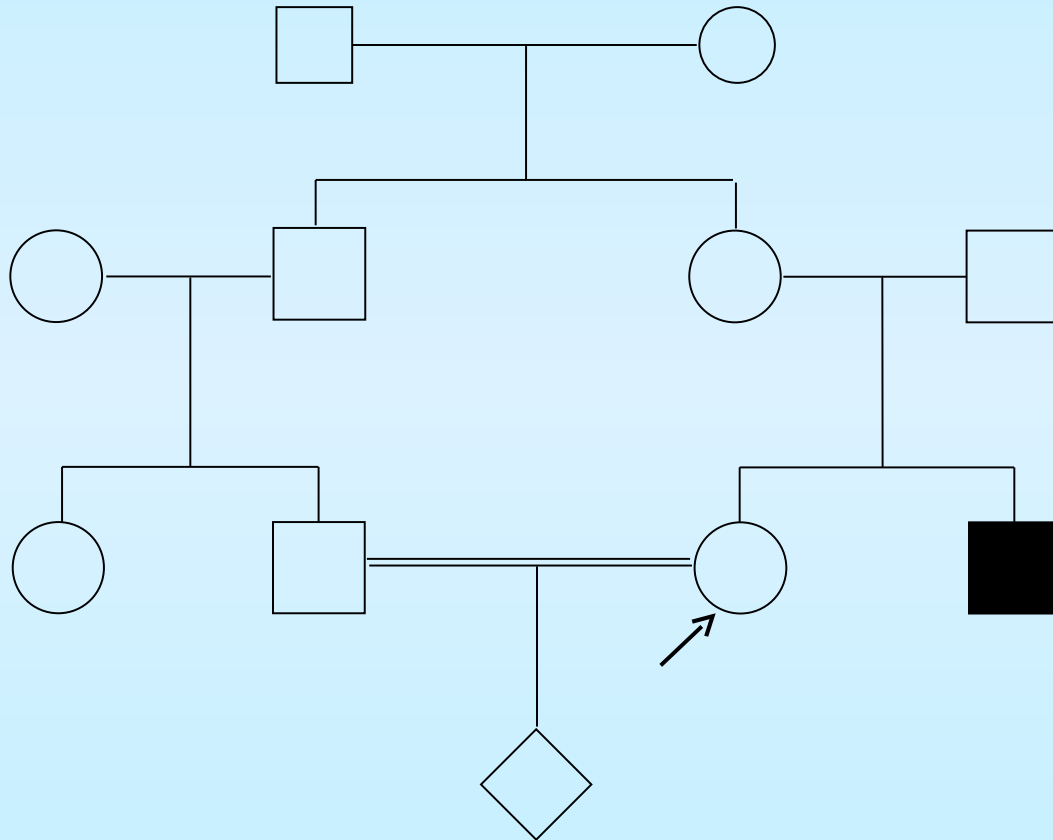
p. 21, task 17 – risk of consanguineous marriage
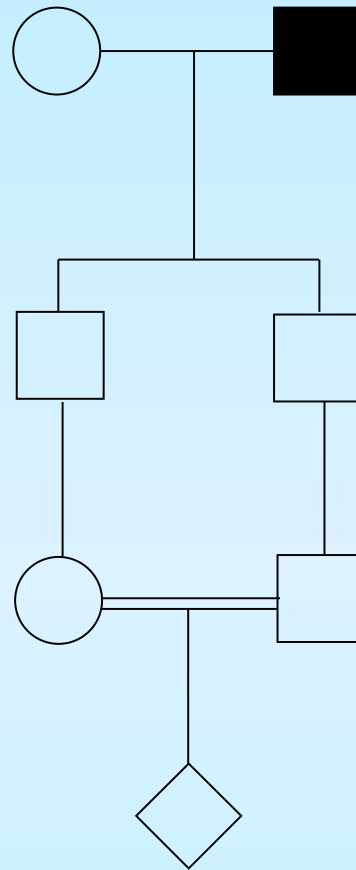
a) PKU (AR) - risk: $1/2 \cdot 1/2 \cdot 1/4 = 1/16$

b) brachydactyly (AD) - risk $\cong 0$

c) haemophilia A (GR) - risk $\cong 0$

probability: 2/3 . 1 . 1/2 . 1/2 . 1/4 = 1/24 (cca 4%)