ZAPOROZHYE STATE MEDICAL UNIVERSITY
DEPARTMENT OF MEDICAL BIOLOGY

Lecture

Human genetics

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QUESTIONS

- -Why man requires a special methods for studies of inheritance?
- -Characteristics the basic methods of Human genetics.

Man is not a very favorable subject for studies of inheritance because:

- Members of Homo sapiens are heterozygous for many genes.
- Controlled matings are impossible.
- Man has a long time between generations and a small number of progenies.
- Man has a lot of chromosomes and genes.

The basic Human's genetics methods are:

- **Pedigree analysis**
- **Twins method**
- **Cytological method**
- **Populative – statistic method**
- **Cell culture**
- **DNA analysis and other.**

Pedigree Analysis

- Used to determine individual genotypes;
- Used to predict the mode of transmission of single gene traits:

  dominant and recessive,

  X-linked and autosomal.

Goals of Pedigree Analysis

1. Determine the mode of inheritance:

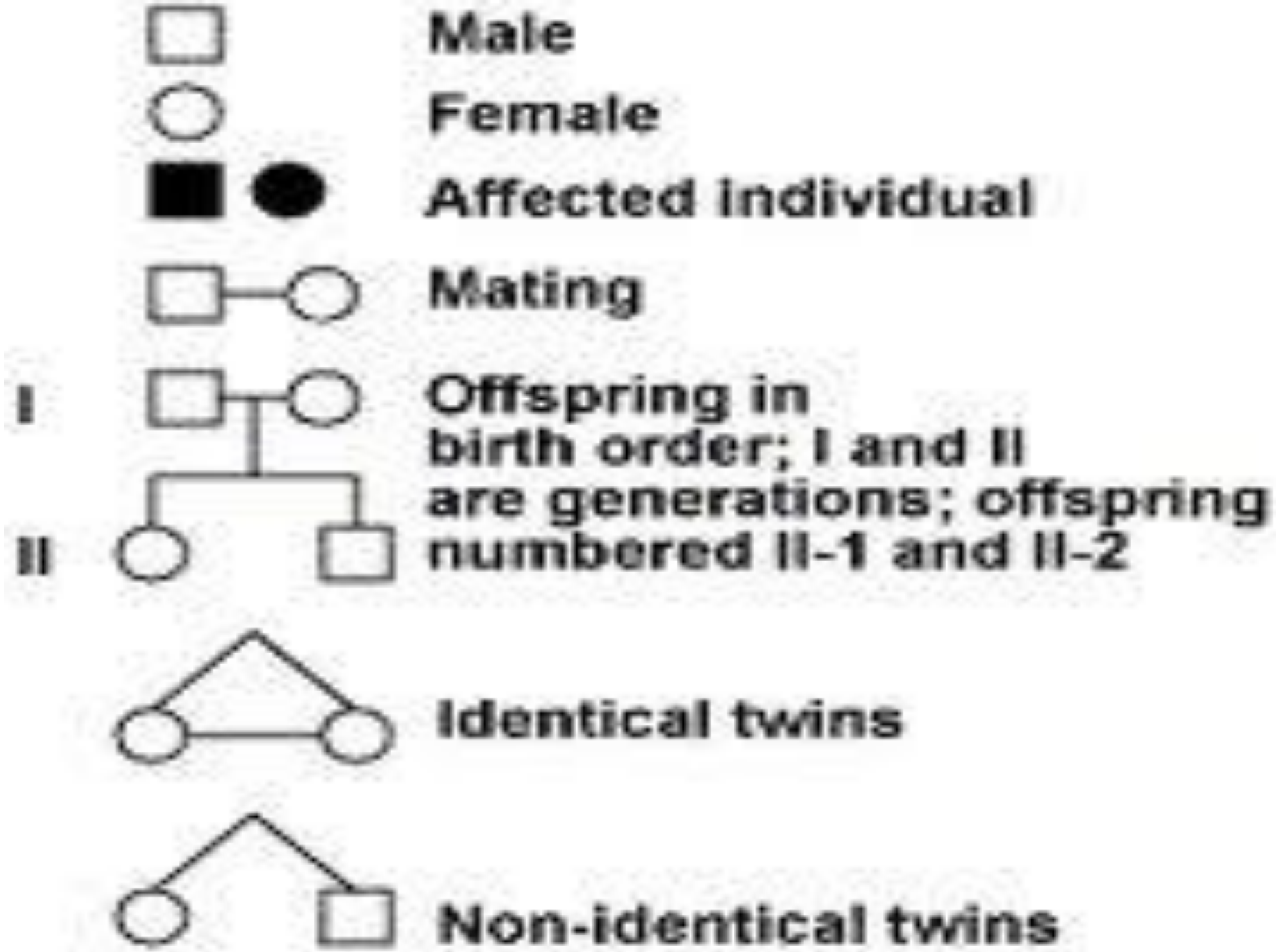
   dominant, recessive, partial dominance, sexlinked, autosomal, mitochondrial, maternal effect.

2. Denermine the probability of an affected offspring for a given cross.

Genealogical method or Pedigree analysis.

It has the following stages:

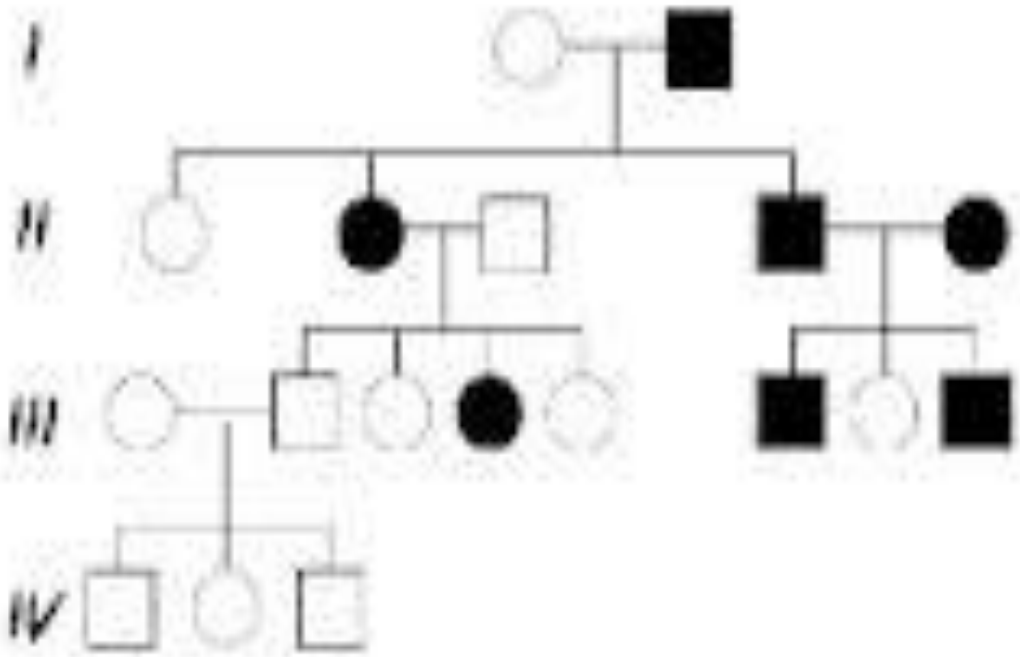
1) Gathering the information.
2) Construction of the pedigree chart.
3) Genealogical analysis.


Male
Female
Affected individual
Mating
I
II
Offspring in birth order; I and II are generations; offspring numbered II-1 and II-2
Identical twins
Non-identical twins

1. Autosomal – dominant inheritance
2. **The autosomal dominant traits appear in every generation. There is no skipping of generation.**
3. **Every affected person has at least one affected parent. A disease in homozygotes shows a severe form.**
4. **The trait is transmitted by an affected person to half of his offspring on an average.**
5. **Both sexes have equal chances of having the trait and transmitted it.**
6. **The penetrance of the gene is 50% - 100%:**

- **50% - if one of the parents heterozygous,**
- **75% - if both of the parents heterozygous,**
- **100% - if one of the parents homozygous.**

Pedigree of brachydactyly

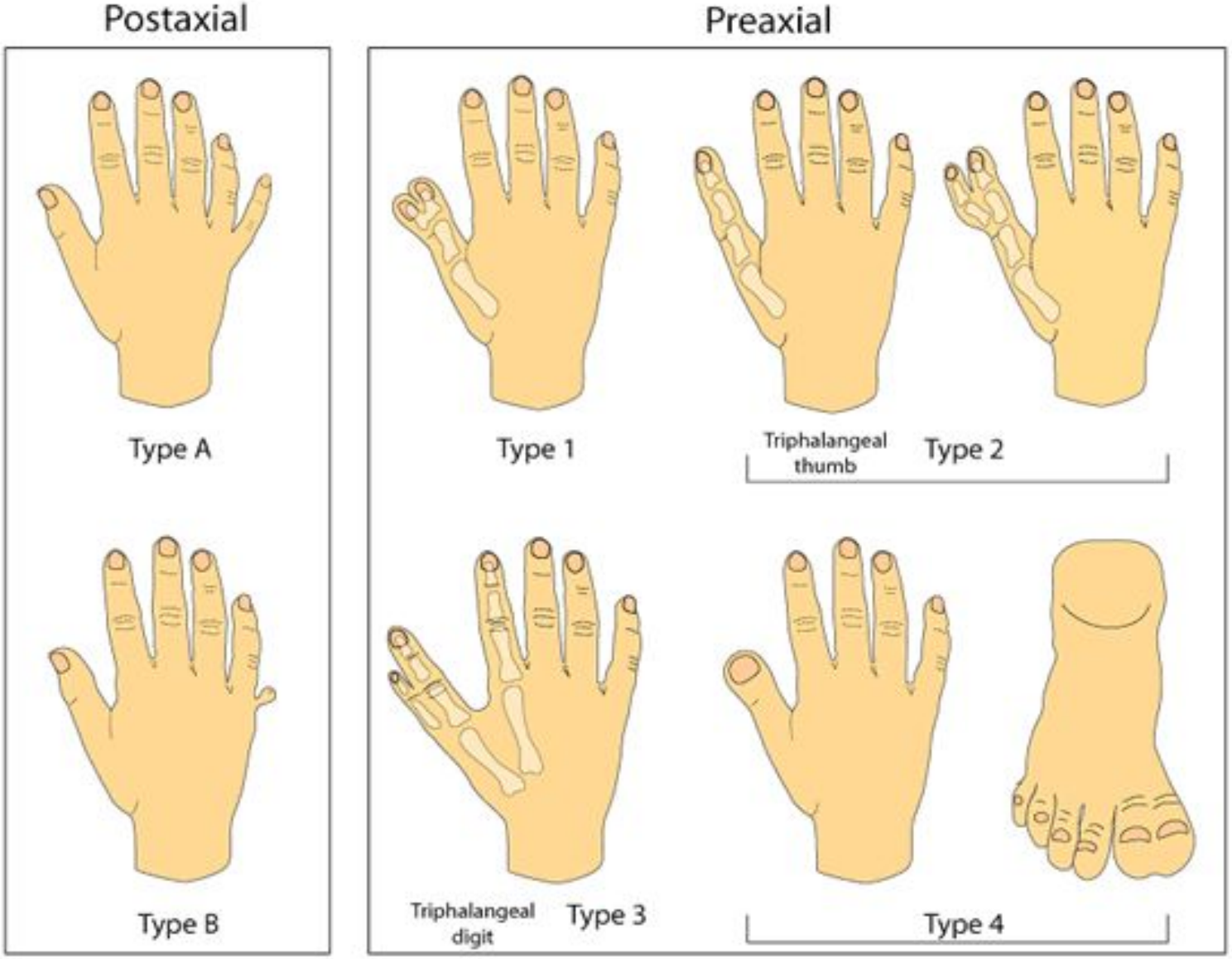


after Suzuki et al. 1961

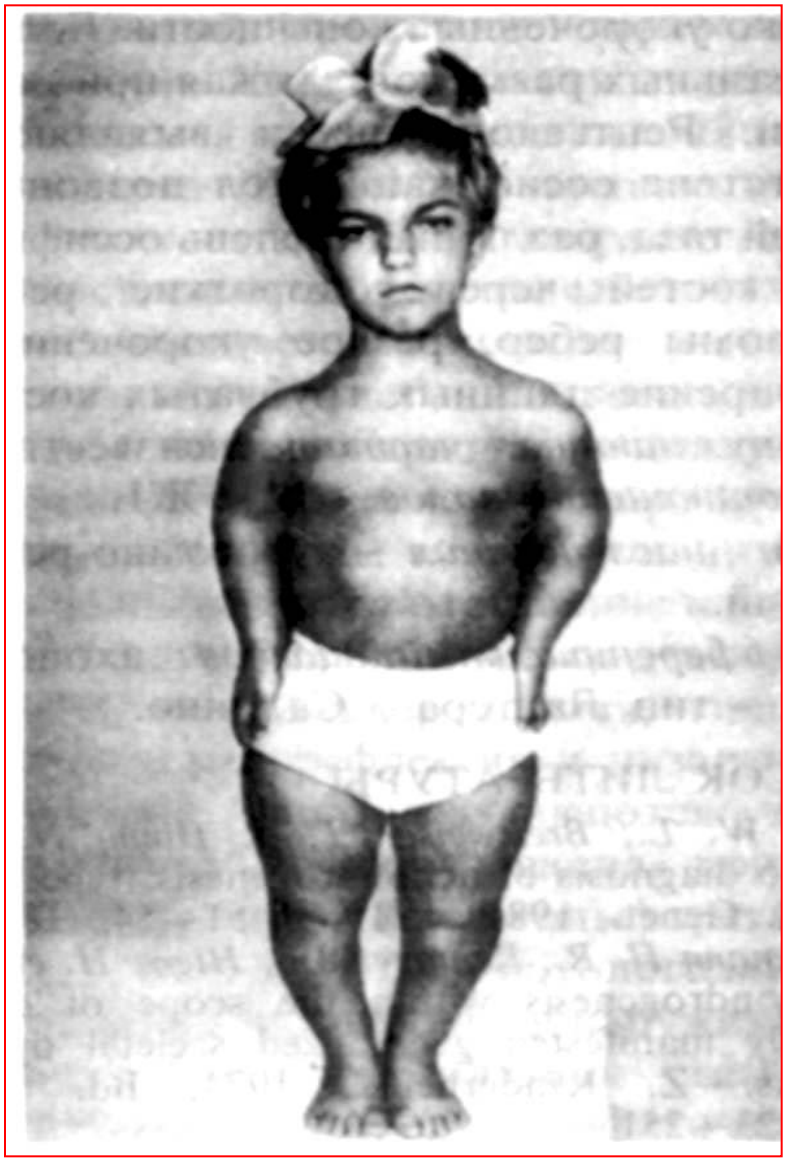
Autosomal Dominant *traits:*

- **Osteogenesis imperfecta**
- **Brachydactyly (short fingers)**
- **Achondroplasia (dwarfism in which the link bones fail to grow)**
- **Marfan's Syndrome**
- **Polydactyly**
- **Syndactyly**

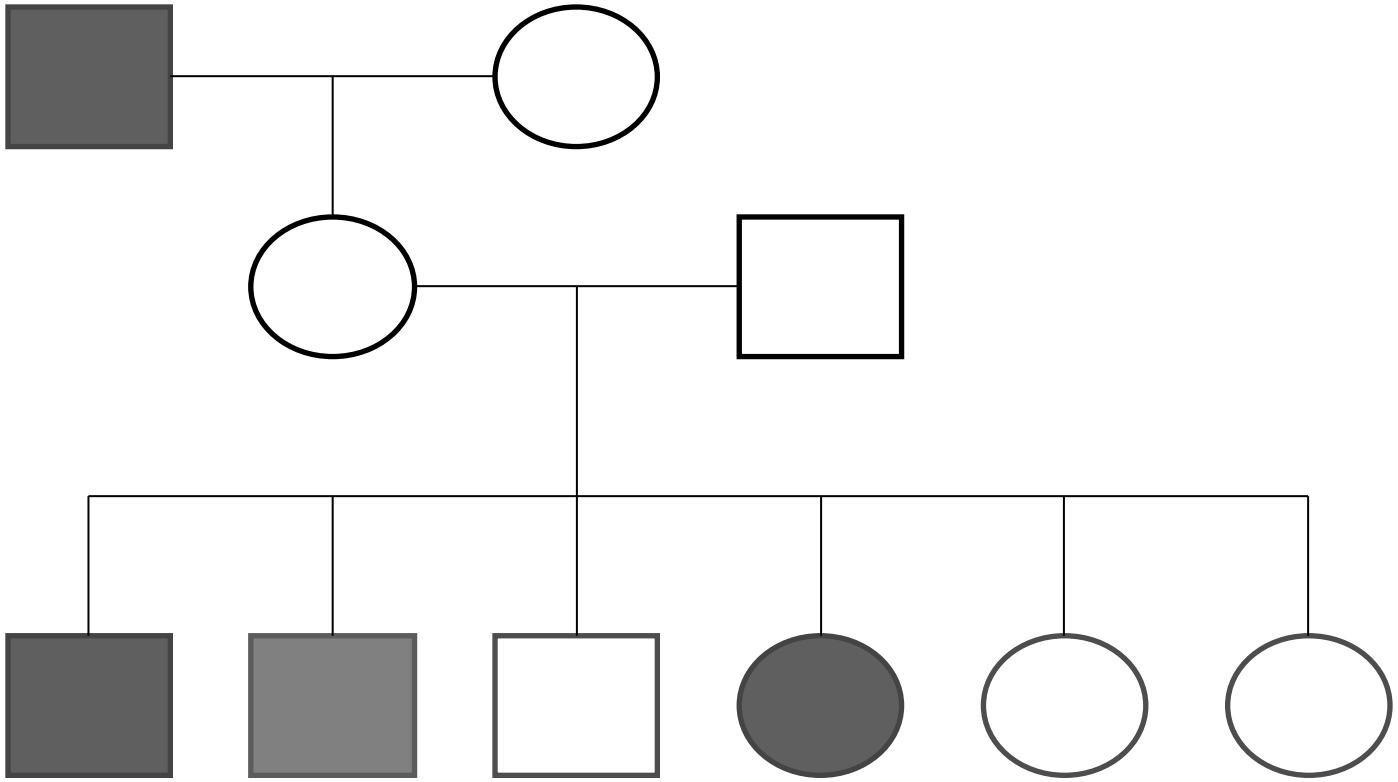
POLYDACTYLY



Achondroplasia



Autosomal recessive inheritance:



1. An affected individual has two normal parents, both of whom are heterozygous.
2. The autosomal recessive condition is typically seen only in the sibs (brothers and sisters). It is not seen in the parents, offsprings or other relatives.
3. Both sexes are equally affected and transmit the trait equally.
4. The recessive trait express itself phenotypically only in homozygous condition.
5. The ratio of affected carrier and non – affected is 1:2:1 in the sibs. The recurrence risk in such a family is 1 in 4 for each birth.

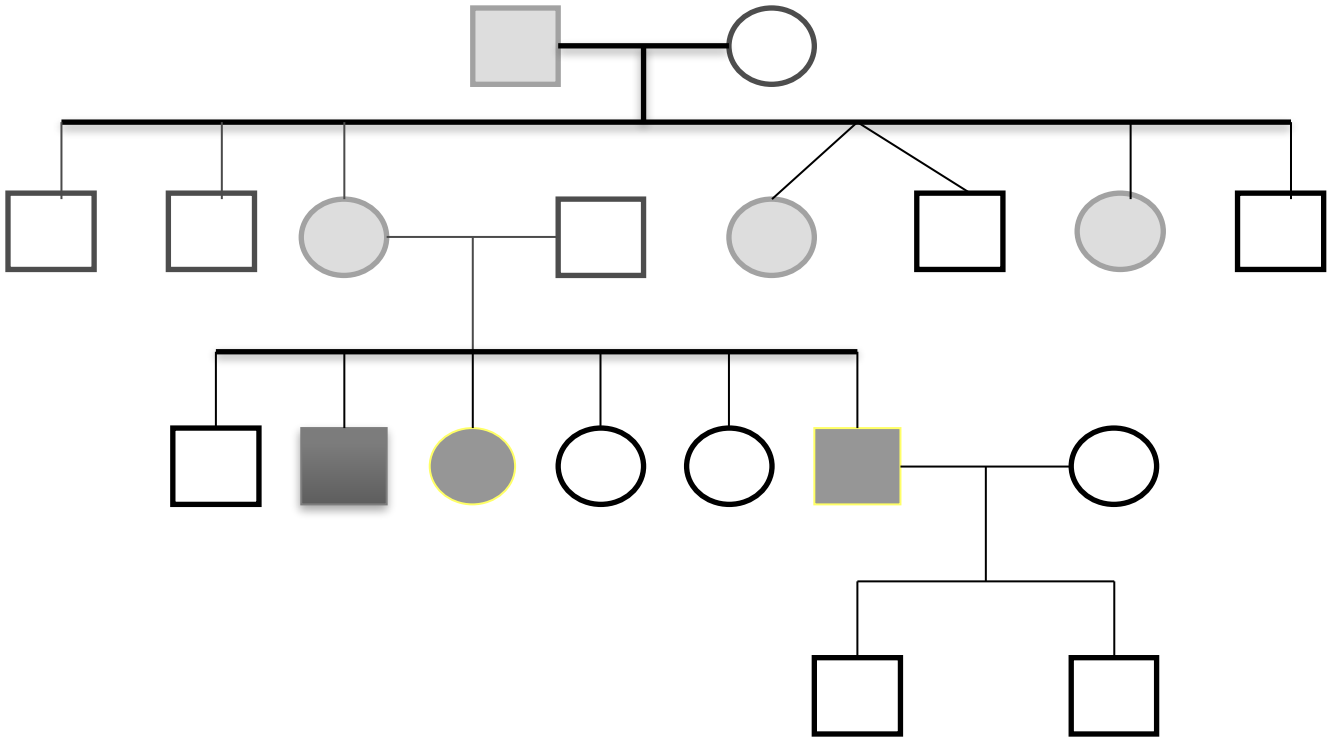
Autosomal recessive traits:

- **Albinism,**
- **Phenylketonuria,**
- **Galactosemia,**
- **Gaucher's disease,**
- **Wilson's disease (hepatolenticular degeneration),**
- **Porphyria.**

Albinism

Sex linked inheritance

X – linked dominant inheritance:

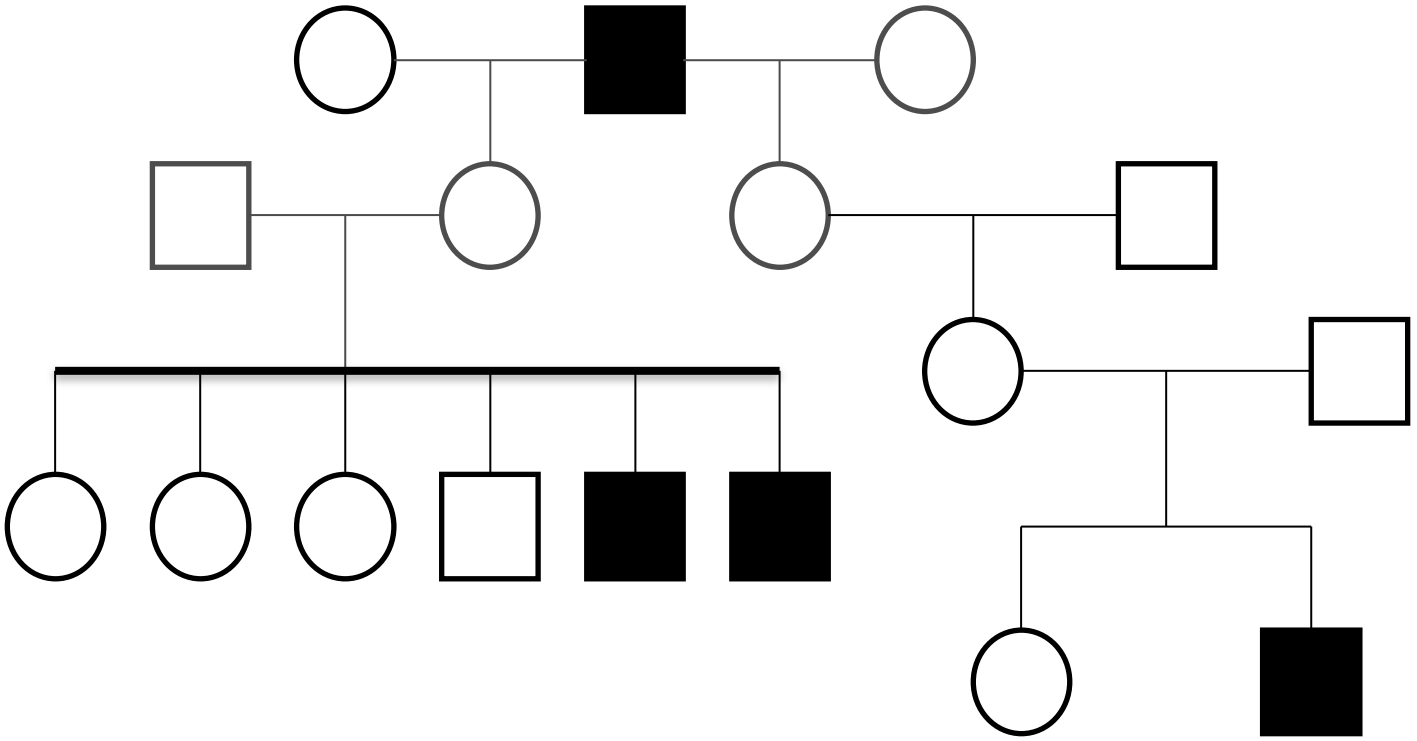


1. **The X – linked dominant conditions are very rare and affected females are twice as common as affected males.**
2. **The affected males pass on the trait to all their daughters. None of their sons would be affected.**
3. **The heterozygous females transmit the trait to half of their children of both sexes.**

The examples of these traits:

- **Vitamin D resistant rickets**
- **Brown – coloured teeth (defective tooth enamel)**

X – linked recessive inheritance

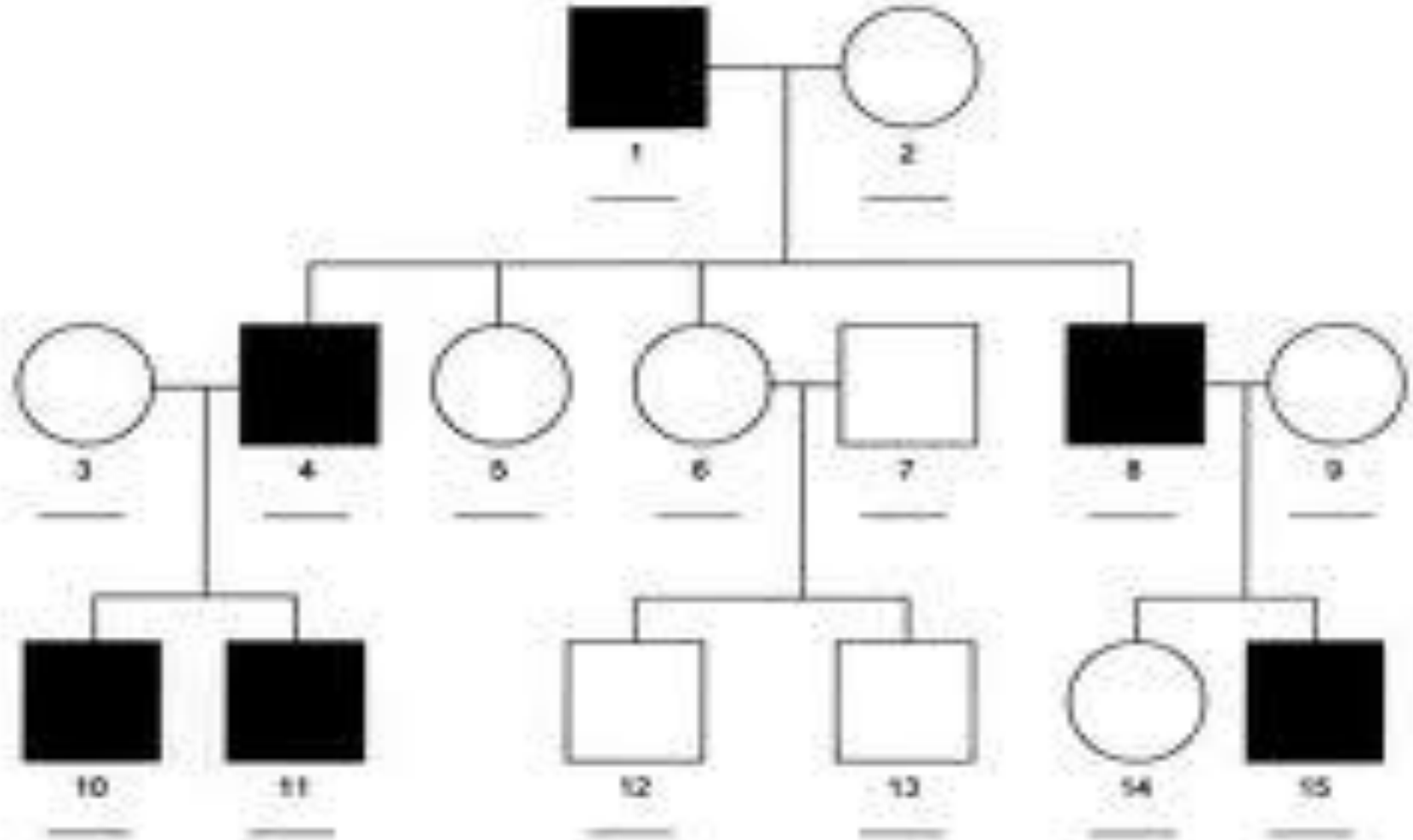


- **X – linked recessive trait is most commonly seen in males.**
- **The father can transmit the trait to half of his grandsons through his daughters – carries.**
- **The trait is transmitted from mother (she is a carrier) to all of her sons.**

The common examples of these traits are:

- **Haemophilia;**
- **Colour blindness;**
- **Duchane muscular dystrophy.**

Y-linked Inheritance




CONCEIVE
EASY


wiseGEEK

Twin's method allows to determine:

role of heredity and environment in the expression of some phenotypic traits.

It is necessary to calculate the following coefficient:

Coefficient of pair concordance (K) :

$K = C/(C + D) \cdot 100\%$

Where: C – number of concordant twin pairs; D – number of disconcordant (differ) twin pairs.

Coefficient of heredity (H)

$$H = \frac{Kmt - Kdt}{100\% - Kdt} \times 100\%$$

Kmt – for monozygotic twins

Kdt – for dizygotic twins

Coefficient of environmental influence

E = 100% - H

H = 0,7 – 1 trait is mainly determined by genotype (ex. blood groups).

H = 0,4 – 0,6 trait is determined by combined action of genotype and environment (ex. hypertension, diabetes).

H = 0 – 0,3 trait is determined by environmental factors (ex. infectional diseases: cholera, malaria).