Mendelian inheritance

Introduction

Genetics : Is the branch of biology that deals with study of heredity and variations among related organisms .

1st Mendel law (Principle of Segregation):

Each organism contains two factors for each trait, and the factors segregate during the formation of gametes so that each gamete contains only one factor for each trait, one from each parent. For example, flower color: P = purple (dominant) , *p* = white (recessive) .If cross a homozygous Purple (PP) with a homozygous white (*pp*):

P P × *p p*

P *p*

F_1 monohybrids are allowed to self-pollinate

P *p* × P *p*

Genotypes: 1 PP: 2 Pp: 1 pp

Phenotypes: 3 Purple : 1 White

Both Punnett square and laws of probability can be used to calculate the expected phenotypic ratio of a cross . In humans do not produce a large number of offspring, its best to use the law of probability

A **Punnett square** can be used to determine the genotypes of potential offspring from a given mating.

The probability of 2 or more independent events occurring together is the product (multiplication) of their chance of occurring separately, for example:

In the cross Tt x Tt What is the chance of obtaining either an T or p from a parent:

- The chance of T=1/2
- The chance of t=1/2
- Therefore ,the probability of receiving these genotypes is as follows:
- The chance of TT= ½ x ½ = ¼

- The chance of Tt= ½ x ½ = ¼
- The chance of tT= ½ x ½ = ¼
- The chance of tt= ½ x ½ = ¼

So the chance of an event that occurs in two or more independent way (phenotypic ratio) is the sum of the individual chances.

- So the chance of offspring with tall (TT, Tt, tT) is ¾
- The chance of offspring with short (tt) is ¼

Test cross

Test cross ,can indicate whether an individual who has dominant phenotype is heterozygous or homozygous dominant ,or use to determine unknown genotypes.

Test cross: Cross with a homozygous recessive individual. For example, a plant with **purple** flowers can either be **PP** or **P*p***…

therefore, cross the plant with a *pp* (white flowers, homozygous recessive)

Mendel's 2nd law (Principle of Independent Assortment)

"Members of one pair of factors segregate independently from other pair of factors during gamete formation"

Mendel performed a second series of experiments in which he crossed true-breeding plants that differ in 2 traits such crosses called dihybrid crosses. For example, flower color:
P = purple (dominant) and *p* = white (recessive)
and stem length: T = tall and *t* = short

TT PP × *tt pp*

(tall, purple) (short, white)

F1 Generation: (T*t* P*p*) All tall, purple flowers

If F_1 generation is allowed to self-pollinate, Mendel observed 4 phenotypes:

T*t* P*p* × T*t* P*p*

(tall, purple) (tall, purple)

	TP	Tp	tP	tp
TP	TTPP	TTPp	TtPP	TtPp
Tp	TTPp	TTpp	TtPp	Ttpp
tP	TtPP	TtPp	ttPP	ttPp
tp	TtPp	Ttpp	ttPp	ttpp

Phenotype Ratio = 9:3:3:1

Genotype ratios (9): 1 TTPP :2TTPp : 2Tt PP: 4Tt Pp:1TTpp: 2Tt pp:1tt PP:2 tt Pp: 1tt pp

phenotype ratios (4): (9)Tall, purple : (3)Tall, white : (3)Short, purple:(1)Short, white

Genetic Terminology

Chromosomes: Carry the hereditary information (genes)

Trait : Any characteristic that can be passed from parent to offspring

Heredity : Passing of traits from parent to offspring.

Gene : Is the fundamental unit of heredity .

Genome :The entire set of genes in an organism

Alleles :The alternative forms of a gene found at one locus.(dominant &recessive) . Different alleles usually determine the production of polypeptides.

Locus : Is a particular location on a chromosome , the two genes at homologous loci are called gene pair

Dominant: The allele of a gene that masks or suppresses the expression of an alternate allele; the trait appears in the heterozygous condition

Recessive: An allele that is masked by a dominant allele; does not appear in the heterozygous condition, only in homozygous

Homozygous: Having identical alleles for one or more genes **called pure**.

Heterozygous: Carrying two different alleles for one or more genes **also called hybrid** .

Genotype: The genetic makeup of an organism.

Phenotype: Is the physical appearance of an individual **(e.g. red, white)** .

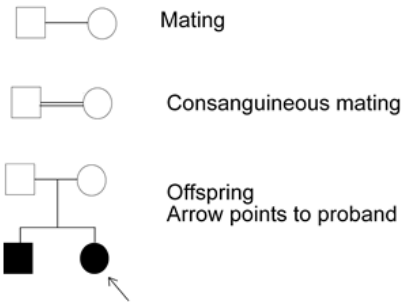
Monohybrid cross : Cross involving a single trait e.g. flower color

Dihybrid cross : Cross involving two traits e.g. flower color & plant height

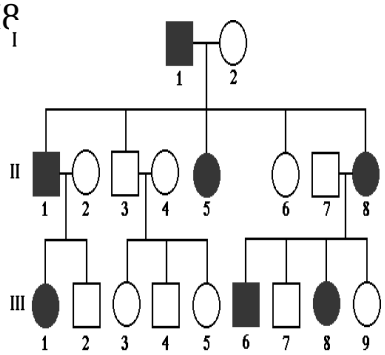
Pedigree Analysis : Pedigrees are family trees that explain genetic history.

Goals of Pedigree Analysis

1. **Determine the mode of inheritance: dominant, recessive, partial dominance, sex-linked, autosomal, mitochondrial, maternal effect.**
2. **Pedigrees are used to find out the probability of a child having a disorder in a particular family.**



The generations are identified by Roman numerals, while an individual in each generation is identified by Arabic numerals numbered from the left. Therefore the affected individuals are I1, II1 and II5,II8 ,III1,III6 and III8



Interpreting a Pedigree Chart

1. Determine if the pedigree chart shows an autosomal or X-linked disease.
   - If most of the males in the pedigree are affected the disorder is X-linked
   - If it is a 50/50 ratio between men and women the disorder is autosomal.
2. Determine whether the disorder is dominant or recessive.
   -If the disorder is dominant, one of the parents must have the disorder.
   -If the disorder is recessive, neither parent has to have the disorder because they can be heterozygous.

Mode of Mendlian inheritance in man

A trait that is determined by a gene one of the autosomal chromosomes is said to be inherited as an autosomal trait, or traits determined by genes on one of the sex-chromosomes may be either dominant or recessive such as traits are said to be sex-linked or x-linked.

Autosomal dominant inheritance

The criteria for diagnosis of autosomal dominant:

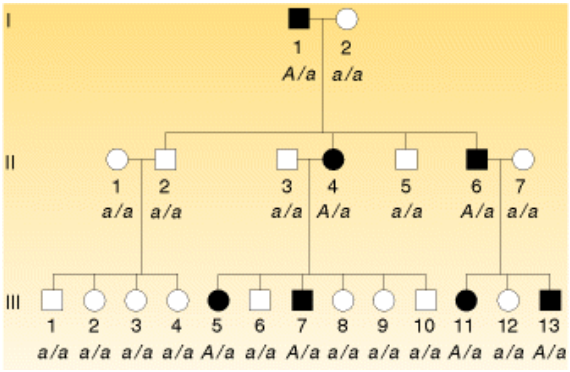
1- Males and females equally likely to be affected
2- The trait appears in every generation with no skipping
3- The trait is transmitted by an affected person to half his children
4- Unaffected individuals do not pass the trait to their children
5- Every affected child has an affected parent

For example: Huntington's disease and Polydactyly

Huntington's Disease:

Is a degenerative disease of the nervous system. Once the deterioration of the nervous system begins the condition is irreversible and fatal and cause choric movement disorder

Pedigree of a dominant phenotype determined by a dominant allele *A* . In this pedigree, all the genotypes have been deduced.

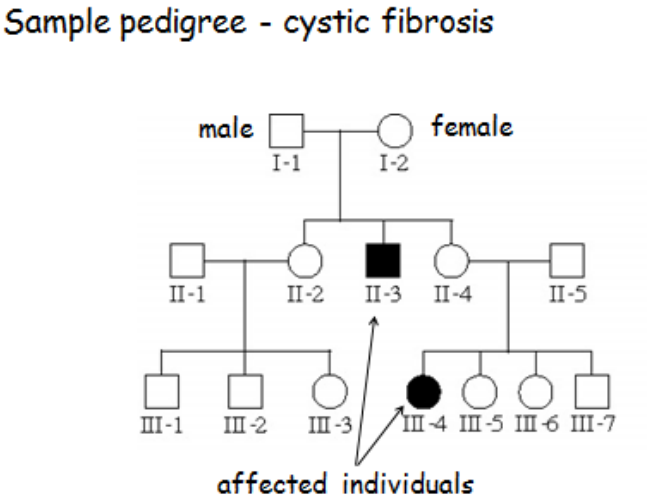


Autosomal recessive inheritance

1. The trait characteristically appears only in some of the sibs, not in their parents.
2. On average, one-fourth of sibs are affected.
3. The parents of the affected child may be consanguineous.
4. Males and females are equally likely to be affected.

For example:

Cystic fibrosis, Sickle cell anaemia, Phenylketonuria (PKU) and Tay-Sachs disease

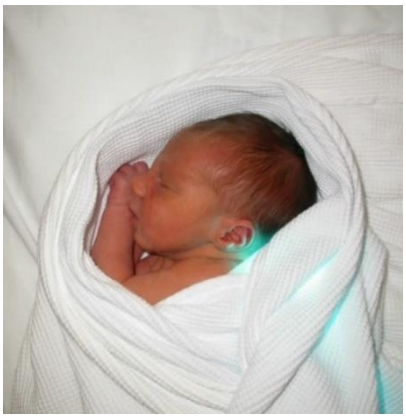


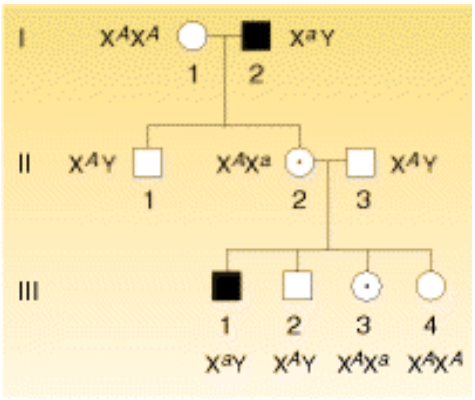
X-linked recessive inheritance:

1. The incidence of the trait is much higher in males than in females
2. The trait is passed on from an affected man through all his daughters to half their sons
3. The trait is never transmitted directly from father to son.
4. The trait may be transmitted through a series of carrier females.

For example:

Hemophilia, Color blindness and Glucose-6-Phosphate Dehydrogenase deficiency



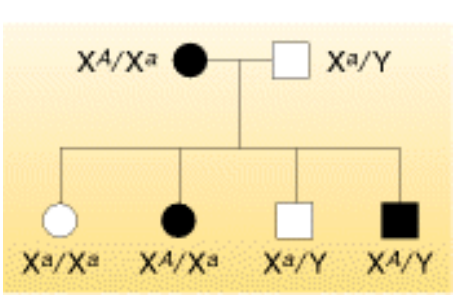


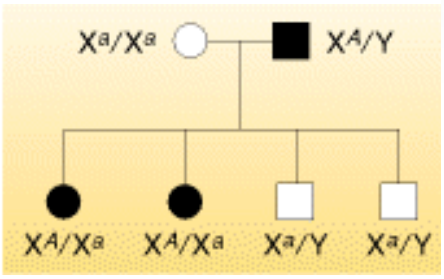
X-linked dominant inheritance:

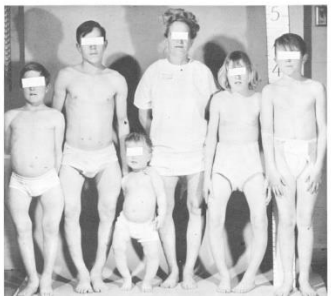
1. Affected males transmit the trait to all their daughter none of their sons.
2. Affected females who are heterozygous transmit the condition to half their children of either sex.
3. Affected females who are homozygous transmit the trait to all their children.

4. Sex-linked dominant inheritance cannot be distinguished from autosomal dominant inheritance by the progeny of affected females, but only by the progeny of affected male. **For example:**

There are few examples of X-linked dominant phenotypes in humans. One example is **hypophosphatemia**, a type of vitamin D-resistant rickets. Rickets is a disorder caused by a deficiency of phosphate (Phosphate is an electrolyte that helps your body with energy production and nerve function). It leads to softening and weakening of bones in children (bones become bent and distorted).



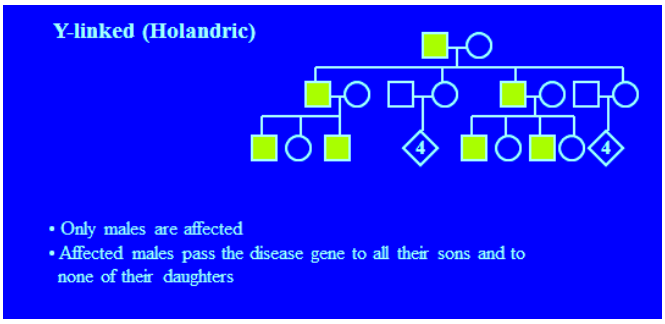




Y-linked (Holandric inheritance):

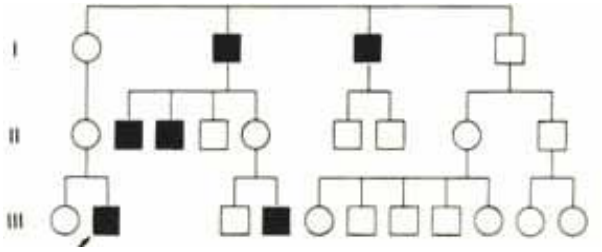
1. Y-linked of a trait would mean that only males are affected.
2. An affected man would transmit the gene on his only Y-chromosome to all his sons but none of his daughters.

There are two dozen Y-linked traits that have been discovered such as **testes determining factor (TDF)**, which is involved in determining maleness, as well as the gene responsible for the **hairy ear**. Another example: **Male infertility**.



Sex- limited trait :

A trait that is autosomally transmitted but expressed in only one sex either males and females. For example, facial hair distribution and sperm production in man and ovary formation in the female are sex-limited traits. Sex-limited expression (trait only found in males or females)



Sex influenced:

The trait which is controlled by genes present on autosomes but whose expression is influenced by the sex of individual are called sex-influenced traits. Sex influenced traits are those that are dominant in one sex but recessive in the other. This is due to the different cellular environments in males and females provided by sex hormones (estrogen, progesterone, testosterone etc. Example: baldness is caused by an autosomal allele that is dominant in males due to the presence of testosterone, the male sex hormone. In the presence of high levels of testosterone, the baldness allele has a very powerful influence. In the presence of low levels of the hormone, this allele is quite ineffectual. All humans have testosterone, but males have much higher levels of this hormone than female do. The result is that in males, the baldness allele behaves like a dominant allele, while the female it behaves like a recessive allele.

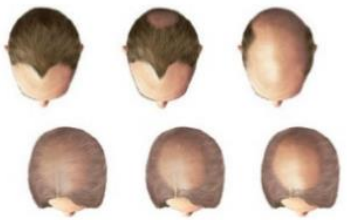
For example:

B represents the non-bald

BB genotype: non-bald in both sexes

bb genotype: bald in both sexes

Bb genotype: bald in men; non-bald in females



Male and Female Pattern Baldness