Characteristics of Organisms Used For Genetic Studies

General Features of an Organism Used for Genetic Studies

1. good genetic background
2. easy to grow
3. controlled mating possible
4. can be genetically engineered
5. funding available
6. variation

Viruses

- easily grown in culture
- rapid generation time
- encode only a few the proteins, which permit a detailed analysis of well-defined system
- some viruses have control mechanisms found in eukaryotic species
- can be genetically engineered
- lack organized chromosome
- not all genetic features can be extrapolated to other organisms

Escherichia coli

- easily grown in culture
- rapid generation time
- many mutants available
- many genes involved
- life cycle very well defined
- relatively simple to generate new mutants
- can be genetically engineered
- contain a rudimentary chromosome
- not all genetic features can be extrapolated to higher organisms

Human

- intense public interest and funding available
- relatively well-mapped for most eukaryotic species
- many diseases (mutant phenotypes) understood clinically
- well-defined cytogenetic system
- long generation time
- many traits can only be studied in cell culture
- cannot make controlled crosses
- cannot be genetically engineered

Drosophila

- short generation time for a eukaryotic organisms (two weeks)
- availability of many mutants controlling specific phenotypes
- large chromosomes with well-defined cytogenetic system
- survives well in the lab and matings are easily performed
- currently, the best organism to study developmental genetics
- transposable elements can be manipulated to clone genes
- can be genetically engineered

Maize

- best mapped plant species
- many mutants are available that control seed traits
- well-defined cytogenetic system
- transposable elements well understood and can be used to clone genes
- matings tedious, but produce many (hundreds) of progeny
- only three generations per year
- cannot be easily genetically engineered
- not well funded

Arabidopsis thaliana

- small genome with little repetitive DNA
- short generation time (six weeks)
- many mutants rapidly becoming available
- matings tedious (مزعج), but produce many (thousands) of progeny
- currently, an intensely investigated organisms; the Drosophila of plant species
- can be easily genetically engineered

Although these species have been studied intensely, these are by no means the only species analyzed for genetic studies. Mouse has been used for many genetic studies because it is a mammal that is a relatively good model for human gene expression. A good example is the globin genes, protein that carry oxygen in blood cells. The genetic and molecular organization of the mouse and human genes are quite similar: they have the same number of exons and have the introns located at the same amino acid residue in the transcript. Furthermore, the order of the multigene family is conserved. Yeast has also been well studied. Although this species is a good model system, this is not the reason that it was first investigated. The primary reason was because of its role in brewing.

Methods of genetic study

1- Planned breeding

Such as Mendel experiment used in descriptive studies, depends on some versus trait and then inbreeding between them.

2- Pedigree analysis

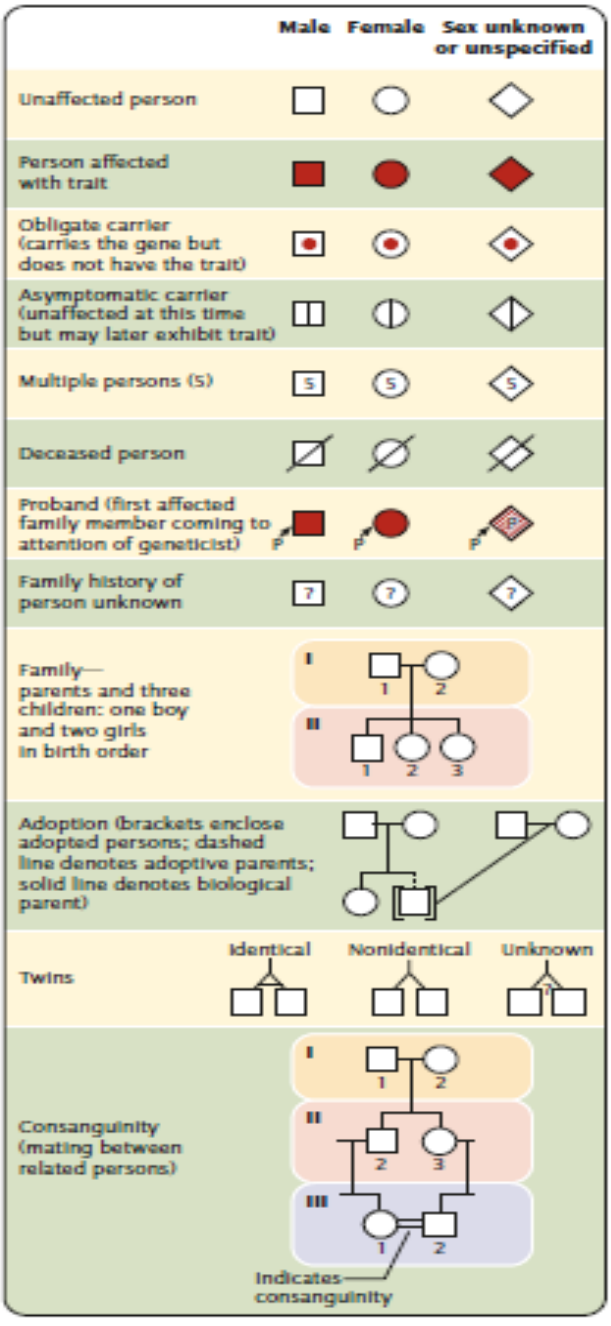
Why do Pedigrees?

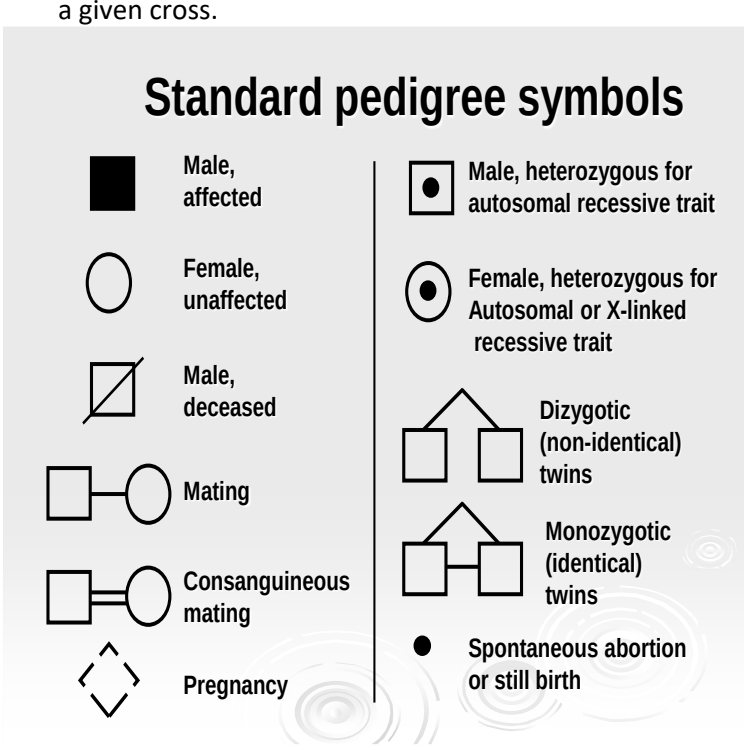
Punnett squares and chi-square tests work well for organisms that have large numbers of offspring and controlled matings, but humans are quite different:

1. Small families.
   Even large human families have 20 or fewer children.
2. Uncontrolled matings, often with heterozygotes.
3. Failure to truthfully identify parentage.

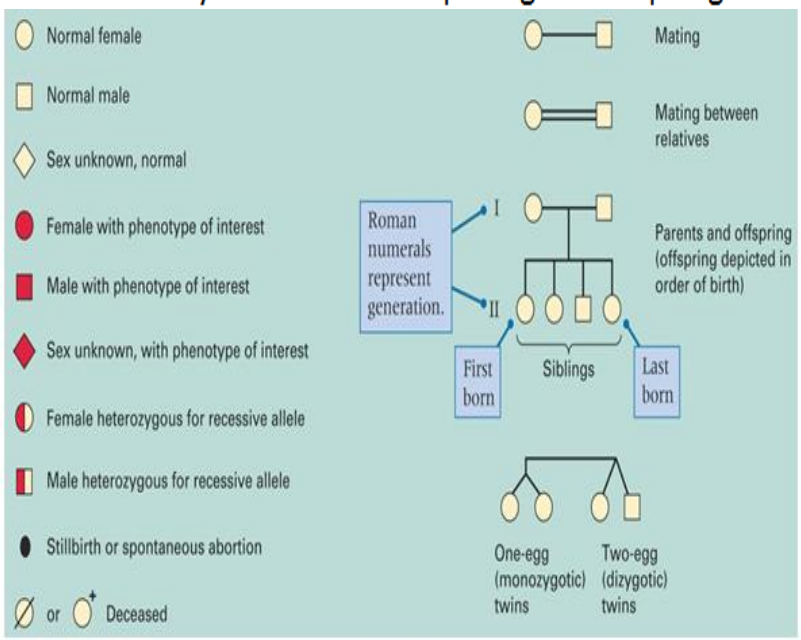
Goals of Pedigree Analysis

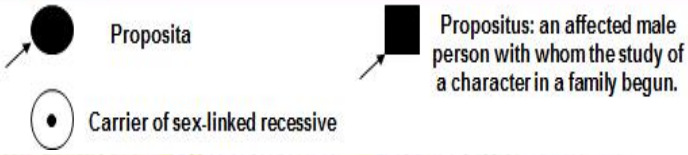
1. Determine the mode of inheritance:
   dominant, recessive, partial dominance,
   sex-linked, autosomal, mitochondrial, maternal effect.
2. Determine the probability of an affected offspring for a given cross.

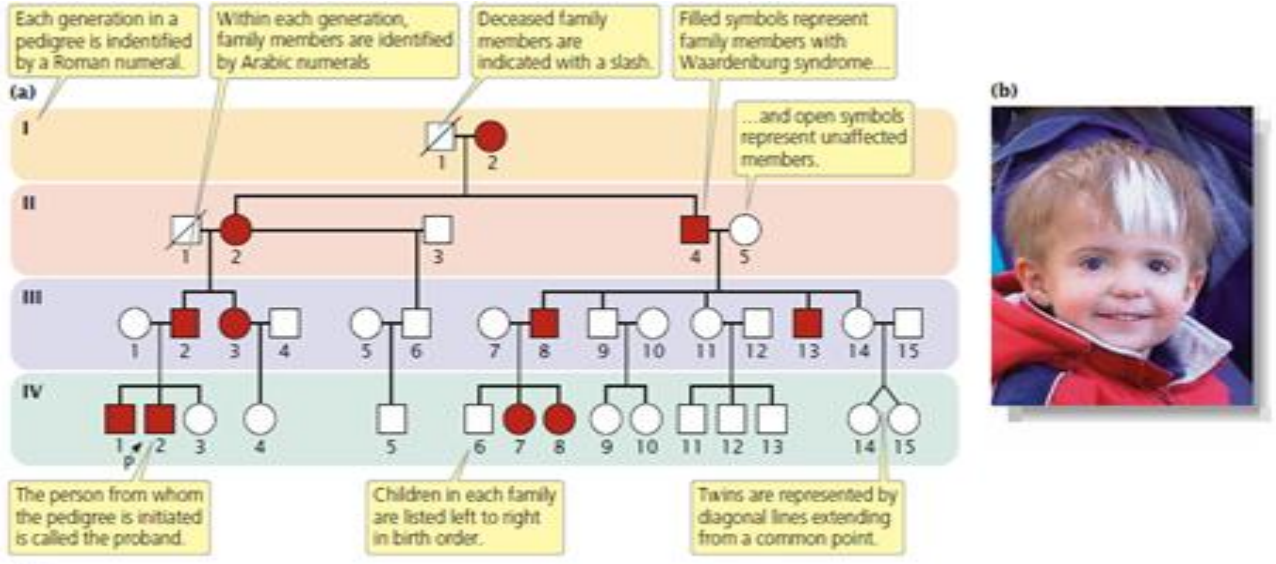




Conventional symbols used in depicting human pedigrees.

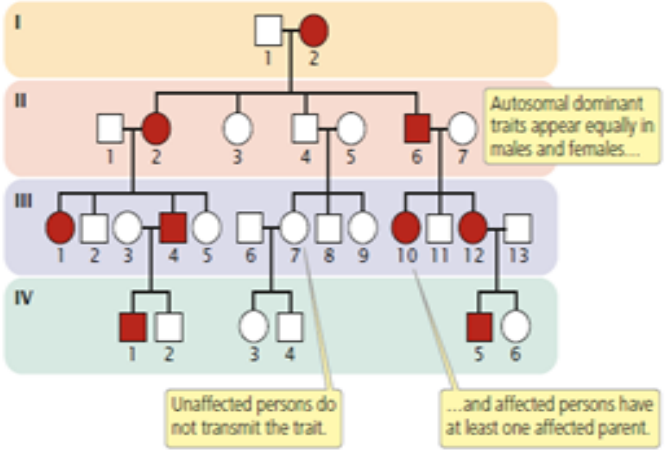






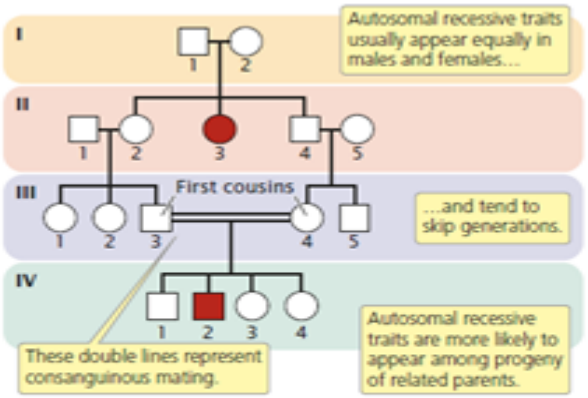
6.3 Waardenburg syndrome is (a) inherited as an autosomal dominant trait and (b) is characterized by deafness, fair skin, visual problems, and a white forelock.

Autosomal dominant traits appear in both sexes with equal frequency. An affected person has an affected parent (unless the person carries new mutations), and the trait does not skip generations. Unaffected persons do not transmit the trait.



Autosomal dominant traits normally appear with equal frequency in both sexes and do not skip generations.

Autosomal recessive traits appear with equal frequency in males and females. Affected children are commonly born to unaffected parents who are carriers of the gene for the trait, and the trait tends to skip generations. Recessive traits appear more frequently among the offspring of consanguine matings.

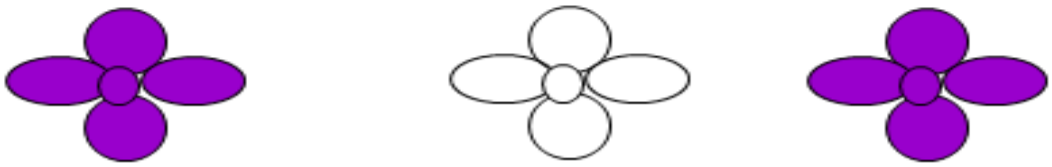


6.4 **Autosomal recessive traits normally appear with equal frequency in both sexes and seem to skip generations.**

1- Dominant/Recessive

- One allele is dominant over the other (capable of masking the recessive allele)

PP = purple **pp = white** **Pp = purple**



Problem: Dominant/Recessive

- In pea plants, purple flowers (P) are dominant over white flowers (p) show the cross between two heterozygous plants.

GENOTYPES:
- PP (1); Pp (2); pp (1)
- ratio 1:2:1

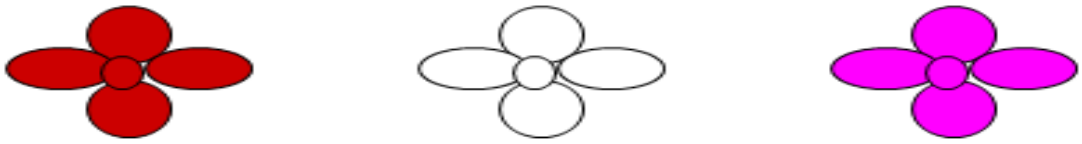
PHENOTYPES:
- purple (3); white (1)
- ratio 3:1

	P	p
P	PP	Pp
p	Pp	pp

2-Incomplete Dominance

- A third (new) phenotype appears in the heterozygous condition.
- Flower Color in 4 O'clocks

RR = red **rr = white** **Rr = pink**

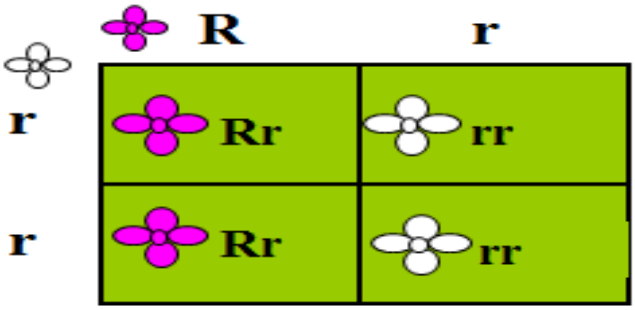


Problem: Incomplete Dominance

- Show the cross between a pink and a white flower.

GENOTYPES:
- Rr (2); rr (2)
- ratio 1:1

PHENOTYPES:
- pink (2); white (2)
- ratio 1:1



3-Codominance

- The heterozygous condition, **both** alleles are expressed equally
- Sickle Cell Anemia in Humans

NN = normal cells

SS = sickle cells

NS = some of each

Problem: Codominance

- Show the cross between an individual with sickle-cell anemia and another who is a carrier but not sick.

GENOTYPES:
- NS (2) SS (2)
- ratio 1:1

PHENOTYPES:
- carrier (2); sick (2)
- ratio 1:1

4-Overdominance

- Overdominance is the phenomenon in which a heterozygote is more vigorous than both of the corresponding homozygotes. It is also called **heterozygote advantage**
- The phenotype of the heterozygote individuals being more pronounced than in either homozygote.
- Overdominance is related to a common mating strategy used by animal and plant breeders
- Two different highly inbred strains are crossed. The hybrids may display traits superior to both parents. This phenomenon is termed **hybrid vigor** or **heterosis.**
- **Heterosis** is used to improve quantitative traits such as size, weight and growth rate
- **Heterosis is different from overdominance, because it typically involves many genes**

Hybrid corn

- Nearly all field corn (maize) grown in most developed nations exhibits heterosis. Modern corn hybrids substantially out yield conventional cultivars and respond better to fertilizer.

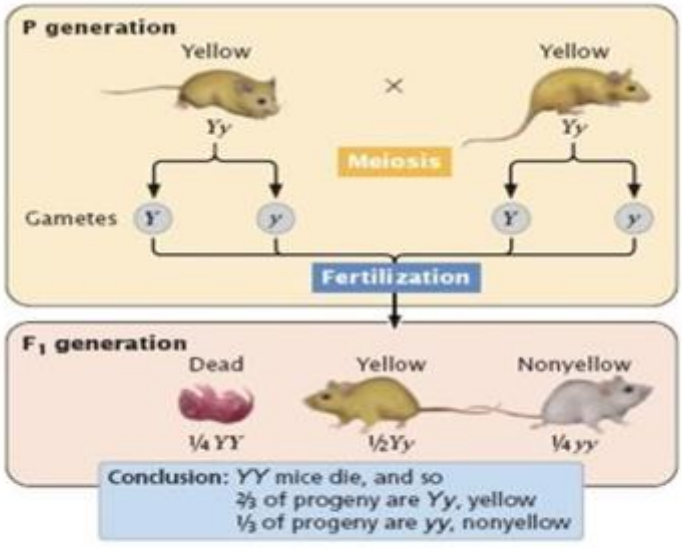
5-Lethal genes

- Lethal allele: An allele that results in the death of an organism, and the gene involved is called Essential gene (**an allele that gives rise to the death of an organism). The gene that is responsible is called an ESSENTIAL GENE (Can be either Dominant or Recessive)**
- Essential gene: are genes that, when mutated, can result in a lethal phenotype.
- If the mutation is caused by a dominant lethal allele, the homozygote and heterozygote for the allele will show the lethal phenotype. If the mutation is caused by a recessive lethal allele, the homozygote for the allele will have the lethal phenotype. Most lethal genes are recessive.

a- Dominant genes with recessive lethal effect.

Examples of diseases caused by recessive lethal alleles are cystic fibrosis, Tay-Sachs disease, sickle-cell anemia, and brachydactyly. Huntington's disease is caused by a dominant lethal allele and even though it is not described as lethal, it is invariably lethal which means that the victim experiences gradual neural degeneration and mental deterioration for some years before death occurs. One coat color of mice is caused by recessive lethal gene. This gene causes a death if both recessive alleles are possessed by the same individual.

Recessive lethal alleles don't cause death in the heterozygous form because a certain threshold of protein output is maintained. In the homozygous form, the protein output doesn't meet the threshold, causing death.



Y allele is completely dominant on y phenotypically, but in vitality case y is dominant on Y.

b- Recessive gene with recessive lethal effect.

In case of corn plant that contains a nodules in the edge part of chromosomes called “Satellite”(contain genes responsible for chlorophyll production).

If two satellites found (full green color). If one satellite found (bright green). No satellite (no color, dead) Then one gene is sufficient for chlorophyll production.

6- Multiple alleles

Many genes have more than two alleles (even though any one diploid individual can only have at most two alleles for any gene), such as the ABO blood groups in humans, which are an example of multiple alleles. Multiple alleles result from different mutations of the same gene. Coat color in rabbits is determined by four alleles. Human ABO blood types are determined by alleles A, B, and O. A and B are codominants which are both dominant over O. The only possible genotype for a type O person is OO. Type A people have either AA or AO genotypes. Type B people have either BB or BO genotypes. Type AB have only the AB (heterozygous) genotype.

- Another example: eye color in *D. melanogaster*
- R = red, rw = white, rc= coral, ra= apricot

RR X rara

Rra (Red hybrid)

If rwrw X rara

rwra (Light apricot)

Multiple genes (Polygeny)

There are two types of heritable traits

- **Qualitative genetics**

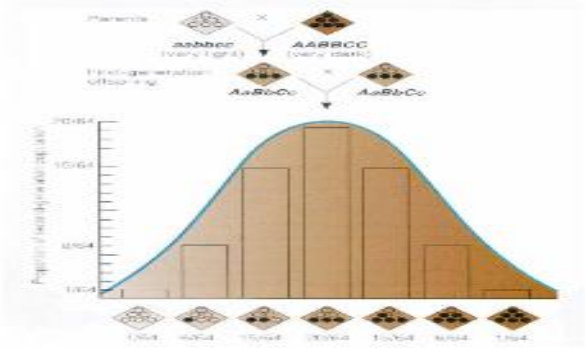
1- With contrasting expression. 2- Discontinuous traits. 3- Mostly controlled by a single genes

- **Quantitative traits**

1- Continuous traits: Controlled by multiple genes / polygenes (several genes located in different chromosomes but are responsible for a single trait)

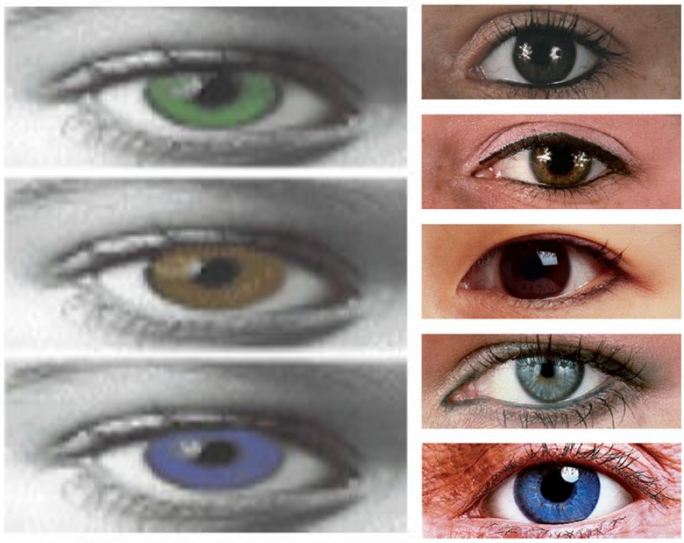
2- Phenotypes in ranges. 3- Phenotypes show normal distribution curve (bell-shaped curve)

- **Polygenic Inheritance**
- Most traits are controlled by more than one gene, (i.e., polygenic).
- For some polygenic traits, a few different alleles can produce a large number of different phenotypes (appearance).
- The example of skin color at the right, illustrates this.



Polygenic Inheritance: Eye Color

- Despite what we all learned in biology, eye color is likely to be a polygenic trait. The early view that blue is a simple recessive has been repeatedly shown to be wrong by observation of brown-eyed offspring of 2 blue-eyed parents.
- Blue-eyed offspring from 2 brown-eyed parents is a more frequent finding.
- At least two separate genes, each with two incompletely dominant alleles, govern human eye color.
- A man and a woman, each heterozygous for both genes, could have children with five different eye colors, ranging from light blue (no dominant alleles) through light brown (two dominants) to almost black (all four alleles dominant).



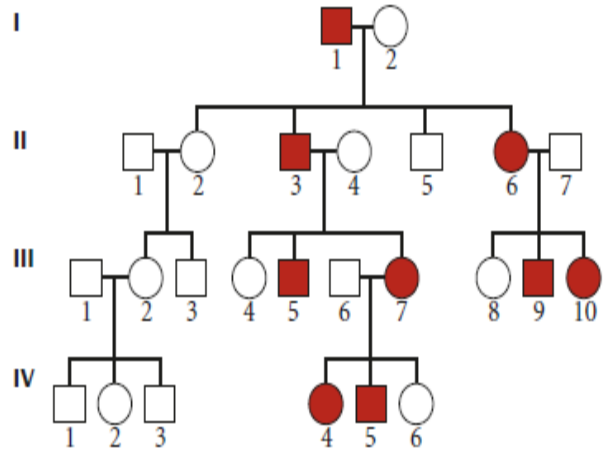
Questions/

1-What three factors complicate the task of studying the inheritance of human characteristics?

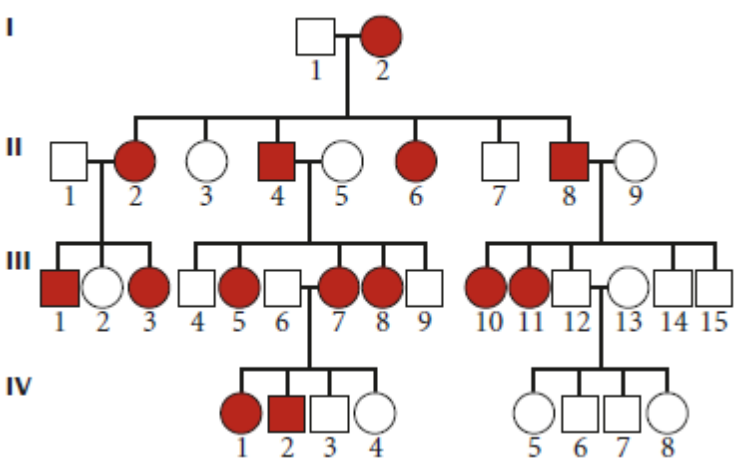
2-

For each of the following pedigrees, give the most likely mode of inheritance, assuming that the trait is rare. Carefully explain your reasoning.

a.



b.



3-What are continuous characteristics and how do they arise?

4-Give the expected genotypic and phenotypic ratios for the following crosses for ABO blood types.

a. I^Ai X I^Bi

b. I^AI^B X I^Ai

c. I^AI^B X I^AI^B

d. ii X I^Ai

e. I^AI^B X ii

5-The type of plumage found in mallard ducks is determined by three alleles at a single locus: M^R, which codes for restricted plumage; *M,* which codes for mallard plumage; and m^d, which codes for dusky plumage. The restricted phenotype is dominant over mallard and dusky; mallard is dominant over dusky $(M^R > M > m^d)$. Give the expected phenotypes and proportions of offspring produced by the following crosses.

a. $M^RM \times m^dm^d$

b. $M^Rm^d \times Mm^d$

c. $M^Rm^d \times M^RM$

d. $M^RM \times Mm^d$

6- The L^M and L^N alleles at the MN blood group locus exhibit codominance. Give the expected genotypes and phenotypes and their ratios in progeny resulting from the following crosses.

a. $L^ML^M \times L^ML^N$

b. $L^NL^N \times L^NL^N$

c. $L^ML^N \times L^ML^N$

d. $L^ML^N \times L^NL^N$

e. $L^ML^M \times L^NL^N$

7- The following pedigree represents the inheritance of a rare disorder in an extended family.What is the most likely mode of inheritance for this disease?