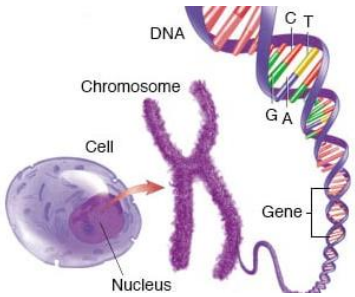
Biology 3201

Unit 2 Part 1

Mendelian Genetics

Name: _____

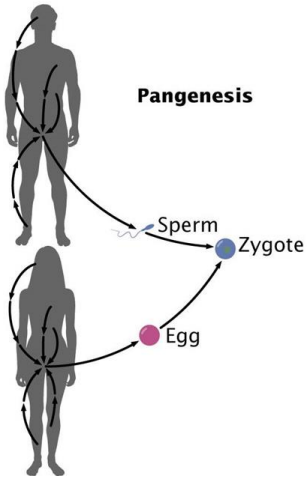
_____ the study of heredity, or the passing of traits from parents to offspring



Historical Explanations of Inheritance

Early experiments included breeding plants and animals in specific ways to produce offspring with desirable characteristics.

Canines, such as the Eurasian wolf (Canis lupus lupus), humans have used special breeding practices to gradually develop breeds of dogs with specific attributes.



The Greek philosopher Aristotle (384–322 B.C.E.) proposed the first widely accepted theory of inheritance, called _____.

According to this theory, egg and sperm consist of particles, called _____, from all parts of the body.

Upon fertilization of the egg by a sperm, the pangenes develop into the parts of the body from which they were derived.

In 1677, the amateur scientist Antony van Leeuwenhoek (1632–1723) discovered living sperm in semen with his exquisitely designed single-lens microscopes.

He believed that he saw a complete miniature person, called a _____, in the head of sperm.

Leeuwenhoek believed that the _____ came from the father but developed in the mother.

During the 1800s, when the breeding of ornamental plants was becoming popular, scientists observed that the offspring had characteristics of both parent plants.

The idea of blending became the working theory of inheritance. Scientists believed that characteristics of the parents blended in the offspring in a way that was irreversible. In other words, scientists believed that the original parental characteristics would not reappear in future generations.

None of the explanations of inheritance proposed prior to the 1850s stood the test of time.

Developing a Theory of Inheritance: Gregor Mendel's Experiments

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Figure 14.2 In this garden, Gregor Mendel planted and tended the pea plants that helped to establish the field of genetics.

The work of a monk and teacher named _____ (1822–1884) laid the foundation for the field of genetics, the science of inheritance.

Between the years 1856 and 1863, Mendel bred, tended, and analyzed over _____ pea plants in the monastery garden. He observed many different traits, or characteristics.

Before doing any experiments, Mendel let the plants self-pollinate to ensure that they were true-breeding.

True-breeding plants exhibit the same characteristics generation after generation.

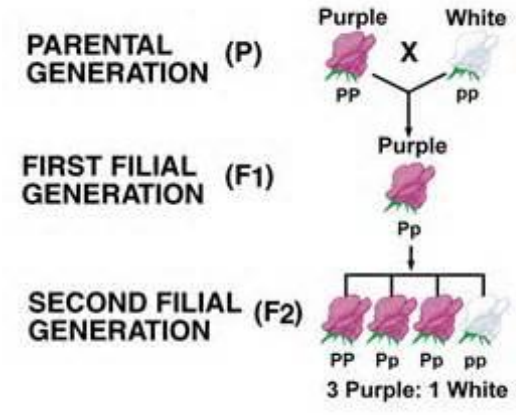
He crossed true breeding purple flowers with true breeding white flowers (_____).

Then he crossed their offspring (_____) with each other to create a new set of offspring (_____)

_____ parent generation

_____ first filial

_____ second filial



Mendel's Peas

Mendel studied seven traits that were expressed in two forms.

Mendel observed that, for every trait, the F1 plants showed only one of the two parental characteristics.

In the cross between plants _____, _____ all the seeds in the F1 generation were _____. Although all the F1 plants had a copy of each form of the factor for seed shape, only one form was shown, or expressed.

Why?

Gene	The set of information that controls a trait
Alleles	The different forms of a gene
Dominant Allele	An allele whose trait always shows up in the organism when the allele is present
Recessive Allele	The allele that is masked when a dominant allele is present
Hybrid	An organism that has two different alleles for a trait

_____ inheritance whereby dominant trait (or allele) conceals presence of recessive trait (or allele)

_____trait (or allele) expressed when present.

_____trait (or allele) not expressed when the dominant form is present

A _____is a portion of DNA that determines a certain trait. Genes are responsible for the expression of traits.

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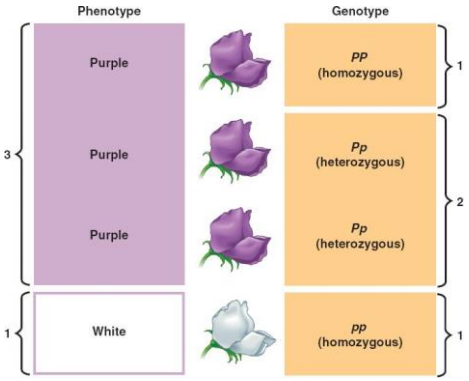
An_____ is a specific form of a gene. Alleles are responsible for the variations in which a given trait can be expressed.



_____combination of alleles for a trait (what the actual genetics/alleles are)

_____ can be written various ways. (e.g., Rr, R1R2, RR', RW, IAIB).

_____ individual with identical alleles for a trait

_____ having two dominant alleles (BB)

_____ having two recessive alleles (bb)

_____ individual with different alleles for a trait

_____ visible expression of a trait (what you see)

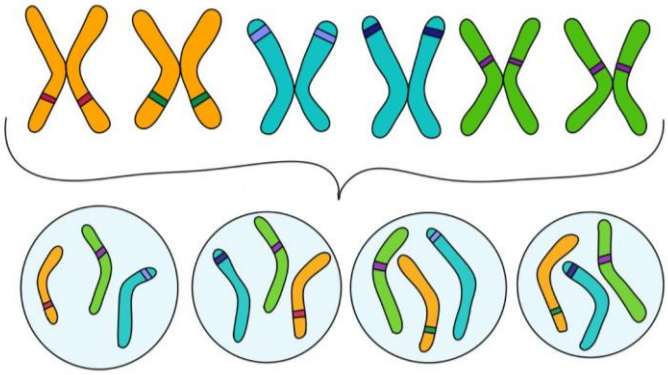
<u>Mendel's First Law -</u> The Law of Segregation

All individuals have two copies of each factor.

These copies segregate (separate) randomly during gamete formation, and each gamete receives one copy of every factor.

3 pairs of chromosomes:

possible gametes:



_____ an inherited trait is determined by pairs of factors (alleles) that segregate so that each gamete contains one copy

_____ grid showing possible results of genetic crosses

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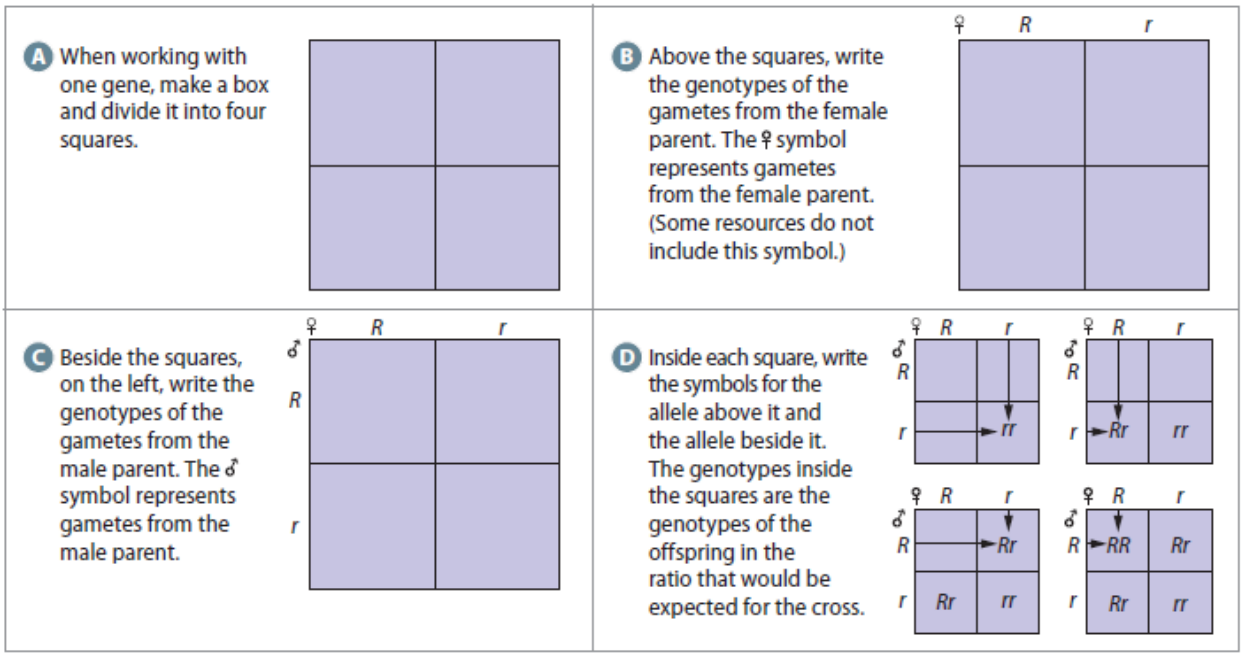
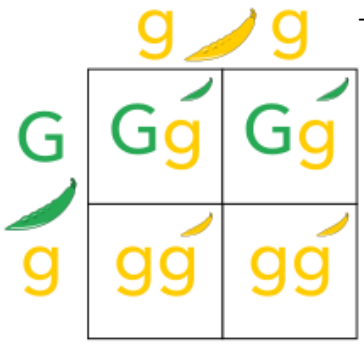


_____ describes the number of times a genotype would appear in the offspring after a cross.



_____

_____

_____ pertains to the relative number of offspring manifesting a particular trait or combination of traits. It can be determined by doing a cross and identifying the frequency of a trait or trait combinations that will be expressed based on the genotypes of the offspring.

_____

_____

Investigation 14.A Modelling a Monohybrid Cross

<u>Mendel's Monohybrid cross</u>

_____

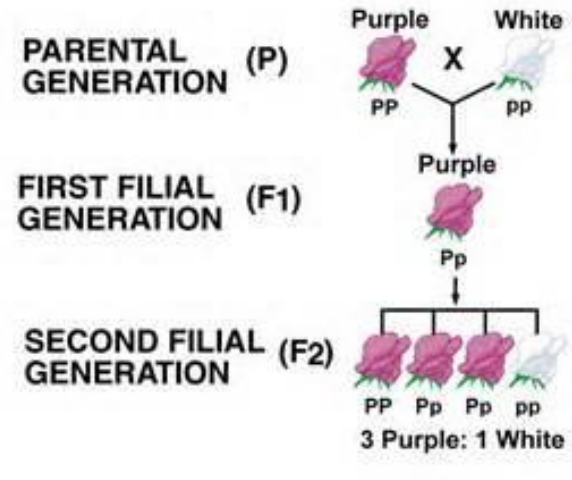
cross between individuals that differ in one trait. They are both homozygous, one is dominant and the other is recessive.

All offspring in the F1 generation will be Heterozygous.

The resulting F2 generation will have a

1:2:1 Genotypic Ratio

3:1 Phenotypic Ratio



<u>One Trait Crosses</u>

In humans, black hair colour is completely dominant to red hair colour. If a homozygous black haired male has children with a homozygous red haired female, what are the genotypic and phenotypic ratios?

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Somewhat Harder One Trait Crosses

In pea plants green seeds are dominant to yellow seeds. If a heterozygous plant is crossed with a homozygous recessive plant, what percent of the offspring will be yellow?

In pea plants green seeds are dominant to yellow seeds. If a heterozygous plant is crossed with another heterozygous pea plant, what percent of the offspring will have the same genotype as the parents?

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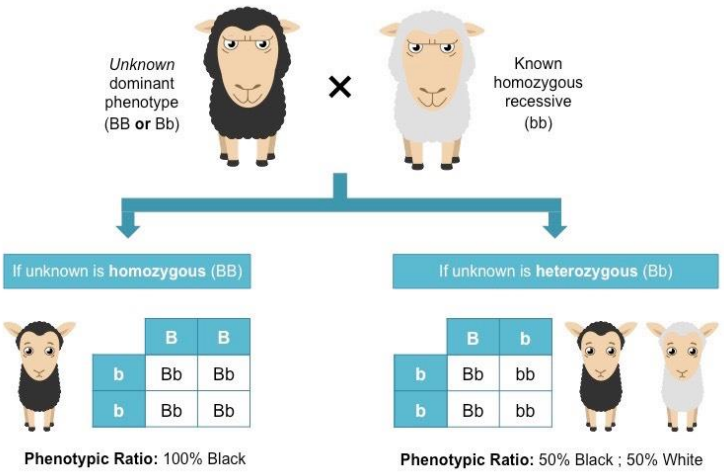
Test Cross



_____ cross between homozygous recessive individual and an individual with unknown genotype

You can use Punnett squares to predict the genotypes and phenotypes of the offspring of the test crosses.

Much Harder One Trait Cross Questions

If tallness is a dominant trait of pea plants, what are the genotypes of two pea plants that produce 146 tall and 52 short plants when mated?

If tallness is a dominant trait of pea plants, what are the genotypes of two pea plants that produce 153 tall and 144 short plants when mated?

Activity 14.1 Working with Punnett Squares

Exit Card #1

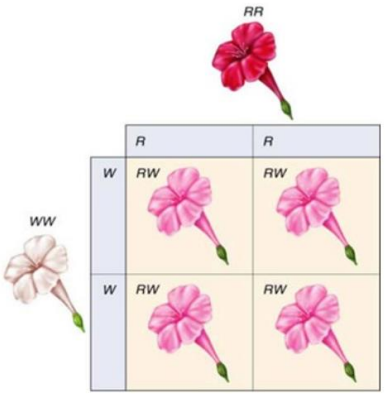
Incomplete Dominance

_____ neither allele for the same gene conceals the presence of the other – blending of the two traits

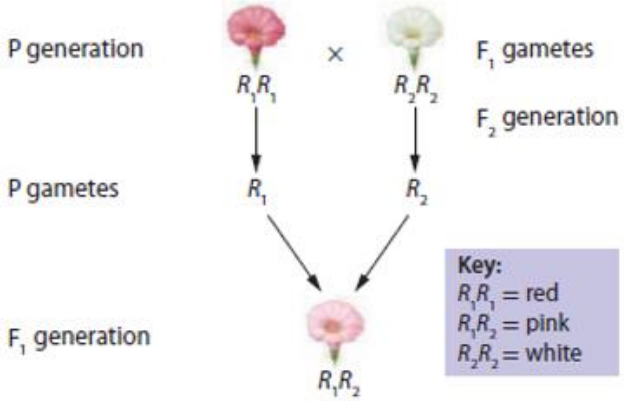
When representing incomplete dominance, upper-case and lower-case letters are not generally used to represent the alleles.

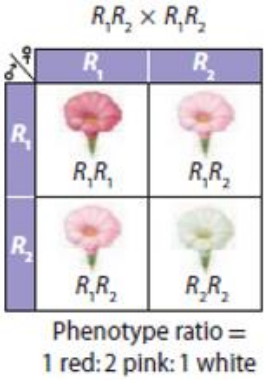
Some geneticists use all upper-case letters, with subscripts to denote the alleles.

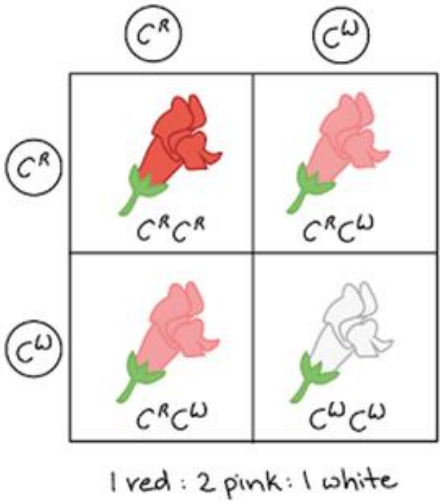
Don't forger about different notations for alleles *R_1R_2, RR', RW, C^RC^W* we will now start to see it appearing.



- Red= RR
- White= WW
- RW= pink- each allele is equally expressed to result in a blended product





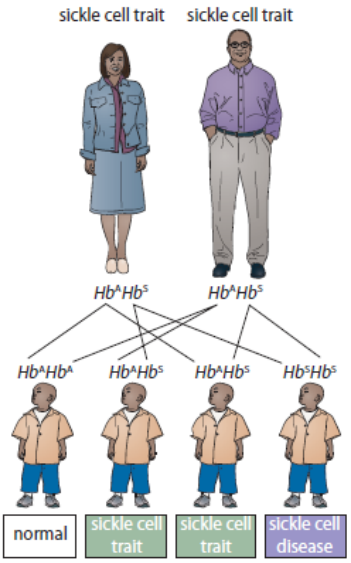


The allele for red flowers in the four o'clock plant directs the synthesis of red pigment.

When only one allele is present, the flower cannot make enough pigment to make the flowers red, resulting in incomplete dominance (pink flowers).

The allele for normal hemoglobin is represented as Hb^A, and the allele for sickle cell hemoglobin is represented as Hb^S.

Individuals who are homozygous (Hb^SHb^S) have sickle cell disease.



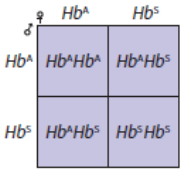


Figure 14.11 When a man and a woman are both heterozygous for the sickle cell gene, there is a one in four chance that they will have a child with sickle cell disease.

In humans, hair is incompletely dominant. The combination of straight hair and curly hair produces wavy hair. What are the phenotypic and genotypic ratios if a male with pure straight hair has a child with a female with wavy hair?

Co-dominance

_____ two alleles for a gene are expressed equally

_____

_____

A roan horse or cow is an excellent, visible example of co-dominance.

A roan animal is a heterozygote in which both the base colour and white are fully expressed.

If you look closely at the individual hairs on a blue roan you will see a mixture of black hairs and white hairs.

One allele is expressed in the white hairs, and the other allele is expressed in the black hairs.

A red roan has a mixture of chestnut-coloured hairs and white hairs.

The roan colouring of a horse usually does not affect the head, mane, and tail.

This horse's body looks blue because black and white hairs are thoroughly mixed.

A blue roan (H^BH^W) is the product of a mating between black (H^BH^B) and white (H^WH^W) parents.

In Rhododendron Flowers, Flower colour is Co-dominant. The combination of a red allele and a white allele produces a red and white flower mix. What are the phenotypic and genotypic ratios if a red flower and a white flower plant are crossed?

Activity 14.3 Analyzing Co-dominant and Incomplete Dominant Inheritance

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Exit Card #2

Multiple Alleles (A type of Co-dominance)

_____ refer to the occurrence of a gene with more than two alleles for a particular gene.

In humans, a single gene determines a person's ABO blood type.

This gene determines the type of antigen, if any, that is attached to the cell membrane of red blood cells.

The gene is designated I, and it has three common alleles: I^A, I^B, and i.

Table 14.2 ABO Blood Types

Genotype	Phenotype	Antigen
ii	O	none
I^Ai	A	A
I^AI^A	A	A
I^Bi	B	B
I^BI^B	B	B
I^AI^B	AB	A and B

_____, also called _____, is a type of protein found on the outside of red blood cells.

The protein is genetically inherited (passed down from your parents).

If you have the protein, you are Rh-positive.

If you did not inherit the protein, you are Rh-negative. The majority of people, about 85%, are Rh-positive.

	A -	A +	B -	B +
Red blood cells				
Antigens present	A antigen	A antigen Rh antigen	B antigen	B antigen Rh antigen

What is the probability that a man who has heterozygous type A blood and a woman who has heterozygous type B blood will have a child with type O blood?

What are the genotypic and phenotypic ratios?

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What is the probability that a man who has homozygous type A+ blood and a woman who has heterozygous type B- blood will have a child with type A+ blood?

Simplified (we will see the complex version soon)

Exit Card #3

Mendel's Second Law

_____ two alleles for a gene assort independently of alleles for other genes during gamete formation (not always true)

If genes are located close to each other during _____ in _____, they often move together.

Two Trait Crosses

A cross that involved two different traits which each have their own pair of alleles

For example Mendel compared the shape of seeds, round (R) vs wrinkled (r) and colour, green (G) vs yellow (g) at the same time.

4x4 = 16 boxes

	RY	Ry	rY	ry
RY	RRYY	RRYy	RrYY	RrYy
Ry	RRYy	RRyy	RrYy	Rryy
rY	RrYY	RrYy	rrYY	rrYy
ry	RrYy	Rryy	rrYy	rryy

However you will not always need to do 16, a lot the time it will be 4 or 8

In rabbits, gray hair is dominant to white hair and , black eyes are dominant to red eyes. What is the phenotypic and genotypic ratio between a male rabbit with homozygous gray hair and red eyes and a rabbit with heterozygous gray hair and heterozygous black eyes?

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An aquatic arthropod called a Cyclops has antennae that are either smooth or barbed. The allele for barbs (B) is dominant over smooth (bb). In the same organism Non-resistance to pesticides (N) is dominant over resistance to pesticides (nn). A Cyclops that is resistant to pesticides and has heterozygous barbed antennae is crossed with one that is smooth and heterozygous for Non-resistance. What is the chance of an offspring that is heterozygous for both traits?

Mendel's Dihybrid Cross

_____ cross between individuals that differ in two traits

Mendel crossed plants that were true-breeding for two different traits with plants that were _____ for the _____ form of the _____ _____.

Mendel crossed _____(_____)

with _____ (_____).

This produced an _____ of plants that were all heterozygous for both traits (_____).

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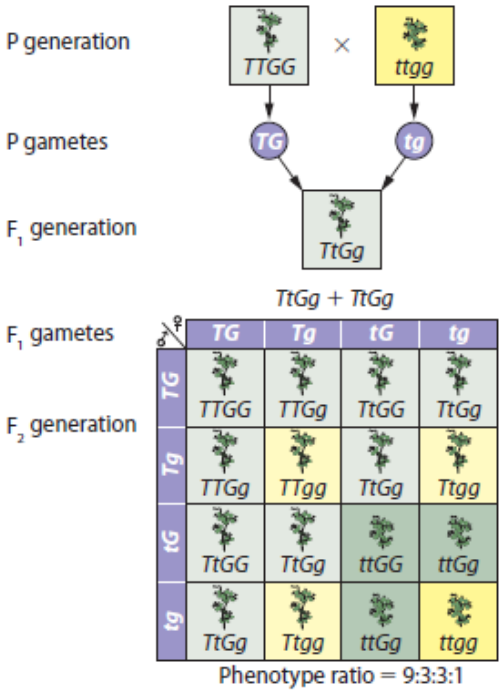
Mendel allowed the _____ and then analyzed the traits of the _____.

The cross _____ produced F2 plants with the phenotypes of tall with green pods, tall with yellow pods, short with green pods, and short with yellow pods in a ratio of _____. Phenotypic Ratio

For _____ cross that Mendel carried out and analyzed, he found the same pattern in the F2 generation.



Activity 14.2 Analyzing a Dihybrid Cross

Exit Card #4

<u>Chromosome Theory of Inheritance</u>

in 1902, _____ (1877–1916), a graduate student at Columbia University in New York, studied sperm development in _____

Sutton examined the processes of _____ and migration of sister chromatids during meiosis I and meiosis II.

Sutton realized that the distribution of chromosomes into developing gametes _____

_____

_____

These factors come in pairs, as do chromosomes. During gamete formation, the factors segregate just as homologous chromosomes do.

Sutton published a paper proposing the theory that the inherited factors described by Mendel are carried on chromosomes.

Around the same time, German biologist _____ (1862–1915) was studying

_____.

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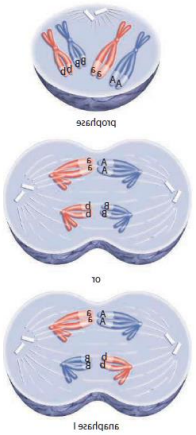
Working independently of Sutton, Boveri proposed the _____ to explain Mendel's observations.



_____

inherited factors (now known as genes) are carried on chromosomes

AKA

genes are located on chromosomes, and chromosomes provide the basis for the segregation and independent assortment of genes.

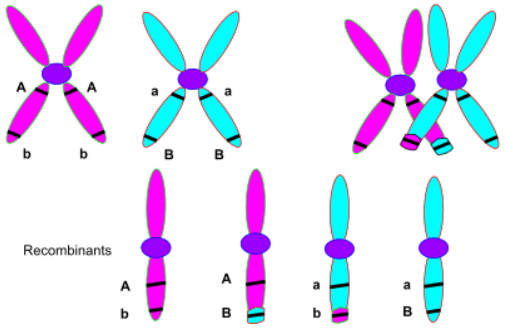
Often referred to as the Sutton-Boveri chromosome theory of inheritance.

Genes On The Same Chromosome

Sutton predicted that when alleles of two different genes are on the same chromosome they do not assort independently. (_____)

_____ genes on the same chromosome

Experimental data show, however, that linked genes segregate on a regular basis.



Morgan and The Chromosome Theory of Inheritance

Thomas Morgan was skeptical of Suttons work and did experiments with _____

_____

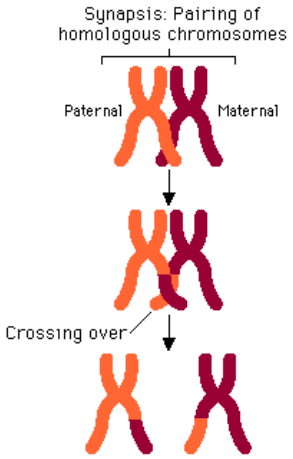
(_____).

He came to the same conclusion that genes were indeed carried on chromosomes.

He developed the theory of _____ by identifying the gene for white eyes in fruit flies was carried on the _____.

One of his students Alfred Sturtevant had assigned genes a numerical value based on location along the chromosome and found that linked genes could separate with predictability because of _____.

Crossing Over



Crossing over is a random event and occurs, with equal probability, at nearly any point on the sister chromatids, except near the centromere. This means that a crossover is more likely to occur between genes that are farther apart on a chromosome than between genes that are closer together.

Morgan and his students came up with a new definition for the chromosome theory of inheritance

_____

_____

_____

Modern Chromosome Theory of Inheritance

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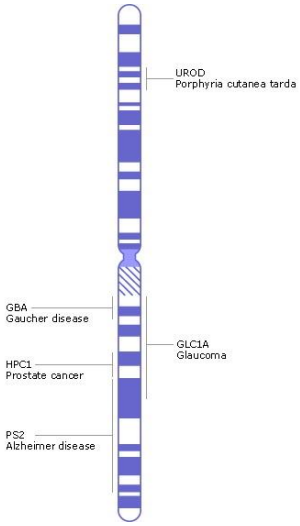


The gene-chromosome theory now states that _____

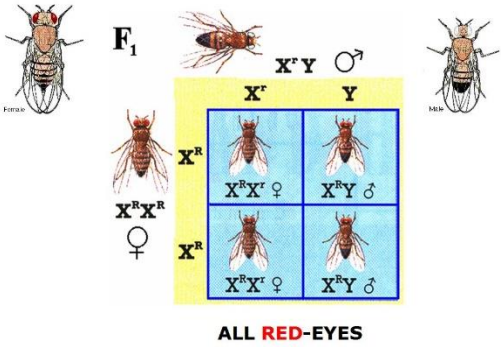
_____

_____

_____

Sex Linkage

_____ trait controlled by genes on X or Y chromosomes



Eye colour in fruit flies is X linked and carried on the X chromosome.

What are the phenotypes of the offspring if you were to cross a homozygous red eyed female and a white eyed male fruit fly?

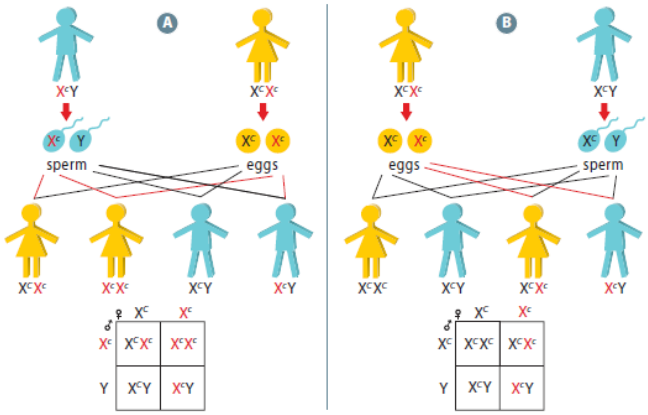


Figure 14.17 An allele for sex-linked colour vision deficiency is passed to the next generation. Males can pass the X-linked recessive trait only to their daughters (**A**). Females who are heterozygous for the condition have a 50 percent chance of passing the recessive allele to a child (**B**). Can a man with normal vision have a child who has the condition?

Sex Linkage and One Trait Crosses

In fruit flies, the gene for red eyes is dominant over the gene for white eyes. The trait is sex-linked on the X chromosome. What are the genotypic and phenotypic ratios if a heterozygous red-eyed female is crossed with a white eyed male?

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Activity 14.4 Sex-linked Inheritance Patterns

Exit Card #5

Polygenic Inheritance

_____ trait for which phenotypes vary between extremes

- Human Height

_____ trait controlled by many genes

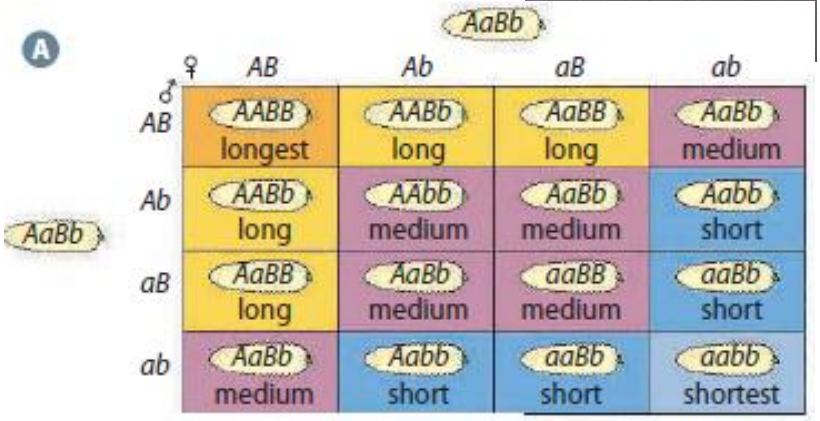
- Corn Ear Length
- Eye Colour

Polygenic Inheritance in Corn

Ear length is controlled by two genes, A and B. In true-breeding corn with the genotype AABB, four dominant alleles contribute to ear length.

As a result, this genotype has the longest ears.

True breeding corn with the genotype aabb has four recessive alleles, none of which contribute to ear length. Thus, this genotype has the shortest ears.

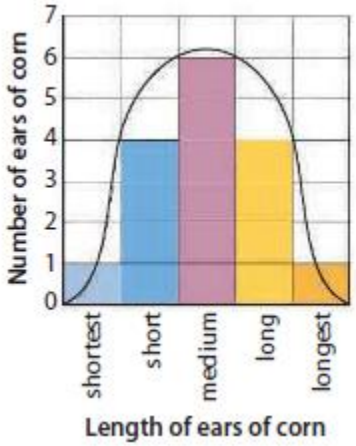


If true-breeding lines for shortest ears of corn and longest ears of corn are crossed, the F1 generation will have medium length ears (AaBb) where two dominant alleles contribute to ear length.

P - AABB × aabb F1 - AaBb

If we cross two of the F1 generation we get the Punnett square to the right.

Phenotypic ratio =
1 shortest : 4 short : 6 medium :
4 long : 1 longest



If there are three genes (A, B, and C), creating a range of zero to six contributing alleles, there is a more continuous distribution of phenotypes.

If three genes control ear length, there is a phenotypic ratio of 1:6:15:20:15:6:1.

Polygenic Inheritance Example

The table below shows the gene pairs involved in determining eye color. If a man with grey-blue eyes is crossed with a woman with green eyes, what are the genotype and phenotype ratios of their offspring?

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Genotype	Eye Colour
AA BB	black-brown
AA Bb	dark brown
AA bb	brown
Aa BB	brown-green flecked
Aa Bb	light brown
Aa bb	grey-blue
aa BB	green
aa Bb	dark blue
aa bb	light blue

Exit Card #6

<u>Genes and the Environment</u>

Environmental conditions often affect the expression of genetic traits.

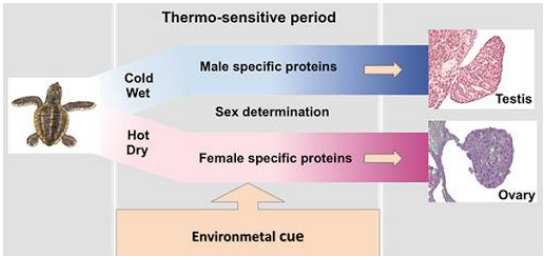
Some genes are influenced by temperature.

Siamese Cats - Their fur is pigmented on the cooler parts of their bodies: the face, ears, tails, and feet. Dark colouring is the result of a gene that is only active below a certain temperature.

Curly Wings Fruit Flies - If flies that are homozygous for curly wings are raised at 25 °C, their wings will be curly. If they are raised at 16 °C, their wings will be straight.

Seasonal Changes in Hares – Snowshow Hares change colour from brown in summer to white in winter with the seasonal temperature changes

Sex determination in reptiles – certain species of reptiles such as turtles – if the eggs are incubated at high temperature they produce females, at low temperatures they produce males.



Some genes are influenced by sunlight

Human Skin – natural skin colour darkens do to exposure to sunlight (tanning)

Hair Colour – The sun bleaches your hair and causes it to become lighter

Some genes are influenced by PH

Hydrangea Plant – a neutral soil will give pink flowers while an acidic soil will give blue flowers.

Some genes are influenced by diet/nutrition

Diet and Nutrition can affect gene expression

STSE CONNECTIONS + SOCIAL AND ENVIRONMENTAL CONTEXT Gene Expression and the Environment

<u>Pedigrees</u>

What is a pedigree?

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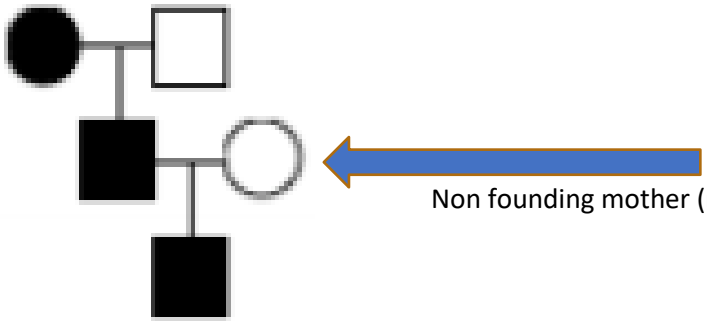
Holy Heart

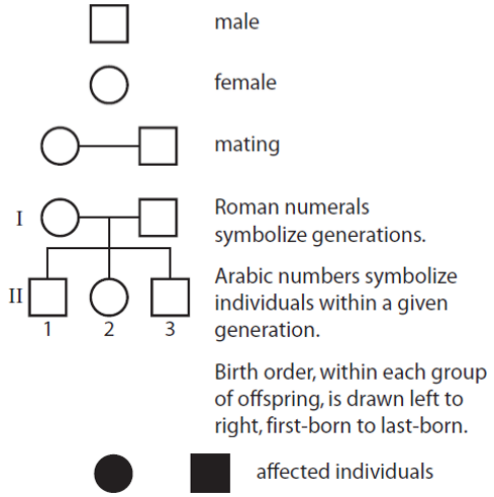
_____ flowchart that uses symbols to show inheritance of a trait within a biological family

_____ are the male and female at the very top of the pedigree.

_____:
one parent is from the original founders and the other is marrying into the family.



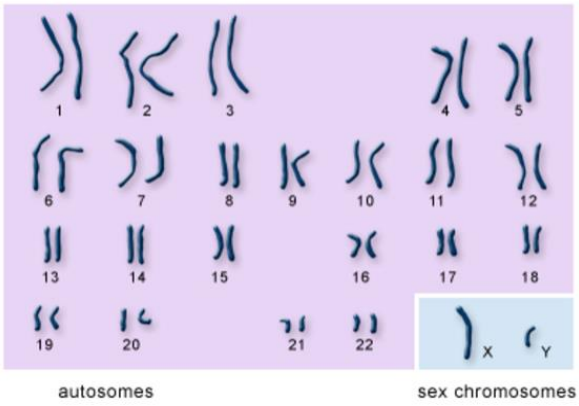
Non founding mother (married in)



Pedigrees Trace Inherited Genetic Disorders

A genetic disorder is an illness that is caused by changes to a person's genetic material. These changes can range from alterations of a single gene to changes to the structure and number of entire chromosomes.

When geneticists want to learn about the inheritance of human traits, they collect as much information as possible about the history of a biological family and use this information to create a diagram called a pedigree.



Review of Terminology

_____ chromosome other than sex chromosome

_____ X or Y chromosome; determines genetic sex

_____ is a pattern of inheritance characteristic of some genetic diseases. Autosomal means that the gene in question is located on one of the numbered, or non-sex, chromosomes. Dominant means that a _____ of the disease-associated mutation is enough to cause the disease. i.e _____ : _____

_____ is one of several ways that a trait, disorder, or disease can be passed down through families. An autosomal recessive disorder means _____

_____ of an abnormal gene must be present in order for the disease or trait to develop.

____:_____

i.e aa: _____

_____

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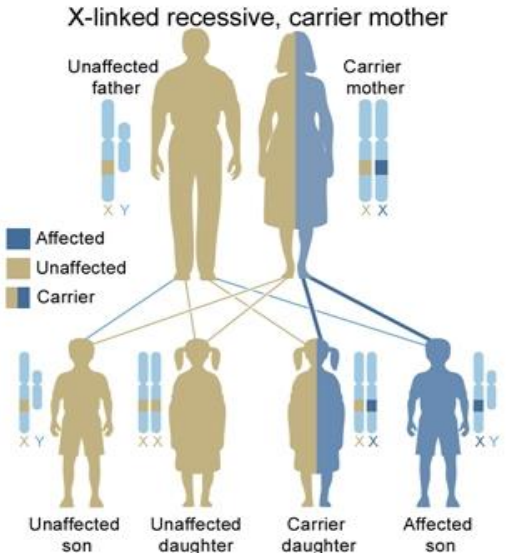
_____ inheritance is a way a genetic trait or condition can be passed down from parent to child through mutations (changes) in a gene on the X chromosome.

In _____(who only have one X chromosome), a mutation in the copy of the gene on the _____ causes the condition.

_____ would need _____ of the gene, one on each X chromosome.



General Assumptions

In the problems that follow, you'll be reasoning about the mode of transmission of genetic traits that are controlled by _____, with _____, a _____.

We also make three simplifying assumptions:

1. An individual in the pedigree will have the disorder (_____ _____) when the individual carries at least one dominant allele of a dominant trait, or two recessive alleles of a recessive a trait.

2. In each problem, the trait in question is rare in the general population. Assume for the purposes of these problems that _____ _____. This does not apply to the founding parents – either or both of the individuals at the top of the pedigree could be carriers.

3. Not-Y-Linked. The causative genes in these problems may be autosomal or X-Linked, but are _____.

Analyzing a Human Pedigree

5 Key Clues

There are five things to remember in reasoning about pedigrees.

(1) A unaffected female (white circle) or male (white square) individual cannot have any alleles of a dominant disorder. (because a single allele of a dominant trait causes an individual to be affected).

(2) Individuals marrying into the family are assumed to not be carriers of recessive traits (because the trait is rare in the population)

(3) An unaffected individual can be a carrier (have one allele) of a recessive trait. (because two alleles of a recessive trait are required for an individual to be affected)

(4) When a trait is X-linked, a single recessive allele is sufficient for a male to be affected. (because the male only has one allele of an X-linked trait)

(5) A father transmits his allele of X-linked genes to his daughters, but not his sons. A mother transmits an allele of X-linked genes to both her daughters and her sons.

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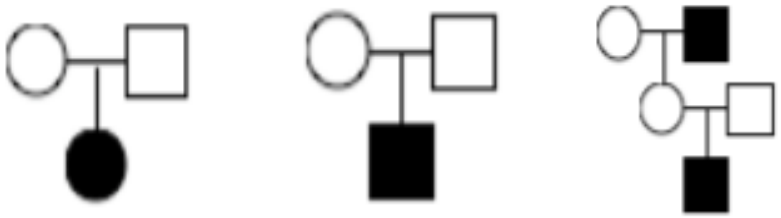
Patterns that Indicate A Recessive Trait

_____

_____

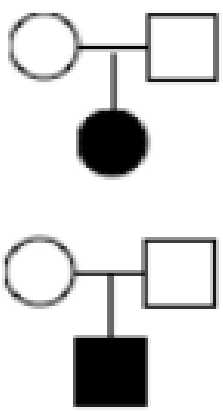


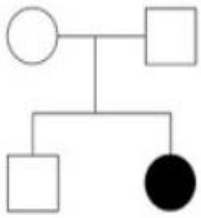
Since this is a genetic disease at least one parent must have an allele for the disease.

If neither parent is affected, the trait cannot be dominant.

_____

_____





Autosomal Recessive

Cannot be dominant as unaffected parents could not have an affected offspring

Parents MUST be heterozygous

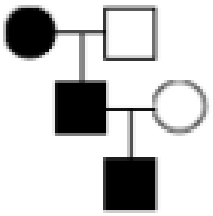
_____

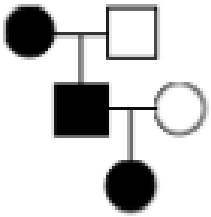
_____

_____.

An affected individual must inherit a recessive allele from both parents, so both parents must have an allele.

If the father had a recessive X-linked allele, he would have to be affected (since he only has one X-linked allele).



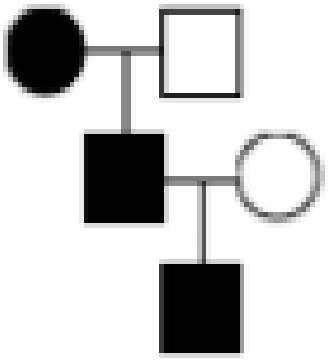


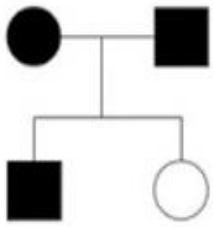
_____

_____

The unaffected mother, who is marrying in, does not carry an allele for the disease; so the affected child inherits an allele only from the affected father.

No child could be affected by a single autosomal recessive allele, or X-linked recessive allele, so the trait is dominant.





Autosomal Dominant

Cannot be recessive as affected parents could not have an unaffected offspring

Parents MUST be heterozygous

_____

_____

A father does not transmit X-linked alleles to a son, so the disease cannot be X-linked dominant.

Lets try some

What type of inheritance pattern is shown by this pedigree?

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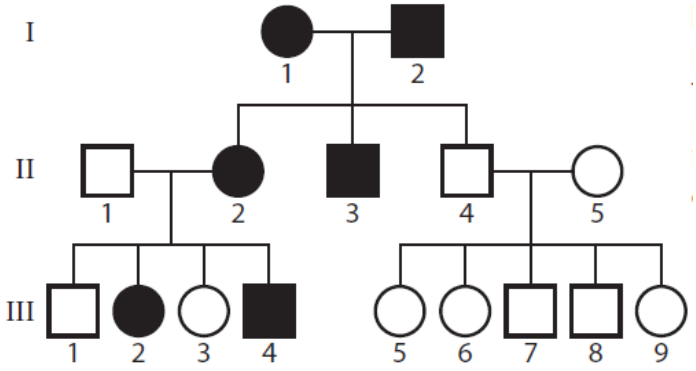
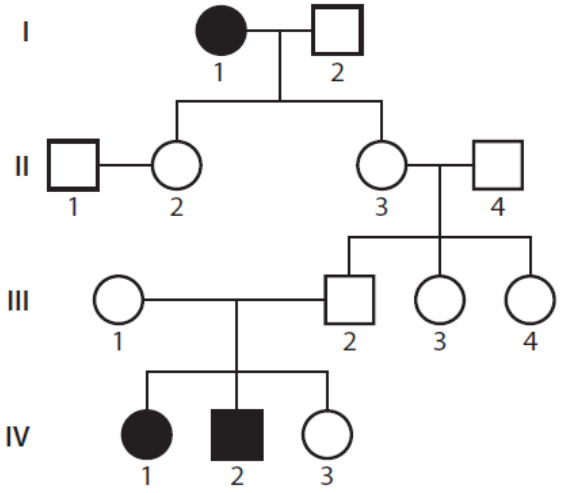
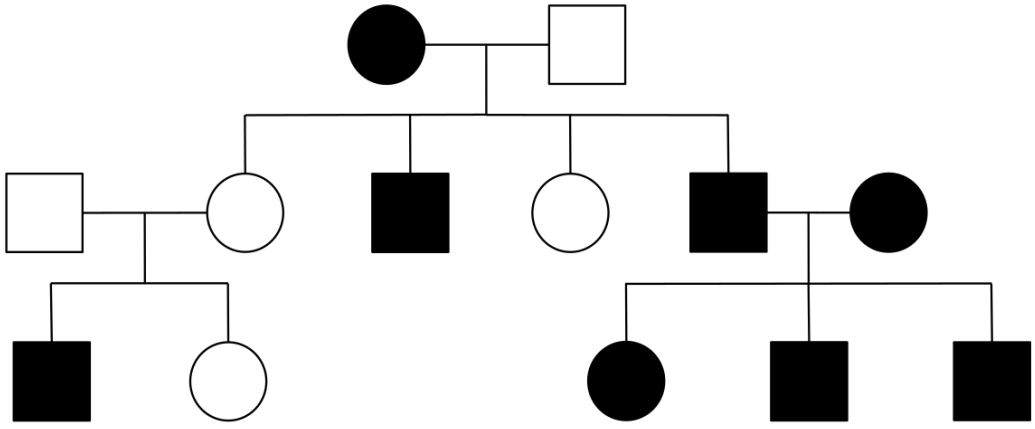


What type of inheritance pattern is shown by this pedigree?



What type of inheritance pattern is shown by this pedigree?



What type of inheritance pattern is shown by this pedigree?

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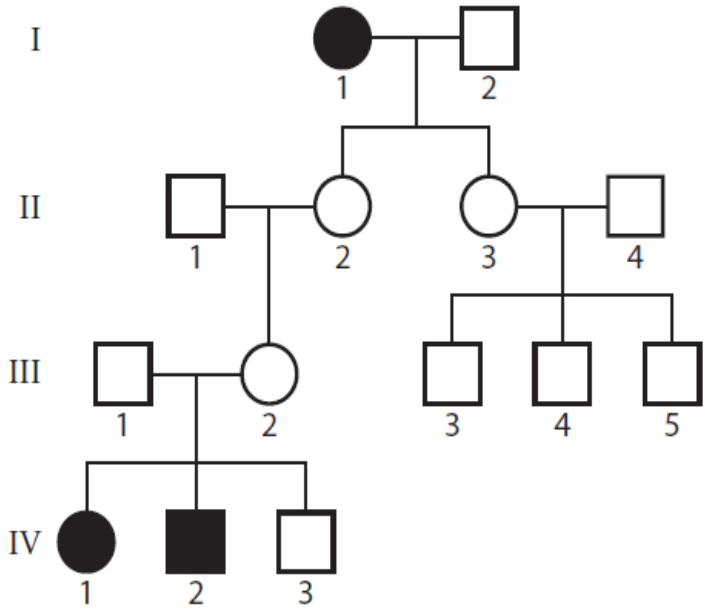
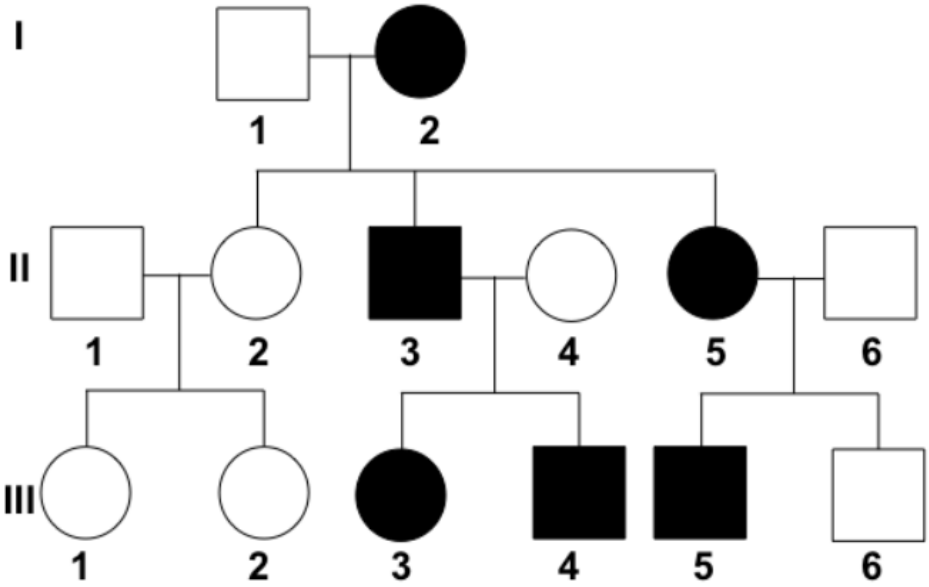


What type of inheritance pattern is shown by this pedigree?



All else fails try them by writing possibilities on them.

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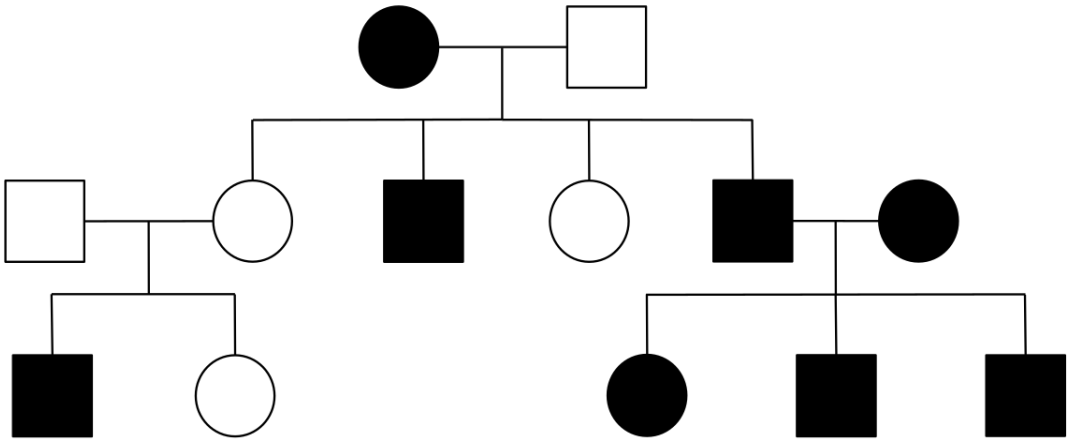
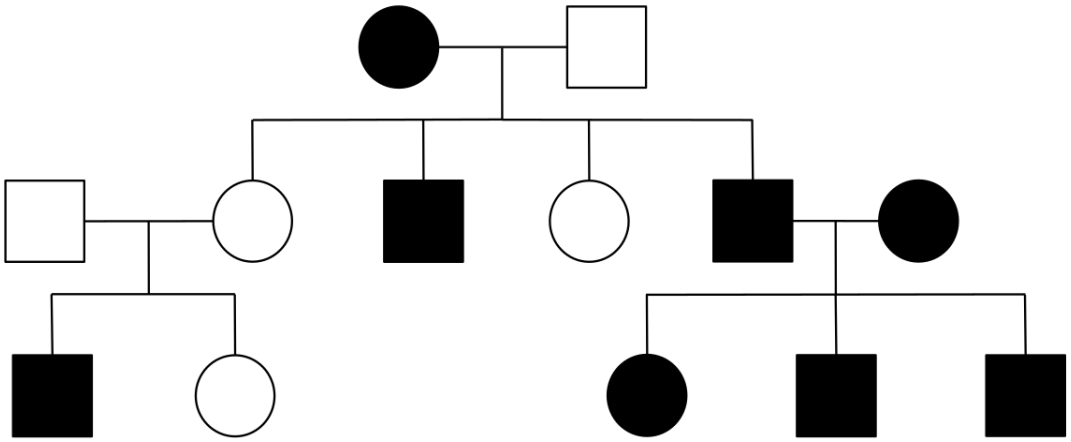
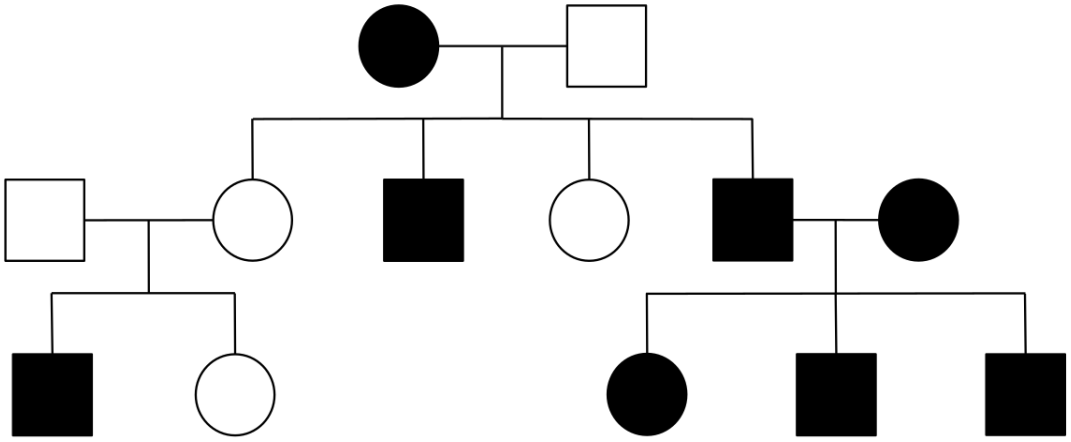


All else fails try them by writing possibilities on them.



All else fails try them by writing possibilities on them.



Blood Types can also be shown in pedigrees. In order to do blood types you will have to work them out.

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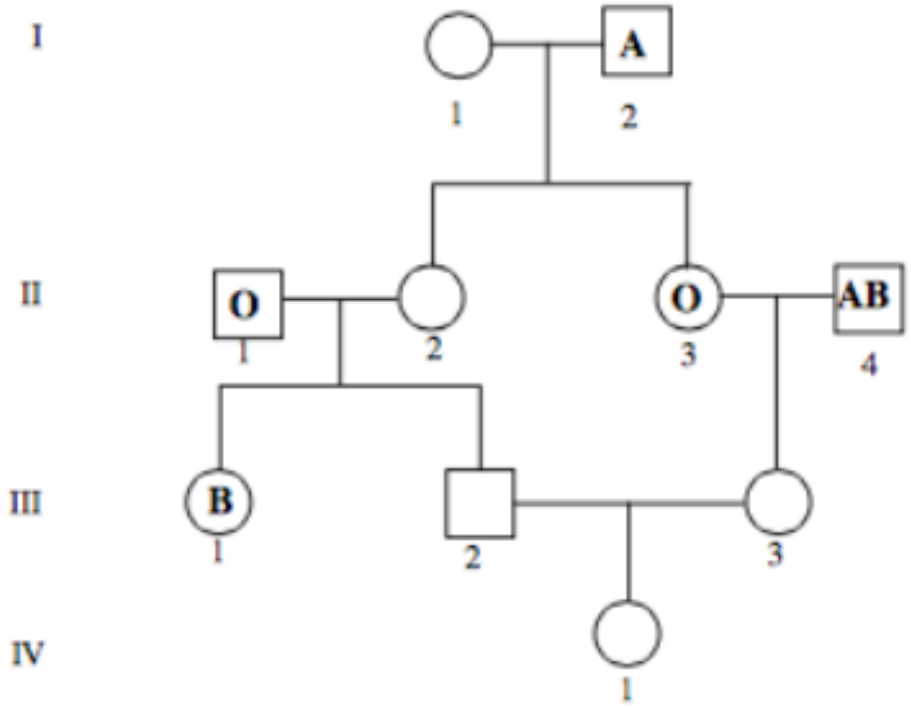

Activity 14.5 Analyzing Pedigrees

Exit Card #7