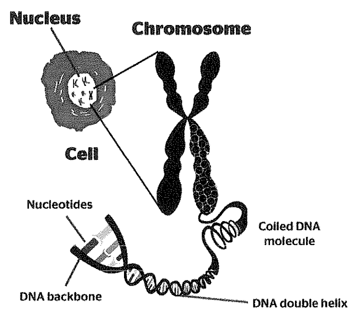
Name: ____________________ Date: __________ Class Period: Copy

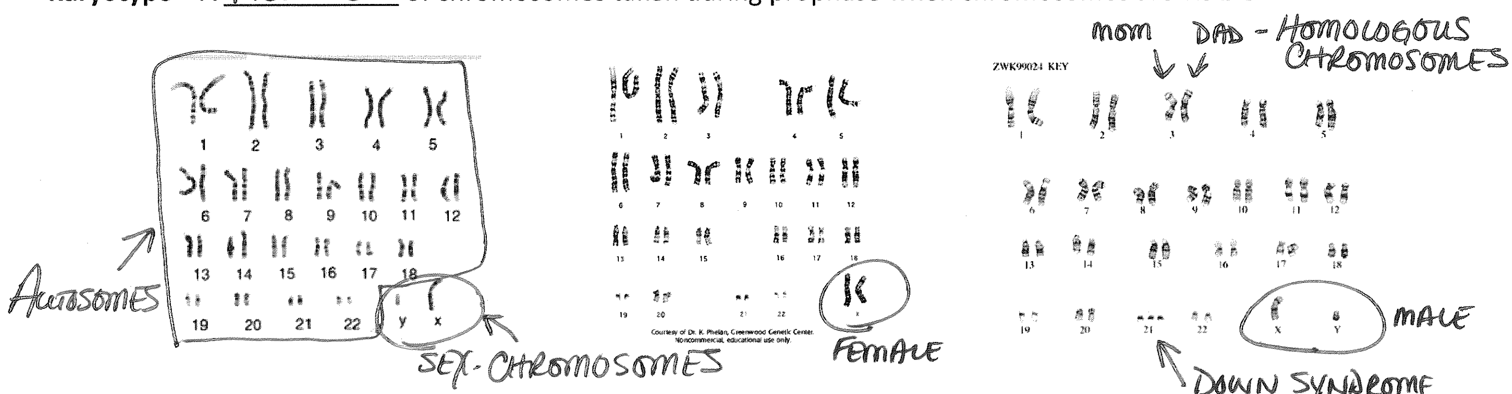
Beyond Mendelian Genetics

Essential Vocabulary

Chromosome – A package of tightly coiled DNA. Most human cells contain 46 chromosomes. Human reproductive cells (gametes) contain 23 chromosomes.



Karyotype – A PICTURE of chromosomes taken during prophase when chromosomes are visible.



Autosome – Any chromosome that is NOT a sex chromosome.

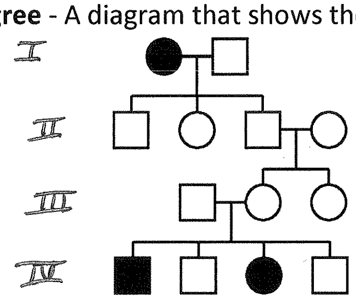
Sex-Chromosome – One of the pair of chromosomes that determines the GENDER of an individual.

Mutation – A change in the STRUCTURE or AMOUNT of genetic material in an organism.

Genetic Disorder - An inherited disease or disorder that is caused by a mutation in a GENE or by a CHROMOSOMAL defect.

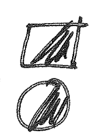
Carrier - An individual who has ONE copy of a RECESSIVE autosomal allele (Bb).

Pedigree - A diagram that shows the occurrence of a GENETIC TRAIT in several generations of a family.



Circle: Female	Horizontal Line: Mating
Square: Male	Vertical Line/Bracket: Siblings
Shaded: Affected by Disorder	GENERATIONS - TOP TO BOTTOM

CARRIERS SOMETIMES SHOWN AS ½ SHADED



Patterns of Inheritance

Single Trait Genetics - Some human genetic disorders are controlled by one gene that can either be determined by the dominant allele or recessive allele.

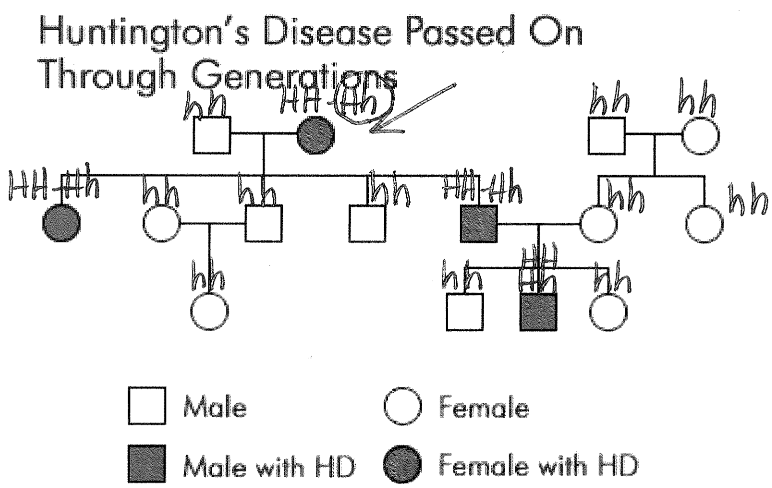
Dominant Trait Example: Huntington's disease, which affects the nervous system, is an autosomal dominant disorder within chromosome 4. Individuals with Huntington's disease must have at least one allele for the dominant trait (H). Therefore an individual with Huntington's disease must have at least one parent with the disease.

Possible Genotypes: Homozygous for Disease: HH Heterozygous for Disease: Hh No Disease: hh

Create a Punnett Square to determine the probability of a male who heterozygous for Huntington's Disease mates with a female without the disorder.

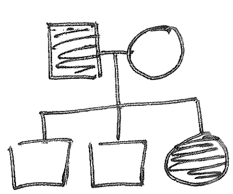
	H	h
h	Hh	hh
h	Hh	hh

What is the probability of this couple having a child with Huntington's Disease? 50%



The pedigree above represents a family with Huntington's Disease. What is the genotype of the unshaded family members? hh What are the two possible genotypes of the shaded members? HH or Hh Write all possible genotypes by the individuals in the pedigree. Refer to the shaded female in the first generation. Knowing that she had children without the disease, what is her genotype? Hh

Mr. Jones has Huntington's Disease but his wife does not. They have two sons without the disorder and one daughter with the disorder. Draw a pedigree for the Jones family below.



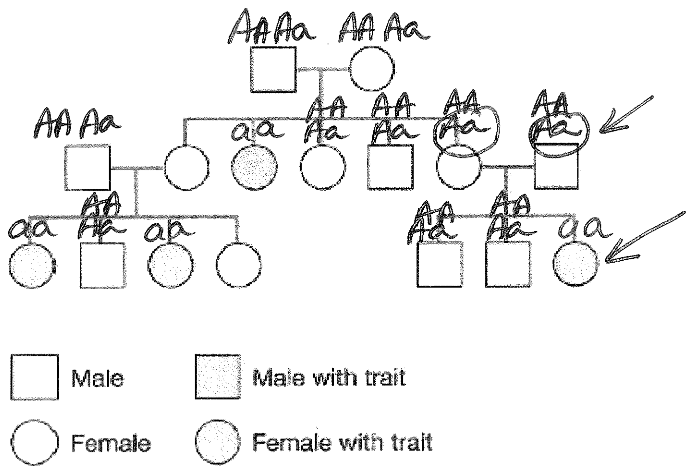
Recessive Trait Example: Albinism, which affects pigmentation (color), is an autosomal recessive disorder found within chromosome 15. Individuals that are carriers for the trait (Aa) do not have the disorder but do have the ability to pass the disorder to their offspring. The only way for an individual to have Albinism is if the recessive allele was passed from both parents.

Possible Genotypes: Have Disease: aa Heterozygous (Carrier) for Disease: Aa No Disease Alleles: AA

Create a Punnett Square to determine the probability of a male who is a carrier for Albinism mates with a female who is also a carrier for Albinism.

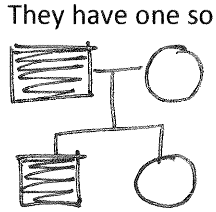
	A	a
A	AA	Aa
a	Aa	aa

What is the probability of this couple having a child with Albinism? 25%

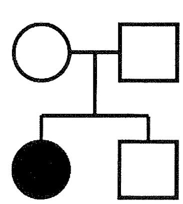


The pedigree above represents a family with Albinism. What is the genotype of the shaded family members with the trait? aa What are the two possible genotypes of the unshaded members? AA or Aa Write all possible genotypes by the individuals in the pedigree. Refer to the last shaded female in the third generation. Knowing that she has Albinism, the genotypes of her parents must be Aa and Aa.

Mr. Burns has Albinism and his wife does not. They have one son with albinism and one daughter without. Draw a pedigree for the Burns family below.



Refer to the pedigree below. Does this represent a trait that is dominant or recessive? RECESSIVE.
Explain how you know? THE PARENTS DO NOT HAVE THE TRAIT BUT MUST BE CARRYING THE RECESSIVE ALLELE WHICH THEY BOTH PASSED TO THE SHADDED OFFSPRING.



Polygenic Inheritance – When several GENES affect one phenotype. Examples of polygenic traits in humans are EYE color, height, and SKIN color. Polygenic traits are difficult to sort out because one phenotype is determined because the multiple genes may even be located on different CHROMOSOMES.

Incomplete Dominance – In some cases, a heterozygous offspring may have an intermediate, or "in between" phenotype. When a snapdragon plant that has red flowers is crossed with a snapdragon plant with white flowers, the offspring have pink flowers; therefore snapdragon color inheritance is much different than the pea plant flower color in Mendel's experiment. You do not use lower case letters because neither allele is dominant. Red (R) and White (W).

Possible Genotypes: Red Flowers: RR White Flowers: WW Pink Flowers: RW

Create a Punnett Square representing the cross between a white flowered plant and a pink flowered plant.

	W	W
R	RW	RW
W	WW	WW

What is the probability of the offspring having white flowers? 50%

In humans, hair texture is a good example of incomplete dominance. Individuals with straight hair are homozygous (SS) and individuals with curly hair are homozygous (CC). Individuals with wavy hair are heterozygous (SC). Create a Punnett Square for a man with straight hair and a woman with curly hair.

	S	S
C	SC	SC
C	SC	SC

What is the probability that their offspring will have straight hair like dad? 0%

Note: Hair textures are also polygenic, so there are always exceptions to the rule!

Multiple Alleles – Some genes have more than two possible ALLELES. For example, more than two alleles exist for rabbit coat color. Still, only two alleles for a gene can be present in an individual. Even though multiple alleles exist for rabbit coat color, there are still laws of dominance in affect. Each allele type is represented in the diagram below.

Rabbit coat color

Allele	Phenotype
C	Rabbit with fully colored coat
c^{ch}	Rabbit with light gray coat
c^{h}	Himalayan rabbit: white with dark ear tips, nose, paws, and tail
c	Albino rabbit

Order of dominance $C \rightarrow c^{ch} \rightarrow c^{h} \rightarrow c$

Refer to the table above. Create a Punnett Square for a male rabbit with a fully colored, light gray coat crossed with a female albino rabbit with dark tipped ear tips, nose, paws, and tail.

C c^{CH}

cc^{h}

	C	c^{CH}
c	Cc	cc^{CH}
c^{h}	Cc^{h}	$c^{CH}c^{H}$

What is the probability of these rabbits having offspring that are solid albino? 0%

cc

In humans, blood type is controlled by multiple alleles. Our blood type is determined by the types of proteins found on our red blood cells. There can be A proteins present, B proteins present, both A and B proteins present, or no proteins present. An individual receives two alleles (one from each parent); therefore the allele combinations can result in four possible blood types. Type A blood, Type B blood, Type AB blood, or Type O blood. Note that Type A and Type B are "codominant" while they are both dominant over Type O (which represents the complete absence of proteins).The chart below represents the multiple allele options.

Blood Types

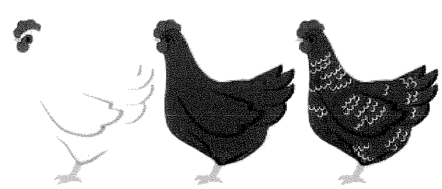
Genotype(s)	Phenotype
ii	O
I^AI^A, I^Ai	A
I^BI^B, I^Bi	B
I^AI^B	AB

Create a Punnett Square for a man that has heterozygous Type A (I^Ai) blood and a female that has heterozygous Type B (I^Bi) blood.

	I^A	i
I^B	I^AI^B	I^Bi
i	I^Ai	ii

What is the probability of the offspring having Type O blood? 25%

Codominance – Two traits that can appear at the SAME time. The phenotype of Type AB blood being caused by both Type A and Type B being present at the same time represents codominance in humans. Codominance also takes place in other animals. Erminette Chickens can have white feathers only (WW), black feathers only (BW), or heterozygous chickens will have both white and black feathers and are called speckled (BW)



Phenotype	WHITE	BLACK	SPECKLED
Genotype	WW	BB	BW

Create a Punnett Square representing the cross between two speckled chickens.

	B	W
B	BB	BW
W	BW	WW

What is the probability of the offspring having white feathers only? 25%

Sex-Linked Traits – Traits that are controlled by genes located on SEX chromosomes. The two possible human sex chromosomes are called the X and Y chromosomes. Females have two X chromosomes, and males have one X and one Y chromosome.

Create a Punnett Square crossing a female (XX) and a male (XY).

	X	Y
X	XX	XY
X	XX	XY

What is the probability that the couple will have a baby boy? 50% baby girl? 50%

Most sex-linked traits are linked to the X-chromosome. Colorblindness is a recessive trait linked to the X-chromosome. Only females can be **carriers** for X-linked traits. Below you can see the possible genotypes for colorblind males and females.

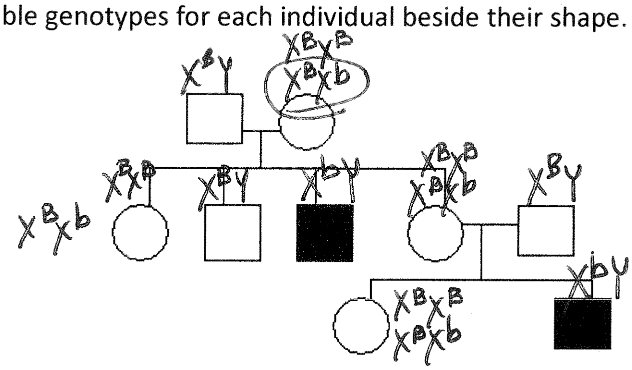
Normal Female	Carrier Female	Colorblind Female	Normal Male	Colorblind Male
X^BX^B	X^BX^b	X^bX^b	X^BY	X^bY

Create a Punnett Square representing the cross between a colorblind female and a normal male. You must keep the allele for the trait (B or b) linked to the X-chromosome. The interpretation of the Punnett Square will be gender specific.

	X^B	Y
X^b	X^BX^b	X^bY
X^b	X^BX^b	X^bY

If this couple has a baby boy, what is the probability that the boy will be colorblind? 100%
If this couple has a baby girl, what is the probability that the girl will be colorblind? 0%

Pedigrees for sex-linked disorders can be tricky. Because the disorder is linked to a sex-chromosome, you must provide the sex-chromosomes and allele for the trait for every individual's genotype. Refer to the pedigree below for colorblindness. Write all possible genotypes for each individual beside their shape.



Knowing that the shaded males are colorblind, what must be the genotype of their mother's? X^BX^b father's? X^BY or X^bY
How did you know? THE MALES GET THE Y CHROMOSOME FROM DAD & X FROM MOM. MUST GET COLOR B. FROM MOM.
Create a Punnett Square for the parents in the first generation. If the couple in the first generation has another daughter, what is the chance that the daughter will be colorblind? 0%
Why are X-linked triats more common in boys than girls? GIRLS HAVE 2 X-CHROMOSOMES. AS LONG AS THEY GET A HEALTHY GENE FROM ONE PARENT THEY WILL NOT HAVE THE RECESSIVE TRAIT. GIRLS CAN BE HEALTHY CARRIERS. BOYS EITHER HAVE IT OR THEY DON'T.

	X^B	Y
X^B	X^BX^B	X^BY
X^b	X^BX^b	X^bY