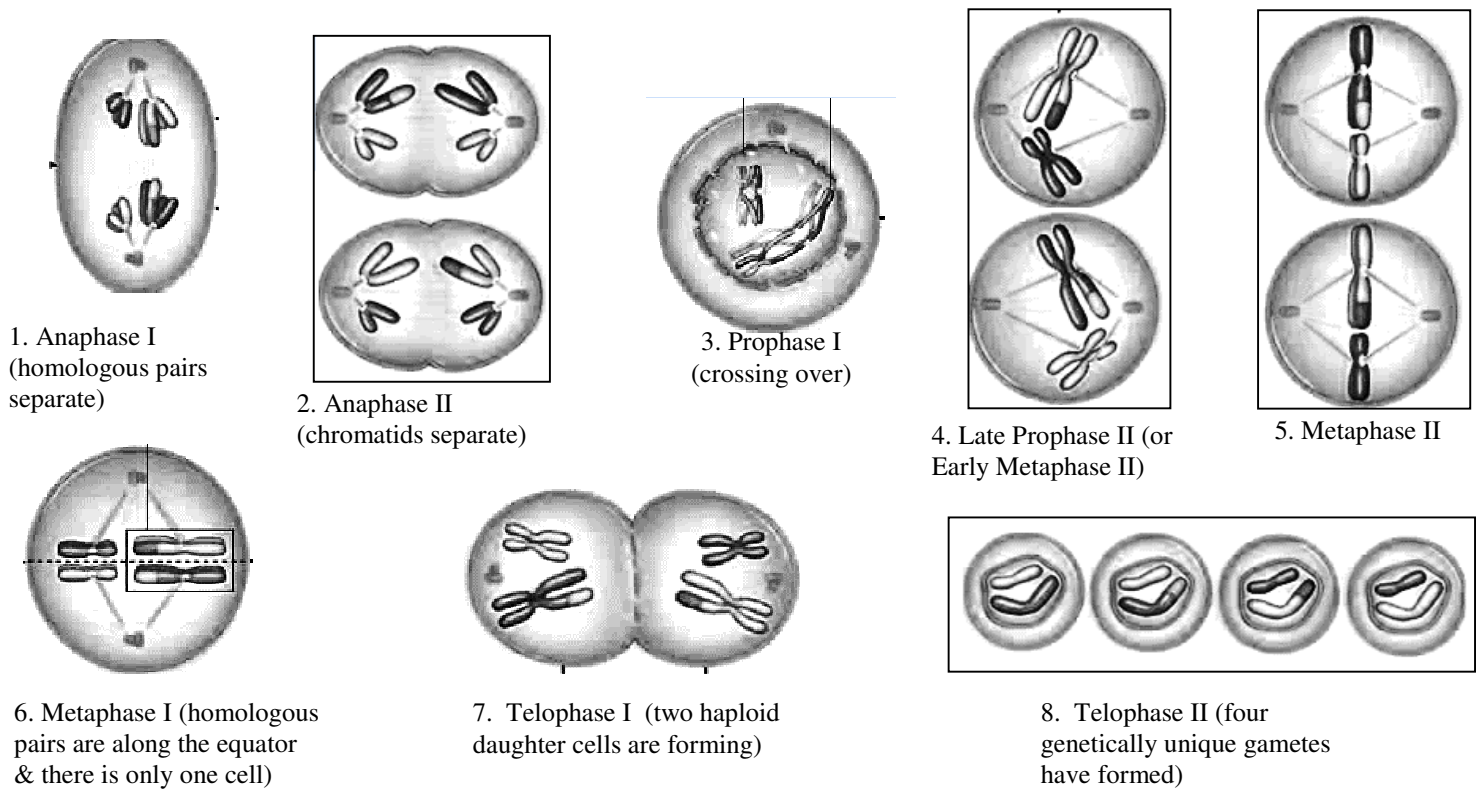


Review for Meiosis and Genetics Unit Test: Theory

1. What is a **karyotype**? What stage of mitosis is the best for preparing karyotypes?
 - a karyotype is a picture of all of the chromosomes in a cell, organized into homologous pairs
 - karyotypes are prepared when the cells are in very late prophase or early metaphase when the chromosomes are condensed and most visible
2. What are **genes** and what is their function?
 - genes are sections of DNA which (chromosomes) which code for a specific trait
 - some genes are the 'recipe' for a protein, others are important in regulating cell functions
3. What are **alleles**?
 - alleles are an alternate form of a particular gene, they produce slightly different proteins
4. Below are the links to two very good reviews of meiosis. They are worth watching:
<http://www.lpscience.fatcow.com/jwanamaker/animations/meiosis.html>
<http://www.sumanasinc.com/webcontent/animations/content/meiosis.html>
5. Regarding meiosis:
 - a) Which type(s) of cells perform meiosis?
 - meiosis takes place in reproductive cells in the gonads
 - b) What are the two main (overall) purposes of meiosis?
 - meiosis is a 'reduction division' so it reduces the number of chromosomes in a diploid cell by one half, producing haploid gametes. When gametes from two individuals join during fertilization, it produces a diploid zygote
 - meiosis introduces genetic diversity. The genes in the gametes are mixed in different combinations (by random segregation and crossing over) so the offspring have different traits than the parents. High genetic diversity is important for the survival of the species.
 - c) Which two processes of meiosis increase the genetic diversity of the offspring? During which stages of meiosis do these processes take place?
 - during Prophase I, the homologous chromosomes in the reproductive cells line up beside one another (synapsis) and non-sister chromatids cross over (at cross-over points called chiasmata). The non-sister chromatids exchange sections of DNA, so all four chromatids have a unique combination of maternal and paternal genes.
 - during Metaphase I, the homologous chromosomes randomly move to either side of the equator so the maternal and paternal chromosomes are mixed up between the two daughter cells. Similarly, during Metaphase II, the sister chromatids are oriented randomly on either side of the equator so the maternal and paternal chromosomes are further mixed up between the gametes
 - crossing over and random segregation ensure that each gamete is genetically unique, so the offspring have maximal genetic diversity
6. Identify the following phases of Meiosis from the description. Include whether it is meiosis I or II.
 - a) Homologous chromosomes pair up and form tetrads Prophase I
 - b) Spindle fibers move homologous chromosomes to opposite sides Anaphase I
 - c) Nuclear membrane reforms, cytoplasm divides, 4 daughter cells form Telophase II
 - d) A haploid number of chromosomes line up along equator Metaphase II
 - e) Crossing-over occurs Prophase I
 - f) Chromatids separate Anaphase II
 - g) Homologous pairs of chromosomes line up along the equator Metaphase I
 - h) Cytoplasm divides, 2 genetically unique daughter cells are formed Telophase I (cytokinesis I)

7. Name the following stages of meiosis. Include whether it is meiosis I or II.



8. Mendel studied the inheritance of traits in the common garden pea and made significant discoveries in genetics.

a) Give three reasons why the garden pea is an excellent species to study:

- garden peas are inexpensive, readily available and easy to grow
- they reproduce quickly (people only produce offspring every 20 years or so)
- they have seven distinct traits that are easy to see and classify
- it is easy to control their reproduction by removing the male parts of the flowers. They can be cross-pollinated or allowed to self-pollinate (for the F_1 and F_2 generations)

b) Define or explain what is meant by a monohybrid cross and a dihybrid cross.

- a **monohybrid cross** is the breeding of individuals that differ for one trait; for example, a tall pea plant bred with a short pea plant
- a **dihybrid cross** is the breeding of individuals that differ in two traits; for example, a tall pea plant with purple flowers bred with a short pea plant with white flowers

c) Mendel used **true-breeding plants** for his F_1 cross. What are two other genetic terms that mean “true-breeding”?

- true-breeding means that the plants are homozygous or pure-bred. They always produce offspring with a known trait because they have two copies of the same allele for that trait

d) The offspring of Mendel’s F_1 cross were **hybrids**. What are two other genetic terms that mean “hybrid”?

- hybrid means that the plants are heterozygous or carriers. They have two different alleles for the trait

e) The traits that Mendel studied showed **complete dominance**. What does this mean?

- complete dominance means that if the individual has one copy of the dominant allele, it will express that trait and it will be visible in the individual. If the individual is heterozygous, there will be no evidence of the trait produced by the recessive allele

- f) What is the **principle of dominance**? Give an example to illustrate this concept.
- the principle of dominance states that when individuals with dominant and recessive traits are crossed, the offspring will only show (express) the dominant trait. This is in contrast to the idea that traits are “blended” in the offspring. For example, if true-breeding plants with red flowers are crossed with true-breeding plants with white flowers, the offspring will produce only red flowers if the red trait is dominant
- g) What is the **law of segregation**? Give an example to illustrate this concept.
- Mendel’s experiments suggested that each parent has two factors (alleles) that determine the presence of that trait in the offspring, but these factors separate during gamete formation so each parent only contributes one factor to the offspring
 - for example, if both parents are heterozygous for a trait, offspring can be homozygous recessive, homozygous dominant or heterozygous for that trait because they get one allele from each parent
 - in the F₂ generation, two heterozygous tall parent plants will produce offspring in the phenotypic ratio 3 tall (dominant) : 1 short (recessive) offspring because they get one allele from each parent
- h) What is the **law of independent assortment**? Give an example to illustrate this concept.
- during his dihybrid cross experiments, Mendel saw that the inheritance of one trait was completely independent of (unaffected by) the inheritance of other traits. Mendel didn’t know this was because each of the traits he studied was on a different chromosome

9. How is the **law of segregation** related to meiosis?

- the law of segregation (see ‘g’, above) states that each trait is determined by two factors and that the offspring get one of each factor from each parent
- during meiosis, the homologous chromosomes in a diploid reproductive cell are separated from one another and one of the homologous chromosomes is randomly assigned to one haploid gamete. So each gamete contains only one allele for each trait and these are passed to the offspring during fertilization

An animation showing how independent assortment during meiosis produces all the different combinations of alleles in gametes:

<http://www.sumanasinc.com/webcontent/animations/content/independentassortment.html>

10. How is the **law of independent assortment** related to meiosis? During what phase(s) of meiosis does the law of independent assortment apply?

- the law of independent assortment (see ‘h’, above) states that the inheritance of each trait is unaffected by the inheritance of other traits. This is true as long as the genes for the traits are found on separate chromosomes
- during metaphase I and II, the maternal and paternal chromosomes line up randomly on either side of the equator and are randomly pulled into the daughter cells. This means that the daughter cells get a random mix of alleles from both parents, so the inheritance of each trait is unaffected by the inheritance of other traits

animation: <http://www.sumanasinc.com/webcontent/animations/content/mendelindassort.html>

11. Explain what is meant by each of the following types of inheritance. Give an example of each to illustrate your answer:

- a) **complete dominance** is when the dominant allele for one trait completely masks the presence of the recessive allele for that trait. Only the dominant trait is expressed; for example, in Mendel’s peas the F₁ generation plants were all tall even though these plants had one tall and one short parent

- b) **incomplete dominance** is when the dominant trait and the recessive trait are ‘blended’ in the individual so the heterozygote has a phenotype that is part way in between the dominant and the recessive trait; for example, a homozygous red-flowered plant crossed with a homozygous white flowered plant would produce offspring with pink flowers
- c) **co-dominance** is when both alleles are dominant so both are expressed in the individual. The traits are not blended, each trait is clearly visible in the individual; for example, a homozygous red-flowered plant crossed with a homozygous white flowered plant would produce offspring with red and white striped flowers
- d) **multiple alleles** means that there are more than two alleles (versions of a gene) for one trait, even though each individual only has two copies of the allele (one maternal and one paternal). For example, multiple alleles control the inheritance of height in humans, this is why people can be such a wide range of heights, not just tall or short. Similarly, skin colour is controlled by multiple alleles, and also multiple genes which is why there is so much variation
- e) **sex-linked inheritance** means that the gene for a trait is carried on one of the sex chromosomes, either the X or the Y chromosome; for example, the gene for hemophilia is carried on the X chromosome so it is X-linked (sex-linked)

12. Explain why boys are much more likely to suffer from X-linked recessive disorders than girls.

- boys are much more likely to suffer from X-linked recessive disorders because they have only one copy of the X chromosome, so if that X chromosome has a recessive allele for a recessive disorder, it will be expressed even though it is recessive
- the Y chromosome is not homologous with the X chromosome, so the Y chromosome does not provide any alternative forms of the alleles on the X
- girls, on the other hand, get two different X chromosomes, one from their father and one from their mother. In order for a girl to develop an X-linked recessive disorder, she must get two copies of the recessive allele which is much more unlikely

13. How is an autosomal genetic disorder different from a sex-linked disorder?

- an autosomal genetic disorder is caused by a gene carried on an autosome (chromosomes 1 to 22) while a sex-linked disorder is caused by a gene carried on a sex chromosome (an X or Y)
- because boys and girls get different sex chromosomes, the expression of sex-linked disorders is different in males and females
- the expression of autosomal disorders is the same in males and females

14. What is a test cross and why is it used? Give an example to illustrate your answer.

- when an individual has the dominant phenotype but unknown genotype for a completely dominant trait, they could be either homozygous or heterozygous for the dominant allele
- in order to determine whether they are homozygous or heterozygous, they are bred with an individual which is homozygous recessive (they have the homozygous trait) and this is called a ‘test cross’

- in the Punnett squares to the left, a test cross was performed to see if a male black lab (B) is homozygous or heterozygous for the dominant black allele. He was bred to a female golden (yellow) lab

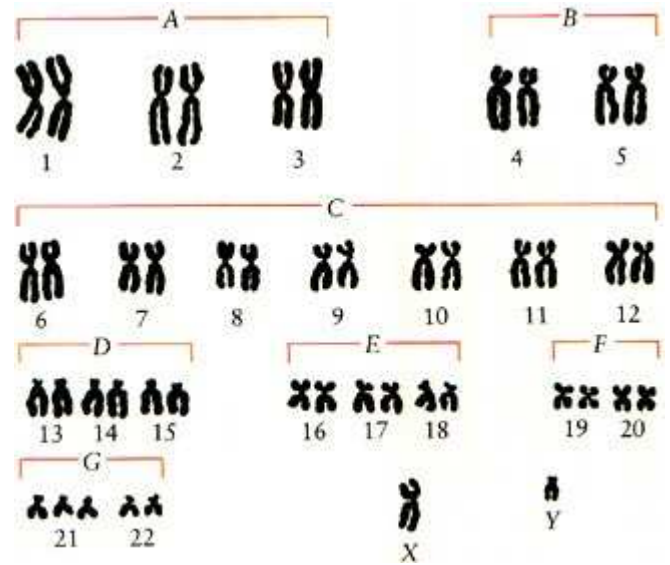
- if all the puppies are black (they have only the dominant trait), there is good evidence that the black male is homozygous for the dominant allele (top Punnett square)
- if any of the puppies are golden (yellow), then the male must be heterozygous for the allele (bottom Punnett square)

		Test Cross		← if unknown parent is homozygous dominant
		B	B	
homozygous recessive	b	Bb	Bb	
	b	Bb	Bb	

		B	b	← if unknown parent is heterozygous dominant
		homozygous recessive	b	
b	Bb		bb	

15. Give one example of each of the following and give a very brief description of the symptoms of this disorder:
- a) an **autosomal recessive disorder** is caused when an individual gets two copies of a recessive allele that is carried on an autosome (chromosomes 1 to 22). An example is cystic fibrosis which is caused by a single point mutation in the gene for a channel protein which is carried on chromosome 7. Approximately 4% of the population (1 in 25 people) carry the recessive allele. If an individual gets two copies of the recessive allele (one from each parent), then the cells lining the lungs, digestive tract and reproductive system can not pump sodium ions out of the cell or chloride ions into the cell. This causes the cells to secrete a very thick, sticky mucous that clogs the airways, intestines and reproductive organs. Bacteria build up in the mucous and cause infections. Patients have frequent respiratory infections and pneumonia is the most common cause of death in people with CF.
 - b) an **autosomal dominant disorder** is a disorder caused by a single copy of a dominant allele that is carried on an autosome. An example is Huntington's disease. It is caused by a gene on chromosome 4 which codes for a protein which is important in nerve cell function. The average age of onset of symptoms is between 30 – 50 years and causes progressive damage to the brain. Symptoms include loss of long and short term memory, changes in mood and muscle coordination. Uncontrollable involuntary muscle spasms and loss of cognitive (mental) function are devastating to the individual and their family. The disease is lethal.
 - c) an **X-linked recessive disorder** is caused by a recessive allele carried on the X-chromosome. Males need only one copy of the recessive allele in order to develop the disorder, while females need two copies (one on each of their X chromosomes) in order to develop the disorder. Examples of X-linked disorders include red-green colour blindness, premature balding and hemophilia. Hemophilia is a bleeding disorder in which a protein involved in making or maintaining a blood clot (scab) is ineffective. Hemophilia patients do not bleed more severely, they bleed longer because the body does not form effective clots to stop the blood from flowing. Even a minor injury can cause prolonged bleeding. An affected individual bruises very easily and may bleed to death from a relatively minor cut or internal bleeding.
 - d) a **non-disjunction disorder** is caused by an error during anaphase I or II of meiosis. During anaphase I, if the homologous chromosomes (tetrads) do not separate, both copies of the homologous pair are pulled into the same daughter cell. This cell ends up with an extra copy of a chromosome (causing a trisomy) while the other daughter cell ends up without a copy of this chromosome (causing a monosomy). During anaphase II, if the sister chromatids do not separate, both copies of the chromatid are pulled into the same daughter cell. This cell ends up with an extra copy of a chromosome (causing a trisomy) while the other daughter cell ends up without a copy of this chromosome (causing a monosomy). Examples of non-disjunction disorders include trisomy 18 (Edwards syndrome), trisomy 21 (Down syndrome), XO (Turner's syndrome), XXY (Klinefelter syndrome) and XYY ('supermale') syndrome. See notes from class or below for descriptions.
 - e) a **trisomy** is a non-disjunction disorder caused when homologous chromosomes or sister chromatids do not separate properly during Anaphase I or Anaphase II, respectively. A gamete gets an extra copy of a chromosome and this is passed to the zygote during fertilization. **Down syndrome** is one of the most common trisomies, caused by three copies of chromosome 21. Individuals with Down syndrome have flat faces, slanted eyes, large tongues, heart problems, thick necks and cognitive (mental) impairment.
 - f) a **monosomy** is a non-disjunction disorder caused when homologous chromosomes or sister chromatids do not separate properly during Anaphase I or Anaphase II, respectively. A gamete is missing a copy of a chromosome and this is passed to the zygote during fertilization. All autosomal monosomies are lethal to the fetus, so these fetuses are miscarried. **Turner syndrome** is one of the few non-lethal monosomies. A girl gets only one X chromosome. Affected individuals are always girls who are usually shorter than average, have webbed necks, thick legs and arms, are infertile and have normal intelligence. They never mature sexually, but they can be given hormone treatments which will help them to develop some secondary sex characteristics.

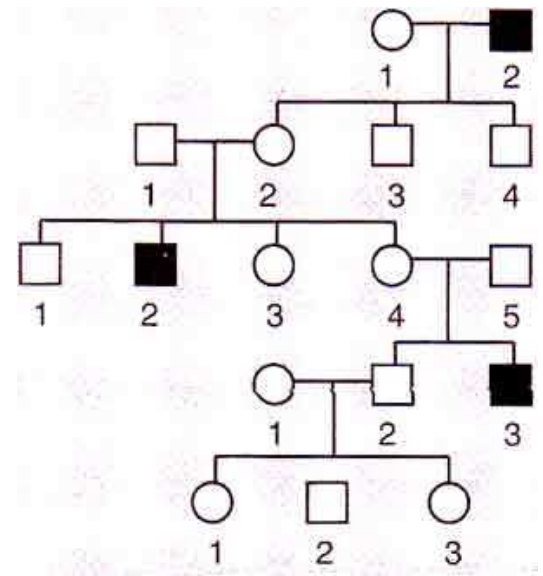
16. Study the karyotype shown to the right.
- What is the sex of this person? **Male**
 - What is the name of the syndrome of this person? **Down syndrome (trisomy 21)**
 - Describe very briefly what happened during meiosis to cause this disorder.
 - the answer to this question is the same as the answer to question 15e) above



Review for Meiosis and Genetics Unit Test: Pedigrees

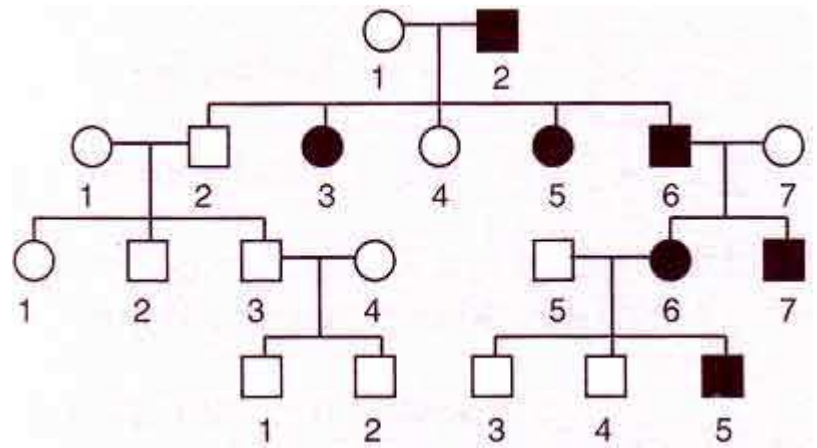
- Study the pedigree shown to the right and answer the following questions:

- How many generations are shown? **Five (5)**
- Is this disorder caused by a dominant or recessive allele? How do you know? Be very specific.
 - this pedigree shows a recessive disorder because individual III – 2 has the disorder, while neither of his parents is affected, so both parents must be heterozygous carriers of the recessive allele
- Could this disorder be X-linked? Explain why or why not.
 - this disorder could be X-linked because it is only seen in males
 - X-linked disorders are much more common in males than females because males only need a single copy of the allele in order to develop the disorder



- What are the genotypes of individuals (use alleles A/a):
 - I – 1 is A? (A-) because we don't know if they are homozygous or heterozygous
 - I – 2 is aa because they have the disorder
 - II – 1 is Aa because he must have the recessive allele in order to produce an affected son
 - II – 2 is Aa because she must have the recessive allele in order to produce an affected son
 - III – 4 is Aa because she must have the recessive allele in order to produce an affected son
 - IV – 2 is A? (A-) because we don't know if they are homozygous or heterozygous
- Could individual IV – 1 be a carrier for the disorder? Explain why or why not.
 - If the disorder is autosomal recessive, then IV – 1 could be a carrier. We don't know anything about her parents. Her children do not have the disorder, but if she and IV – 2 are both carriers, this could just be due to random chance. Their children could be CC or Cc and would not show the disorder
- Could the individuals in the fifth generation be carriers of the disorder? Explain why or why not.
 - if the disorder is autosomal recessive, then the fifth generation could be carriers of the recessive allele. Carriers have a normal dominant allele, so they could have the recessive allele and show no symptoms
- If the disorder is X-linked, from which parent did individual IV – 3 get the recessive allele? Explain.
 - if the disorder is carried on the X chromosome, then IV – 3 got the recessive allele from his mother (III – 4). Because IV – 3 is male, he had to inherit the Y chromosome from his father, so his X chromosome had to come from his mother

2. Study the pedigree shown to the right, then answer the following questions:
- Is this disorder caused by a dominant or recessive allele? How do you know?
 - the disorder is dominant because it is passed to every generation and every affected parent has produced an affected child
 - Is this disorder X-linked?
 - the disorder is not X-linked because it affects males and females equally
 - What are the genotypes of individuals (use the alleles B/b):



I – 1 is bb because they are not affected (all unaffected individuals are bb)

I – 2 Bb because they have the disorder but produced children who do not have the disorder (so not BB)

II – 2 is bb because they are not affected

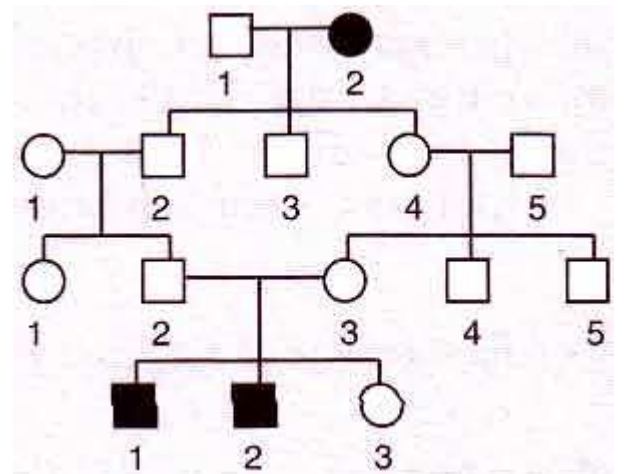
II – 3 is Bb because they are affected but the mother is bb so II – 3 can not be homozygous

III – 6 is Bb because they are affected but the mother is bb so III – 6 can not be homozygous

IV – 5 is Bb because they are affected but the father is bb so IV – 6 can not be homozygous

- Are any of the individuals carriers of the allele for this disorder? Explain why or why not.
 - there are no carriers for this disorder. Because it is caused by a dominant allele, if the individual has one copy of the dominant allele they will be affected by the disease

3. Study the pedigree shown to the right, then answer the following questions:
- Is this disorder caused by a dominant or recessive allele? How do you know?
 - caused by a recessive allele because it “skipped” two generations. Individuals IV – 1 and IV – 2 are affected but neither parent has the disorder
 - Is this disorder X-linked?
 - no, this disorder can not be X-linked. If it was X-linked, then individuals II – 2 and II – 3 would both be affected because they would have received the recessive allele from their mother
 - What are the genotypes of individuals (use alleles C/c)



I – 2 must be cc because she has the disorder

II – 1 is CC or Cc, we don't know if it is she is a carrier

II – 2 is Cc, his mother is cc so he must get one recessive allele from her

III – 3 must be Cc because she produced two affected sons

IV – 2 must be cc because he has the disorder

IV – 3 is CC or Cc, both parents are Cc so there is a 25% chance she is CC and 50% chance she is Cc

- Is it possible to know the genotype of individual I – 1 from this pedigree? Why or why not?
 - no, it is impossible to know if I – 1 is a CC or Cc. He and his wife did not produce any affected children (cc) but this could either be due to him being CC or just random chance producing all Cc children
- Identify two carriers of the allele for this disorder:
 - II – 2, II – 3 and II – 4 must be carriers because their mother is affected (cc) and they each got one allele from her
 - III – 2 and III – 3 both must be carriers because they produced affected offspring.
- What is the relationship between the following individuals:

I – 1 is III – 3's <u>grandfather</u>	II – 4 is II – 5's <u>wife</u>
IV – 2 is III – 4's <u>nephew</u>	III – 2 is III – 4's <u>first cousin</u>

Answers to Genetics Practice Problems:

- Genotypes of offspring: $2 Nn : 2 nn$ Phenotypes of offspring: 2 normal colouring : 2 albino
- Genotypes of offspring: $1 RrPp : 1 Rrpp : 1 rrPp : 1 rrpp$ (or 4 : 4 : 4 : 4)
Phenotypes of offspring: 1 red, plain : 1 red, checkered : 1 grey, plain : 1 grey, checkered
- Genotypes of offspring: $2 BB' : 2 B'B'$ Phenotypes of offspring: 2 brown hair : 2 blonde
- Genotypes of offspring: all Rr Phenotypes of offspring: all rose combed
- Genotypes of offspring: $50\% TtRr : 50\% Ttrr$
 - half of the offspring will have tall vines and yellow fruit (Ttrr)
 - there will be no offspring with dwarf vines because all offspring have the dominant allele for Tall
- What are the sexes and eye colors of flies with the following genotypes?

$X^R X^r$	female red eyes	$X^R Y$	male red eyes	$X^r X^r$	female white eyes
$X^R X^R$	female red eyes	$X^r Y$	male white eyes		
- What are the genotypes of these fruit flies:

white eyed, male	$X^r Y$	red eyed female (heterozygous)	$X^R X^r$
white eyed, female	$X^r X^r$	red eyed, male	$X^R Y$
- The cross is $X^r X^r \times X^R Y$. Genotypes of offspring: $50\% X^R X^r : 50\% X^r Y$
Phenotypes of offspring: 50% red eyed females : 50% white eyed male

7. Human blood types exhibit both co-dominance and multiple alleles. Complete the chart below:

Blood Type	Possible Genotype(s)	Type of Immunoglobulins on their RBCs	Will make antibodies to	Can Donate Blood to	Can Receive Blood from
A	$I^A I^A$ or $I^A i^O$	A	B	A or AB	A or O
B	$I^B I^B$ or $I^B i^O$	B	A	B or AB	B or O
AB	$I^A I^B$	A and B	none	AB only	anyone
O	$i^O i^O$	none	A and B	anyone	O only

- It is impossible for $I^A i^O \times I^B I^B$ to produce a type O child, type O is $i^O i^O$ but the mother has only I^B alleles
- The cross is $I^A i^O Rh+Rh- \times i^O i^O Rh-Rh-$ so there is a 25% chance of a child being O+ ($i^O i^O Rh+Rh-$)
- The cross is $I^A I^B \times i^O i^O$ so the possible genotypes of the children are $I^A i^O$ or $I^B i^O$
The children's blood types are 50% A heterozygous : 50% B heterozygous
- Let H be normal hemoglobin, so h is sickle hemoglobin
The cross is Hh x Hh The genotypes of the offspring are $1 HH : 2 Hh : 1 hh$
 - There is a 50% chance that the children will be carriers of the sickle gene (Hh)
 - There is a 25% chance that a child will be HH and experience severe symptoms
- The cross is $X^H Y \times X^H X^h$ The genotypes of the children are $1 X^H X^H : 1 X^H X^h : 1 X^H Y : 1 X^h Y$
 - 25% chance that they will have a daughter with the disease (or 50% of their girls will have it)
 - 25% chance that they will have a daughter who is a carrier (or 50% of their girls will be carriers)
 - 25% chance that they will have a son with the disease (or 50% of their sons will have it)

13. The cross is $X^H Y \times X^h X^h$ The genotypes of the children are $2 X^H X^h : 2 X^h Y$
 50% of their children may have hemophilia (or 100% of the boys)
- Because the girls will get a normal H gene from their father and a hemophilia (h) gene from their mother, all of the girls will be carriers but not develop the disease
 - Because the sons get the Y chromosome from their father, they will all get a hemophilia (h) gene from their mother so all of the sons will develop the disease. They do not have the protection of a second X^H
14. The cross is $X^N Y \times X^N X^n$ (N for normal)
- The genotypes of the children are $1 X^N X^N : 1 X^N X^n : 1 X^N Y : 1 X^n Y$
 - There is a 50% chance that a son will be affected (half of the boys will be $X^n Y$)
 - There is a 50% chance that their daughter will be affected (half of the girls will be $X^N X^n$)
15. The F1 offspring are all heterozygous and show a blended trait (red + yellow = orange)
 The F2 generation ($RR' \times RR'$) will produce offspring with genotypes $1 RR : 2 RR' : 1 R'R'$
 The F2 phenotypes will be 1 red pepper : 2 orange peppers : 1 yellow pepper
16. Because the male could be BB or Bb, the breeder should do a test cross and breed him to a homozygous recessive female (bb). If any of their puppies have brown spots, then the male is heterozygous (Bb). If the puppies are all black spotted, the male is most likely BB.
17. The cross is $RR \times RW$. The genotypes of the offspring are $2 RR : 2 RW$
 The phenotypes of the offspring are 50% red : 50% roan (both red and white)