Human Blood Types: Codominance and Multiple Alleles

Codominance: both alleles in the heterozygous genotype express themselves fully

Multiple alleles: three or more alleles for a trait are found in the population

Blood Type Alleles:

- I^A – produces antigen A
- I^B – produces antigen B
- i – produces no antigens

Note: replace "produces" with "allele for" to get the key for all blood types problems.

I^A and I^B are not dominant over each other. The allele i is recessive to I^A and I^B.

Genotype	**Phenotype**
I^AI^A or I^Ai	Type A blood
I^BI^B or I^Bi	Type B blood
I^AI^B	Type AB blood
ii	Type O blood

Blood Types:

If you are Type A, you have antigen A on your red blood cells. You also have antibody B in your blood plasma.

Antigen: a type of carbohydrate attached to RBC's
Antibody: part of your body's defense system
→ antibody B "attacks" antigen B

If you are Type B, you have antigen ___B___ on your RBC's and antibody __A__ in your blood plasma.

If you are Type AB, you have antigen __A___ and antigen __B__. (That's codominance – both I^A and I^B are expressed.) You have no antibodies if you're type AB. Why?

If you are Type O, you have no antigens on your RBC's, and you have antibodies A and B.

<u>NOTE: Only the antigens are donated, not the antibodies.</u>

TYPE	Can Donate To:	Can Receive From:
A	A, AB	O, A
B	B, AB	O, B
AB	AB	O, A, B, AB
O	O, A, B, AB	O

Type AB is called the universal recipient, and Type O is called the universal donor. Why?

Reference Chart: Antigens and Antibodies

Antigen A	Antibody A	Antigen B	Antibody B
Type A	Type B	Type B	Type A
Type AB	Type O	Type AB	Type O

Blood Type Problems

1. **What's the probability of a woman with type AB blood and a man with type O blood having a child with type A blood?**

2. **Cross a man heterozygous for type A blood with a woman with type AB blood. Give the genotypic and phenotypic ratios of their offspring.**

3. **A man with type A blood and a woman with type B blood have a child with type O blood. Give the genotypes of the parents and the child and show a Punnett square for the cross.**

Incomplete Dominance

Incomplete dominance:
genetic inheritance in which neither allele is completely dominant or recessive; the heterozygous condition produces a phenotype that is intermediate between the alleles
Example: In snapdragons, there are 2 alleles for flower color: C_1 is the allele for red flowers and C_2 is the allele for white flowers.

P homozygous red X homozygous white
$C_1 C_1$ $C_2 C_2$

F1

	C_1	C_1
C_2	$C_1 C_2$	$C_1 C_2$
C_2	$C_1 C_2$	$C_1 C_2$

KEY:
C_1 -- red
C_2 -- white

All F1 offspring are ___$C_1 C_2$ __ which gives the phenotype of PINK flowers. (Note: snapdragons & four o'clock flowers use the same alleles).
Another example of incomplete dominance is sickle cell anemia in humans. Homozygous recessive produces the disease, homozygous dominant has no disease. If you have the disease, your red blood cells are sickle shaped under low oxygen conditions and can't carry oxygen very well. They also clog up blood vessels, cutting off the blood supply to different areas of the body.

People with the heterozygous ($Hb^S Hb^A$) genotype for sickle cell anemia do NOT have the disease. Only a few of their RBC's are sickle shaped and the normal cells carry enough oxygen. However, heterozygotes have an advantage – they are immune to the disease malaria, which is spread by mosquitoes and is often fatal if not treated properly.

Sickle-Cell Anemia and Hemoglobin
Incomplete Dominance in terms of Red Blood Cell Shape
Codominance in terms of Hemoglobin

Organismic Phenotype	Genotype	Hemoglobin Types Present
Sickle-Cell Trait (few RBC's sickle shaped but only under low oxygen - since this is an intermediate, RBC shape in sickle cell is an example of incomplete dominance)	**Hb^SHb^A** **(heterozygous)**	**S and A** Since both forms of hemoglobin are present, in terms of hemoglobin, sickle cell is actually an example of codominance.
Sickle-Cell Anemia (RBC's are sickle shaped)	**Hb^SHb^S**	**S**
Normal (RBC's never sickle shaped)	**Hb^AHb^A**	**A**

Remember: Sickle-cell *trait* and sickle-cell *anemia* are NOT the same thing.

Sickle-cell *trait* has a few red blood cells that become sickle shaped but only under low oxygen levels. However, people with sickle cell trait (heterozygous) are also immune to malaria. This is a huge advantage for populations existing where malaria is rampant. It is also the reason why sickle cell *anemia* is still common in these same populations. In some areas of West Africa, as much as 40% of the population is heterozygous (sickle-cell trait). It would be a disadvantage to be heterozygous in areas of the world where malaria is not a threat, since being heterozygous introduces the possibility of passing on sickle-cell *anemia* to children and future generations.

Having sickle-cell anemia affects the general health of a person in a lot of ways. For a flow chart illustrating these devastating effects, examine the other side of this paper.

It is important to note that while sickle-cell trait and sickle-cell anemia are extremely rare in Caucasian populations, it is not impossible for a Caucasian person to have either of these.

Sex-Linked Traits

Sex-linked traits:
traits that are controlled by genes located on the X chromosome

Our understanding of sex-linked traits came from studying the fruit fly. Sex is determined the same way in fruit flies as it is in humans → _____XX_____ = female _____XY______ = male
___Thomas Hunt Morgan_______ : year: ___1909____ : While studying fruit flies, he noticed a male fly with white eyes (red eyes is the normal phenotype). He crossed this male with a red eyed female. All of the F1 had red eyes, showing that ___red_______ eyes were dominant. So far, the cross fit the normal pattern.

P red eyed female X white eyed male
_____RR_____ _____rr______

KEY:
R = red eyes
r = white eyes

F1

	R	R
r	Rr	Rr
r	Rr	Rr

Note: This cross later turned out to be incorrect!!

All F1 flies have the genotype _______Rr______ and the phenotype _________red eyes________.
Morgan crossed the F1, expecting the 3:1 ratio that a monohybrid cross would normally give
(3 ____red eyed_________ : 1 ________white eyed________). He got that ratio, but with a twist. All of the white eyed flies were males! Morgan explained that his results were from the gene for eye color being located on the ____X___ chromosome. If you look at his complete experiment through the F2, you can see how it works out. With sex-linked traits, we also keep track of the sex chromosome in addition to the gene for the trait. Check out the revised key.

Key:
X^R -- gene for red eyes
X^r -- gene for white eyes
Y -- males, no gene for trait

P red eyed female X white eyed male
$X^R X^R$ $X^r Y$

F1

	X^R	X^R
X^r	$X^R X^r$	$X^R X^r$
Y	$X^R Y$	$X^R Y$

F1 are all ___red eyed_____ for phenotype, with the genotypes ____$X^R Y$____(Male) or ____$X^R X^r$____ (Female).
(Remember, to get the F2, cross the male and female F1.)

red eyed male X red eyed female
____$X^R Y$___ ____$X^R X^r$______

F2

	X^R	Y
X^R	$X^R X^R$	$X^R Y$
X^r	$X^R X^r$	$X^r Y$

# of offspring	Genotypes	Phenotype	Sex
1	$X^R X^R$	red eyes	female
1	$X^R Y$	red eyes	male
1	$X^R X^r$	red eyes	female
1	$X^r Y$	white eyes	male

In sex-linked traits, there is no corresponding gene on the Y chromosome in males, so whatever gene is on the X chromosome is expressed, whether it is dominant or recessive.

The recessive condition of sex-linked traits is mostly found in males (although it is still possible to have a female with the recessive condition). Why is this true?

Male only need one recessive allele for the recessive form to be expressed. Females must have TWO recessive alleles.

There are several sex-linked genes in humans. Colorblindness, Duchenne Muscular Dystrophy, and Hemophilia (a disease in which a person's blood has extreme difficulty in clotting) are a few of them. The genes for these diseases are recessive. However, the symbols are different. For hemophilia, we use the symbols X^H -- normal clotting and X^h – hemophilia.

Cross a man with hemophilia with a woman who is heterozygous. Give the genotypes and phenotypes of the possible children, including the sex of each possibility.

P	**hemophilia**	*	**no disease**	**KEY:**
	X^h Y		**X^H X^h**	**X^H – no disease**
				X^h - hemophilia
				Y – no gene

F1

	X^h	Y
X^H	$X^H X^h$	X^H Y
X^h	$X^h X^h$	X^h Y

Geno.	Pheno.	Sex
$X^H X^h$	no disease	female
X^H Y	no disease	male
$X^h X^h$	hemophilia	female
X^h Y	hemophilia	male

Sex – Linked Traits: Problem Keys for Common Diseases

Hemophilia:

X^H – no disease
X^h – hemophilia
Y – no gene

Duchenne Muscular Dystrophy:

X^D - no disease
X^d – DMD
Y – no gene

Colorblindness:

X^C – no disease → **Please make sure there is a physical difference between your capital "C" and your lowercase "c" !!!**
X^c – colorblind
Y – no gene

Hypophosphatemia: → **Note that the allele for this disease is is <u>dominant</u>, yet the alleles for all of the other diseases listed above are recessive!**

X^H – hypophosphatemia
X^h – no disease
Y – no gene

Don't forget eye color in fruit flies:

X^R – red eyes
X^r – white eyes
Y – no gene

Sex-Limited Trait

Sex-limited traits:
<u>Traits that are only expressed in the presence of sex hormones and are only observed in one sex or the other</u>

Sex-limited traits are controlled by genes located in the autosomes (non-sex chromosomes). Although both males and females carry these genes, they are only expressed in one sex. Because you need large amounts of the proper sex hormone for the genes to be expressed, most sex-limited traits are not expressed in children. Examples: In many birds, male plumage is more colorful than female plumage. In humans, beard growth in men is a sex-limited trait as well as milk production in women.

GENE + HORMONE = SEX LIMITED TRAIT

Sex-Influenced Traits

Sex-influenced traits:
<u>Traits expressed in both sexes, but they are expressed differently</u>

Example: Baldness

1. **In the presence of male sex hormones, the allele for baldness is dominant.**
2. **Female sex hormones cause the allele to be recessive. A woman may lose hair if her genotype is <u>homozygous recessive</u>.**

Polygenic Traits

Polygenic trait:
<u>Trait that is controlled by more than one gene</u>

→ polygenic traits are especially prevalent in genes that control body __shape_____ or __form___.
Inheritance of polygenic traits can be very complicated.

Example: parakeet color → see handout

Some human traits that are considered polygenic:

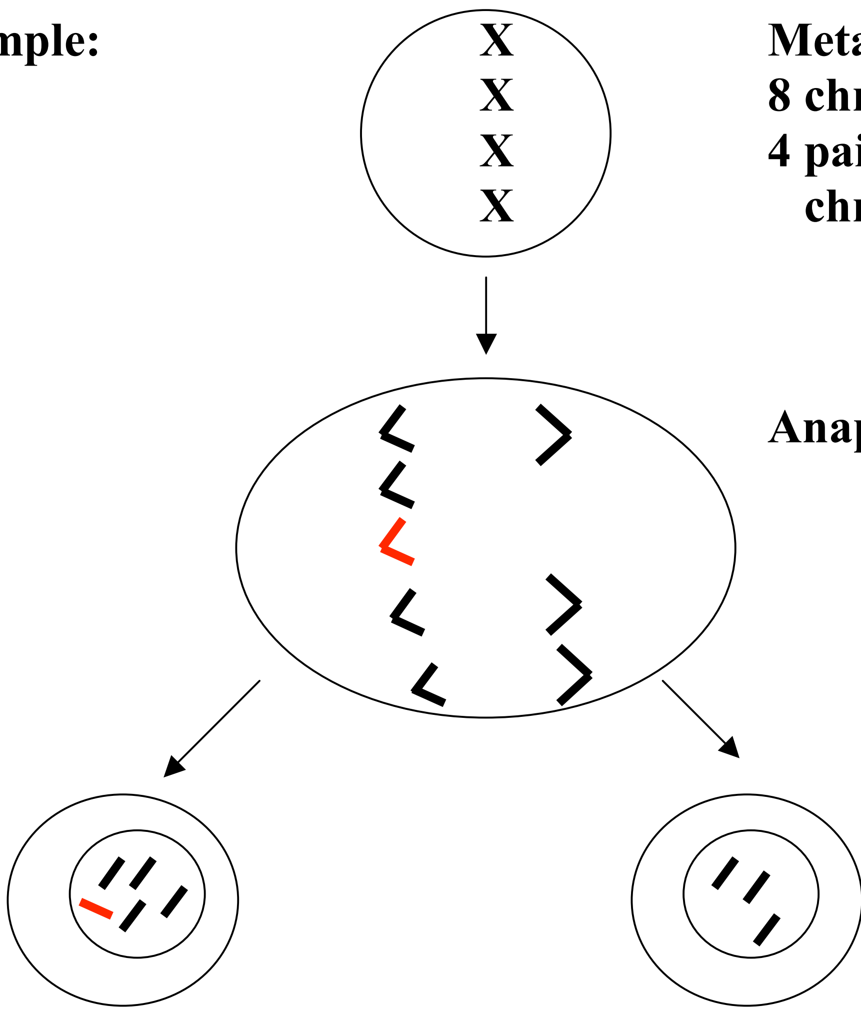
- a. **skin color (numerous genes control the amount of melanin in the skin)**
- b. **height**

Nondisjunction

Nondisjunction: <u>"not coming apart"</u>

Recall from meiosis that during anaphase, the chomosomes are pulled apart to opposite ends of the cell. If this doesn't happen properly during oogenesis or spermatogenesis, the egg or sperm will end up with too few or too many chromosomes.

Example:



Metaphase II
8 chromosomes
4 pairs of homologous chromosomes

Anaphase II

5 chromosomes (vs. normal 4)

3 chromosomes (vs. normal 4)

Many human genetic disorders are a result of nondisjunction.

1. **Down's Syndrome: trisomy 21 → there are ____3____ copies of chromosome number 21 instead of the normal ____2___ copies. Some of the characteristics of people with Down's syndrome include:**
   a. **reduced mental capacity (varies greatly between individuals)**
   b. **lack of muscle tension ("floppy" appearance, tongue often lolls)**
   c. **ears are placed lower on the head (often problems with ear infections as infants)**
   d. **larger head and facial features are closer together**
   e. **life expectancy is greatly reduced (late twenties to early thirties was lifespan; now closer to 40's, 50's)**
   f. **often sterile, but not always**

2. **Turner Syndrome – "XO" – a sperm or egg is produced without a sex chromosome; the O indicates a missing sex chromosome**

   →people with Turner's syndrome are females; the sex organs do not fully develop and therefore they are sterile; lack secondary sex characteristics

3. **Klinefelter Syndrome – "XXY" – an extra X is present from either the sperm or the egg**

   →people with Klinefelter syndrome are usually male, may have reduced mental capacity, and are often sterile; lack secondary sex characteristics

Notes on XYY Syndrome: (aka __Richard Speck Syndrome__)
sex: _male_ characteristics: tall; lanky; severe adult acne; aggressive/violent tendencies ___
→ a study done on XYY syndrome showed a proportionately larger population of XYY males in prison than in the general population

Environmental Effects on Gene Expression

Gene expression can be affected by both the external environment and the internal environment inside an organism. Phenotype is generally a combination of genetic and environmental influences.

Example: Himalayan rabbits (temperature)

1. **The rabbits are normally covered with _white_ fur.**
2. **However, its _ears_, nose, and _feet_ are _black._**
3. **This pattern occurs because most of the rabbit's body is generally warmer than its extremities.**
4. **_Body temperature_ affects the expression of genes that code for fur color.**
5. **When researchers remove a patch of hair from the body and cool the skin as new fur grows, the new fur is _____black______. When they shave hair from the feet or ears and keep those areas warm, the fur that grows back is ____white_________.**

Example: Japanese goby fish (social environment)

1. **The goby fish can change its ___sex______ back and forth in response to changes in its social environment.**
2. **Goby fish exist in schools of many females and only a few males.**
3. **If a large male goby leaves a population, a female goby will ____become male_____.**
4. **If another large male enters that goby population, this new male turns back into a female.**

Example: Human height (internal environment)

- → **While human height is a ____polygenic________ trait, it is also affected by the nutrients in your diet.**