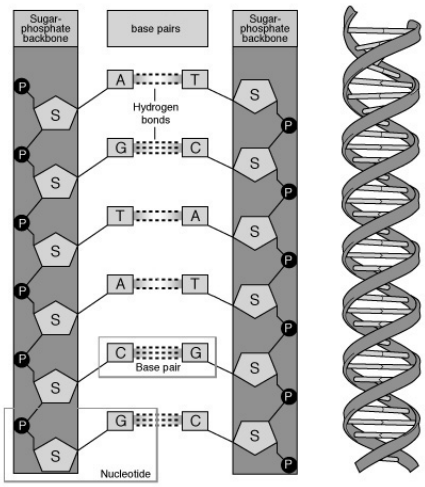
SCIENCE 10 Unit 1- GENETICS Review

Part I- DNA and Protein Synthesis

- the **nucleus** contains a master set of instructions that determines what each cell will become, how it will function, and how long it will live before being replaced
  - these instructions are carried in **chromosomes**—a thread like structure made mostly of DNA
- every plant and animal species has a specific number of chromosomes in the nucleus of each cell
  - humans have **46** chromosomes (there are 23 types—1 from each parent)
- in the cells of most plants and animals, chromosomes come in **pairs**—one of each pair comes from each parent when an egg and sperm unite
  - in humans, 23 from the mother, 23 from the father
- chromosomes are made of a material called DNA (**deoxyribonucleic acid)**
  - each chromosome consists of a single molecule of DNA
- DNA is the material found in the cell nucleus that contains genetic information
- DNA is divided into segments, called **genes**
  - a gene is a relatively small part of a DNA molecule—each DNA molecule consists of hundreds of thousands of genes
  - genes are segments of DNA that control **protein production** (the type of protein, and when they're made), thus controlling the cell's activities and structure
  - proteins are important as building blocks of cells, cell messengers, and catalysts for chemical reactions (= enzymes- they speed up chemical reactions)

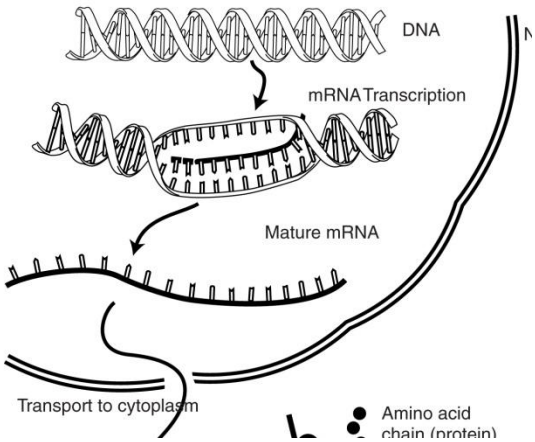


- each rung of a DNA molecule, along with the piece of the ladder's side to which the rung is attached, is a building block molecule
- there are 4 types of building block molecules:
  - **A (adenine), C (cytosine), G (guanine), and T (thymine)**
  - the order in which the A, C, G, T building blocks are strung together, is called the **genetic code** (building blocks for DNA)
  - ***A binds to T*** and ***C binds to G*** (apple tree, car garage)
  - the genetic code is different for every individual (except identical twins)
- DNA controls many features, like hair and eye colour, and your ability to digest certain foods (lactose intolerant) through genes, which control the type of proteins your cells can make
- proteins are made through the process called **PROTEIN SYNTHESIS** and has 2 steps:

1. Transcription- occurs in the nucleus

- DNA unzips
- At the start codon of one strand of DNA, a single strand of messenger RNA (mRNA) is copied
- RNA bases are different. They are:
- **A (adenine), C (cytosine), G (guanine), and U (Uracil)**
- Paring rule as follows:

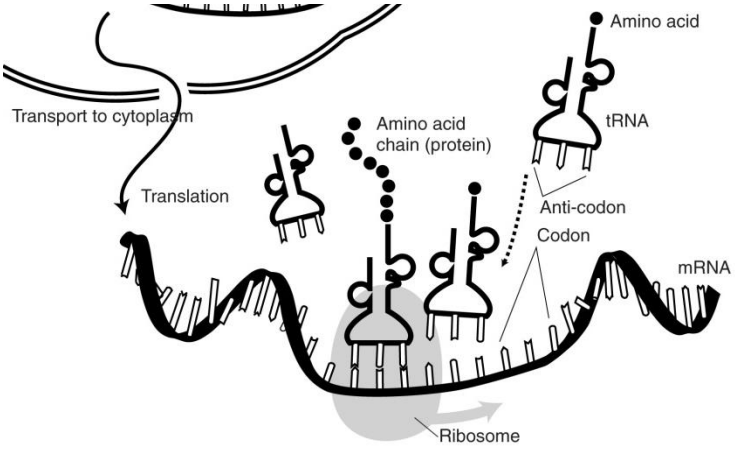
DNA	RNA
A	U
T	A
C	G
G	C

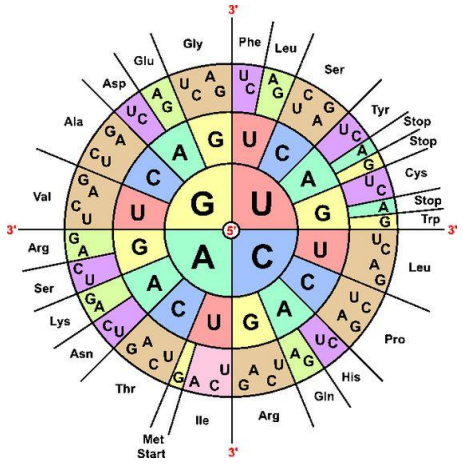


- mRNA leaves the nucleus and goes to the ribosome (=rRNA)

2. Translation- occurs at the ribosome in the cytoplasm

- the ribosome attaches to mRNA and starts to read the code
- transfer RNA (tRNA) brings in amino acids and chains them together
- the type of amino acid depends on the code- it is in order!
- the long chain of amino acids then goes to the Golgi body where it is packaged and shaped into a protein

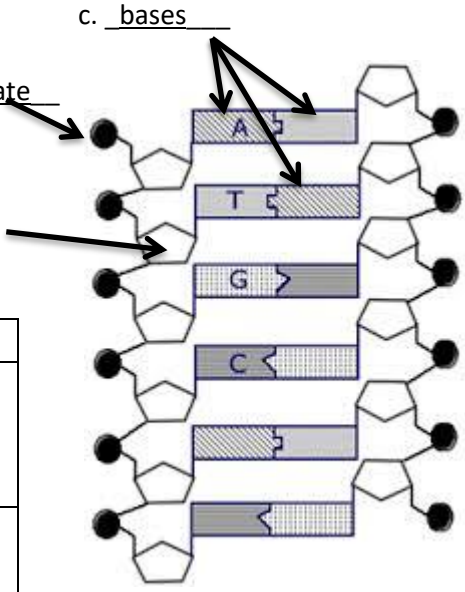




Key terms you should know:

Term	Definition
Deoxyribonucleic acid	
nucleotide	
Sugar	
Base	
Protein synthesis	
Start codon	
Transcription	
mRNA	
translation	
tRNA	
Ribosome	
Amino acid	
Protein	
gene	
chromosome	

Part I- DNA and Protein Synthesis Review Questions



1. What is the full name for DNA? Label the diagram to the right.

Deoxyribonucleic acid

2. How is DNA similar/different from RNA?

	DNA	RNA
Similar	*Sugar/phosphate backbone, 4 bases* *Adenine, Guanine, Cytosine,*	
Different	*Double helix* *Thymine*	*Single strand* *Uracil*

3. Why is DNA called the "blueprint of life"?

It holds the instructions for controlling the cell, when to divide, when to grow, what to do, and when to die.

4. Give 3 important functions for proteins.

#1	*building blocks of cells*
#2	*cell messangers*
#3	*catalysts for chemical reactions*

4. What is the genetic code of DNA? Give the full names for each base.

The genetic code consists of 4 bases- adenine (A), guanine (G), thymine (T) and cytosine (C)

5. What are the base pairing rules for DNA?

A binds with _T_ **C** binds with _G_

6. Give the matching DNA strand for each of the following:

TAC AAA CTT	TAC CAT TAA	TAC AAA TAT
ATG TTT GAA	ATG GTA ATT	ATG TTT ATA

7. What is protein synthesis, and what does it have to do with genes?

Protein synthesis is the making of proteins. Genes give the instructions on how to make a particular protein, such as skin pigment.

8. How are transcription and translation different?

Transcription happens in the nucleus and involves making mRNA from an unzipped strand of DNA

Translation happens in the cytoplasm at a ribosome and makes an amino acid chain using tRNA and mRNA

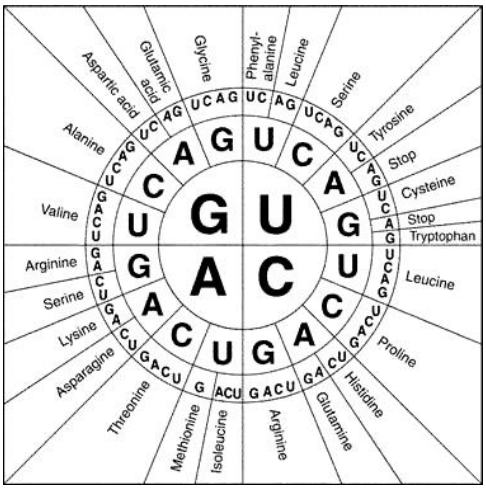
9. What would happen if a desired segment of DNA that needed to be copied did not have a start codon?
Without a stop codon, no transcription would occur. No protein would be made.

10. Describe the function of each:

a. mRNA – *messenger that carries gene instructions to the ribosome*

b. tRNA- *transfers an amino acid to the ribosome to make protein*

c. ribosome (rRNA)- *the ribosome, reads the mRNA and facilitates specific tRNA to come in and start building the amino acid chain*



11. Give the amino acid sequence for the following:

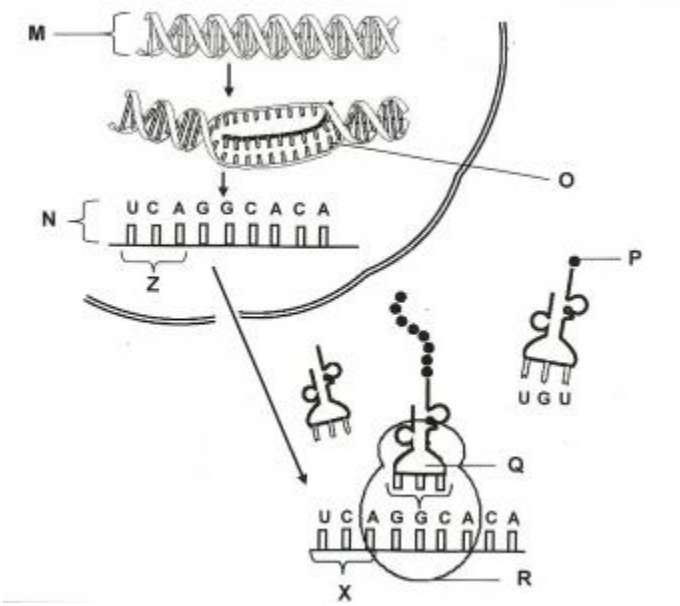
TAC AAA CTT CAT

AUG UUU GAA GUA
Start-Phen-Glu-Val

TAC AAA TAT TTT

AUG UUU AUA AAA
Start-Phen-Iso-Lys

12. Label each part below and describe what is happening at each step.

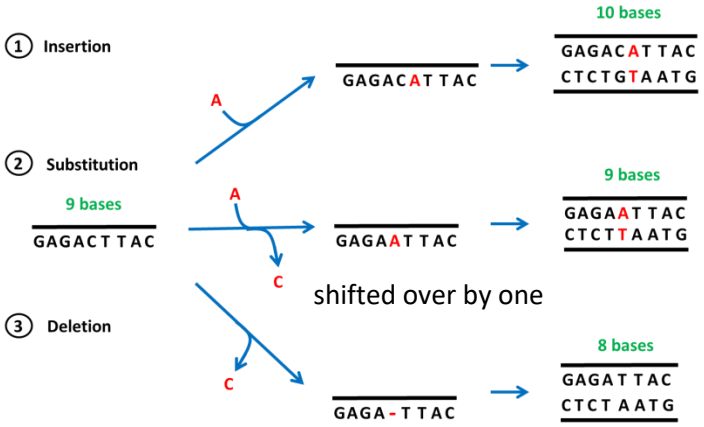


Letter	Name	Function
M	*DNA*	*Control center of the cell, holds all instructions for all proteins*
N	*mRNA*	*Holds instructions for making one particular protein and carries it from the DNA to the ribosome*
O	*Gene*	*Region of DNA being copied to make a particular protein*
P	*Amino Acid*	*Building block of proteins*
Q	*tRNA*	*Transfers amino acids to the ribosome for the making of protein*
R	*Ribosome*	*Reads the instructions of mRNA and facilitates the incoming tRNA to build an amino acid chain (protein)*

Part II- MUTATIONS AND GENETIC ENGINEERING

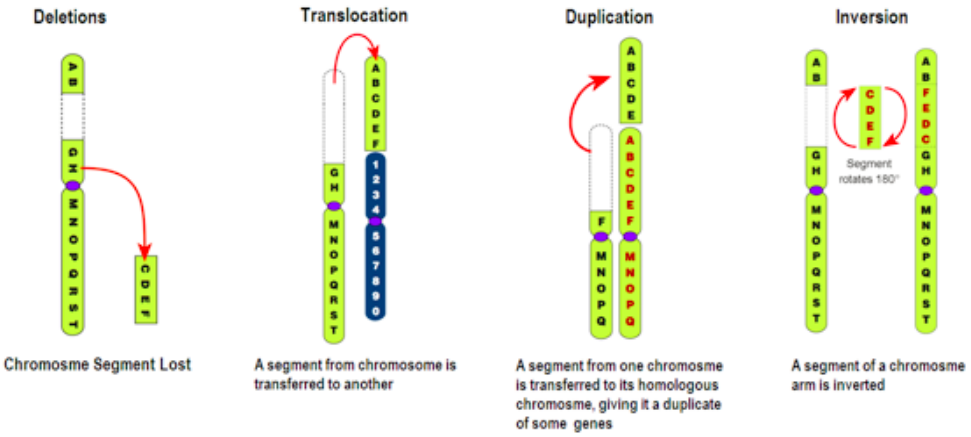
MUTATIONS

- a **mutation** is the change in the DNA of an organism and may alter how well the protein can do its job
- because genes code for specific proteins based on the order of the DNA building blocks, a change in the order of A, T, C, G is known as **gene mutation**



- there are 3 types of gene mutations
  - **insertion**- a base is added
  - **deletion**- a base is removed
  - **substitution**- a base is replaced by a different base
  - addition and/or deletion can cause a **FRAMESHIFT** where the genetic code is base- this can result in a totally non-functional protein

- **Chromosomal mutations** involve modifying the chromosome. The main types include **deletion**, **translocation**, **duplication**, and **inversion**.



- mutations are caused by **mutagens**—substances/factors that cause a DNA mutation (damages DNA)
  - example: sun's UV rays, x-ray, chemicals (mercury), cigarette smoke, hotdogs
  - mutagens that cause cancer are known as **carcinogens**
  - **cancer** occurs when several mutations "break" the cell cycle, causing cells to reproduce out of control. A mass of cancer cells is called a **tumour**.
- **POSITIVE MUTATIONS**- are beneficial and increase survivability- ex. rabbits with longer ears, can hear better and avoid predators
- **NEGATIVE MUTATIONS**- are harmful and decrease survivability - ex. rabbits without ears, can't hear predators and are more likely to get eaten
- **NEUTRAL MUTATIONS** may cause a change but have no effect on survivability- ex. white spirit bears

PLANT AND ANIMAL BREEDING AND GENETIC ENGINEERING

- **Natural Selection**- the process in which traits of an organism are selected for/against by "nature"- giving them a mating or survival advantage. A slow process resulting in an organism well adapted to its environment.

- **Artificial Selection**- the process in which traits of an organism are selected for/against by humans. A fast process resulting in an organism suited to human need- not necessarily adapted to its environment.

- Scientists have developed the technology to manipulate genes. This is known as **genetic engineering.**
- **Transgenic organisms** are organisms whose genetic information has been altered with the insertion of genes from another species
- The steps of genetic engineering are:
  1. Identifying and isolating the desired gene and cutting it out
     -**restrictive enzymes** are used to cut the DNA at specific spots
     -the cut out DNA segment has **"sticky ends"** and will stick to other segments of DNA
  2. Putting the desired gene into a **vector**- a virus that will carry the desired gene to the desired location
  3. Infecting the target genome (cell) with the vector and allowing the vector to insert the gene into the target genome
- There are many uses for genetic engineering:
  - Grow human hormones, antibiotics, and antigens in transgenic bacteria
  - Make artificial sweeteners
  - Study human disease by inserting human DNA into mice
  - **Gene therapy**- correct a genetic disorder in a human by inserting a "healthy" gene
  - Create plants and animals that are resistant to disease/insect attack
  - Improve nutrition and extend ripening time of fruit and vegetables
  - Assist in human reproduction (fix "broken" genes in sperm/egg) and human organ transplant
- There are also some unknowns and risks for genetic engineering
  - The long term health risks are not known- although it is unlikely to be problems
  - Introduced genes may "escape" into the environment- ex. Cross pollination with non-GMO'd plants
  - Pesticide resistant crops may cause excessive pesticide use
  - People with allergies may inadvertently be exposed ex. Eat a tomato with a peanut gene
- **cloning** is the process of creating identical genetic copies of an organism
- The steps of cloning are:
  1. Remove an egg cell from a female egg cell donor. Remove DNA from the egg.
  2. Remove a somatic cell from a somatic cell donor. Remove DNA from the somatic cell.
  3. Insert somatic cell DNA into the egg
  4. Insert the egg into a surrogate mother. Allow egg to develop.

Key terms you should know:

Term	Definition
Gene mutation	
Insertion	
Deletion	
Substitution	
frameshift	
Chromosomal mutation	
Deletion	

Term	Definition
Translocation	
Duplication	
inversion	
Mutagen	
Carcinogen	
Cancer	
tumour	
Positive mutation	
Negative mutation	
Neutral mutation	
Natural selection	
Artificial selection	
Genetic engineering	
Transgenic organism	
Restrictive enzyme	
Sticky end	
Vector	
cloning	

Part II- MUTATIONS AND GENETIC ENGINEERING Questions

1. List the 3 types of gene mutations and give an example of each.

	Type of Mutation	Example
#1	*insertion*	*An extra "A" is inserted into a DNA sequence* *CAT TAA → CAT ATA A*
#2	*deletion*	*An "A" is removed from a DNA sequence* *CAT TAA → CAT TA*
#3	*substitution*	*An "A" is replaced with a "T" in a DNA sequence* *CAT TAA → CAT TTA*

2. What is a frameshift mutation? Give an example using the code CAT TAA TTT

A frameshift occurs with insertion or deletion gene mutations. This is when an extra base is added or removed, shifting the genetic code over such that a very different set of instructions are made, often resulting in a faulty protein.

Ex. CAT TAA TTT → CAA TTA ATT T

3. What is the difference between a gene mutation and a chromosomal mutation? Which do you think has the potential of being the most harmful?

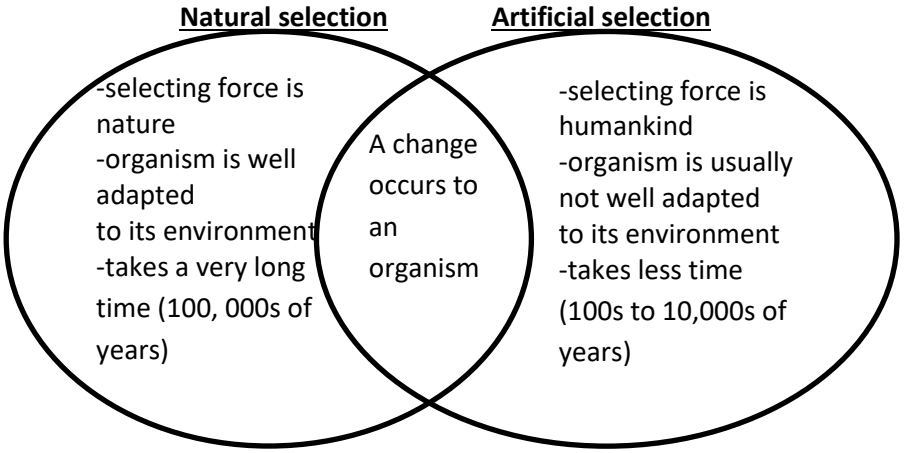
A gene mutation occurs at the gene level and involves one or more bases. A chromosomal mutation is much bigger and occurs with entire segments of chromosome, often having several to many genes on them.

Chromosomal mutations can affect more than one gene and more than one protein, and therefore can be more harmful.

4. How are translocation and duplication chromosomal mutations different?

In translocation, a segment of chromosome is moved to a new region. In duplication, a segment of chromosome is copied one or more times, resulting in more than one copy of genes.

5. Use the Venn Diagram below to compare and contrast artificial and natural selection. Which do you think is "best" for humans? Which is "best" for the species?



It can be argued that natural selection is most beneficial to a species, whereas artificial selection is most beneficial to humans.

6. Describe the steps required to make a transgenic organism.
    1. Identify and isolate the desired gene and cutting it out
        -restrictive enzymes are used to cut the DNA at specific spots
        -the cut out DNA segment has "sticky ends" and will stick to other segments of DNA
    2. Put the desired gene into a vector- a virus that will carry the desired gene to the desired location
    3. Infect the target genome (cell) with the vector and allow the vector to insert the gene into the target genome

7. What is a GMO? Are GMO's necessarily bad?

A GMO is a genetically modified organism that has had its genetic information altered by humans. This could include the insertion of new genes, the deletion of undesirable genes, or the modification of genes. Not all GMO's are necessarily bad, although there are some potential risks if they are not regulated carefully.

8. Explain how genetic engineering may be used in gene therapy to treat certain genetic diseases like cystic fibrosis.

Scientists can genetically engineer a vector to carry a functioning gene and infect a person who has a disease caused by a broken gene. The hope is that the functioning gene will be inserted into the person's genome and the disease will be "fixed".

10. What is a clone? Describe the steps in making a cloned curly haired sheep from a frizzy haired egg donor mother and a curly haired somatic cell donor.

A clone is an exact genetic copy of an individual. It has identical DNA. To make a cloned curly haired sheep, you would first remove the egg from the frizzy haired egg donor and take the DNA out of the egg. You would then insert the DNA from the somatic cell of the curly haired donor and then place the egg in a surrogate mother. The offspring produced from this would be a clone of the curly haired somatic cell donor, since they would have identical DNA.

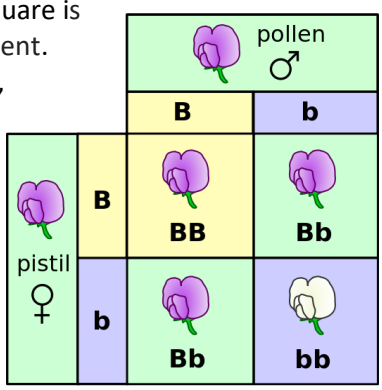
Part III- GENETICS

- Children often resemble parents due to the inheritance of genetic material- genes
- One set of genes is from the mother, one set of genes is from the father
- Gregor Mendel, an Austrian monk who lived in the 1800's, noticed patterns of inheritance in pea plants
- He noticed that certain physical traits were predictable in offspring. Some traits were always passed on to offspring, some appeared to be "hidden".
    - A **dominant trait** is fully expressed in the offspring
        - Can be expressed as AA or A_
    - A **recessive trait** is not expressed in the offspring if a dominant gene is present
        - Is expressed as aa
- The patterns that Mendel observed are due to the expression of **genes**.
- Each gene has an **allele**, or one possible version of the gene, or trait. The alleles for eye colour, for example are blue or brown or green.
- Alleles are designated letters, often capitalized if dominant lowercase if recessive.

- An individual is said to be….
    - **Homozygous Dominant** if they have two dominant alleles, ex. AA
    - **Homozygous Recessive** if they have two recessive alleles, ex. aa
    - **Heterozygous** if they have a dominant and recessive allele, ex. Aa

- Homozygous means "the same", or having matching alleles, and heterozygous means "different", or having alleles that are not the same

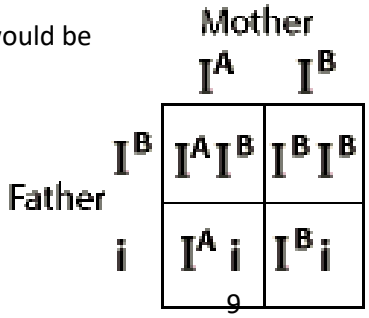
- It is possible to predict the outcome of offspring using a **Punnett Square**. A Punnett square is a diagram that is used to predict an outcome of a particular cross or breeding experiment. For example, if you were to cross a male red flower (Bb) with a female red flower (Bb), you would construct the table to the right.
- This table shows the genotypic ratio and phenotypic ratio of offspring.
    - **Genotypic ratio**- looks at genes ex. 1 BB: 2Bb: 1 bb
    - **Phenotypic ratio**- looks at physical characteristics ex. 3 Red: 1 white
- The **genotype** refers to the genetic makeup of an organism
- The **phenotype** refers to the appearance of a particular physical characteristic



- This table is an example of **complete dominance**, where the presence of a single dominant allele results in phenotypic presentation. If a pea plant is BB or Bb, it will be red. Red is the dominant trait.

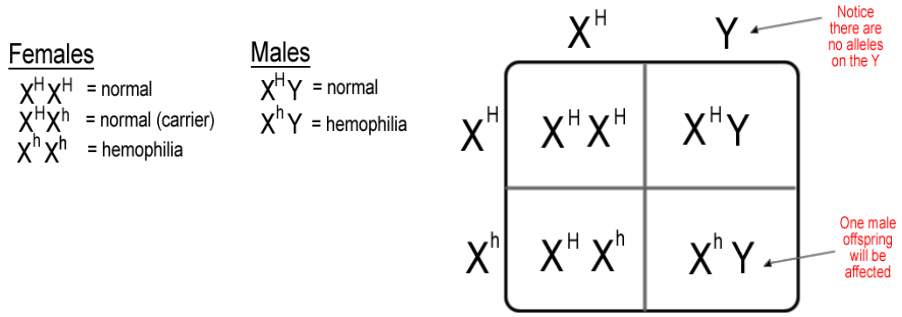
- In **incomplete dominance**, the offspring can express a mixture of the trait. For example, a pea plant that is Bb will be pink (a mixture of red and white). Using the pea plant Punnett square up above…
    - **Genotypic ratio**- 1 BB: 2Bb: 1 bb
    - **Phenotypic ratio**- 1 Red: 2 Pink: 1 white

- In **codominance,** both traits for a characteristic are expressed. An example of this would be human blood type AB.
    - **Genotypic ratio**- 1 I^AI^B: 1 I^BI^B: 1I^Ai: 1I^Bi
    - **Phenotypic ratio**- 1 type AB: 2 type B: 1 type A



- Some traits are **sex-linked**, or found on the sex chromosome X but no the Y. In this case, males are more likely to show recessive traits, as they only have one copy of the gene.
    - Female- XX
    - Male- X (and Y)

- Consider the Hemophilia - this disease is caused by an allele on the X chromosome



- **Genotypic ratio**- 1 X^HX^H: 1 X^HX^h: 1 X^HY: 1X^hY
- **Phenotypic ratio**- 2 healthy females: 1 healthy male: 1 diseases male

Key terms you should know:

Term	Definition
gene	
allele	
Punnett Squares	
Complete dominance	
Incomplete dominance	
Codominance	
Sex-linked	
heterozygote	
homozygote	
phenotype	
genotype	
pedigree	

Genetics Review Sheet

1. Define Genetics and Heredity?

2. What is the difference between the dominant and recessive forms of alleles?
Dominant traits are expressed, whereas recessive traits may be "hidden" if a dominant allele is present.

3. Define the terms homozygous, heterozygous, genotype and phenotype.

4. Draw Punnett squares for the crosses below. State the ratio of the genotype produced.

AA x aa

	A	A
a	Aa	Aa
a	Aa	Aa

All Aa

Rr x Rr

	R	r
R	RR	Rr
r	Rr	rr

1 RR: 2 Rr: 1 rr

TT x Tt

	T	T
T	TT	TT
t	Tt	Tt

2TT: 2 Tt *or* 1TT: 1Tt

5. In humans, the allele that codes for an ability to taste PTC is dominant (T), and the allele that codes for an inability to taste this chemical is recessive (t). A male who is heterozygous for this trait marries a female who cannot taste PTC.
   a. What are the genotypes of the male and female? *Male= Tt Female= tt*
   b. Draw a Punnett square to show the possible genotypes of their offspring.

	T	t
t	Tt	tt
t	Tt	tt

   c. What is the predicted percentage of their offspring that will be able to taste PTC?
   50% will be able to taste PTC

   d. What is the percentage that will not be able to taste PTC? *50% will not be able to taste PTC*

6. Human eye color is inherited as brown eyes are dominant and blue eyes are recessive. Use Punnett squares to solve the following problems. Pick your own letters to represent eye color traits.
   a. A man with blue eyes marries a woman with brown eyes, whose mother had blue eyes. What proportion of the children would be expected to have blue eyes?

Let B- Brown and b- blue

Woman

Man

	B	b
b	Bb	bb
b	Bb	bb

Half the children would have brown eyes and half the children would have blue eyes

b. A brown eyed man marries a blue eyed woman. The first child is blue eyed. What is the man's genotype?

Let B- Brown and b- blue

Woman

Man		b	b
	B	Bb	Bb
	b	bb	bb

The man must be heterozygous dominant (Bb) to be able to produce blue eyed children. He is a carrier.

7. Define and state the difference between complete dominance, co-dominance and incomplete dominance? Use examples of each.

complete dominance	*=If the dominant allele is present, it will be expressed, even if heterozygous*	*AA or Aa= red flower*
co-dominance	*=If one dominant and once recessive allele is present, then both will be expressed*	*Blood type AB, both A and B are expressed*
incomplete dominance	*=If one dominant and once recessive allele is present, then a mixture will be expressed*	*Aa= pink flower (mixture of red(A) and white (a))*

8. Describe the difference between the male and female karyotype (types of chromosomes).
The male has an X and a Y chromosome, whereas the female has 2 X chromosomes.

9. A father is who is ***homozygous dimpled***, and a mother who is ***heterozygous dimpled*** have children.

a) Show the two alleles carried by the father. (Use "D" for dimpled and "d" for smooth) ....._DD_

b) Show the two alleles carried by the mother. (Use "D" for dimpled and "d" for smooth) ....._Dd_

c) Fill in the following Punnett Square showing the cross and show the combinations of genes possible in the children.

Possible gametes from Female Parent

Possible gametes from Male	D	d
D	DD	Dd
D	DD	Dd

d) According to chance, what fraction of their children will have dimples? *4/4 or 100%*

e) What fraction of the children should be ***homozygous smooth***? (if any) *none*

f) What fraction of the children should be ***heterozygous dimpled***? (if any) ½ or 50%

g) What fraction of the children should be ***homozygous dimpled***? (if any) ½ or 50%