Living Environment Name _____

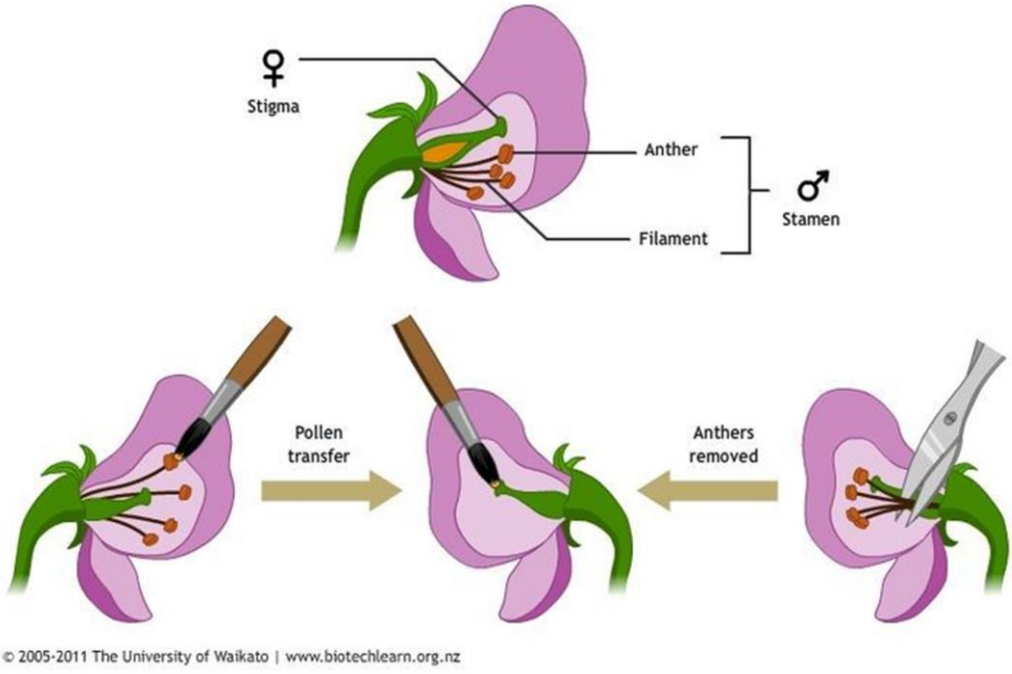
Period _____

Mendelian Genetics

Genetics – scientific study of heredity

Chromosome pairs carry alleles for the same trait. We all have two alleles for each gene. One comes from each parent, one on each member of the homologous pair.

Gregor Mendel was an Austrian monk. He is known as the "father of genetics" because of his early work with pea plants. He used pea plants because the plants reproduce sexually, and produce a fair amount of offspring in a short period of time. The pea flowers contain the male and female gametes. During fertilization these cells join to produce a new cell.



Gamete – sex cell

Sperm – male sex cell

Egg – female sex cell

Fertilization – process in sexual reproduction in which male and female reproductive cells join to form a new cell

Trait – specific characteristic of an individual

Mendel only studied one trait at a time to control variables. He analyzed his data mathematically.

Seed		Flower	Pod		Stem	
Form	Cotyledon	Color	Form	Color	Place	Size
						
Round	Yellow	White	Full	Green	Axial pods	Tall
						
Wrinkled	Green	Violet	Constricted	Yellow	Terminal pods	Short
1	2	3	4	5	6	7

Through his work Mendel concluded that an individual's characteristics are determined by factors that are passed from one parental generation to the next.

Alleles – one of a number of different forms of a gene (represented with capital and lowercase letters)

Mendel's first experiments were monohybrid crosses.

The parent generation of plants were called the P_1 generation. The offspring of that cross are called the first filial F_1 generation.

Mendel observed that each trait is inherited independently. This became Mendel's Principle of Independent Assortment.

Independent Assortment – one of Mendel's principles that states that genes for different traits can segregate independently during the formation of gametes

Purebred – an organism that always produces offspring with the same form of a trait as the parent (the offspring look like the parent)

Segregation – separation of alleles during gamete formation

During gamete formation, the alleles for each gene segregate from each other, so that each gamete carries only one allele for each gene.

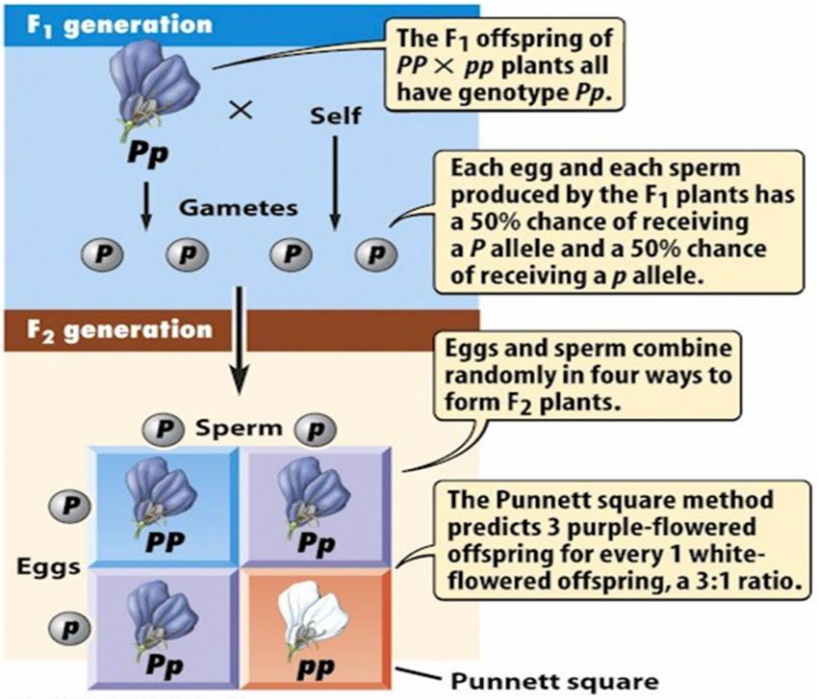


Figure 10-6 Discover Biology 3/e

Homozygous (or purebred) – having two identical alleles for a particular gene (ex. TT, tt)

Heterozygous (or Hybrid) – having two different alleles for a particular gene (ex. Tt)

Phenotype – physical characteristics of an organism (tall plant, short plant)

Genotype – genetic makeup of an organism (allele combination, ex. TT, tt, Tt)

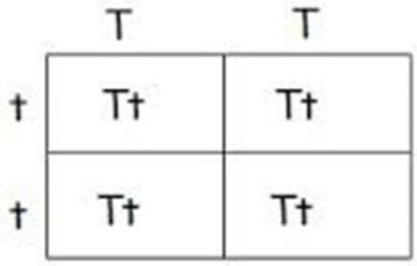
Punnett Square – a diagram that can be used to predict the genotype and phenotype combinations of a genetic cross

Punnett squares show all the possible outcomes of a genetic cross, they can determine the probability of a particular outcome.

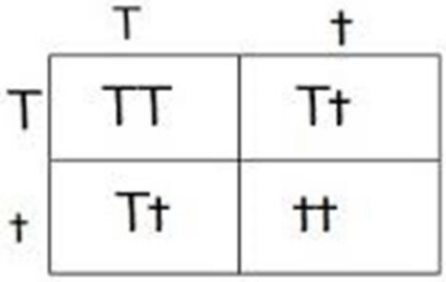
Probability – the likelihood that a particular event will occur.

Mendel's first genetic cross was with purebred tall and purebred short pea plants.

Here is the Punnett Square for that cross:



A homozygous (purebred) tall pea plant was crossed with a homozygous (purebred) short pea plant, these were the P1 generation. All (100%) of the offspring for this cross were heterozygous (hybrid). The shortness phenotype disappeared! All of the pea plants were tall! Mendel then crossed the offspring (of the F1 generation). Here is the Punnett Square for that cross:



Mendel noted that the shortness gene reappeared in the F2 generation. 75% of the offspring were tall, however 25% of the offspring were short. The shortness gene did not disappear, it was just hidden. This is Mendel's Principle of Dominance.

Dominant Allele - an allele that produces the same phenotype whether its paired allele is identical or different (represented with capital letters)

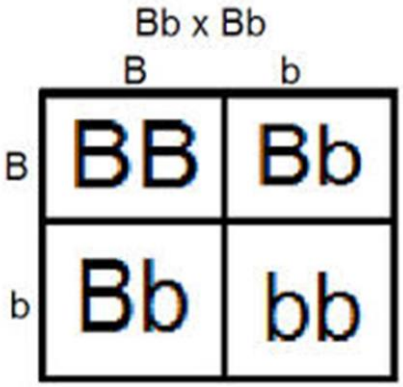
Recessive Allele - an allele that produces its characteristic phenotype only when its paired allele is identical (represented with lowercase letters)

A dominant allele will mask a recessive allele in most cases when it is present.

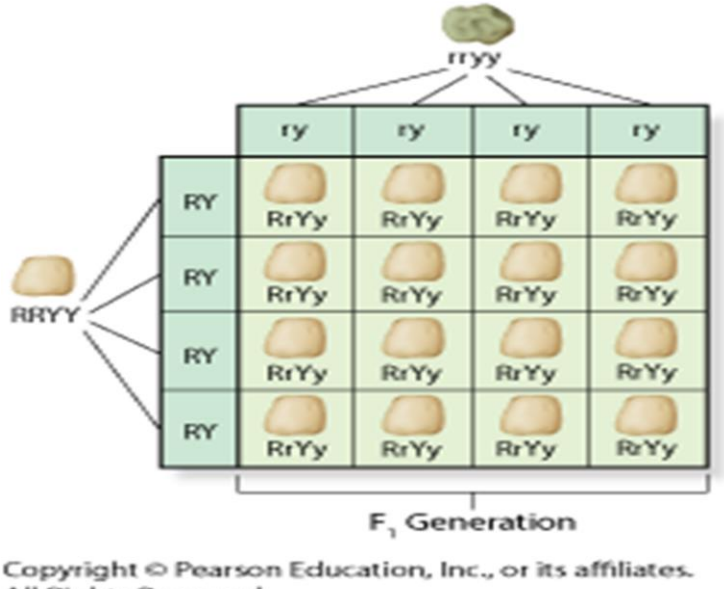
Punnett Square One-Factor Crosses – These can be used in simple dominant and recessive allele crosses as well as for some codominance and incomplete dominance.

Example – beak size in birds, where large beak size is dominant over small beak size

Two heterozygous big beaked birds



Punnett Square Two-Factor Crosses – These can be used to show independent assortment and more than one trait.



Principle of Independent Assortment states that genes for different traits can segregate independently during the formation of gametes.

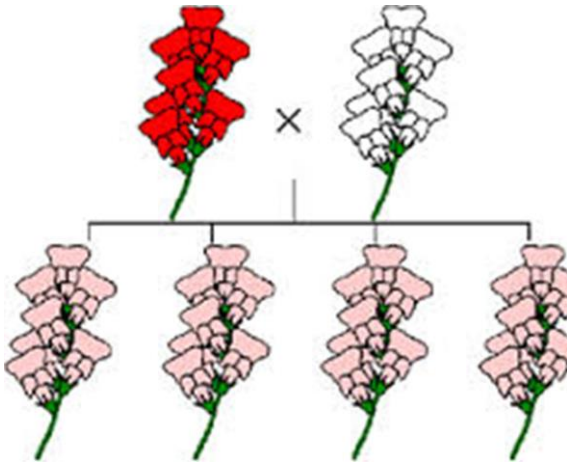
Through Mendel's studies he developed several principles of inheritance.

- Biological characteristics are inherited through units called genes. Genes are passed from parents to offspring.
- Sometimes there are two or more forms (alleles) of a gene for a single trait. Some forms of a gene may be dominant and others may be recessive.
- In most sexually reproducing organisms, each adult has two copies of each gene. Organisms get one copy from each parent. These alleles segregate from each other when gametes are made (meiosis).
- Alleles for different genes usually segregate independently of each other.

Dominant alleles do not ALWAYS mask the recessive allele. Sometimes there is not a dominant allele, this is called incomplete dominance.

Incomplete Dominance – situation in which one allele is not completely dominant over another allele

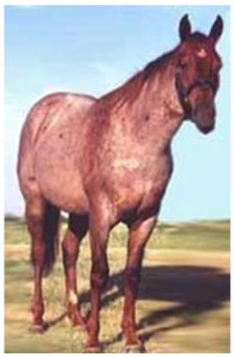
Example – snapdragon flower colors, when a red snapdragon is crossed with a white snapdragon flower plant, pink offspring are produced



Sometimes the phenotypes of **both** alleles are present in an organism. This is called codominance.

Example: Roan coloring in cows and horses

x



Rr: roan

Mix of both
red and white

Codominance – situation in which the phenotypes produced by both alleles are completely expressed

Not all crosses are simple monohybrid crosses involving only two alleles. Many genes have more than two alleles for a trait.

Multiple Alleles – a gene that has more than two alleles
Example – blood type

Some human, plant and animal traits show a large number of phenotypes because the traits are controlled by many genes. These genes act together to produce a single trait.

Polygenic Traits – trait controlled by two more genes
Examples – hair color, skin color

Chromosomal Inheritance

Chromosomal Theory of Inheritance: simply states that chromosomes are carriers of genetic information (Walter Sutton)

Among the 23 pairs of chromosomes in each human body cell there is a single pair of chromosomes called the sex chromosomes. This pair of chromosomes determines if a person is male or female.

Autosome – a chromosome that is not an allosome
Allosome – a sex chromosome

Humans have 22 pairs of autosomes, 1 pair of sex chromosomes (allosomes)

Sex chromosomes do not always match.

Male sex chromosomes X and Y

Female sex chromosomes X and X

Punnett Square for gender determination:

	X	Y
X	X X	X Y
X	X X	X Y

All female eggs carry one X chromosome.

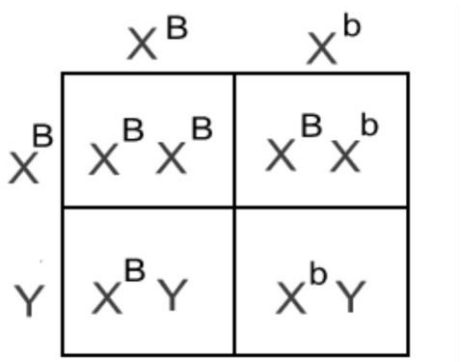
Half of the male sperm cells carry an X chromosome while the other half carry a Y chromosome.

How can a fertilized egg develop into a male?

The egg must receive a Y chromosome from the father in order to develop into a male.

Sex-linked genes - genes carried on the X and Y chromosome

Example: Red-Green Colorblindness



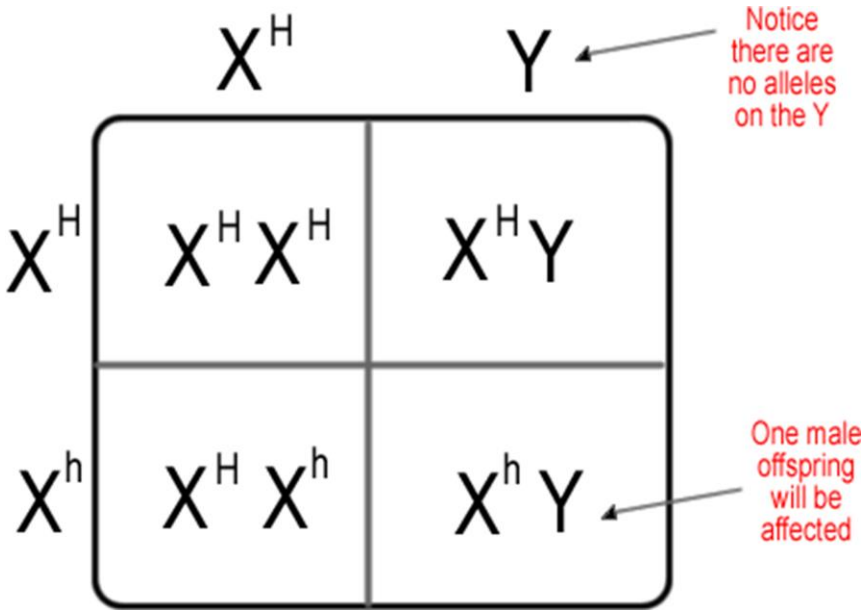
1/2 of the females will be carriers
1/2 of the females will be normal

1/2 of the males will be normal
1/2 of the males will be colorblind

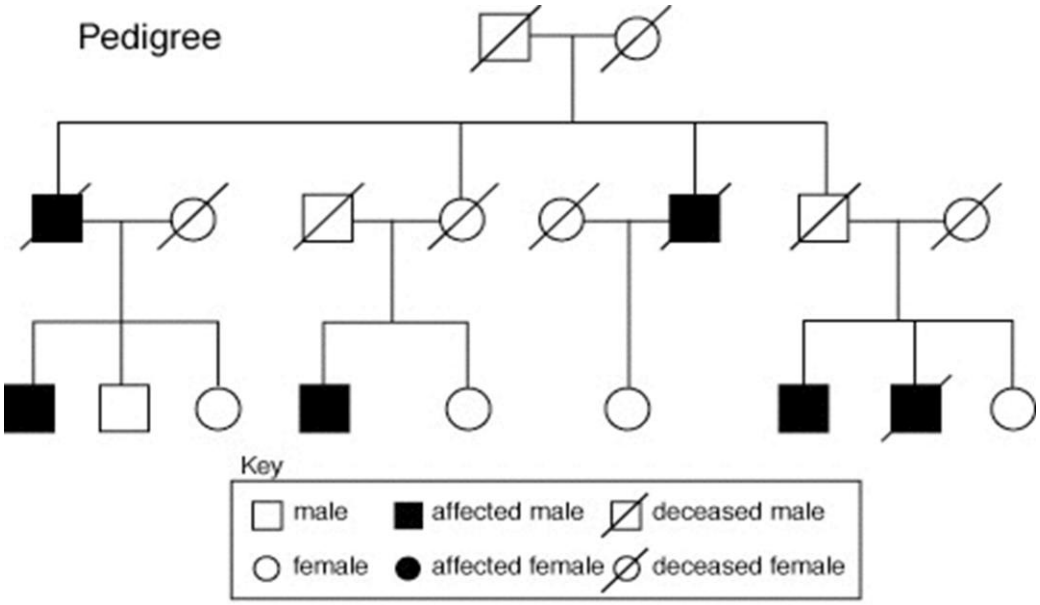
Why are males more likely to be red-green colorblind?

A male is more likely to be colorblind because males have only one X chromosome. Therefore males are more likely than females to have a sex-linked trait that is controlled by a recessive allele like colorblindness.

Another Example Hemophilia



Pedigree - a chart or "family tree" that tracks which members of a family have a particular trait



DNA Structure, Function, and Replication

DNA – deoxyribonucleic acid

Main Function of DNA

Storing Information – The main job of DNA is to store the genetic information of the cell. Genes control all the patterns of development for living things. They contain all the instructions that cause a single cell to differentiate and form into a plant, animal, or even a fungus.

Copying Information – A cell must be able to copy all of its genetic material before it divides. This is VERY important, if the genetic information did not copy before dividing each cell would not contain the proper genetic information to carry out the cell's functions.

Transmitting Information – Genetic information must be carefully sorted and passed on from parents to offspring. The sorting of genes is especially important during meiosis in reproductive cells. Any mistake in meiosis might cause a loss of genetic information that is vital for the offspring's survival.

Structure of DNA

There were many scientists that contributed to identifying the structure of DNA.

Erwin Chargoff discovered that the amount of adenine and thymine were equal in samples of DNA. The same was true for cytosine and guanine. He concluded that guanine pairs with cytosine, and adenine pairs with thymine. This base pairing became known as Chargoff's Rule.

Rosalind Franklin was a scientist that began to study DNA in the 1950s. She used x-ray diffraction to get pictures of DNA that showed the strands were twisted around each other in a shape known as a helix. She also found that DNA us made of two strands, and even had the idea that the nitrogen bases were found near the center of the DNA molecule.

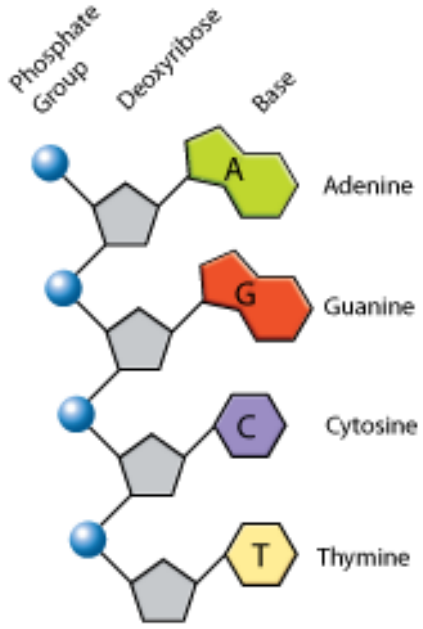
James Watson and Francis Crick also studied DNA in the 1950s. In 1953 Watson say a copy of Franklin's x-ray picture. When he shared what he saw with Crick

they solved the puzzle of the structure of DNA, they realized not only was it two stranded, but the two strands ran opposite of each other (this is known as being **antiparallel**).

DNA is a nucleic acid made up of nucleotides joined into long strands or chains by covalent bonds. DNA forms a double helix. It looks like a twisted ladder.

Each nucleotide is made up of three parts.

1. Phosphate Group
2. Sugar Molecule (deoxyribose)
3. Nitrogenous Base



DNA has four different nitrogenous bases.

Adenine - A

Guanine - G

Cytosine - C

Thymine - T

The bases pair in a specific way.

Guanine pairs with Cytosine.

Adenine pairs with Thymine

GC/AT

DNA Replication

Before a cell divides it must make a copy of its DNA.

Replication – the process of copying DNA prior to cell division

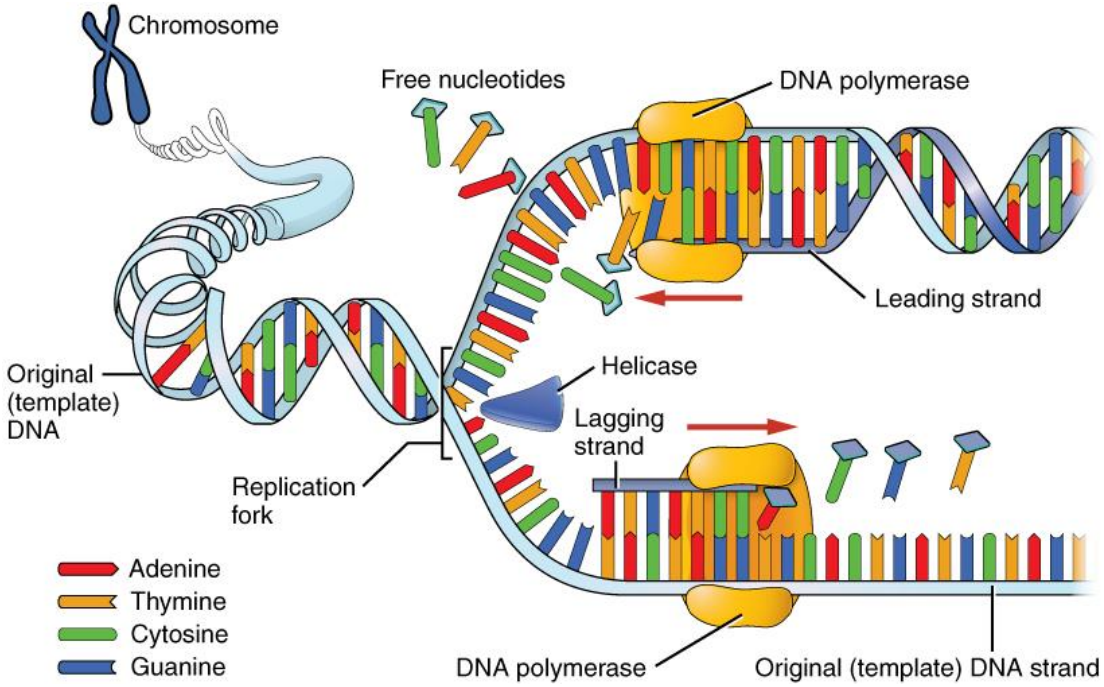
During replication the DNA double helix unwinds and separates into two strands. Then, the process makes two new strands following the rules of base pairing (Chargoff's Rule). Each stand of the double helix of DNA is a template, or model, for making the new strand.

Enzymes are essential in DNA replication. The enzymes pull apart the molecule of DNA by breaking the hydrogen bonds between the base pairs. They then unwind the two strands.

DNA polymerase – the principle enzyme involved in DNA replication

The DNA at the tips of the chromosome region is difficult to replicate. Cells use a special enzyme called telomerase to fix this problem. The telomerase make it less likely that genes will be damaged or lost during replication. Telomerase is often switched off in normal adult cells. However, in cancer cells, telomerase may be switched on, which may be why cancer cells can grow and divide so rapidly.

Telomere – repetitive DNA at the end of a eukaryotic chromosome



RNA and Protein Synthesis

RNA – ribonucleic acid

Three main differences between DNA and RNA

1. The sugar in RNA is ribose, not deoxyribose
2. RNA is usually single-stranded, not double-stranded
3. RNA has the nitrogenous base uracil instead of thymine (so instead of adenine binding to thymine it binds to uracil, guanine and cytosine still bind together, GC/AU)

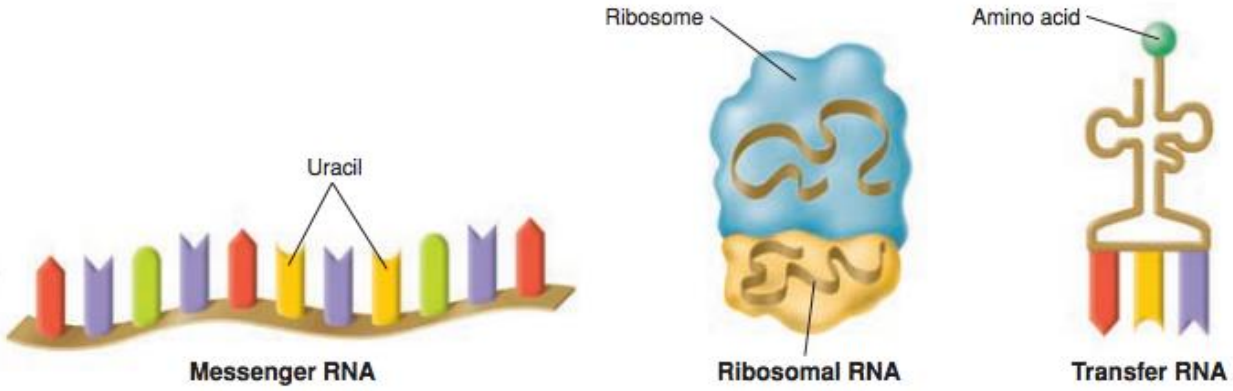
The function of RNA is mainly to make proteins in a process called protein synthesis. RNA controls how the amino acids are made into proteins.

Types of RNA:

Messenger RNA – mRNA – a type of RNA that carries copies of instructions for the assembly of amino acids into proteins from DNA to the rest of the cell.

Ribosomal RNA – rRNA – a type of RNA that combines with proteins to form ribosomes

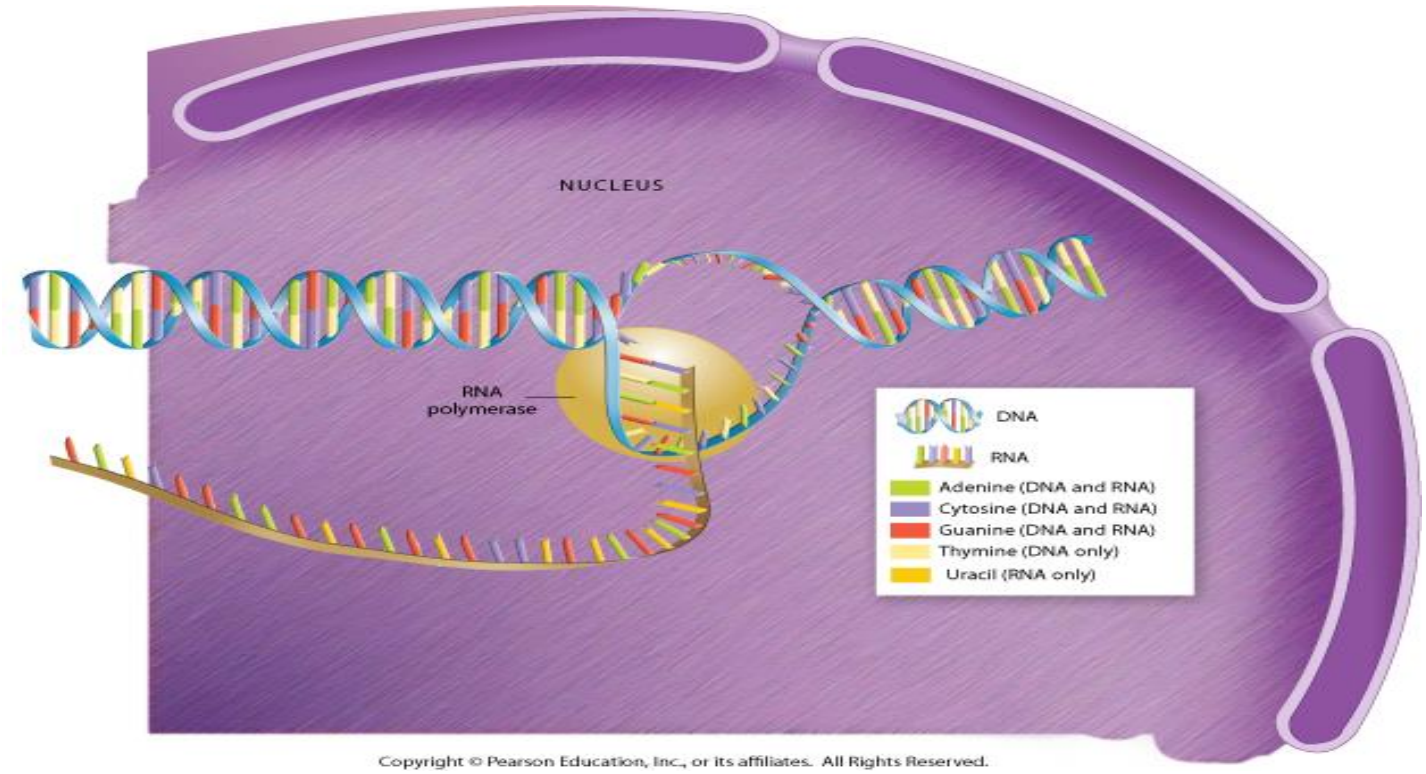
Transfer RNA – tRNA – a type of RNA that carries each amino acid to a ribosome during protein synthesis



Types of RNA The three main types of RNA are messenger RNA, ribosomal RNA, and transfer RNA. Ribosomal RNA is combined with proteins to form ribosomes.

RNA Synthesis

During transcription, segments of DNA act as templates, or patterns, to make complementary RNA molecules. The enzyme RNA polymerase uses one strand of DNA as a template to put together nucleotides to make a strand of RNA.



Ribosomes and Protein Synthesis

The first step in decoding genetic messages is to use transcription to make RNA from a sequence of DNA. The RNA holds the code for making proteins. The proteins are made up of long chains of amino acids.

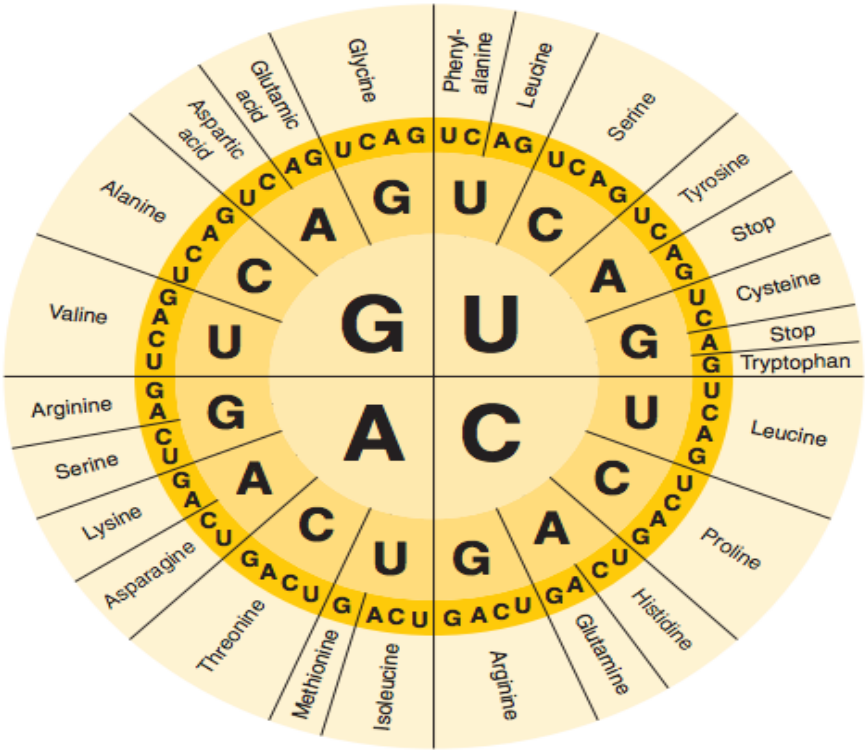
Polypeptide – a long chain of amino acids that makes proteins

Genetic code – a collection of codons of mRNA, each of which directs the incorporation of a particular amino acid into a protein during protein synthesis

Codon – a group of three nucleotide bases in mRNA that specify a particular amino acid to be incorporated into a protein.

The shape and function of a protein is determined by its amino acids and their sequence. Each three-base set in the genetic code is called a codon. A codon specifies one amino acid.

Reading Codons:



Genetic Code The genetic code shows the amino acid to which each of the 64 possible codons corresponds. To decode a codon, start at the middle of the circle and move outward. **Interpreting Graphics** ***For what amino acid does the codon UGC code?***

There are certain codons that tell the cell where to start and stop translating the RNA. AUG acts as the start codon for protein synthesis. After the start codon, mRNA is read three bases at a time. Translation then continues until one of three different stop codons is reached. Then translation stops and the polypeptide is complete.

Translation – a process by which the sequence of bases of an mRNA is converted into the sequence of amino acids of a protein. mRNA is transcribed in the nucleus and then enters the cytoplasm

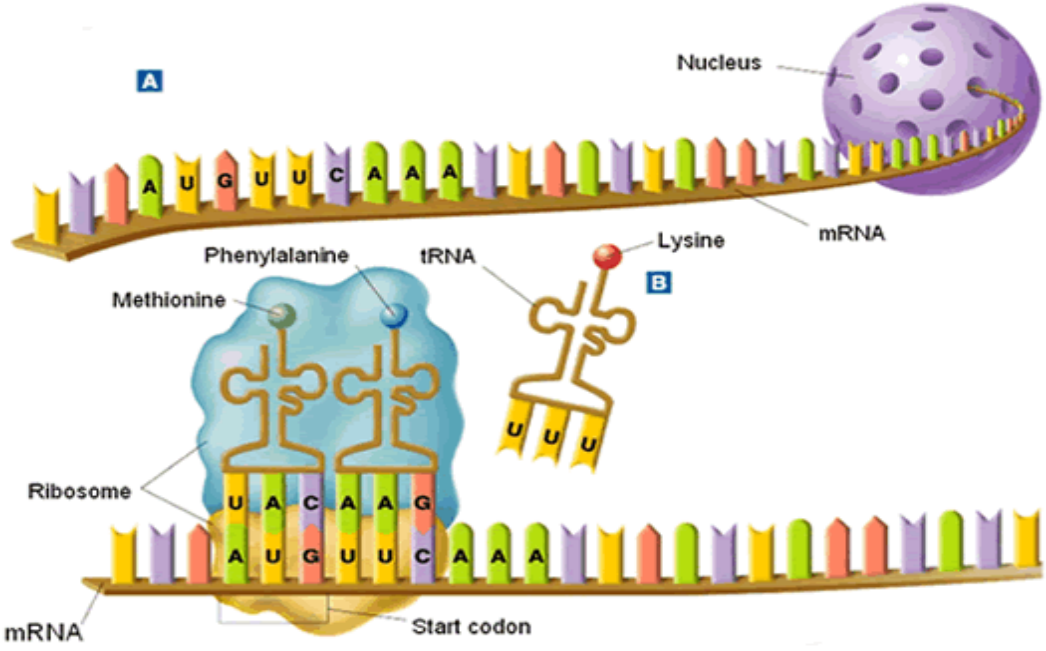
Steps of Translation

1. Transfer RNA – translation begins at AUG, each tRNA has an anticodon whose bases are complementary to the bases of a codon on the mRNA strand.

   Anticodon – a group of three bases on a tRNA molecule that are complementary to the three bases of a codon of mRNA

2. Joining Amino Acids – the ribosome helps make a peptide bond between the first and second amino acids, while at the same time the bond holding the first tRNA molecule onto its amino acid is broken. That tRNA then leaves the ribosome and another tRNA molecule enters

3. Completing the Polypeptide – the polypeptide chain continues to grow until the stop codon is reached on the mRNA, at this time the ribosome releases the polypeptide and the mRNA molecule and translation is complete



Information is transferred from DNA to RNA to protein.

Mutations

Mutation – a change in the genetic material of a cell

Types of Mutations

Gene Mutations

Point Mutation – a gene mutation in which a single base pair of DNA has been changed

Substitution – one base is changed to a different base

usually only affects a single amino acid, sometimes they have no effect at all

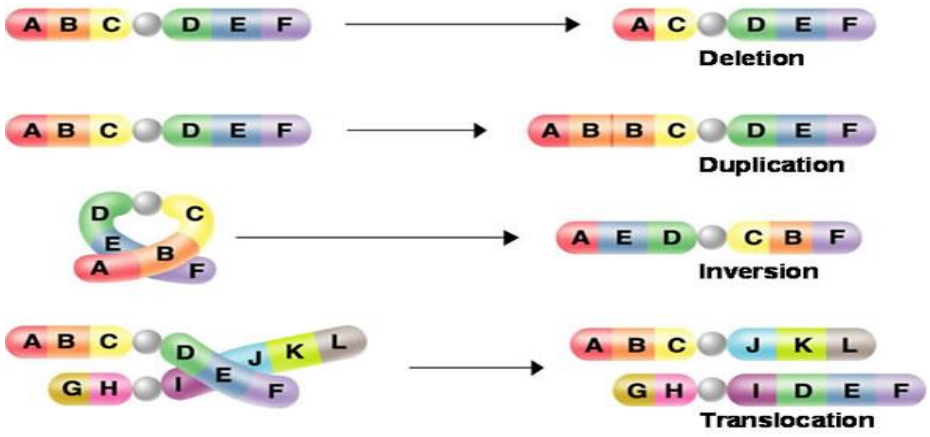
Insertion and Deletions (frameshift mutation) – a mutation that shifts the reading frame of the genetic message by inserting or deleting a nucleotide

The effects of insertions and deletions can be dramatic; it shifts every codon that comes after that mutation.

Chromosomal Mutations

Four Types of Chromosomal Mutations

1. Deletion
2. Duplication
3. Inversion
4. Translocation



Mutagen – a chemical or physical agent in the environment that interacts with DNA and may cause a mutation

Ex. – smoking, x-rays, ultraviolet light

Polyploidy – a condition in which an organism has extra sets of chromosomes

Ex. – seedless fruit, larger plants

The effects of mutations on genes vary widely. Some have little or no effect, and some produce beneficial variations. Some negatively disrupt gene function.

Gene Regulation and Expression

Gene Expression – the activation of a gene that results in the formation of a protein, this happens when transcription occurs

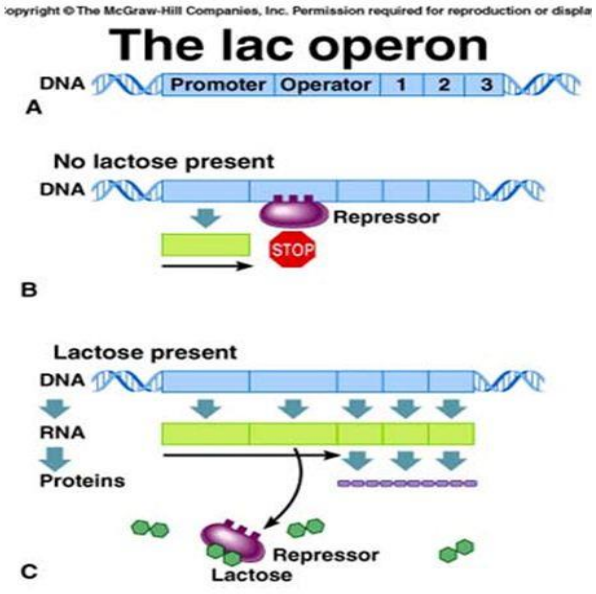
Cells don't always need to produce all of the proteins that their genes are coded for, they can control which genes are expressed.

Gene Expression in Prokaryotes

Structural genes, promoter, and operator collectively form an operon, which is a series of genes that code for specific products and the regulatory elements that control these genes.

An **inducer** is a molecule that initiated gene expression

A **regulator gene** codes for the production of the repressor protein

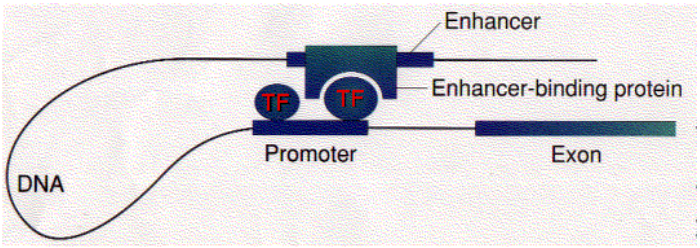


DNA-binding proteins in prokaryotes regulate genes by controlling transcription.

Gene Expression in Eukaryotes

In eukaryotes, gene expression is partly related to the coiling and uncoiling of DNA within each chromosome. The degree of uncoiling indicates the degree of gene expression.

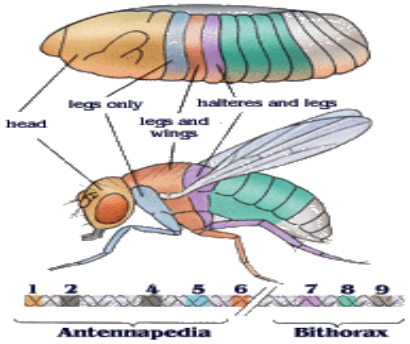
Transcription factors control the expression of eukaryotic genes by binding DNA sequences in regulatory regions.



Controlling gene expression helps shape the way a multicellular organism develops. Remember each specialized cell in and adult begins from the same fertilized egg cell. Different sets of genes are turned off and on as the embryo develops.

Cell Differentiation – the process in which cells become specialized in structure and function

Homeotic gene – a class of regulatory genes that determine the identity of body parts and regions in an animal embryo, mutations in these genes can transform one body part into another



An organism's environment also plays a role in cell differentiation.

Ex. Metamorphosis can be influenced by environmental factors, such as temperature, or change in moisture.

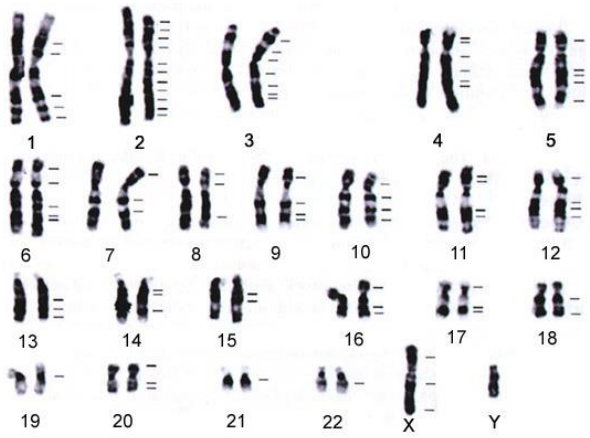
Human Genetics

Human Heredity

Genome – the entire set of genetic information that an organism carries in its DNA

The human genome has been mapped out. The Human Genome Project began working in 1990 and completed their work in 2003. Anyone that has access to the internet can view the human DNA sequence.

Another way to view information about human DNA is by looking at a karyotype. Scientists take pictures of cells during mitosis, then they arrange them in order. They take the pictures during mitosis because the chromosomes are easy to see because they are condensed.

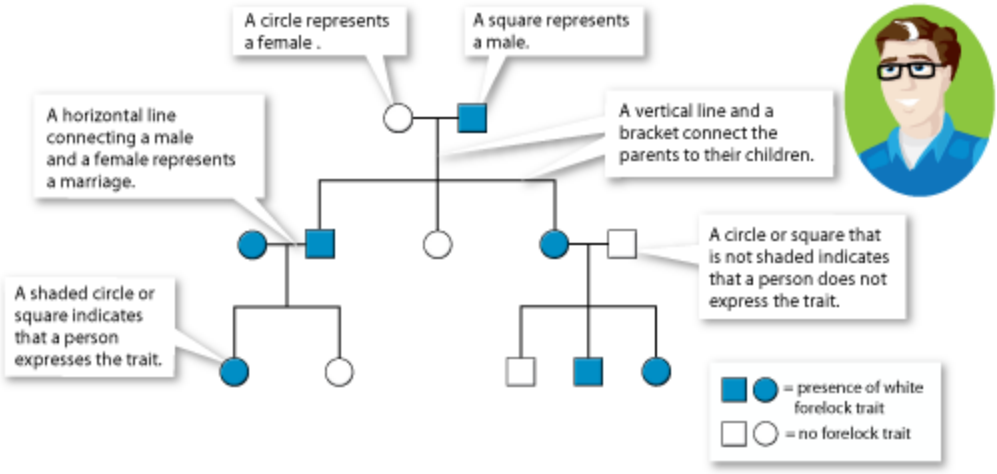


Karyotype – a photograph of the complete diploid set of chromosomes grouped together in pairs, arranged in order

There are 22 pairs of autosomes in a human karyotype and one pair of sex chromosomes. The genes for all the human traits can be found on these chromosomes.

Pedigrees are used to show how a trait is passed along from parents to their offspring. You can think of it as a genetic "family tree".

Pedigree – a chart that shows the presence or absence of a trait according to the relationships within a family across several generations



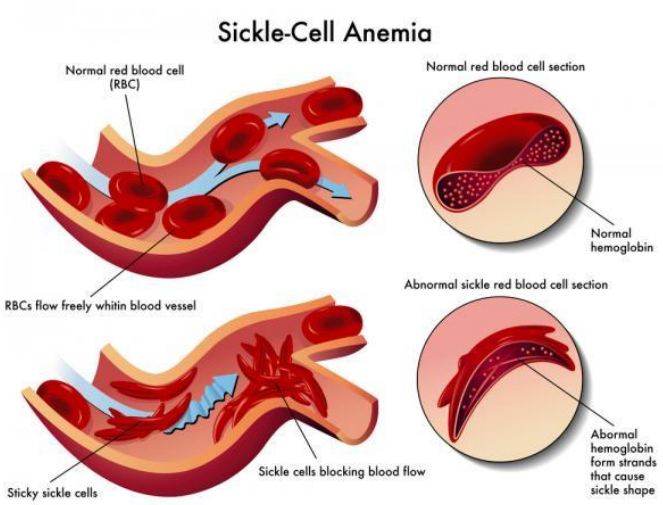
Human Genetic Disorders

Human Genetic Disorders Caused by Individual Genes

Sickle-Cell Disease – a disorder caused by a flawed allele for a polypeptide in hemoglobin.

Hemoglobin – the oxygen-carrying protein in red blood cells

The flawed polypeptide makes hemoglobin molecules stick together because of their sickle shape.



Huntington's Disease – a genetic disorder caused by a dominant allele for a protein found in brain cells.

The allele for this disease has a long string of bases. The codon CAG repeats over and over again, more than 40 times. CAG codes for the amino acid glutamine. No one knows for sure why this long string of glutamine causes the disease. People with Huntington's disease suffer from decreasing mental abilities and uncontrollable movements. Symptoms usually do not appear until middle age.

Cystic Fibrosis – a genetic disorder caused by the deletion of three bases in one gene. The loss of these three bases remove one amino acid from the protein called CFTR. This causes the protein to fold incorrectly, so it cannot do its job.

The allele for cystic fibrosis is recessive, so two copies of the defective allele are needed in order for someone to have the disorder.

Someone with cystic fibrosis may have abnormally thick mucus in their lungs and intestines. Children with cystic fibrosis may also have trouble digesting food.

Chromosomal Disorders

Meiosis usually works fine. Each human gamete usually gets 22 autosomes and 1 sex chromosome. Sometimes something goes wrong in meiosis, the most common error in meiosis is nondisjunction.

Nondisjunction – an error in meiosis in which the homologous chromosomes fail to separate properly

When nondisjunction occurs in meiosis gametes can end up with the wrong number of chromosomes.

Down Syndrome – a condition in which an individual has three copies (a trisomy) of chromosome 21

People with Down syndrome often have some degree of mental retardation and other birth defects.

Turner's Syndrome – a condition when a female only inherits one X chromosome\

Females with Turner's syndrome often do not have sex organs that develop properly, so she cannot have children.

Klinefelter's Syndrome – a condition when a male inherits two X chromosomes

Males with Klinefelter's syndrome usually can't reproduce because the extra X chromosome interferes with meiosis.

There is no evidence of a human being born without an X chromosome, this tells us that the genes on this chromosome are necessary for an embryo to survive and develop.

Studying the Human Genome

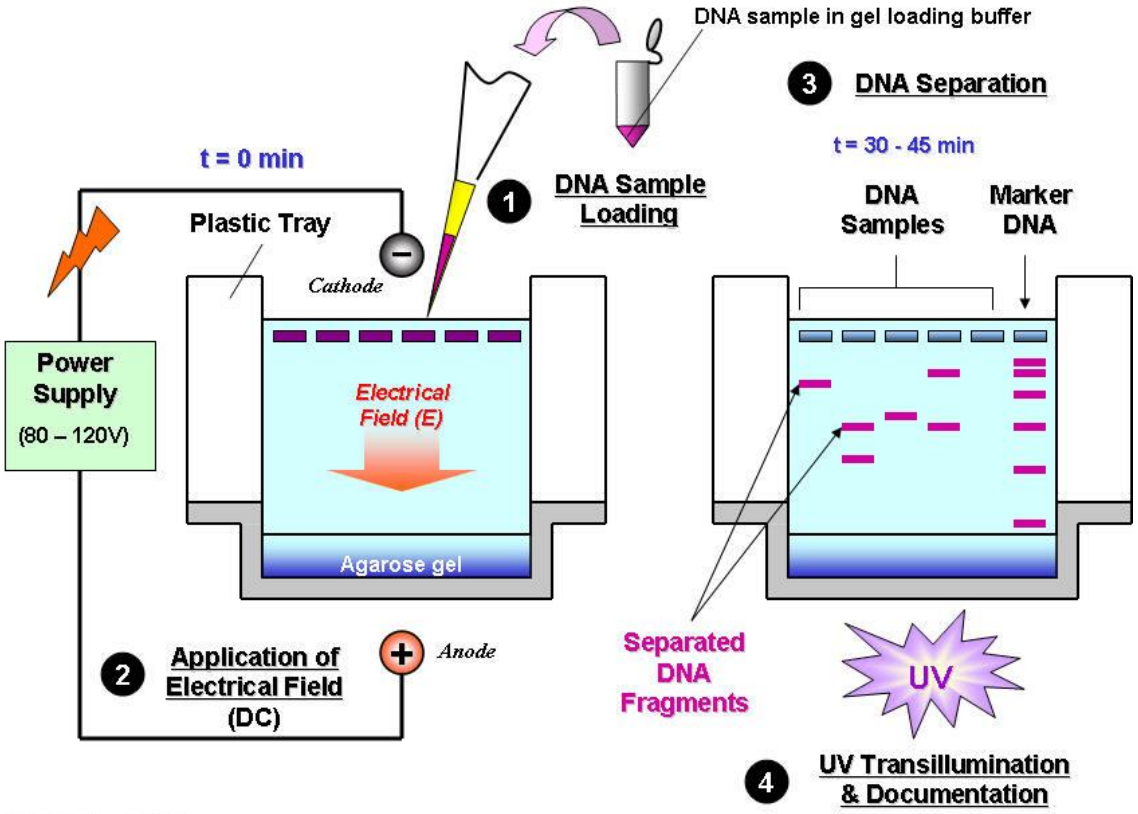
Ever since scientists discovered how genes were passed on, and where they were found on chromosomes they have wanted to read the human genome. Scientists then discovered tools that allow them to cut, separate, and replace DNA. This allows them to read the base sequence in DNA from any cell.

In order to study DNA scientists first need to cut it into smaller segments. They use various restriction enzymes to do this.

Restriction Enzyme – an enzymes that cuts DNA at a sequence of nucleotides.

After the DNA is cut, scientists need to separate and analyze the segments of the DNA. This is done using gel electrophoresis.

Gel Electrophoresis – a procedure used to analyze DNA fragments by placing a mixture of DNA fragments at one end of a porous gel and applying an electrical voltage in the gel



The Human Genome Project is complete and we know the sequence of human DNA (all 3 BILLION base pairs), and all human genes have been identified. This has opened a new field in biology, genomics.

Genomics – the study of whole genomes, including genes and their functions

Now scientists are going to study genomic variation (the 1000 Genome Project) in 1000 people. They will then use that data to study development and disease. The information gathered can be used for new treatments and drugs for people.

Genetic Engineering

Humans have been breeding many different organisms for their own benefits for many years. They have selected which traits they find desirable, and nearly eliminated the unwanted traits. Genetic engineering makes this all possible.

Selective Breeding – a method of breeding that allows only those organisms with desired characteristics to produce the next generation
Ex – Corn

Hybridization – a breeding technique that involves crossing dissimilar individuals to bring together the best traits of both organisms

Pretty Horse + Fast Horse = Pretty Fast Horse

Inbreeding – the continued breeding of individuals with similar characteristics to maintain the derived characteristics of a kind of organism
Ex – Dog Breeds

Selective breeding takes advantage of naturally occurring genetic variation. Humans use selective breeding to pass wanted traits on to the next generation of organisms.

Biotechnology – the process of manipulating the organisms, cells, or molecules to produce specific products

Breeders can use radiation and chemicals to mutate bacterial genes. This has allowed scientists to develop hundreds of useful kinds of bacteria.
Ex – bacteria that can clean up radioactive substances and metal pollution

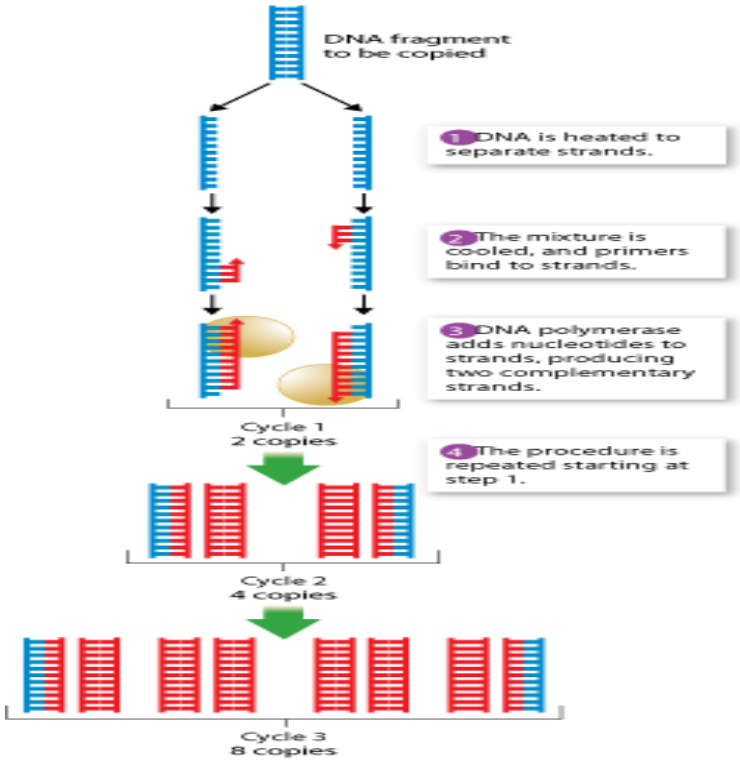
Another way breeders can introduce variation is by introducing certain drugs during meiosis. This causes the chromosomes to not separate in meiosis, creating a new chromosome number. This is called polyploidy. Polyploidy is common in plants, not in animals.

Ex – plants that are larger, and stronger than their diploid relatives in fruits such as bananas and citrus fruits

Scientists are now able to transfer genes for a particular trait from one organism to another. In order to do this scientists need to find the particular gene that they want to transfer, cut it out of the DNA, copy it, and insert it where they want it.

Scientist need many copies of the gene in order to make the appropriate changes to it before they insert it where they want it. In order to do this they use a technique called PCR.

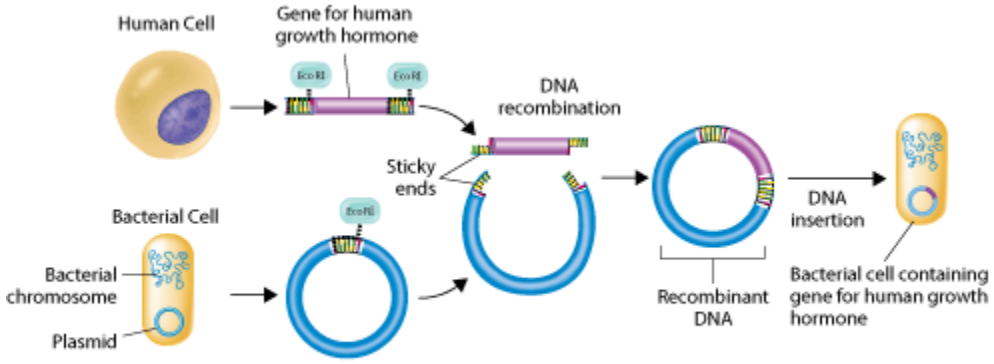
Polymerase Chain Reaction (PCR) – the technique used by biologists to make many copies of a particular gene



Recombinant DNA – DNA produced by combing DNA from different sources

Scientists often use plasmids in recombinant DNA studies.

Plasmid – small, circular piece of DNA located in the cytoplasm of many bacteria

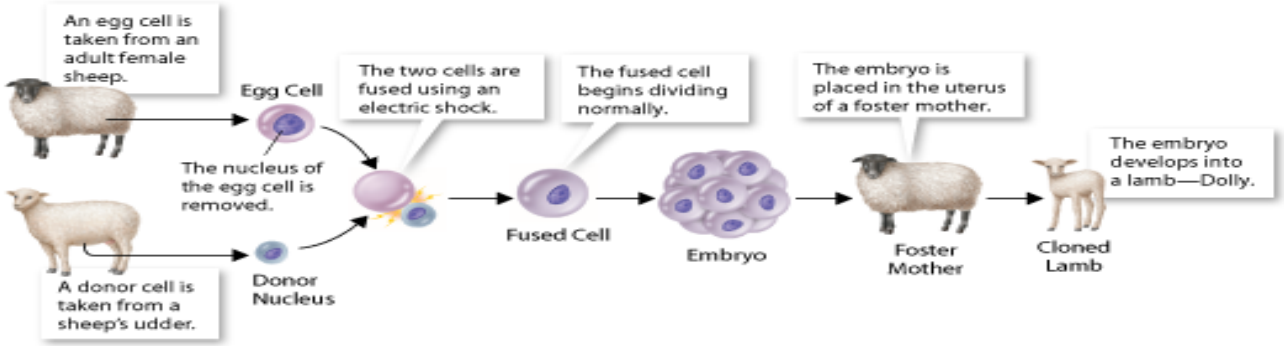


Most living things share the same genetic code, this allows scientists to make organisms that are transgenic. They can be made by putting recombinant DNA into the genome of a host.

Transgenic – a term used to refer to an organism that contains genes from other organisms

Clone – a member of a population of genetically identical cells produced from a single cell

Cloning uses a single cell from an adult organism to grow a new organism. That new organism is then genetically identical to the organism from which the cell was taken.



Applications/Uses of Genetic Engineering

GM Crops – genetically modified crops
Ex – plants resistant to viruses, Bt toxin (harmless to humans, kills insects that eat it, no pesticides needed)

GM Animals – genetically modified animals
Ex – hormones that help cows produce more milk, transgenic salmon that grow quicker

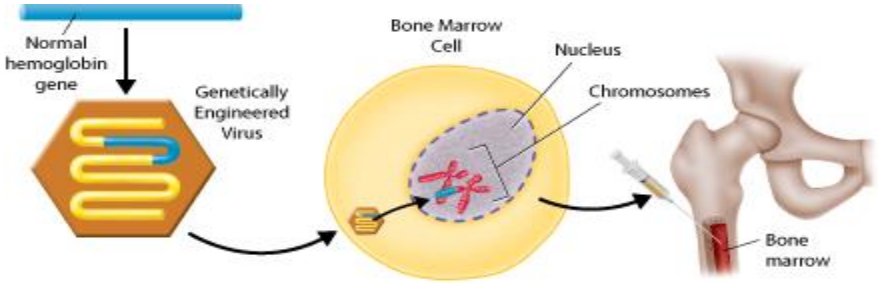
Health and Medicine

Preventing Disease – Scientists are making transgenic plants and animals that make human antibodies to fight disease.
Ex - golden rice with increased amounts of provitamin A (lack of pvA causes infant blindness and other problems)

Medical Research – Some transgenic animals are used as test subjects for medical research. The can model human diseases. They can then use the animals to test new drugs that can treat the disease.

Treating Disease – Recombinant DNA can be used to make important proteins that can save human lives.
Ex – bacteria making insulin, HGH, and the blood clotting protein for hemophiliacs

Gene therapy – a process by changing a gene to treat a medical disease or disorder. An absent or faulty gene is replaced by a normal working gene



Genetic Testing – Many tests can be performed to see if someone is carrying a gene that may predispose them to a disease. Couples may also undergo genetic testing before having a baby to see the probabilities that their child will have certain genetic abnormalities.

Examining Active Genes – When scientists study active genes they can learn how cells work.

DNA microarray – the glass slide or silicon chip that carries thousands of different kinds of single-stranded DNA fragments arranged on a grid

Personal Identification – The human genome is so complex that no individual has the exact DNA of another individual. The exception to this are identical twins.

DNA fingerprinting – a tool used by biologists to determine whether two samples of genetic material are from the same person

Forensics – the scientific study of crime scene evidence

Ethics

Profits and Privacy

Do you have exclusive rights to your DNA? Should you, like patent holders, be able to keep your genetic information private? Why or why not?

Safety of Transgenics

Pros of GMOs – higher yields, use less land, cheaper, use less energy, insect resistant

Cons of GMOs – no long-term studies have been done to test for safety, may hurt helpful insects, GM seeds are expensive

Ethics of the New Biology

Just because we have the technology to modify an organism's characteristics, are we justified in doing so?