DNA

- **Deoxyribonucleic Acid**

- DNA is the molecular basis for heredity. It is found in all organisms and contains the specific instructions to specify characteristics.

- DNA contains the genetic code that holds the instructions for producing proteins. These Proteins are essential for life.

- **Genes** are segments of DNA molecules that code for a specific protein.
Gene Locations

Cell

Chromosome

DNA

Nucleus
Building Blocks of DNA

A DNA molecule is made up of thousands of smaller repeating subunits called NUCLEOTIDES.

The structure, function, and composition of DNA is the same for all organisms. The only difference is the sequence of NUCLEOTIDES that make up the DNA.
NUCLEOTIDE STRUCTURE

- Composed of three parts:
  1. Phosphate Group
  2. 5 Carbon Sugar
     - DNA = Deoxyribose
     - RNA = Ribose
  3. Nitrogenous Base
     - 4 Different bases
       - A - Adenine
       - T - Thymine
       - C - Cytosine
       - G - Guanine

- The number and sequence of nucleotides determine the chemical and structural properties of DNA.
How are Nucleotides Assembled?

- Watson and Crick Developed the double helix model of DNA in 1953. They won the Nobel Prize in 1963.
DNA CONTROVERSY

- Rosalind Franklin was a scientist in the Lab with Watson and Crick. She used a technique called X-Ray Crystallography to photograph the double helix structure of DNA.
  - Watson and Crick stole the information Rosalind had discovered and submitted it before she had a chance to.
  - As a result, Watson and Crick were credited with the discovery while Rosalind did not receive credit until years after her death.
THE DOUBLE HELIX

- The model of DNA resembles a ladder
- Uprights are alternating phosphate groups
- Rungs are composed of alternating bases held together by weak hydrogen bonds
  - These two chains of nucleotides are called complimentary chains
  - The order of nucleotides on one strand determines the order of nucleotides on the other strand.
BASE PAIRING RULES

- **ADENINE** always pairs with **THYMINE**
- **CYTOSINE** always pairs with **GUANINE**
Purines = Pyrimidines
**RNA vs. DNA**

**RNA**
- The sugar is RIBOSE
- URACIL (U) is substituted for (T) Thymine
  - A=U
  - C=G
- RNA is a single stranded chain of nucleotides

**DNA**
- The sugar is DEOXYRIBOSE
- THYMINE is present
  - A=T
  - C=G
- DNA is double stranded
RNA

- mRNA – messenger RNA
  - Reads the information on the DNA and carries the MESSAGE from the nucleus to the ribosome in the cytoplasm.
  - RIBOSOME – Sight of protein synthesis

- tRNA - Transfer RNA
  - Acts as an interpreter. Translates the mRNA sequence into an amino acid
  - Transports amino acids within the cytoplasm to the ribosome.

- rRNA - Ribosomal RNA
  - Makes up the structure of the ribosome
DNA REPLICATION

- Exact duplication of genetic material before mitosis.
- Steps of Replication
  - Double stranded DNA unwinds and unzips between the weak bonds between the base pairs
    - This forms a replication fork
    - The bases are now exposed
    - Each unzipped strand is a template, or pattern, for a new molecule of DNA to form
DNA REPLICATION

- Free DNA nucleotides present in the nucleus bond to the exposed complimentary bases on the two DNA templates
  - This forms 2 new DOUBLE STRANDED DNA Molecules both identical to one another.
DNA REPLICATION

Diagram showing the process of DNA replication, with labels for parent DNA, new DNA strands, sugar-phosphate backbone, base pair (joined by hydrogen bonding), old strands, nucleotides about to be added to a new strand, and new strands.
Protein Synthesis

- DNA carries the codes to synthesize the thousands of proteins that each cell needs. This code is contained in the sequence of DNA base pairs. A sequence of three different bases will ultimately represent the code for a single amino acid.

- **Codon** - Three nucleotide sequences located on the mRNA sequence that codes specifically for an amino acid
Protein Synthesis
Part 1: Transcription

- Transcription: Synthesis of mRNA. The information in the DNA is transcribed (rewritten) to mRNA
  - DNA serves as a template for free RNA nucleotides
  - Nucleotides attach according to the base rules A=U, C=G
  - Specific base sequences of DNA determine the code of the mRNA
  - mRNA moves from the NUCLEUS to the CYTOPLASM
    - It contains codons - 3 nucleotide sequence that codes for 1 amino acid
Step 1: DNA to RNA (Transcription)
The DNA template is used to make a single stranded RNA. RNA nucleotides base-pair with DNA nucleotides on the template strand. RNA has the base U which pairs with A in DNA.
Step 2: mRNA leaves the nucleus
Protein Synthesis
Part 2: Translation

- The process by which the information coded for in the mRNA is used for the assembly of a particular amino acid sequence
  - Translation occurs in the RIBOSOME
  - mRNA “hooks up to” the ribosome
  - tRNA molecules bring amino acid molecules to the ribosome
  - tRNA has a special structure, which allows it to be an interpreter molecule.
    - Carries the *anti-codon-*3 nucleotide sequence that is complimentary to one of the codons on the mRNA
    - *Anticodons* of tRNA align with the codons of mRNA
    - Amino acids carried by tRNA bond together in a sequence determined by the mRNA information
  - Resulting Chain of Amino Acids form a polypeptide
    - The cell translates the bas sequence of the polypeptide protein
Step 3: mRNA and tRNA join in the cytoplasm at the RIBOSOME
Step 4: Amino Acids are carried to ribosome and joins according to the triplet code.
Step 5: The protein chain is created
GENETIC MUTATION

- Mutation: A sudden change in the structure or amount of genetic material
  - Most mutations are harmful
  - However some may be beneficial or have no effect
    - How Do you think a mutation will be beneficial?
    - Answer: Can be the raw materials to drive evolution
  - In order for a mutation to be inherited it must be present in the DNA of the gamete (for a sexually reproducing organism)
  - If a mutation occurs in a somatic cell (body cell) it cannot be passed on to new cells within the organism as a result of mitosis.
MUTAGENS

- Mutagen: Any agent that causes a mutation at higher than spontaneous level.
  - Radiation
    - X-rays
    - UV rays
    - Radioactivity
  - Chemicals
    - Asbestos
    - Formaldehyde
    - Mustard Gas
Mutations

- Classified as
  - GENE MUTATIONS - a mutation occurring in the sequence of bases
  - CHROMOSOMAL MUTATIONS – a mutation that occurs effecting a chromosome
Chromosomal Alterations

- A change in the NUMBER or STRUCTURE of Chromosomes in an Individual
  - Usually many genes are involved and the effects can be seen in the PHENOTYPE.
Change in Chromosome Number

- **Nondisjunction**: Homologous chromosomes do not separate during meiosis.

- **Down Syndrome**: Trisomy 21 – Extra Chromosome #21.
  - Individual will have 47 chromosomes in stead of 46
Patau Syndrome

- Patau Syndrome: serious eye, brain, circulatory defects as well as cleft palate. 1:5000 live births. Children rarely live more than a few months.
Edwards Syndrome

- almost every organ system affected 1:10,000 live births. Children with full Trisomy 18 generally do not live more than a few months.
Klinefelter Syndrome

CRIMINAL MALES: Fact or Fiction

- **47, XYY males**: Individuals are somewhat taller than average and often have below normal intelligence. At one time (~1970s), it was thought that these men were likely to be criminally aggressive, but this hypothesis did NOT stand up to testing and has been disproven over time. (An interesting personal essay from a man who is likely XYY: "The Stereotype of the Karyotype")
Turner Syndrome

- **Monosomy X (Turner's syndrome):** 1:5000 live births; the only viable monosomy in humans - women with Turner's have only 45 chromosomes!!! XO individuals are genetically female, however, they do not mature sexually during puberty and are sterile. Short stature and normal intelligence. (98% of these fetuses die before birth)
Polyploidy

- Possession of extra sets of chromosomes
  - Nondisjunctions of a complete set of chromosomes
    - Common in Plants
    - Rare in Animals
Chromosomal Mutations

- Permanent changes in chromosome structure, which may result from random breakage or recombination of chromosome parts.
Chromosomal Mutations

- **Translocation**: The transfer of a section of one chromosome to a non-homologous chromosome.

- **Addition (Duplication)**: The addition of a piece of chromosome to a homologous chromosome.

- **Deletion**: When a piece of a chromosome breaks off resulting in the loss of some genes.

- **Inversion**: A piece of the chromosome is rotated reversing the order.
Gene Mutations

- Point Mutation: A mutation in which one nucleotide in a gene is changed
  - When one nucleotide is added or removed, the effects can be drastic.

WHY???
Point Mutation

- THE FAT CAT RAN FOR THE RAT

- Addition: THE GFA TCA TRA NFO RTH ERA T
- Deletion: THE ATC ATR ANF ORT HER AT

  - All triplet codes passed the addition or deletion are altered
  - This may result in the formation of protein that is not functionally normal.
Cloning

- A genetically identical copy of an individual.
  - Must occur within a lab
  - Horse, cats, pigs, sheep, cows, dogs and the list is growing of animals already cloned.
Genetic Engineering

- Methods used to improve, produce, and maintain new varieties of organisms
  - **Selective Breeding**: Breeding specific individuals for desired traits
    - **Inbreeding**: The mating of closely related individuals to obtain desired characteristics
      - Ex. Pure bred dogs and cats
      - Decreases variation by increasing homozygous traits however it can result in undesirable effects.
      - HOW???
**Out Breeding (hybridization):**

Breeding organisms not closely related.

- Introduces beneficial traits
- **HYBRID VIGOR** - Superior characteristics resulting from out breeding
  - Ex. Mule (Horse x Donkey)
  - Hybrids are usually sterile

**You can’t foal Mother Nature**

Mules are considered sterile and unable to breed because they do not have an even number of chromosomes, but a mule on a ranch near Colbran has given birth to a foal. What may have happened:

**How to make a mule**

Female horse

64 chromosomes

Male donkey

62 chromosomes

Mammals need an even number of chromosomes to divide into pairs for reproduction.

Female mule

Male foal

The sire of the mystery foal is reportedly a donkey, but who the father is, as well as the exact makeup of the mystery foal, won’t be known until DNA testing is completed.

Genetic testing has proved the foal is the offspring of the mule — not from a horse/donkey mating.

*Source: Denver Post research*

*Thomas McKay | The Denver Post*
Gene Splicing: Transfer of genetic information from one organism to another

- Recombinant DNA: Two pieces of DNA from separate organisms are COMBINED.
  - Genes from one organism are inserted into the DNA of another

This process is carried out through the use of plasmids
Plasmid

Plasmid - Bacterial DNA
Recombinant DNA

1. The desired gene is identified.
2. The gene is inserted into a plasmid.
3. The recombinant plasmid is put into a bacterial cell.
Recombinant DNA

- How is Recombinant DNA made?
  - DNA containing the desired gene must be "cut" out of a much longer DNA molecule.
  - DNA is cut with restriction enzymes.
Transfer of the Insulin gene

Cloning the Insulin Gene

Transfer and cloning of the Insulin gene
Figure S-2: Gel Electrophoresis

1. Restriction enzymes cleave DNA into smaller segments of various sizes.

2. DNA segments are loaded into wells in a porous gel. The gel floats in a buffer solution within a chamber between two electrodes.

3. When an electric current is passed through the chamber, DNA fragments move toward the positively-charged cathode.

4. Smaller DNA segments move faster and farther than larger DNA segments.
Using this technique criminal investigators can determine relationships between a victim and a perpetrator.
Back to Bacteria

- **Conjugation:** Bacteria are able to exchange genetic information (from 1 bacterial cell to another).

- **Transformation:** One strain of bacteria is changed by a gene from another strain of bacteria
  - **WHAT** is the significance of conjugation and transformation?
    - Increased variation – ultimately evolutionary changes
GENETIC DISORDERS

- Tay-Sachs Disease
  - Recessive Trait
  - Fatal genetic disorder in which harmful quantities of a fatty substance accumulate in the nerve cells of the brain
  - Causes the nervous system to stop functioning normally
  - Individual usually dies by age 5
Genetic Disorders

- Sickle Cell Anemia
  - Abnormally shaped red blood cells due to a deformity in hemoglobin
  - Sickle Shaped cells cause damage to vessels and clotting
  - Recessive Disorder
Genetic Disorders

- Huntington’s Disease
  - Autosomal Dominant condition
  - Appears later in life (usually in 30’s and 40’s)
  - Loss of motor skills, decreased mental capacities
PKU- Phenylketonuria

- Inability to utilize the essential amino acid phenylalanine
- High levels of phenylalanine in the blood usually leads to brain damage
- Recessive disorder

The half-solid symbols represent heterozygous carriers of phenylketonuria; the double line between I-1 and I-2 signifies a consanguineous mating.

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Cystic Fibrosis

- Cystic fibrosis is an inherited chronic disease that affects the lungs and digestive system of about 30,000 children and adults in the United States (70,000 worldwide). A defective gene and its protein product cause the body to produce unusually thick, sticky mucus that:
  - clogs the lungs and leads to life-threatening lung infections; and
  - obstructs the pancreas and stops natural enzymes from helping the body break down and absorb food.
HOW CAN WE DETECT GENETIC DISORDERS?

- Amniocentesis: The removal of amniotic fluid for chemical and or cellular analysis.
CVS-Chorionic Villi Sampling

- A sample of the chorion (part of the placenta) is removed for examination.