DNA analysis:

· PCR: PCR is a method to amplify a few copies of DNA to produce millions of copies. It is primarily used to generate enough copies of a DNA sequence for analysis. It relies on "thermal cycling", a process in which the solution containing the dsDNA is repeatedly brought to the temperature point in which it two strands separate, (after lowering temperature a small amount) allowing primer molecules to anneal and polymerase to replicate the strands. The process is repeated, allowing the copies of the initial DNA molecules to grow exponentially. The temperature of the solution can be brought down further to store and halt the reaction. In most cases, the Taq polymerase is used in PCR, because of its high optimum operating temperature.

· RFLP analysis: RFLP analysis is a technique that exploits the variations in homologous DNA samples. Small differences in homologous DNA sequences can generate different "cutting" sites for restriction enzymes, thus allowing DNA sequences that have been cut using restriction enzymes to be separated based on size in a gel electrophoresis. Though it is being replaced by full genome sequencing, it is still used in paternity tests, genome mapping, and gene localization.

· in situ hybridization: mRNA in an organism is tagged with fluorescent tags to examine gene expression.

· DNA sequencing: Read rows top to bottom

· Y-chromosome analysis: The Y-chromosome is analyzed to determine certain paternal relationships.

Chromosomes: A chromosome is a organized structure of DNA and proteins (chromatin) that resides in the nucleus of a cell. Chromosomes replicate during Mitosis and Meiosis 1. There are 46 (44 autosomes, 2 sex chromosomes) chromosomes in human diploid cells and half that in human haploid cells. When designating chromosomes, the number of the chromosome is stated followed by the arms *(the long arm(s) are referred to as "q-arms", and the short ones as "p-arms")*.

Retrotransposon: A RNA subclass of transposons that can amplify themselves in a genome. They are often present in eukaryotic organisms.

Operon: A functional unit of genomic DNA consisting of a cluster of genes controlled by a single promoter or regulator. They are most commonly found in prokaryotes.

Genetic disorders:

· Down syndrome: Down syndrome is caused by the presence of all or part of a third copy of the 21st chromosome. People with the disorder have deformed facial characteristics and severe mental retardation.

· Cystic Fibrosis: Cystic Fibrosis is caused by a mutation in the CFTR gene. People with cystic fibrosis will often develop lung problems, general growth problems, reproductive disorders, and cysts on their pancreas.

· Sickle cell anemia: Sickle cell disease is caused by a point mutation in beta-hemoglobin chain at chromosome 11, causing Glutamic acid to be replaced to Valine. This replacement causes hemoglobin proteins to become malformed, significantly decreasing their oxygen carrying capacity and acquiring the ability to crystallize (giving the cells their shape). People with sickle cell disease have shortened lives (40-50 years) due to renal failure and various other secondary disorders.

· Tay-Sachs disease: Tay-Sachs disease is caused by a mutation in the HEXA gene in chromosome 15. This mutation causes components of cell membranes to accumulate inside neurons, eventually reaching harmful levels and causing death. In most cases, infants with this disease will develop normally for six months, then go through rapid cognitive deterioration followed by physical deterioration. Most infants will die by the age of four.

Lab Procedures:

· Southern Blot: A technique used to analyze DNA samples. DNA is first cut into small pieces with restriction enzymes, and then put through electrophoresis. The DNA is transferred to nitrocellulose paper (and exposed to UV radiation to permanently bind it there, and then hybridized with a DNA probe.

· Northern Blot: A technique used to analyze gene expression. RNA is extracted from cells, and then mRNA is isolated. Then, the process continues like Southern blotting.

· Western Blot: Like the other blots, but with proteins.

· Eastern Blot: Deals with post translational modifications of proteins (e.g. lipids, phosphomoieties, and glycoconjugates).

· Southwestern Blot: Deals with DNA-binding proteins.

· Far-Western Blot: Deals with protein-protein interactions.

· Far-Eastern Blot: Deals with lipids.

Definitions:

Inducible Enzymes: An enzyme that is normally present in minute quantities within a cell, but whose concentration increases dramatically when a substrate compound is added.

Regulatory Genes: A regulator gene, regulator, or regulatory gene is a gene involved in controlling the expression of one or more other genes. A regulator gene may encode a protein, or it may work at the level of RNA, as in the case of genes encoding microRNAs.

Enhancers: An enhancer is a short region of DNA that can be bound with proteins (namely, the trans-acting factors, much like a set of transcription factors) to enhance transcription levels of genes (hence the name) in a gene cluster. While enhancers are usually cis-acting, an enhancer does not need to be particularly close to the genes it acts on, and sometimes need not be located on the same chromosome.

Oncogene: An oncogene is a gene that plays an active role cancer. Oncogenes are "activated" when proto-oncogenes are overexpressed or mutated. In many cases, oncogenes can cause affected cells, that would normally to die from apoptosis, to survive and proliferate instead. The resultant protein may be termed an oncoprotein. In normal function, an proto-oncogene is used to promote the expression or function of other gene complexes.

Inducible operon: Repressor is on by default

Repressible operon: Repressor is unbound (off) by default.

Positive gene regulation: Further induced by a activating protein, such as CAP.

Bacterial Conjugation: Bacterial conjugation is a method of bacterial gene transfer that uses a cell-cell contact or a bridge known as a “pilus”. Results in a extra plasmid in the “recipient”.

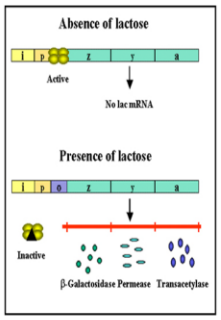
Transformation: The genetic alteration of a cell through uptake of the incorporation and expression of external genetic material, obtained from the environment. Most common in bacteria.

Transduction: The genetic alteration of a cell through injection of foreign DNA from Bacteriophages/viruses.Most common in bacteria.

Lac Operon

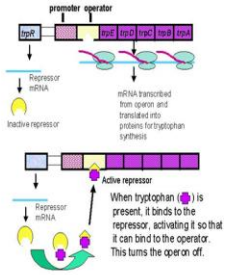
The genes that code for the enzymes needed for lactose catabolism are clustered on the same chromosome in what is called the **Lac Operon**

The *E. coli* only express the genes and make these enzymes when lactose is available to be metabolized. This is an **inducible operon** where genes are expressed in the presence of a substance



Trp Operon

- The genes for the five enzymes in the Trp synthesis pathway are clustered on the same chromosome in what is called the **Trp Operon**
- If the amino acid tryptophan (Trp) is added to a culture of *E coli*, the bacteria soon stop producing the five enzymes needed to synthesize Trp from intermediates produced during the respiration of glucose so the presence of the products of enzyme action represses enzyme synthesis
- This is a **repressable operon** where the operon are **turned off** in the presence of a substance



Second letter

First letter	U	C	A	G	Third letter
U	UUU Phe, UUC Phe	UCU Ser, UCC Ser, UCA Ser, UCG Ser	UAU Tyr, UAC Tyr	UGU Cys, UGC Cys	U
	UUA Leu, UUG Leu		UAA Stop	UGA Trp	C
			UAG Stop	UGG Trp	A
					G
C	CUU Leu, CUC Leu, CUA Leu, CUG Leu	CCU Pro, CCC Pro, CCA Pro, CCG Pro	CAU His, CAC His	CGU Arg, CGC Arg, CGA Arg, CGG Arg	U
			CAA Gln, CAG Gln		C
					A
					G
A	AUU Ile, AUC Ile	ACU Thr, ACC Thr, ACA Thr, ACG Thr	AAU Asn, AAC Asn	AGU Ser, AGC Ser	U
	AUA Met		AAA Lys	AGA Stop	C
	AUG Met		AAG Lys	AGG Stop	A
					G
G	GUU Val, GUC Val, GUA Val, GUG Val	GCU Ala, GCC Ala, GCA Ala, GCG Ala	GAU Asp, GAC Asp	GGU Gly, GGC Gly, GGA Gly, GGG Gly	U
			GAA Glu, GAG Glu		C
					A
					G