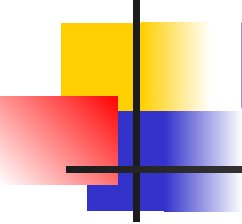


Exploring the World of Science

Designer Genes (C)-2026

KAREN LANCOUR

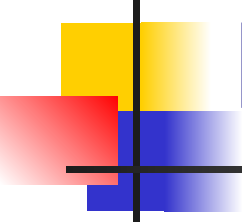
karenlancour@charter.net



Event Rules – 2026

DISCLAIMER

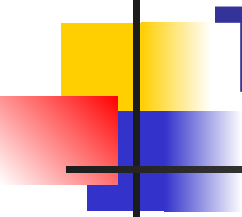
This presentation was prepared using draft rules. There may be some changes in the final copy of the rules. The rules which will be in your Coaches Manual and Student Manuals will be the official rules.



Event Rules – 2026

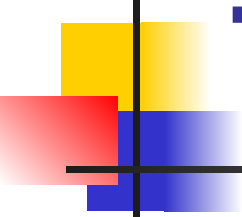
- **BE SURE TO CHECK THE 2020 EVENT RULES FOR EVENT PARAMETERS AND TOPICS FOR EACH COMPETITION LEVEL**

SOSI



TRAINING MATERIALS

- **Training Power Point** – content overview
- **4 Training Handout** – overview, general principles, molecular genetics & biotechnology
- **2 Practice Activities** – molecular genetics & biotechnology sample problems & lab evaluations
- **Sample Tournament** – sample problems with key
- **Event Supervisor Guide** – prep tips, event needs, and scoring tips



SO WEBSITE TRAINING MATERIALS

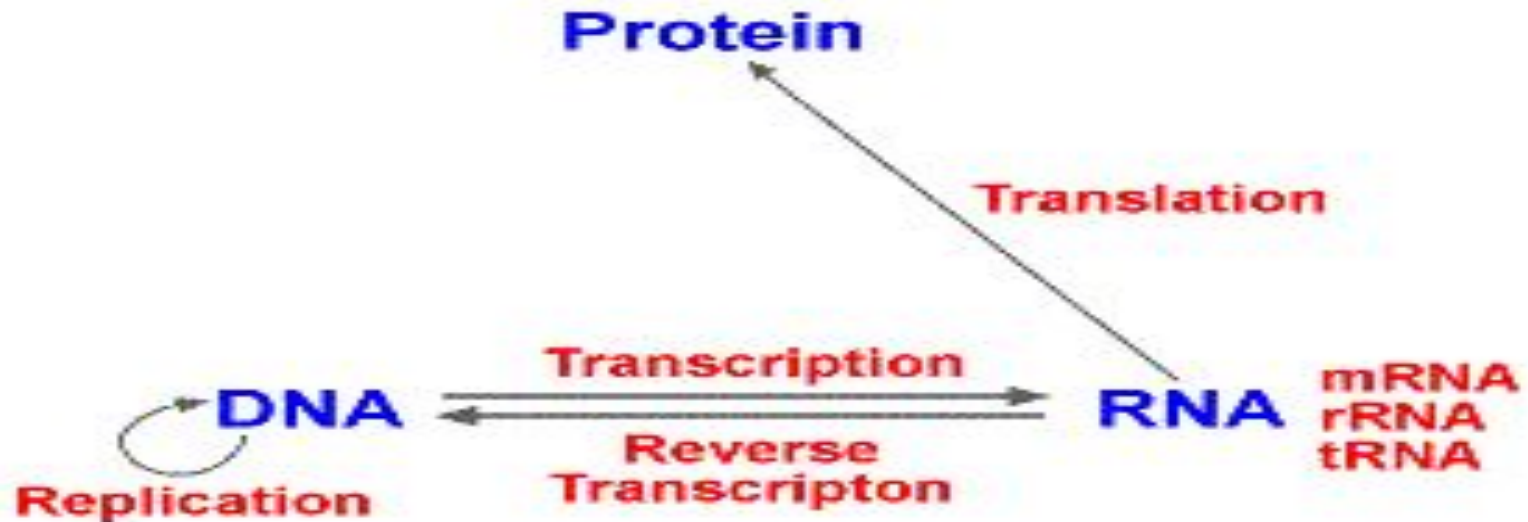
EVENT RESOURCES – the Science Olympiad website www.soinc.org
under Event Information-will be posted throughout the fall

- **Training Power Point** – content overview
- **Internet Resource** – links to good resources for learning content
- **Training Handouts** – overview, general principles
- **Sample Tournament** – sample problems with key
- **Event Supervisor Guide** – prep tips, event needs, and scoring tips

Training PDFs – the Science Olympiad Store at www.soinc.org

- **Biology-Earth Science (BECD)** – current year topics for all bio events with training materials and extra resources
- **Genetics (GNCD) 2019** – all content, extra resources, exams for Heredity and Designer Genes
- **Division B and Division C Test Packets** – national exams from the previous year

CENTRAL DOGMA OF MOLECULAR GENETICS



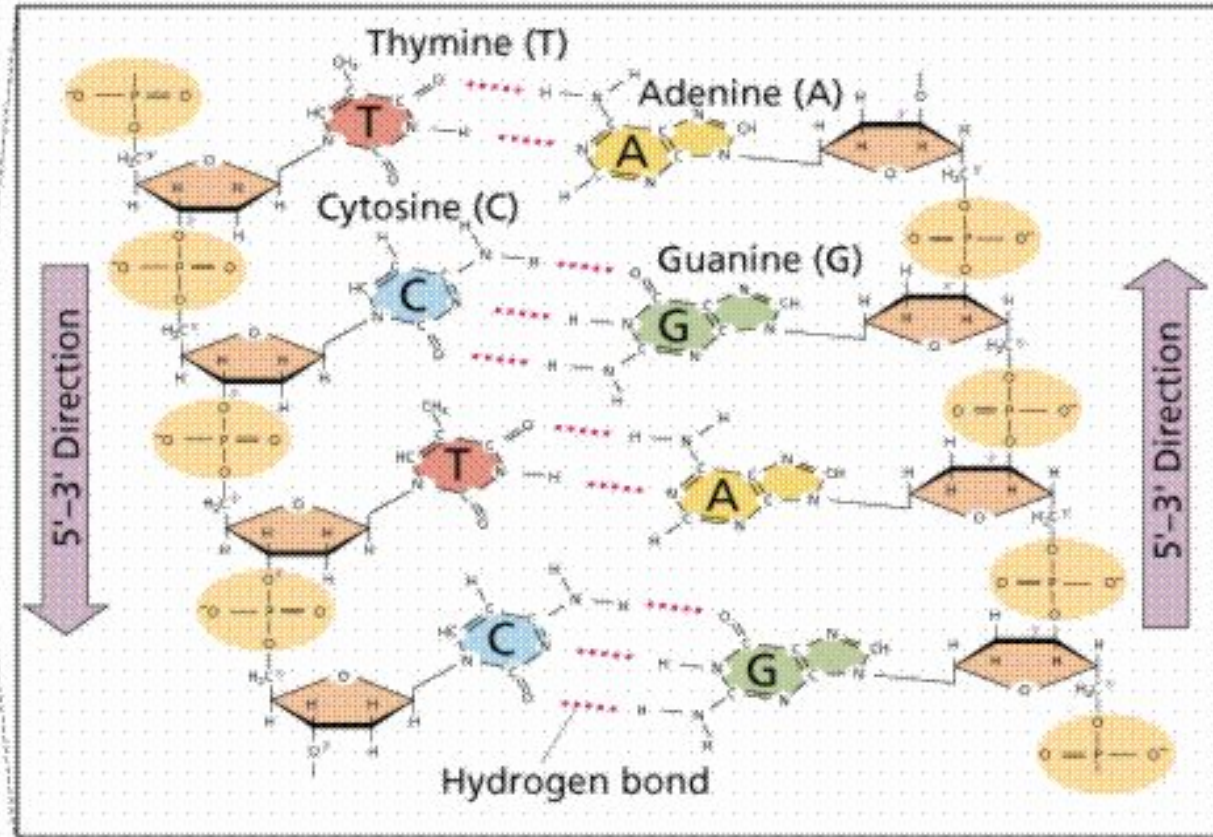
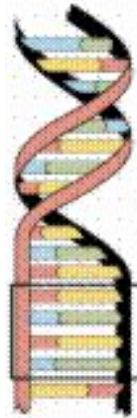
DNA ----□ RNA ---□ PROTEIN SYNTHESIS

REPLICATION□ TRANSCRIPTION□ TRANSLATION

Exceptions among viruses – RNA to DNA
(retroviruses)

DNA Structure

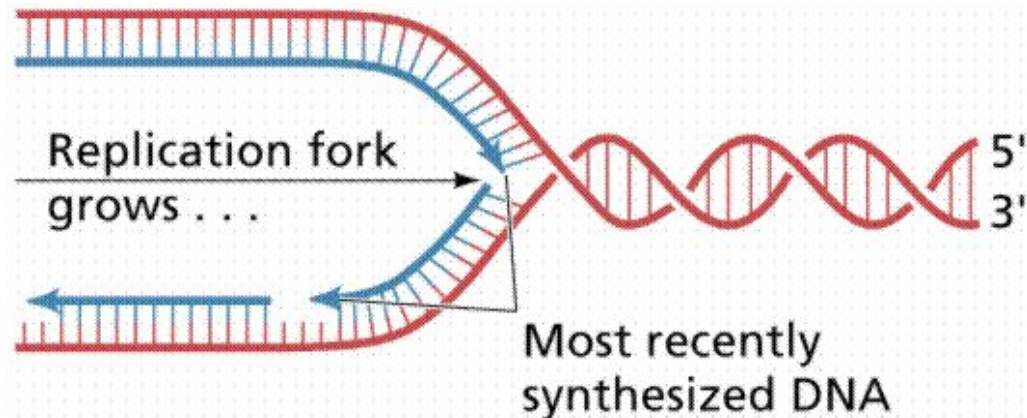
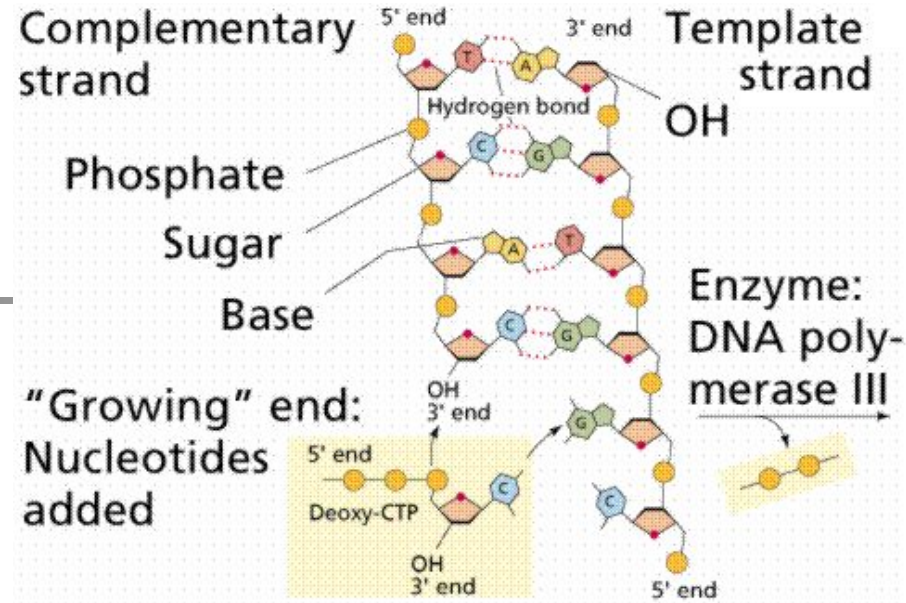
- **Double helix**
- **Antiparallel**
- **Nucleotide**
- ◆ **Deoxyribose**
- ◆ **Phosphate**
- ◆ **Nitrogen bases**
 - **Adenine**
 - **Thymine**
 - **Guanine**
 - **Cytosine**



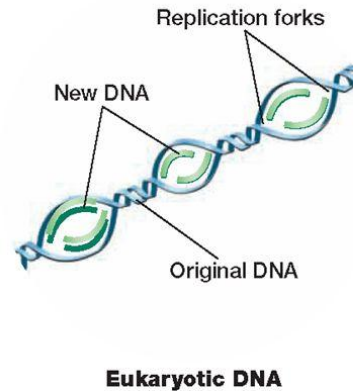
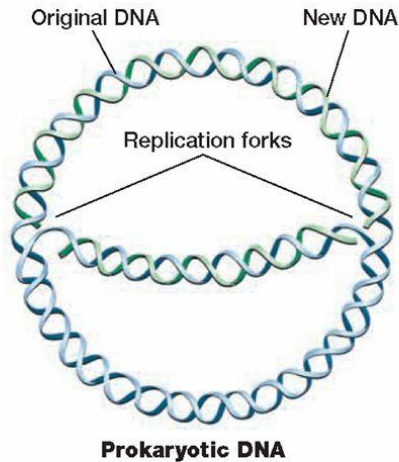
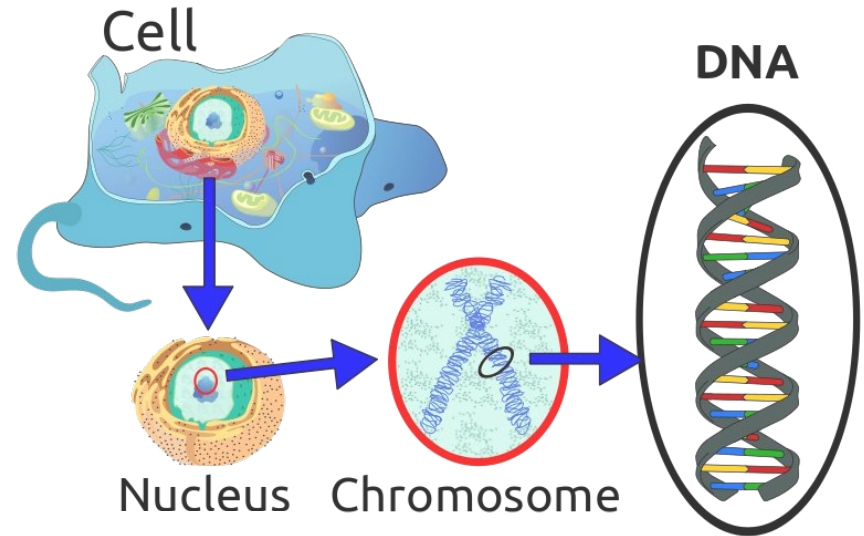
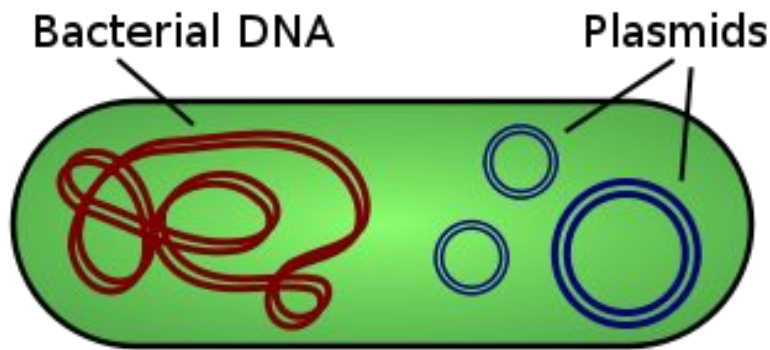
DNA

Replication

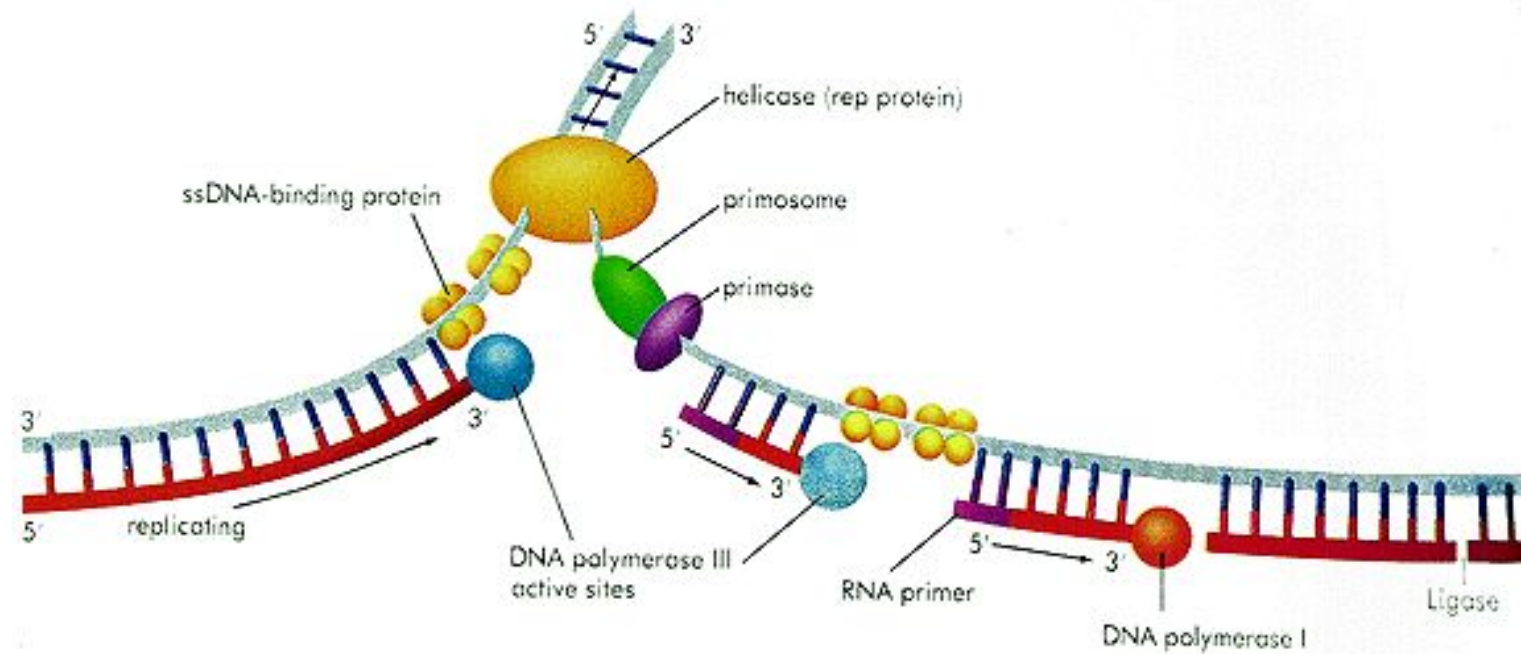
- **Replication** (in nucleus)
- **DNA** uncoils & splits
- **Reads** 3' to 5'
- **Assembles** 5' to 3'
- **4 types** of nucleotides
- **Okazaki** fragments in lagging strand



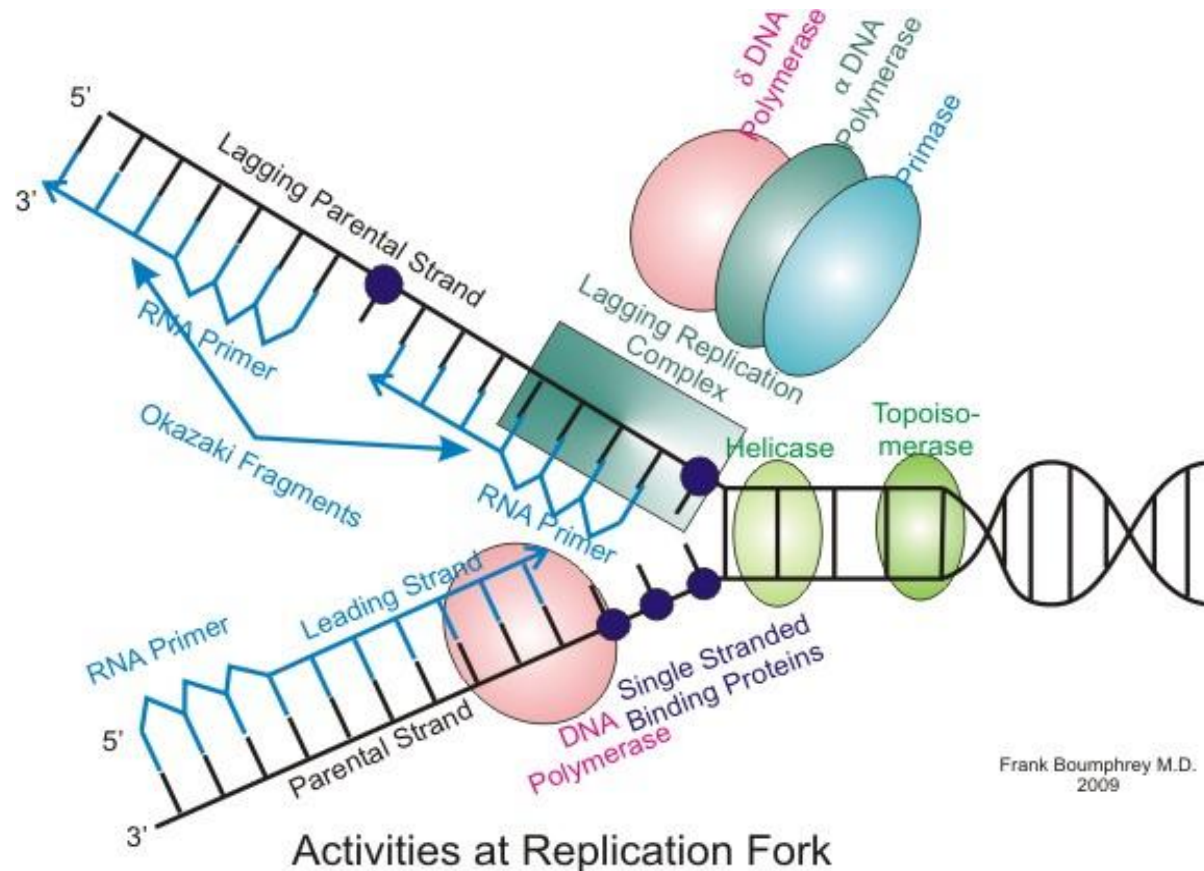
Prokaryotic vs. Eukaryotic



PROKARYOTIC REPLICATION

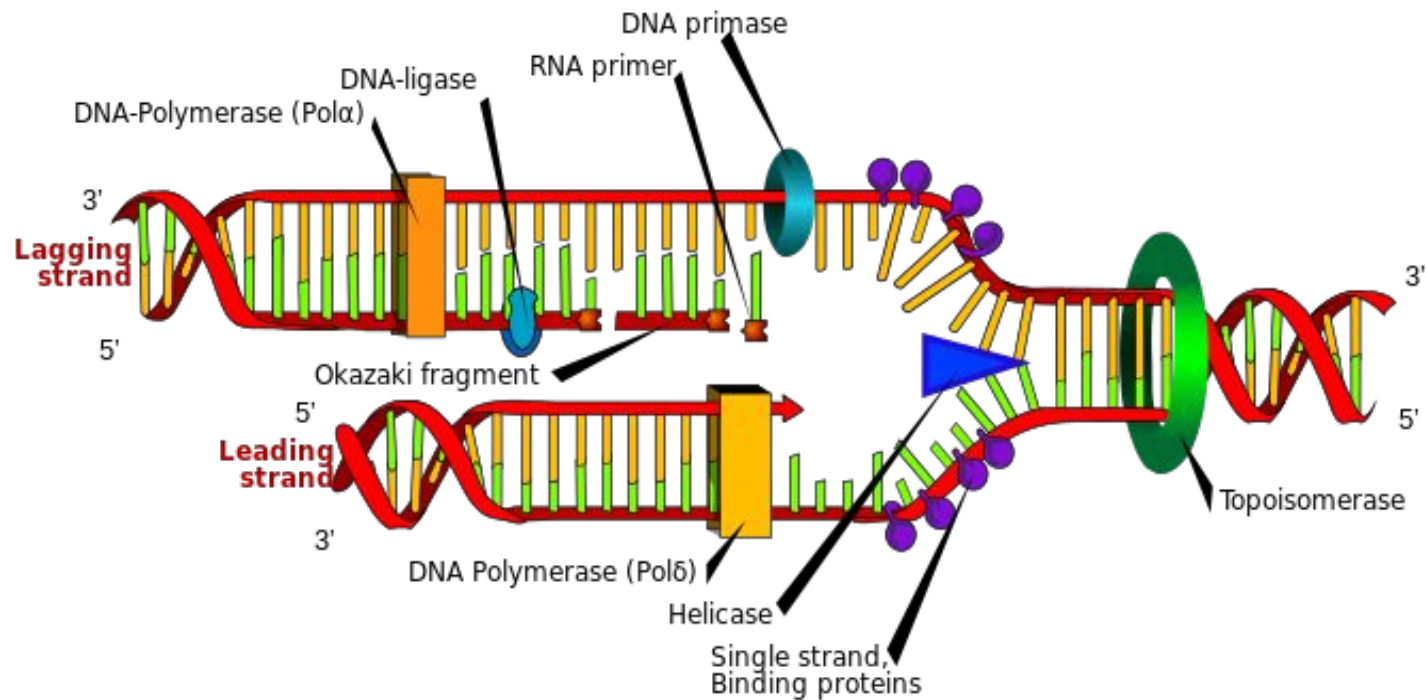


Eukaryotic replication



Frank Boumphrey M.D.
2009

ENZYMES FOR REPLICATION

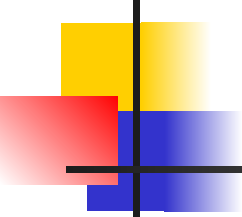




DNA Repair

- Genes encode proteins that **correct mistakes** in DNA caused by incorrect copying during replication and environmental factors such as by-products of metabolism, exposure to ultraviolet light or mutagens
- The **DNA repair process** must operate constantly to correct any damage to the DNA as soon as it occurs

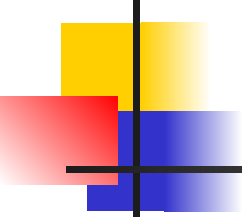
Gene Expression



- **Transcription** – DNA is template for making RNA (in nucleus)
- **Translation (protein synthesis)** - in cytoplasm at the ribosome. **m-RNA** has blueprint, **t-RNA** transfers amino acids, and **Ribosome (r-RNA)** allows T-RNA to attach to M-RNA at appropriate site

Making RNA from DNA template





Types of RNA

- **Differences between RNA & DNA**
 - ❖ RNA is single strand - DNA is double strand
 - ❖ RNA has Ribose – DNA has Deoxyribose
 - ❖ RNA has Uracil – DNA has Thymine
- **Messenger RNA** – carries blueprint from nucleus to cytoplasm – acts as template
- **Transfer RNA** – brings amino acids
- **Ribosomal RNA** – part of ribosome (also protein) reads code and allows m-RNA and t-RNA to connect

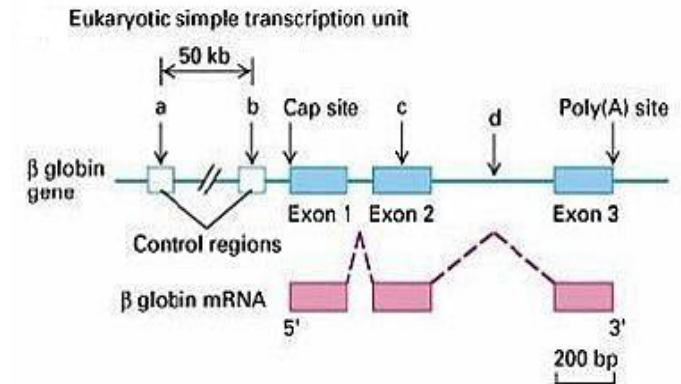
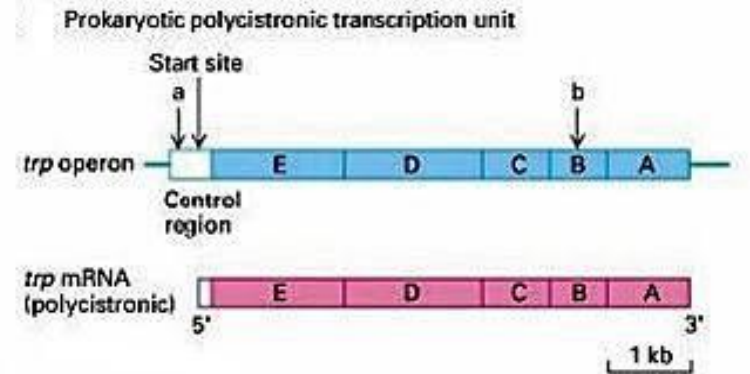
Polycistronic mRNA vs. Monocistronic mRNA

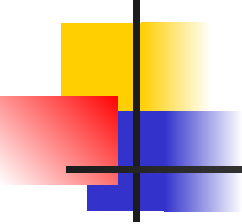
Polycistronic mRNA

- contains for more than one cistron
- codes for fore than one protein
- is transcribed from more than one gene (cistron) and has as many initiation and termination codes
- is present in prokaryotes

Monocistronic mRNA

- contains codons of a single cistron
- codes for a single protein
- is transcribed from a single gene (cistron) and has one initiation and termination codon
- is present in eukaryotes

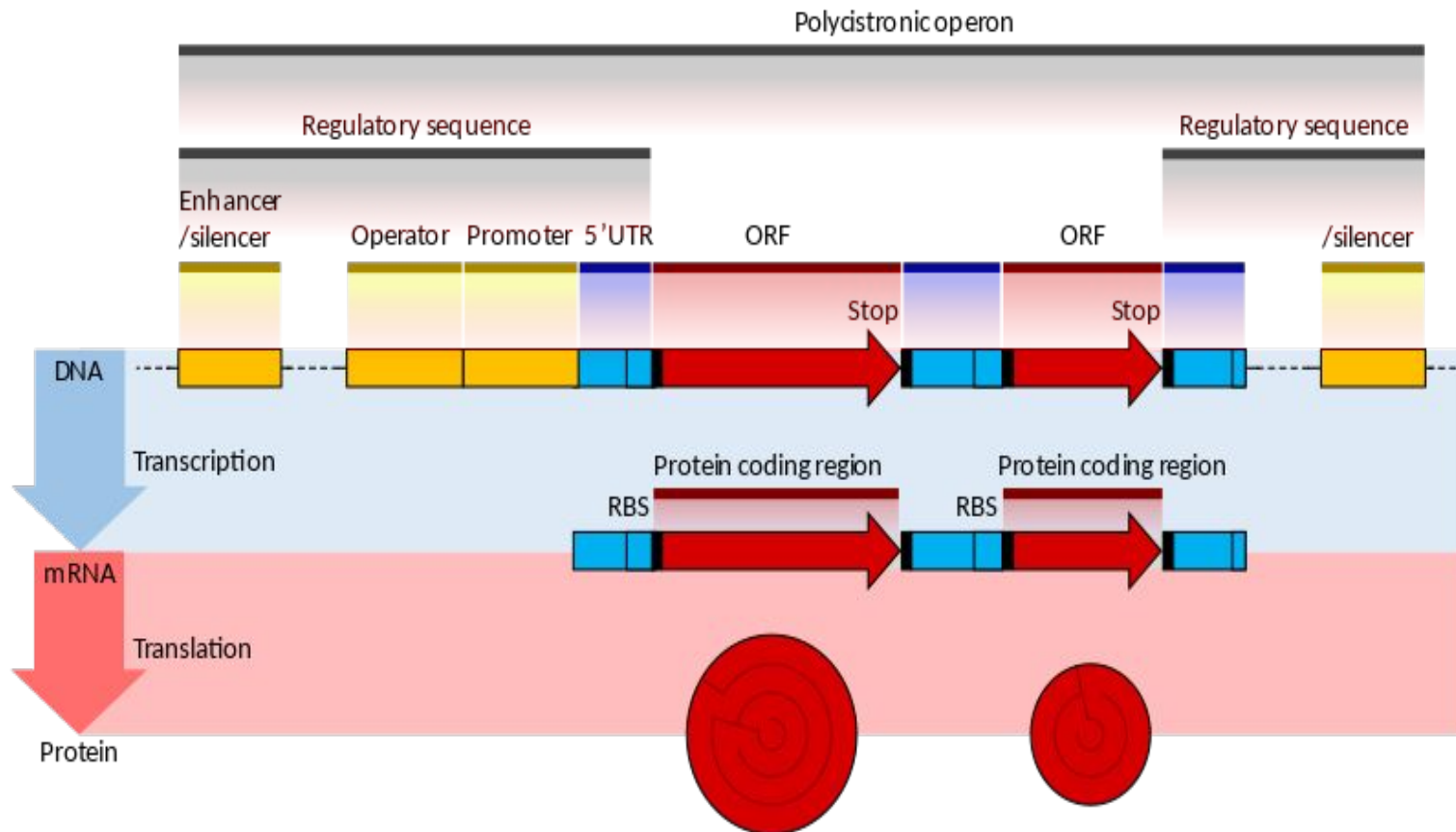




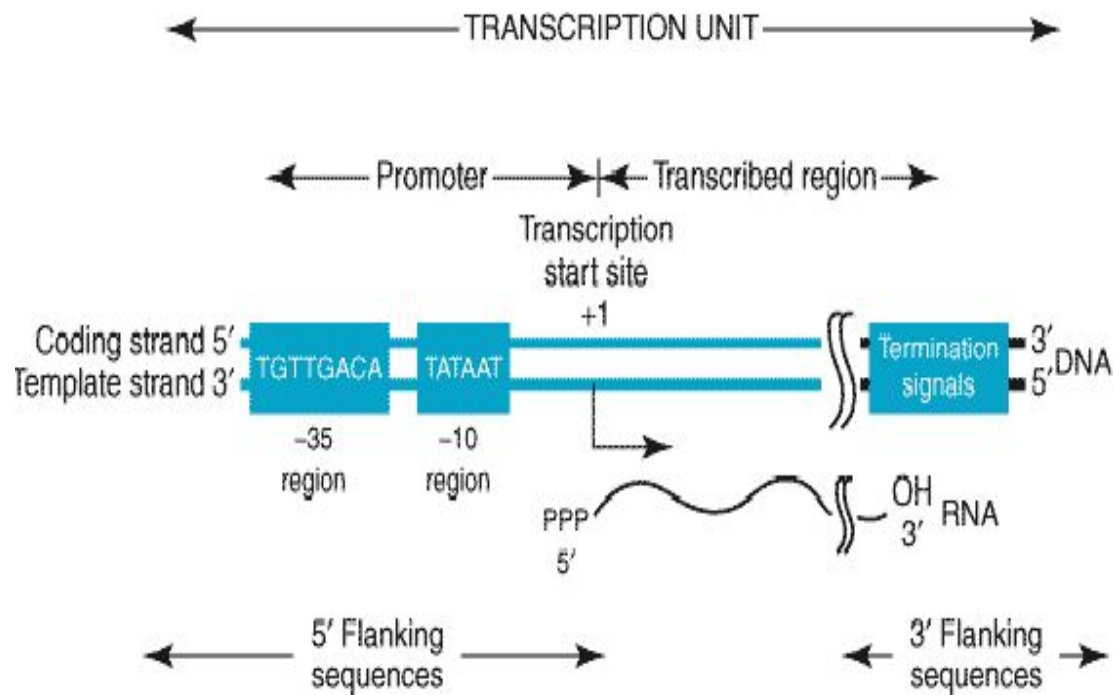
Promoters

- **region of DNA that initiates transcription of a particular gene**
- **located near the genes they transcribe, on the same strand and upstream on the DNA (towards the 3' region of the anti-sense strand**
- **also called template strand and non-coding strand)**

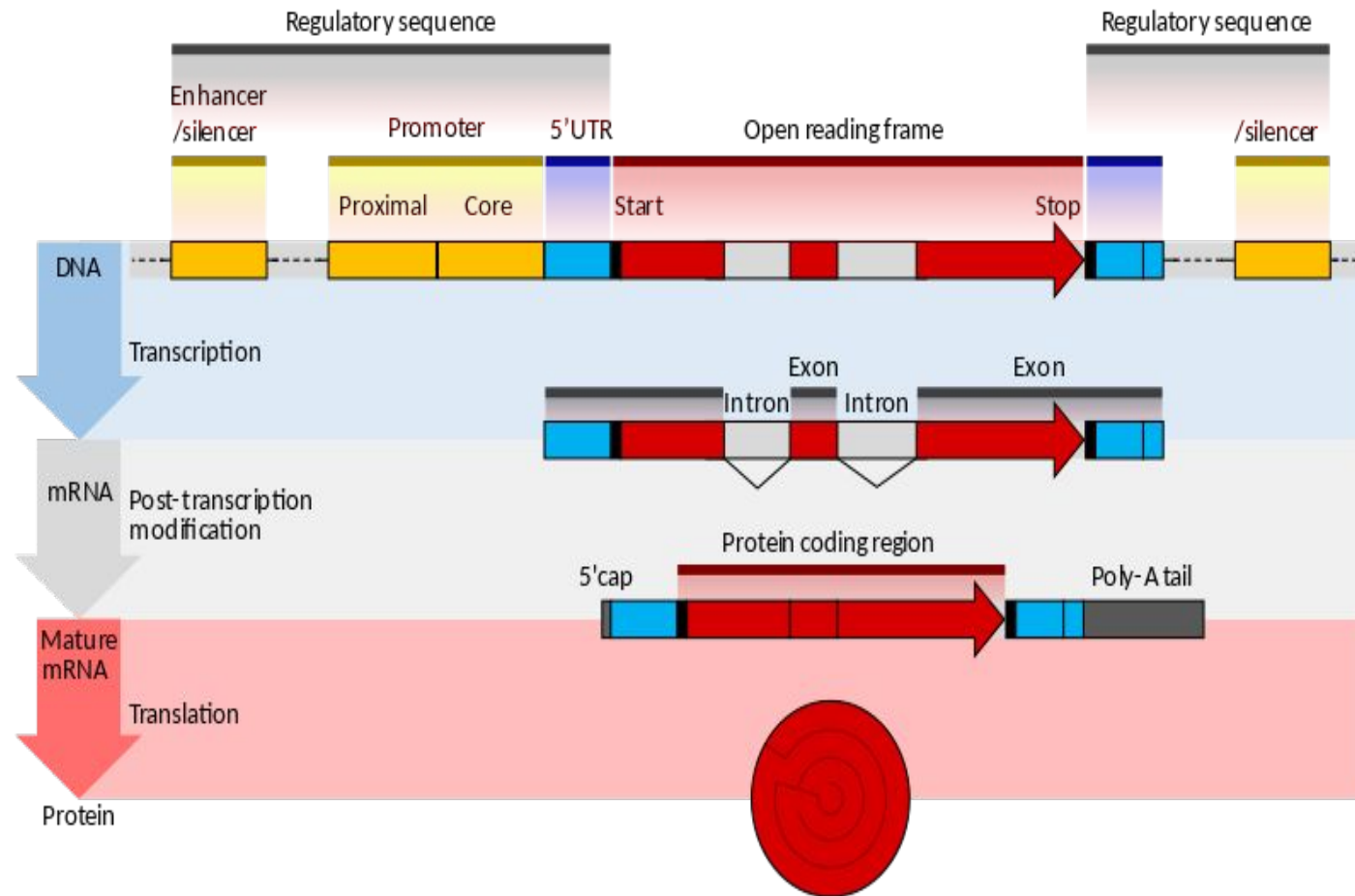
Prokaryotic Gene Organization



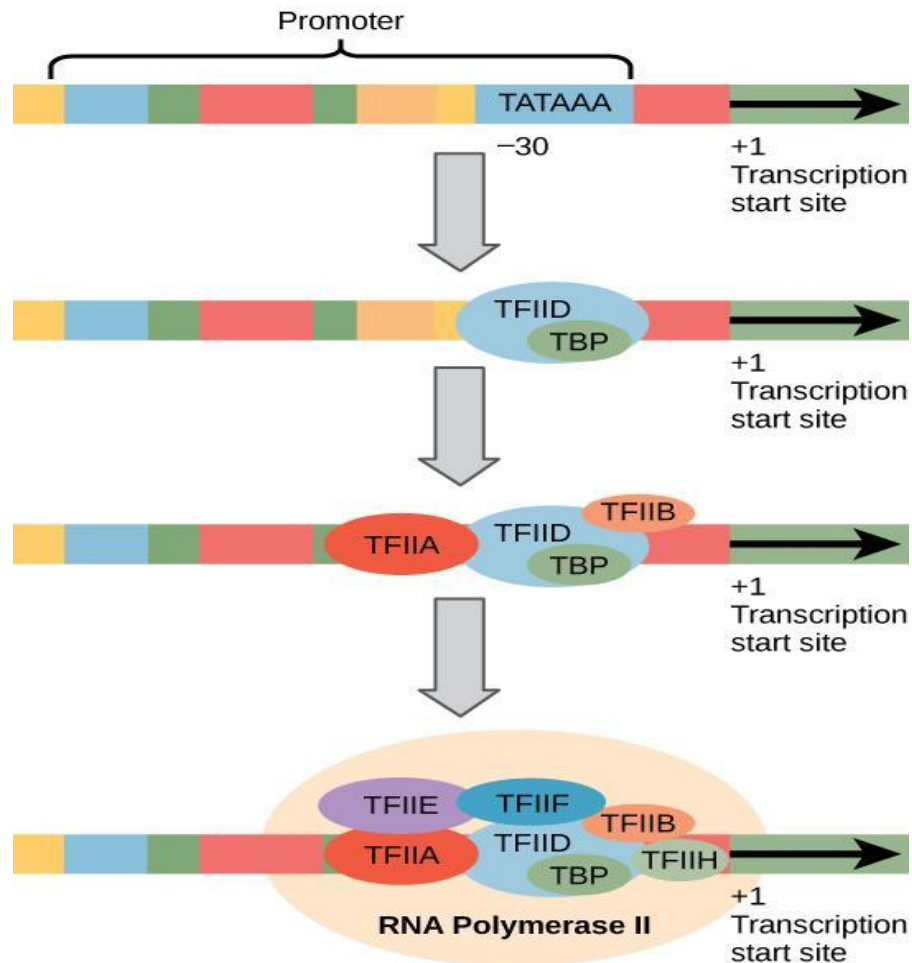
Prokaryotic Promoter

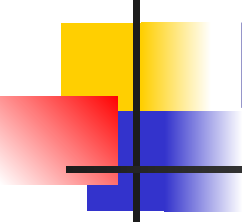


Eukaryotic Gene Organization



Eukaryotic promoter



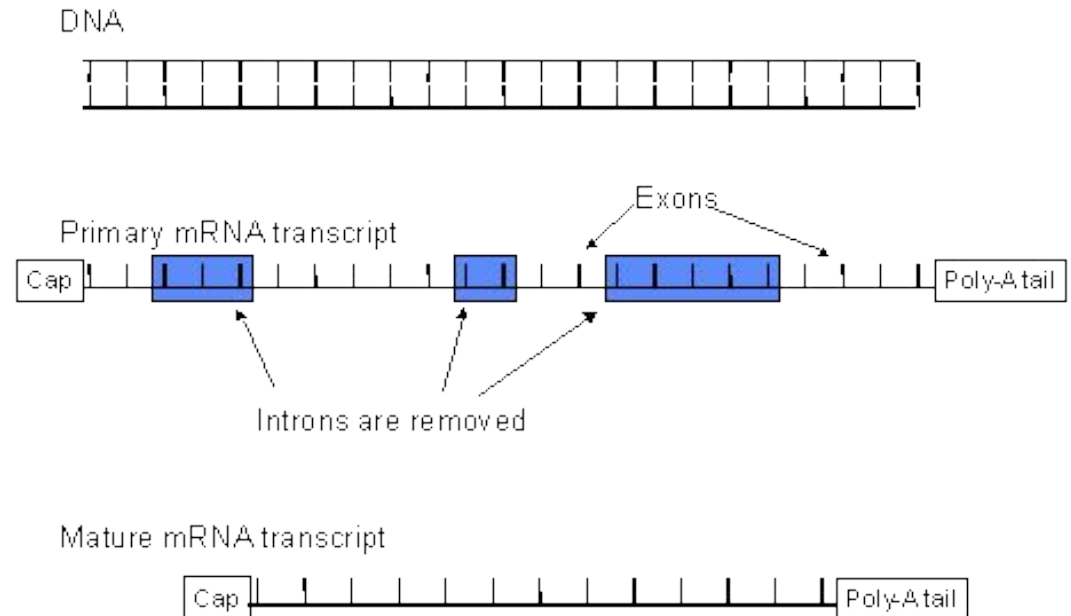


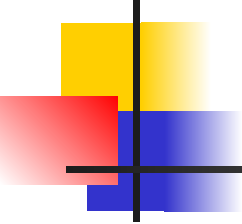
RNA Processing

- **mRNA in prokaryotic cells is to function after transcription but in eukaryotic cells it is modified after transcription**
- **RNA Processing includes 5' capping for RNA stabilization and ribosome binding; splicing for removing intron sequence and 3' polyadenylation for protecting mRNA from 3' exonuclease, extending the half life of mRNA**
- **Eukaryotic pre-mRNA is converted into**

Post-transcription Modifications

- **Introns and exons** at transcription
- **Introns** removed
- **Exons** are coding pieces for protein synthesis
- **Cap and PolyA tail** are added

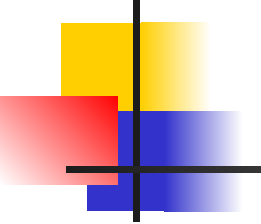




Universal Genetic Code

- **Special start codon (AUG) and three stop codons (UAA, UAG and UGA)**
- **Many codons may code for same amino acid**
- **Third position of the codon, it is more likely the nucleotide is different but it still may code for same amino acid (wobble)**

Universal Code (Codon = Amino Acid)



	U	C	A	G	
First position (5' end)	U UUU Phe UUC UUA Leu UUG	UCU Ser UCC UCA UCG	UAU Tyr UAC UAA Stop UAG Stop	UGU Cys UGC UGA Stop UGG Trp	U C A G
	C CUU Leu CUC CUA CUG	CCU Pro CCC CCA CCG	CAU His CAC CAA Gln CAG	CGU Arg CGC CGA CGG	U C A G
	A AUU Ile AUC AUA AUG	ACU Thr ACC ACA ACG	AAU Asn AAC AAA Lys AAG	AGU Ser AGC AGA Arg AGG	U C A G
	G GUU Val GUC GUA GUG	GCU Ala GCC GCA GCG	GAU Asp GAC GAA Glu GAG	GGU Gly GGC GGA GGG	U C A G
					Third position (3' end)

Amino acid names:

Ala = alanine

Arg = arginine

Asn = asparagine

Asp = aspartate

Cys = cysteine

Gln = glutamine

Glu = glutamate

Gly = glycine

His = histidine

Ile = isoleucine

Leu = leucine

Lys = lysine

Met = methionine

Phe = phenylalanine

Pro = proline

Ser = serine

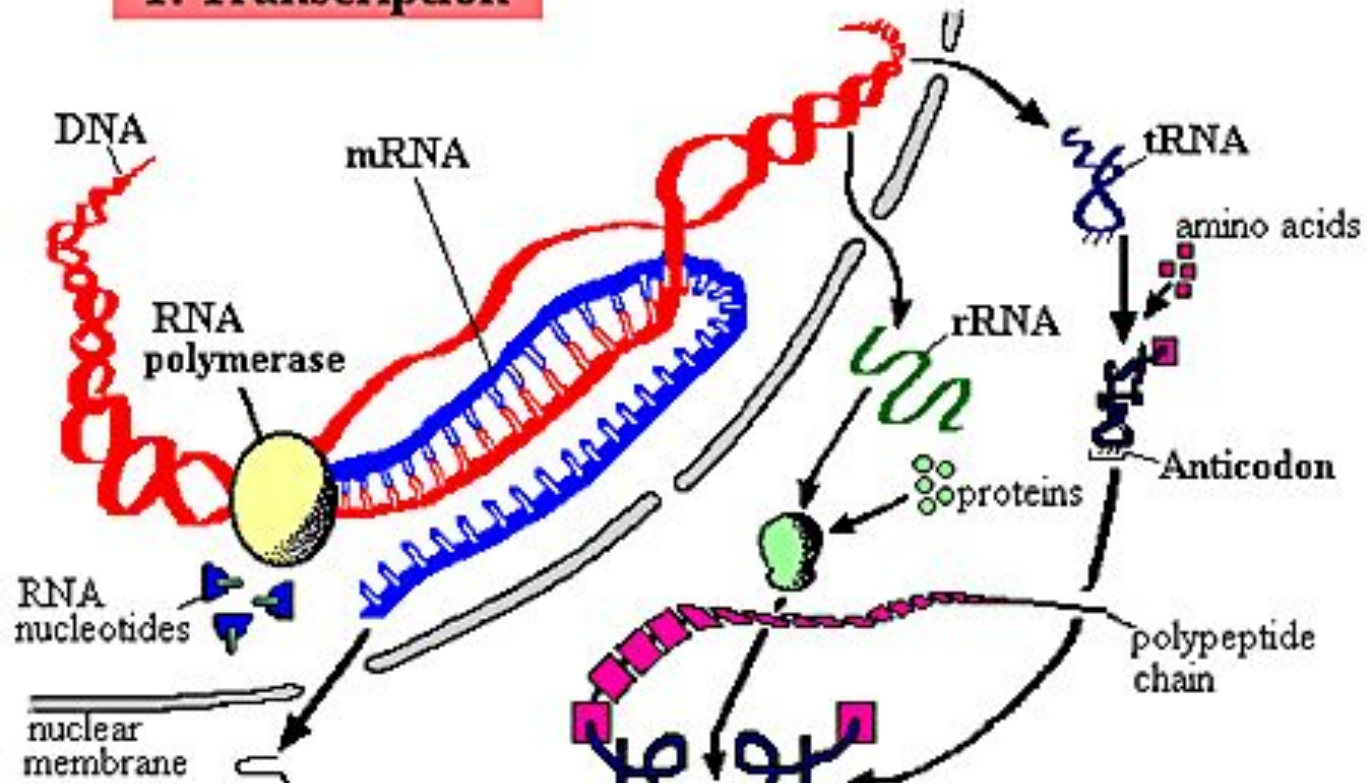
Thr = threonine

Trp = tryptophan

Tyr = Tyrosine

Val = valine

1. Transcription



2. Translation

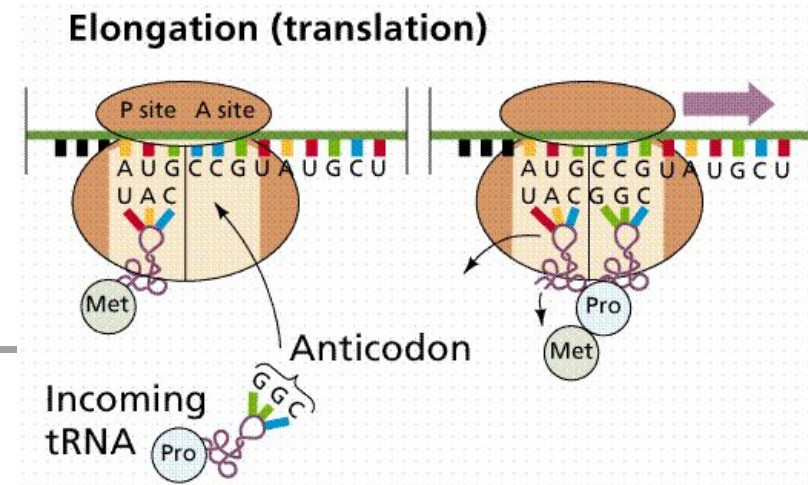
Protein synthesis

Translation (Protein Synthesis)

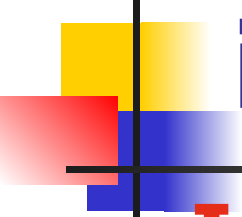
The steps of translation: Eukaryotic

- 1. Initiation:** mRNA enters the cytoplasm and becomes associated with ribosomes (rRNA + proteins). tRNAs, each carrying a specific amino acid, pair up with the mRNA codons inside the ribosomes. Base pairing (A-U, G-C) between mRNA codons and tRNA anticodons determines the order of amino acids in a protein.
- 2. Elongation:** addition of amino acids one-by-one:
As the ribosome moves along the mRNA, each tRNA transfers its amino acid to the growing protein chain, producing the protein
- 3. Termination:** when the ribosomes hits a stop codon - UAA, UGA, or UAG - the ribosome falls apart

Note: The same mRNA may be used hundreds of times



Control of Gene Expression in Prokaryotes

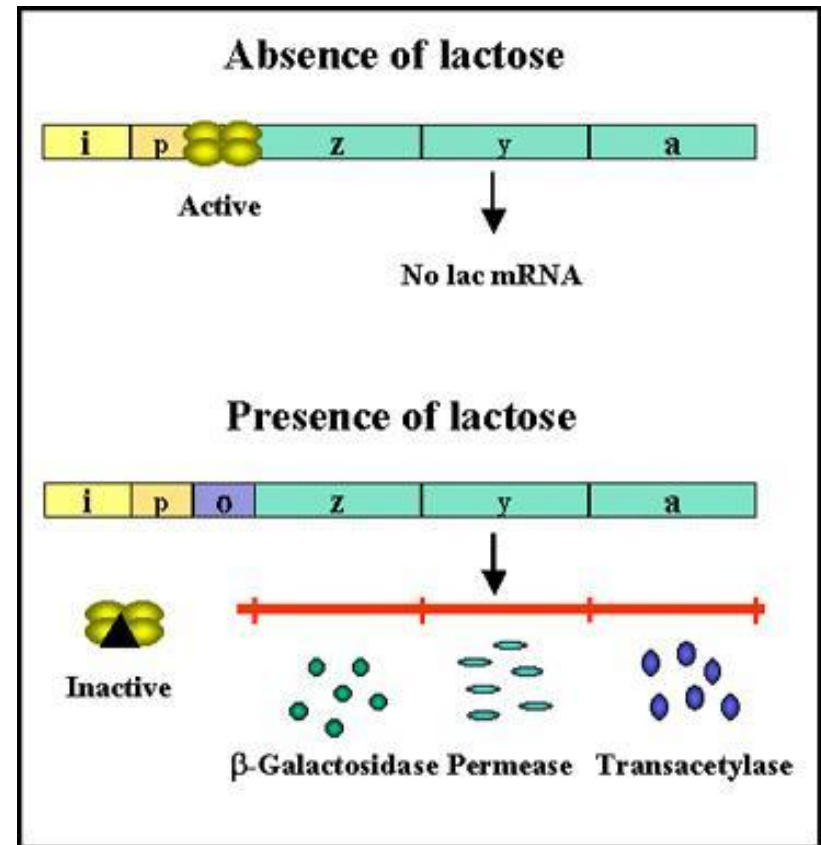


- Important for single celled organisms who depend on environment for all activities
- Bacteria use **operons** - many functional-related genes are clustered and transcribed under the same types of regulation
- **Lac & Trp Operons** - examples of prokaryotic gene regulation

Lac Operon

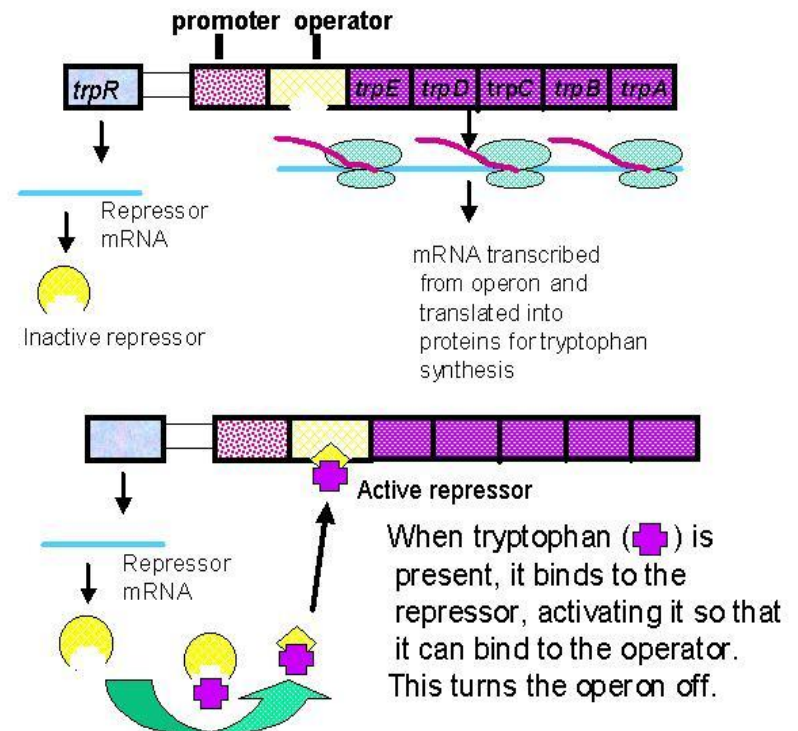
The **genes** that code for the enzymes needed for lactose catabolism are clustered on the same chromosome in what is called the **Lac Operon**

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**
- This is a **repressable operon** where the operon are **turned off** in the **presence of a substance**

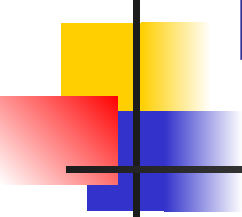




Regulatory Components in Eukaryotes

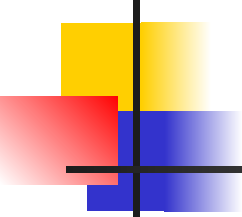
- **Enhancers** - short regions of DNA that can be bound with proteins to *promote expression* of a distal or a proximal gene.
- **Promoters** - proximal DNA sequences that binds to RNA polymerase *for regulating gene expression* .
- **TATA Box** - binds to transcription factor *for regulating gene expression* , usually within 30bp of the transcription start site

Control of Gene Expression in Eukaryotes



- **Transcriptional Control**
- **Post transcriptional Control – assembling proteins**
- **Cell differentiation and specialization**
- **Turning genes “on” and “off”**
- **Chemical Signals – Hormones**
- **Chemical Modifications**
- **Relocation of DNA – transposons**
- **Abnormal Expression of Genes**

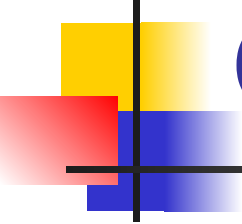
Nuclear vs Cytoplasmic DNA in Eukaryotic Cells



- **Nuclear DNA** – in chromosomes within the nucleus of the cell
- **Cytoplasmic or Organelle DNA** – in chloroplasts and mitochondria
 - **Chloroplast DNA (cpDNA)**
 - **Mitochondrial DNA (mtDNA)**

Features:

- **Maternal inheritance**
- **Resemble prokaryotic DNA**
- **Slow accumulation of mutations**

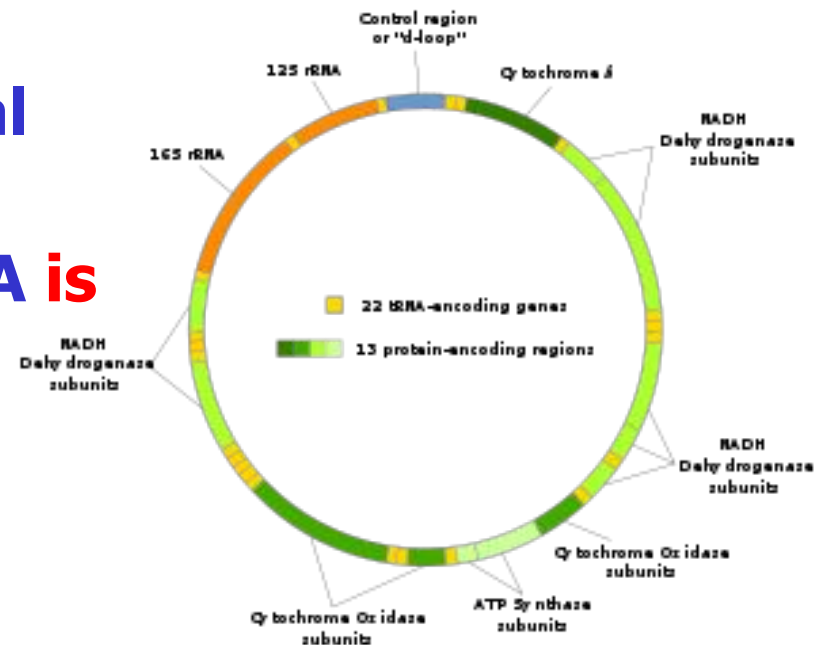


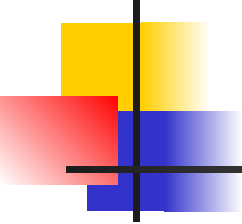
Organelle DNA

- **Mitochondria and Chloroplasts have DNA similar to Prokaryotic cells**
- **It is believed that these organelles were once independent prokaryotes who took up residence and formed a mutualistic relationship**
- **They are involved in energy transfer-photosynthesis & respiration**

MITOCHONDRIAL INHERITANCE

- **The inheritance of a trait encoded in the mitochondrial genome**
- **Mitochondrial DNA or mtDNA is inherited from the mother**
- **The mtDNA is circular and resembles prokaryotic DNA**
- **The mitochondria are responsible for energy production – cellular respiration**





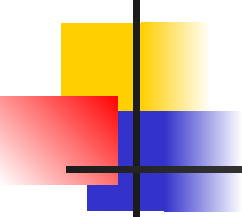
Mutations

- **Mutation** – any change in the DNA blueprint for making protein or RNA
- **Gene mutation**
- **Chromosomal mutation**
- **Agents causing mutations** – radiation, chemicals, excess heat



Genetic Disorders

- **Nondisjunction** – extra or missing chromosomes as Down's Syndrome
- **Trinucleotide repeats** – triplet nucleotides repeated too often as Huntington's
- **Defective genes** – does not produce correct protein as sickle cell anemia (A & T traded places)
- **Human genetic disorders** – can be dominant, recessive, sex-linked, epistatic, variable expressed

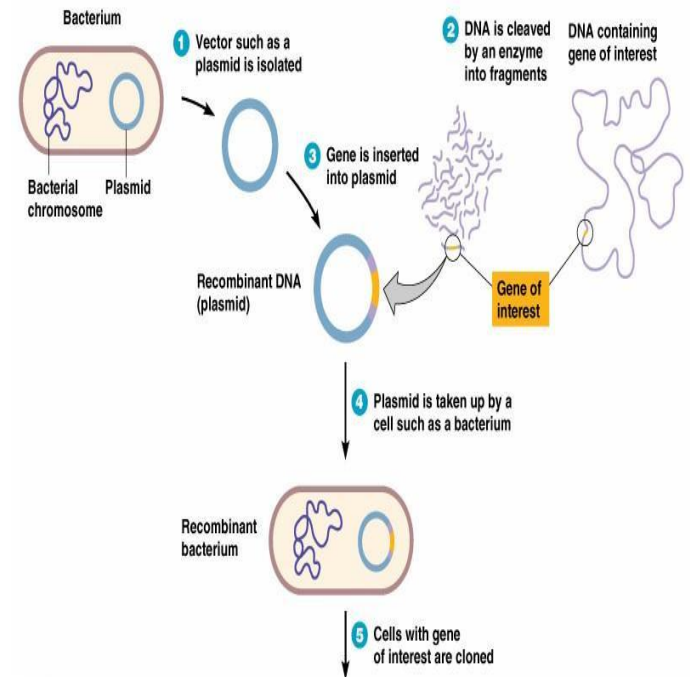
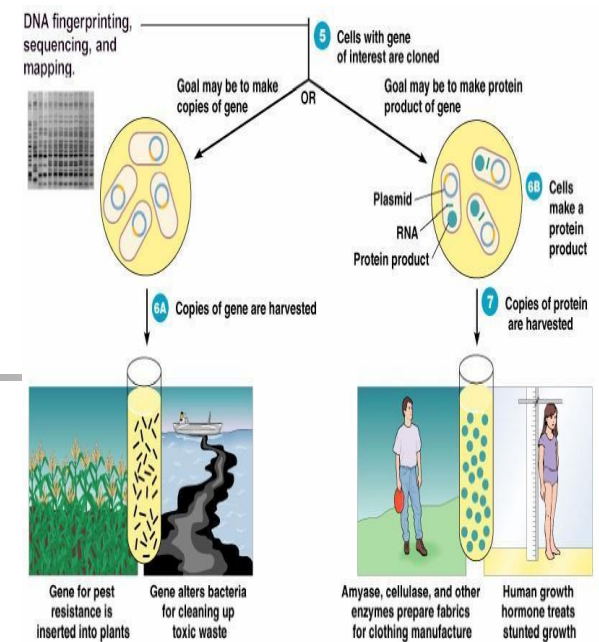


Biotechnology

- Technology used to **manipulate DNA**
- Procedures often called **genetic engineering**
- **Recombinant DNA** - DNA from two sources
- **Transgenic** individuals have DNA from another organism
- Often involve putting genes into viruses or bacteria.
- **Vectors** are the pieces of DNA used to transfer genes into a **host cell** – often plasmids of bacteria

Overview of Biotechnology

- identifying – recognizing desired DNA fragment or plasmid using **radioactive probes**
- cutting DNA - using desired **restriction enzymes** or “enzymatic scissors”
- making hybrids of DNA using **Hybridization techniques**
- Storing, analyzing and using **clones in biotechnology processes**





Basic Tools of DNA Technology

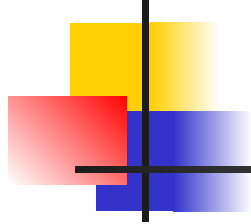
- **Identifying** desired DNA
- **Cutting** DNA with **Restriction Enzymes**
- **Inserting** DNA into **Vector** as **Plasmid**
- **Connecting** DNA pieces with **Ligase**
- **Inserting** **Vector** into **Host Cell** as **bacterium**
- **Cloning** desired **DNA** and **Vectors**
- **Storing** clones in **DNA Libraries**
- **Identifying** cloned genes with **Radioactive Probes**
- **Analyzing** DNA by cutting fragments and separating by **Electrophoresis**



Techniques

- **Cloning** within cells and with **PCR**
- **Storing** clones in **DNA Libraries**
- **Identifying** cloned genes with **Radioactive Probes**
- **Analyzing** DNA by cutting fragments and separating by **Electrophoresis/nucleic acid hybridization/DNA probes**
- **Transferring** DNA from gel by **Blotting**
- **Imaging** with **autoradiography**
- **DNA Sequencing** to determine exact sequence
- **Microassays** to analyze gene function

Cloning Methods Currently in Use



Traditional Restriction digestion cloning

Plasmids and inserts are digested with the same restriction enzymes and then ligated together.

Gateway Recombination

Regions of homology between insert DNA and plasmid are used for a recombination event that transfers the insert DNA into the plasmid.

Gibson assembly

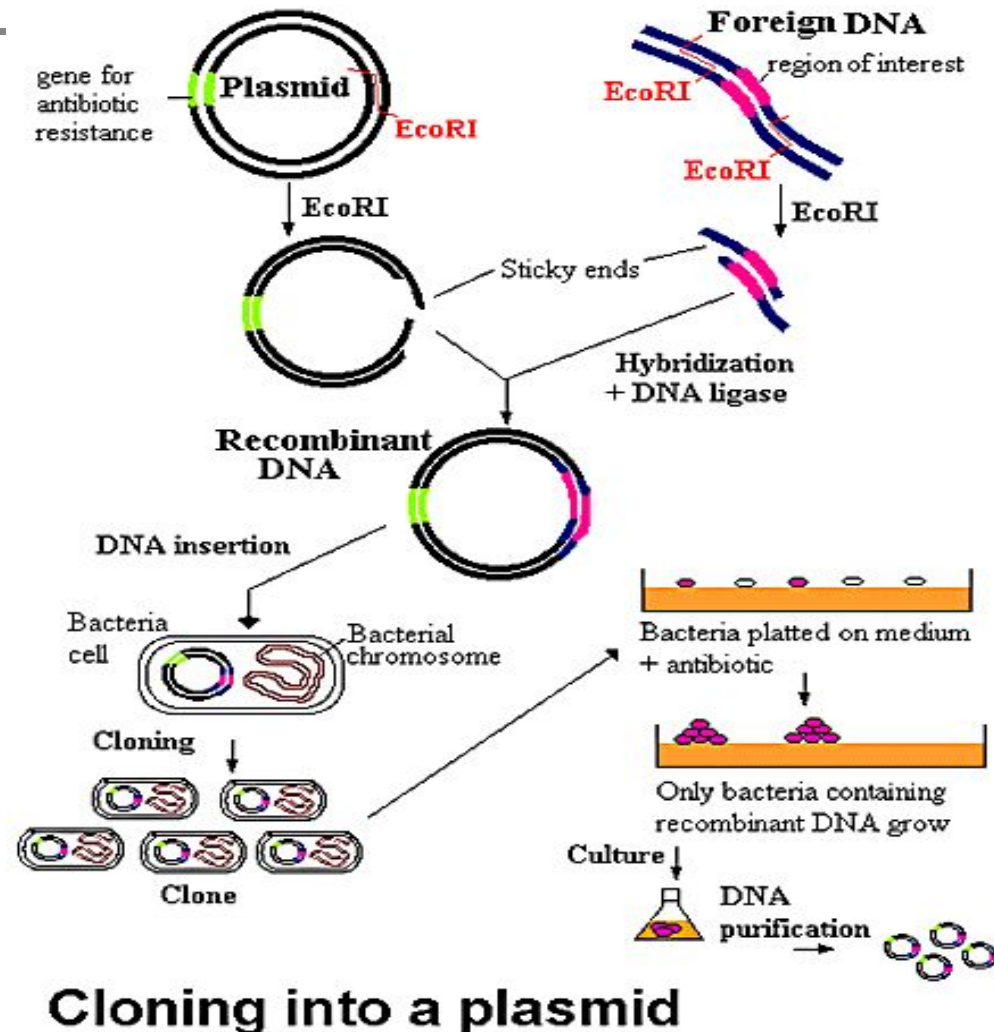
DNA fragments containing homologous overlapping ends are ligated together in one reaction.

TA cloning

Linearized plasmids engineered to have single T overhangs are ligated together with a PCR product insert. Most DNA polymerases leave an A overhang on PCR products, which allows them to base pair with the T overhangs on the TA cloning plasmid.

Cloning into A Plasmid

- Gene selection
- Plasmid selection
- Putting pieces together
- Insert into host bacteria
- Clone the bacteria

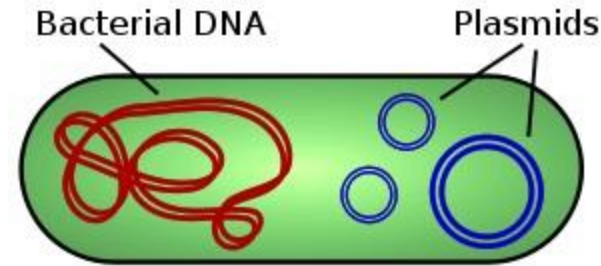




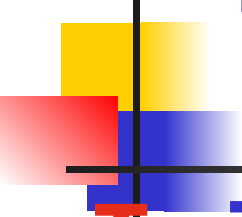
Gene Selection and cDNA – complementary DNA

- Eukaryotic genes contain introns but bacteria **do not contain** the necessary **enzymes to remove introns**
- Eukaryotic genes that are inserted into bacteria must be **inserted without introns.**
- Use *reverse transcriptase* (from retroviruses) and modified M-RNA to produce **cDNA with introns already**

Plasmid Selection and Isolation



- A small DNA molecule **that is physically separate from, and can replicate independently of, chromosomal DNA within a cell as a bacterium**
- **When used in genetic engineering – called vectors**
- **Several methods to isolate plasmid DNA from bacteria**



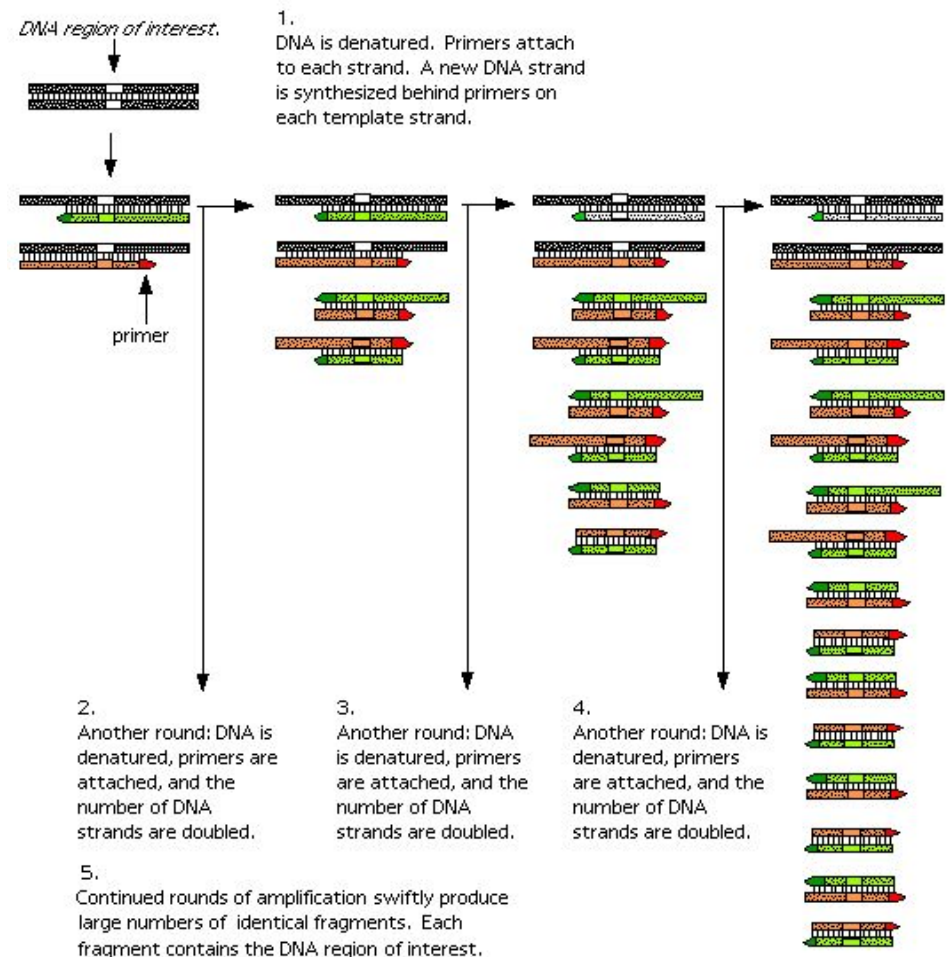
Molecular Cloning

- **To isolate a piece of DNA and amplify it via recombinant DNA technology.**
- **The DNA source can be genomic DNA, cDNA, or PCR amplified DNA fragments.**
- **These DNA pieces are cut by restriction enzymes to create compatible DNA ends with the vectors.**
- **They are then grown in E. coli for amplification.**

Polymerase Chain Reaction (PCR)

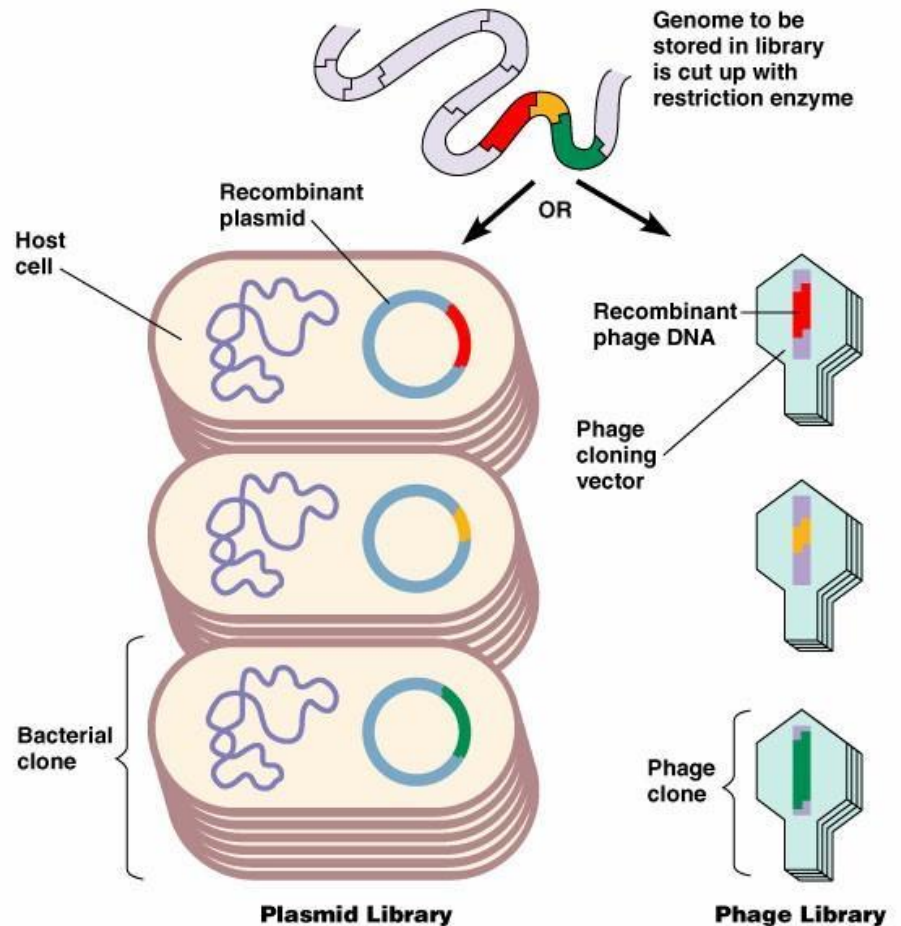
- **Technique for quickly making an unlimited number of copies of any piece of DNA**
- **Sometimes called "molecular photocopying"**

POLYMERASE CHAIN REACTION



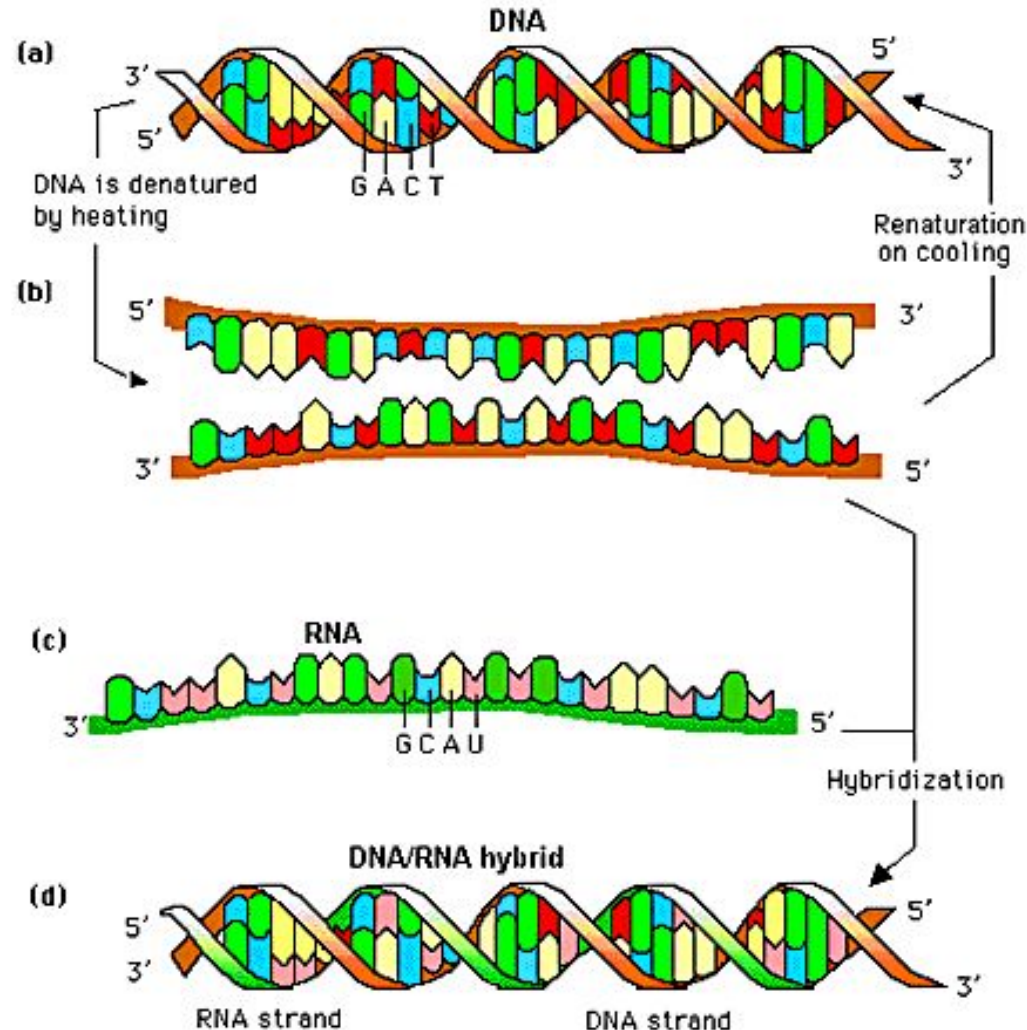
DNA Libraries

- **Genomic** – normal DNA
- **cDNA** – modified to remove introns
- Fragments stored
- Stored in plasmids or bacteriophages

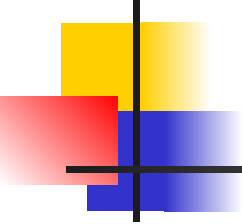


DNA Hybridization

- Base pairing of two single strands of DNA or RNA.
- Can be DNA-DNA, DNA-RNA
- Can be a radioactive probe



Nucleic Acid Hybridization

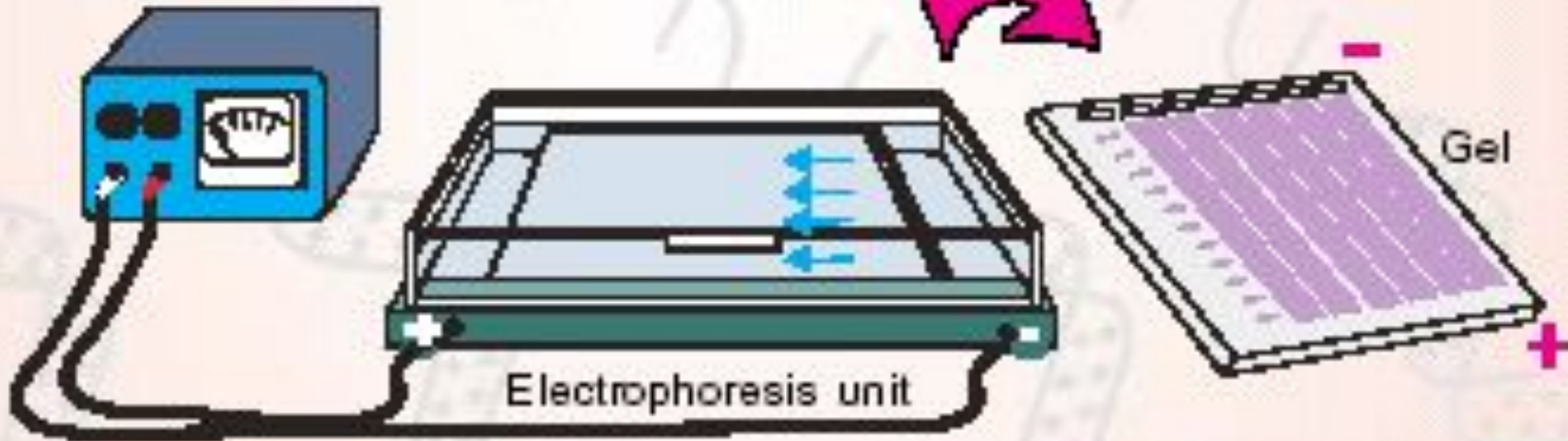


Radioactive Probes

- **short, radioactive strands of DNA**
- **will pair up with complementing strands of DNA**
- **fragments that contain the labeled pieces will show up on an x-ray film**

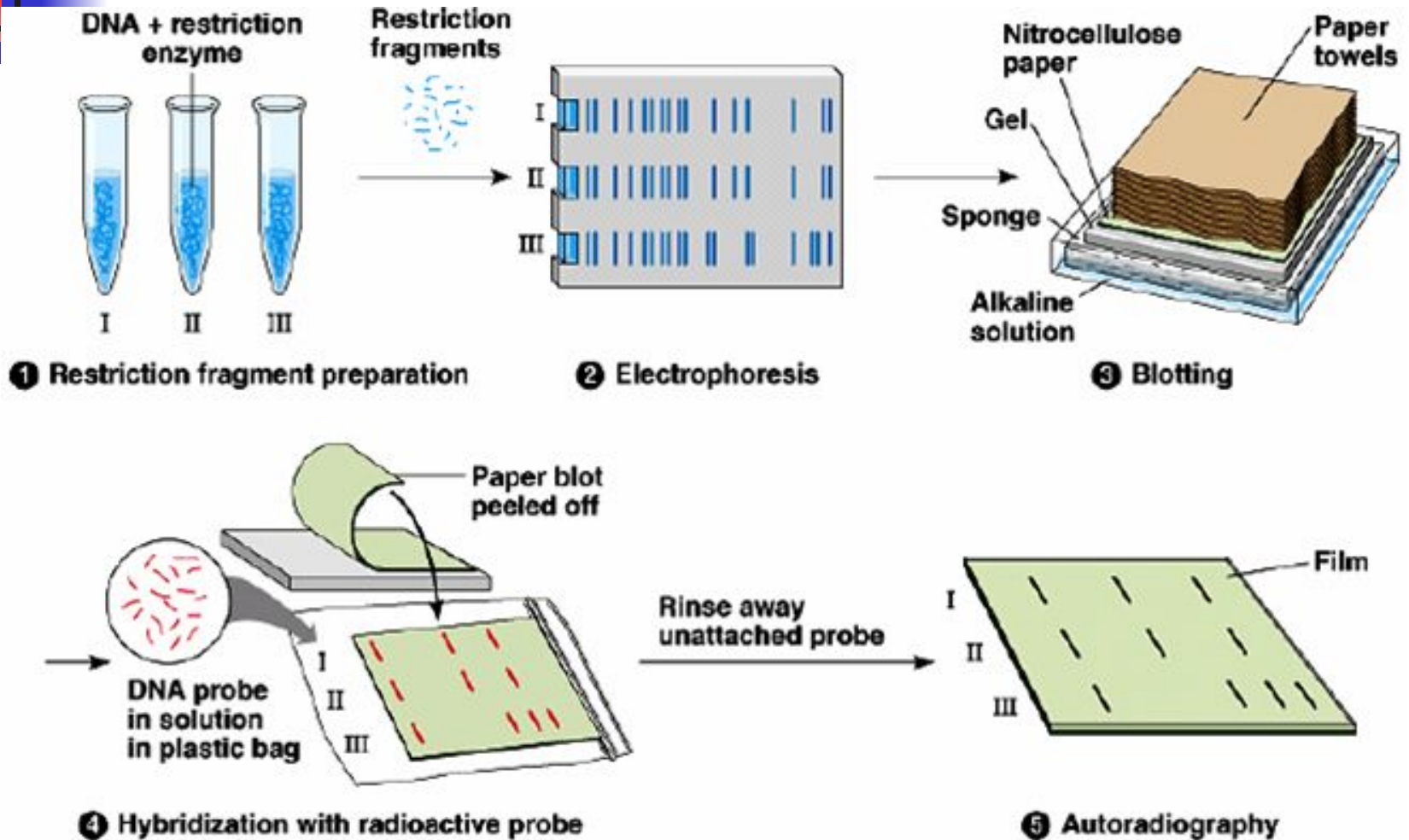
Electrophoresis

Power supply



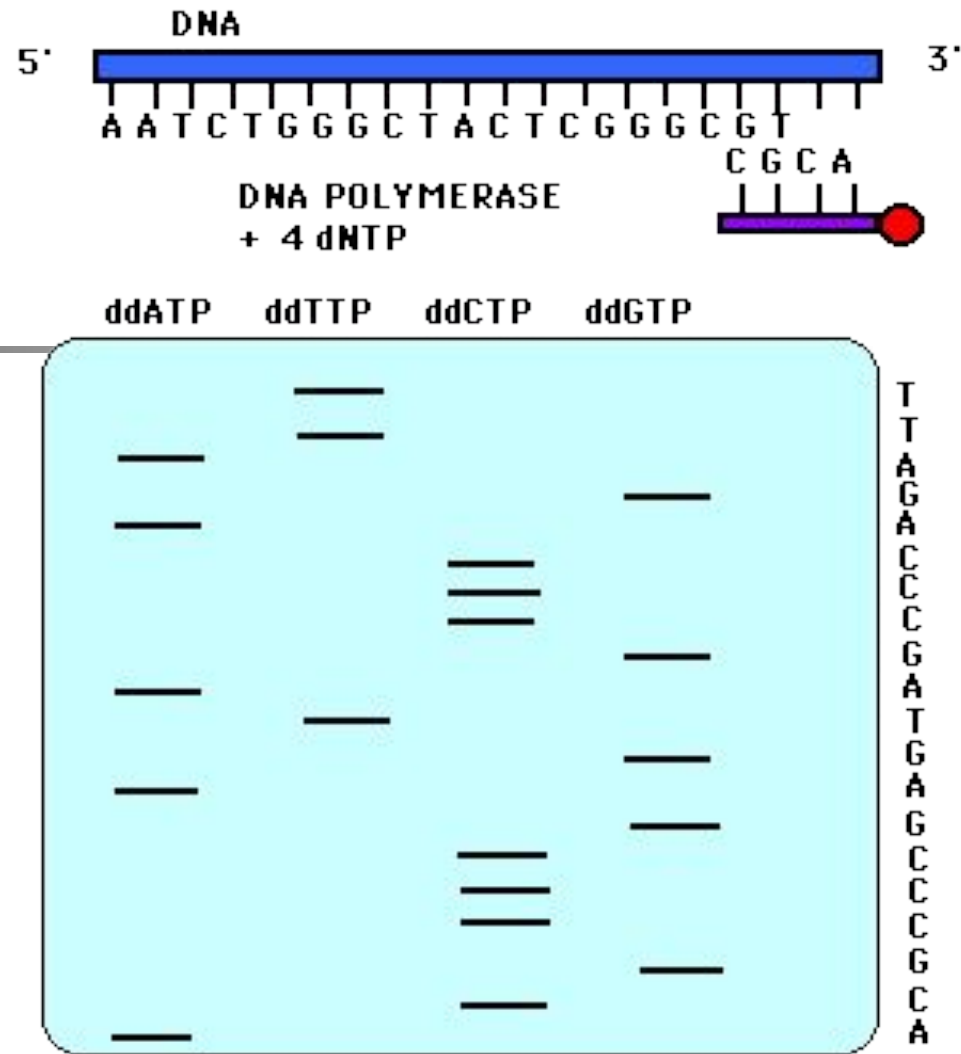
A process in which molecules (such as proteins, DNA, or RNA fragments) can be separated according to size and electrical charge by applying an electric current to them.

DNA Analysis



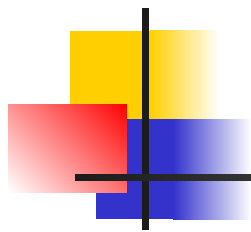
Sanger DNA Sequencing

- Determine the exact nucleotide sequence
- Columns for A, T, C, and G
- Read rows from top to bottom

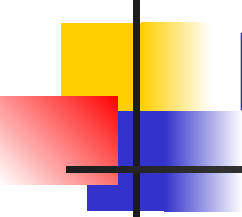


sequenced strand is 5' to 3'
ACGCCCGAGTAGCCCGAGATT

DNA Fingerprinting Techniques



- **RFLP** – Restriction Fragment Length Polymorphism (original)
- **PCR** – Polymerase Chain Reaction
- **VNTRs** – Variable Number Tandem Repeats
- **STRs** - Short Tandem Repeats
- **Ribosomal DNA analysis**
- **Y-chromosome analysis**

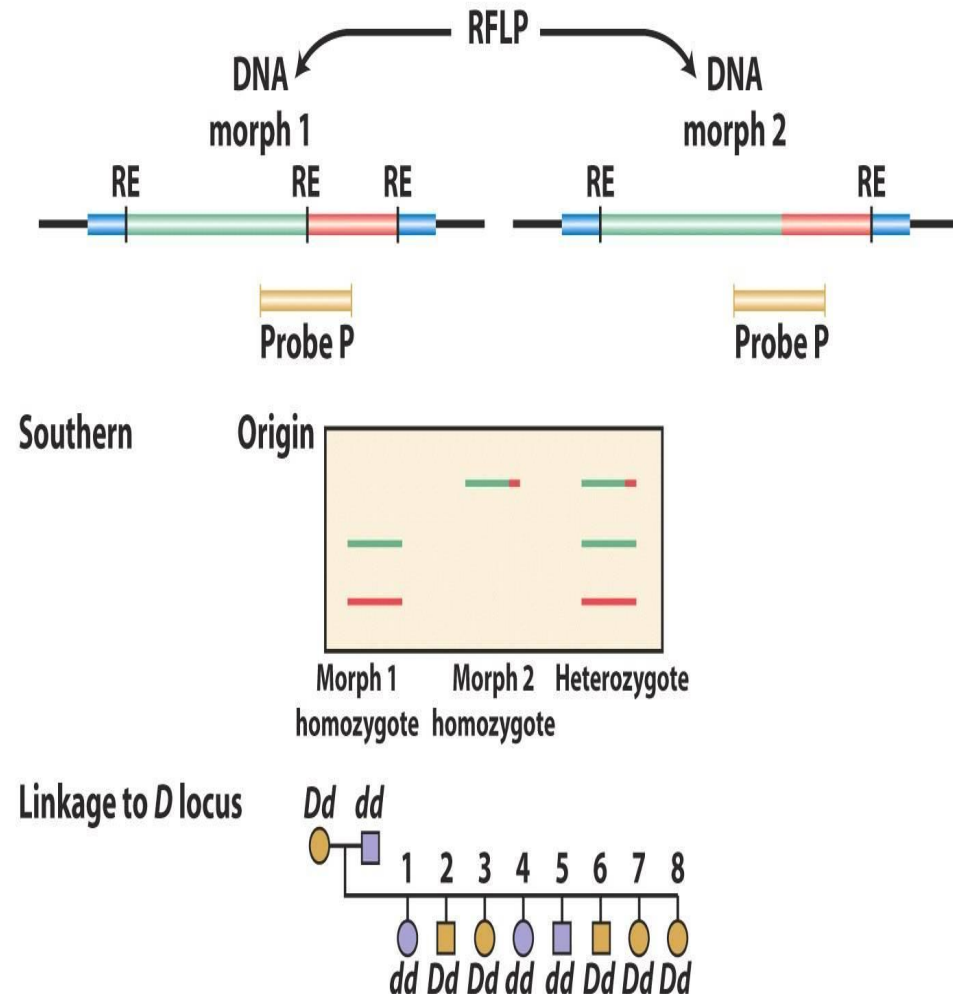


Restriction Fragment Length Polymorphism - RFLP

- The DNA of an organism is **cut up into fragments** using restriction enzymes.
- A **large number of short fragments** of DNA will be produced (RFLP's)
- **Electrophoresis** is a technique used to separate the DNA fragments according to their size
- **Uses-** identification of diseased genes including oncogenes, identification of viral infections, determining family relationships among individuals, and identifying tissue found at a crime scene

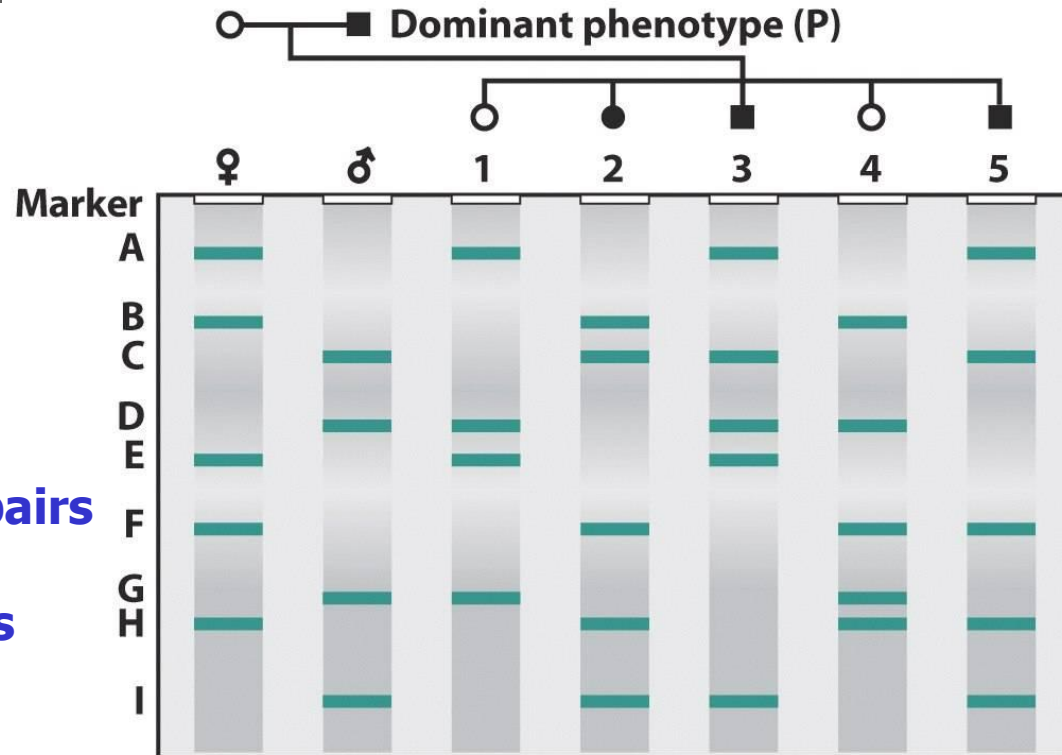
Restriction Fragment Length Polymorphisms (RFLP)

- **Genetic variations** at the site where a restriction enzyme cuts a piece of DNA
- Such variations **affect the size** of the resulting fragments
- These sequences can be **used as markers** on physical maps and linkage maps



VARIABLE NUMBER TANDAM REPEATS (VNTR's)

- Short nucleotide sequences
- Organized in clusters of tandem repeats
- VNTR = 14-100 base pairs
- SNR = 2- 10 base pairs



ANALYSIS EXAMPLES

F and H Always inherited together — linked?

A and B In progeny, always either A or B — “allelic”?

A and D Four combinations; A and D, A, D, or neither — unlinked?

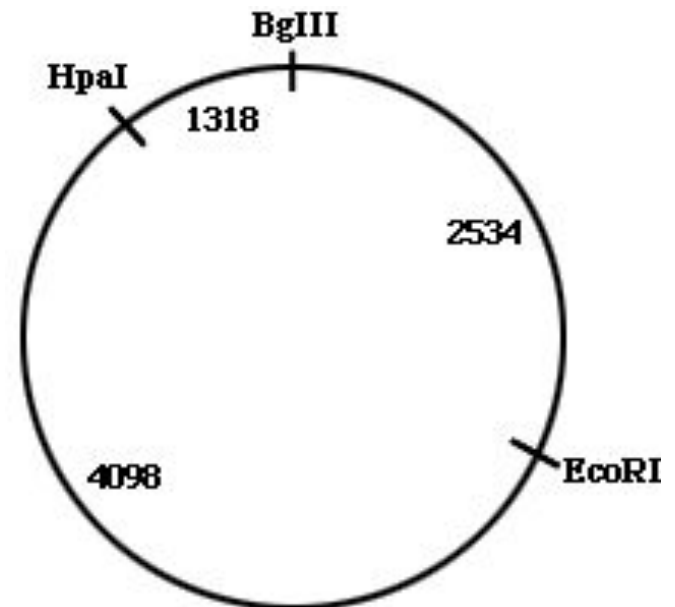
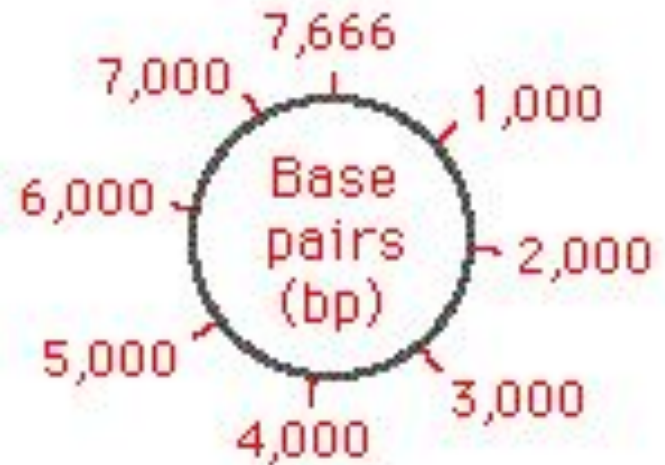
F, H, and E Always *either* F and H *or* E — closely linked in trans?

Allele P Possibly linked to I and C.

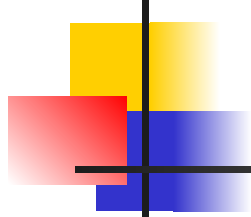
Restriction Mapping

- Description of **restriction enzyme cleavage sites** within a piece of DNA
- Use of **different restriction enzymes** to analyze and generate a **physical map** of genomes, genes, or other segments of DNA

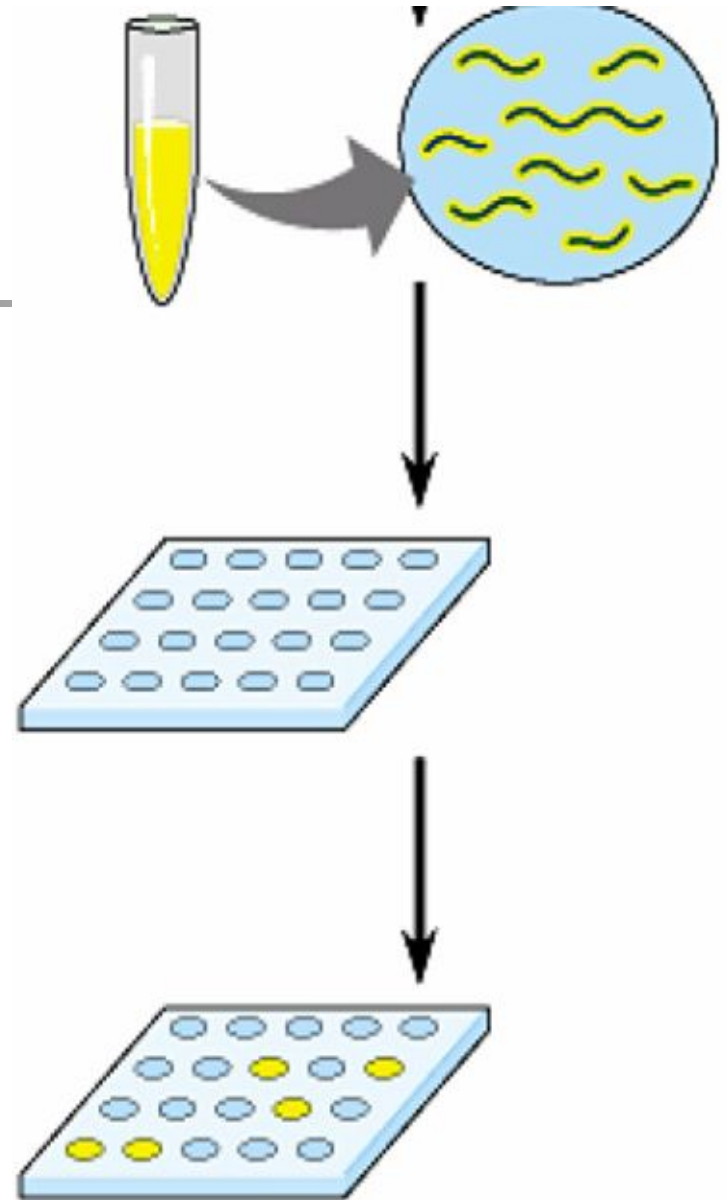
Circular pTut
DNA molecule

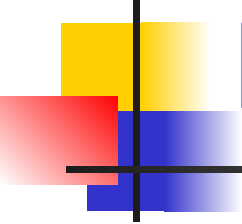


DNA Microassay



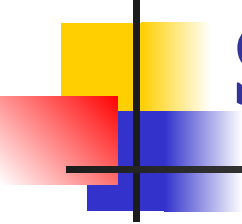
- studying **how large numbers of genes interact with each other**
- precisely apply tiny droplets containing **functional DNA** to glass slides
- attach **fluorescent labels to DNA** from the cell they are studying.
- labeled probes are allowed to bind to complementary DNA strands on the slides
- slides are put into a scanning microscope that can **measure the brightness** of each fluorescent dot
- **brightness** reveals how much of a specific DNA fragment is present, an indicator of **how active it is**





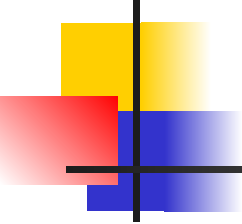
RNA-Seq

- **RNA-seq refers to the method of using Next Generation Sequencing (NGS) technology to measure a set of RNA levels**
- **NGS technology is an ultra-high-throughput technology to measure DNA sequences**
- **Advantages of RNA-seq over microarray include:**
 - 1) **Wider measurable range of expression levels**
 - 2) **Not dependent on known genome**
 - 3) **Free of hybridization artifacts**
 - 4) **Possibility of one platform for all applications**



Next Generation Sequencing Platforms

Platform	Chemistry	Read Length	Run Time	Gb/Run	Advantage	Disadvantage
454 GS Junior (Roche)	Pyro-sequencing	500	8 hrs.	0.04	Long Read Length	High error rate in homopolymer
454 GS FLX+ (Roche)	Pyro-sequencing	700	23 hrs.	0.7	Long Read Length	High error rate in homopolymer
HiSeq (Illumina)	Reversible Terminator	2*100	2 days (rapid mode)	120 (rapid mode)	High-throughput / cost	Short reads Long run time (normal mode)
SOLiD (Life)	Ligation	85	8 days	150	Low Error Rate	Short reads Long run time
Ion Proton (Life)	Proton Detection	200	2 hrs.	100	Short Run times	New*
PacBio RS	Real-time Sequencing	3000 (up to 15,000)	20 min	3	No PCR Longest Read Length	High Error Rate

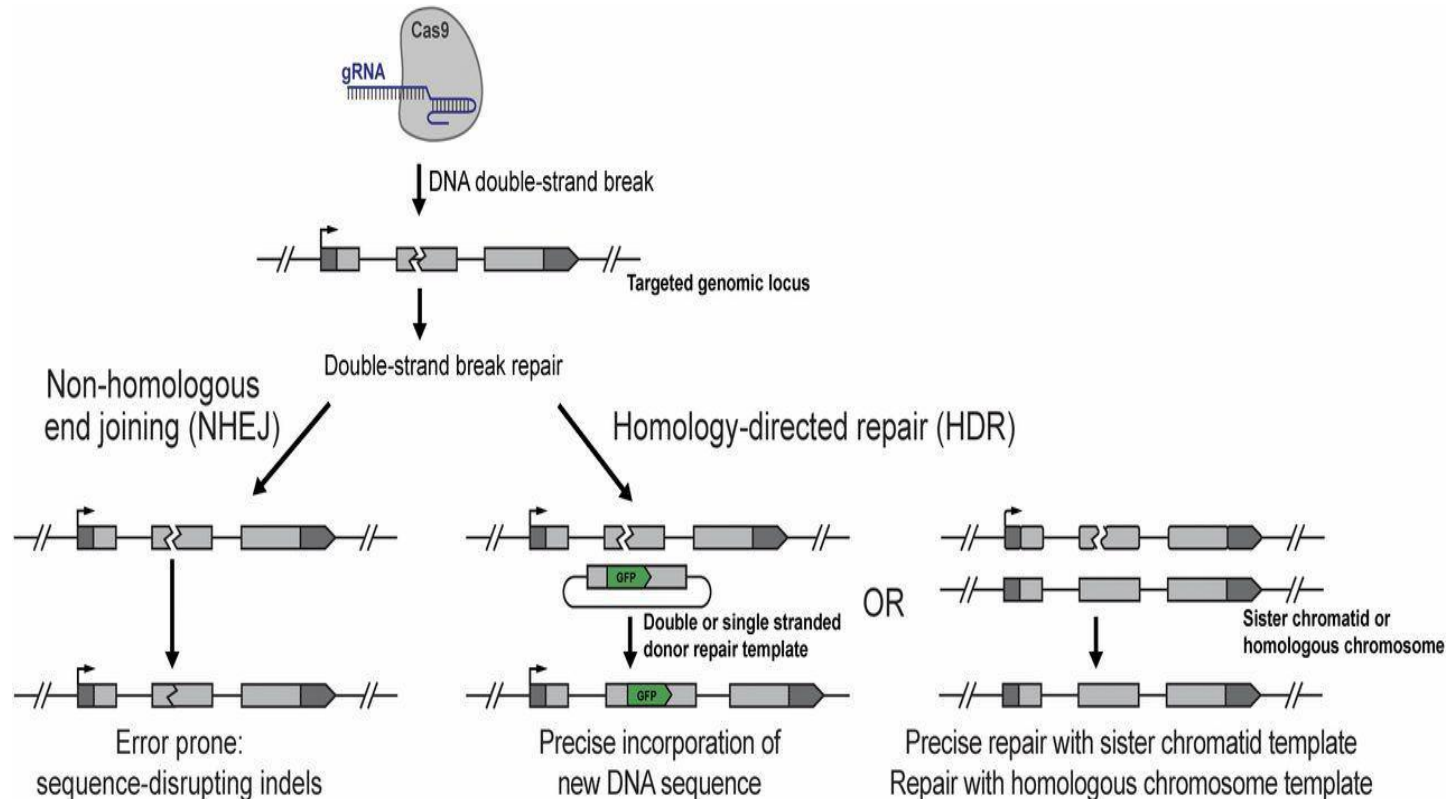


Applications of Biotechnology Techniques

- **Human Genome Project**
- **Diagnosis of Disease – PCR & DNA probes**
- **Human Gene Therapy**
- **Vaccines & Pharmaceutical Products**
- **Forensics – DNA Fingerprints (RFLP & VNTR)**
- **Environmental – Recycling & detoxification**
- **Agricultural – transgenic organisms**

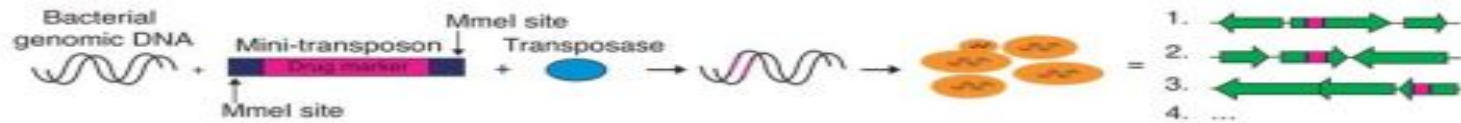
CRISPR Cas9 gene editing

Clustered Regularly-Interspaced Palindromic Repeats.

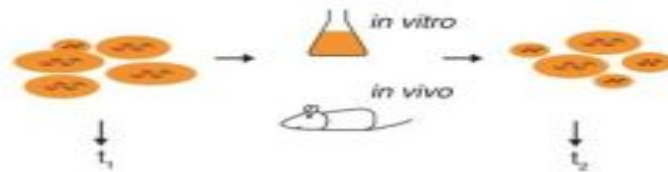


TN-Seq

a.



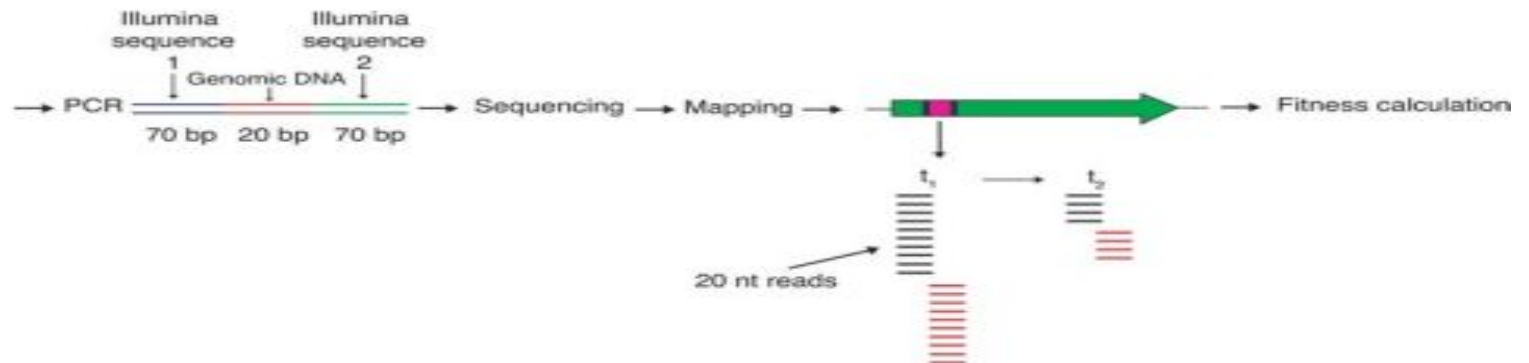
b.

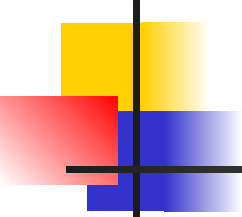


c.



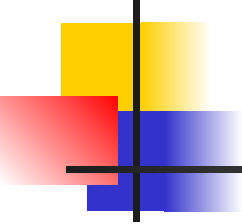
d.





Bioethics

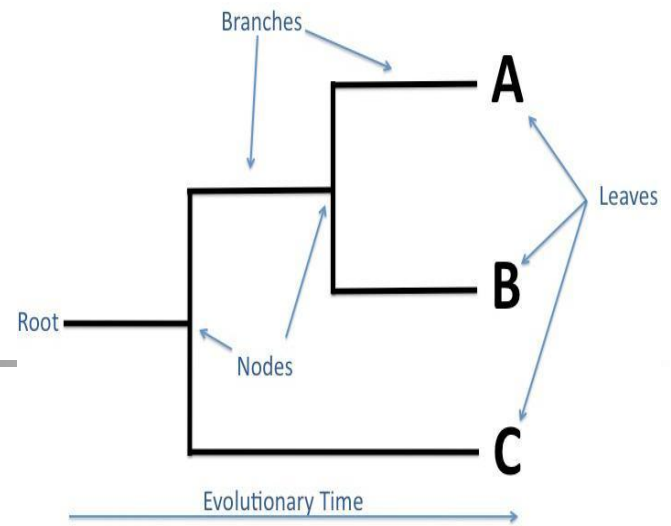
- **Potential Hazards vs. Potential Gains**
- **Concerns:**
 - **genetically modified foods**
 - **genetically engineering microbes**
 - **cloning whole organisms**
 - **embryonic stem cell research**
 - **gene therapy**
 - **genetic testing**
 - **bioterrorism**



Epigenetics

- **The study of heritable changes in gene activity that occur without a change in the sequence of the genetic material. Epigenetics literally means 'in addition to genetics'.**
- **Epigenetic factors can regulate the amount of gene activity, influencing the growth and appearance of an organism**
- **Malfunctions in epigenetic control of gene activity have been implicated in cancer, cardiovascular disease and several inherited genetic conditions**

Phylogenetics



- **Study of evolutionary relationships among groups of organisms based upon their genetics**
 - Relationships between organisms are often represented using phylogenetic trees.
 - Organisms more closely related to each other are physically closer on the tree.
 - Common ancestors are represented by nodes that connect branches together.
- **Has taxonomy folks in a turmoil – they can't agree so we have national lists for our taxonomy events**