UNDERSTANDING GENETICS FOR IMPROVING HEALTH OUTCOMES - CEUGH

Dr. Rahul Kushwah

March - May 2019

AnantLife

COURSE OBJECTIVES:

- Gain an overall understanding of basic cellular and molecular biology to understand how nucleic acids (DNA, RNA) play a role in regulating overall homeostasis and the basis of how changes in nucleic acid can impact cell biology.
- Gain a deep understanding of nutritional genetics, fitness genetics and genetics of chronic metabolic disorders such as diabetes and cardiovascular diseases.
- Gain a deep understanding of detoxification genetics, hormonal genetics, skin genetics, genetics of neurotransmitter synthesis and breakdown along with genetics of endocannabinoid pathways.
- Learn how to not only understand genetic analysis in relation to the above-mentioned genetic pathways but also how to deliver the information to the clients along with nutritional/dietary strategies that can be incorporated to improve the overall health of the clients.
- Gain expertise in analysis of genetic testing report and developing health plans for clients.

LEARNING OUTCOMES:

- Understand the connection between health and genetics and more importantly gain an understanding of how the same dietary/nutritional plan cannot have efficacy for everyone owing to the differences in genetics.
- Detailed understanding of how genes play a role in regulating dietary intake, nutritional profile, response to various fitness regimens, weightloss, detoxification, brain health and overall response to cannabis.
- Learn to apply genetic testing as well as genetic analysis to develop truly personalized health plans for clients to optimize/improve their health with a high success rate for efficacy.

EVALUATION:

- Take home exam – Given on session 5 and due on session 6 (Will cover materials from session 1 – 5) – 50%
- Take home assignment – Given on session 7 and due on session 8 (Will cover materials from session 1-7) – 50%

- PASSING GRADE – 70%

- SUCCESSFUL COMPLETION – Certificate issued by IHN in partnership with Anantlife Canada Inc – CERTIFIED GENETIC TESTING PROVIDER CERTIFICATE

SESSIONS 1 - 8

Session	Topic	Evaluations
1	INTRODUCTION TO MOLECULAR GENETICS, MOLECULAR BIOLOGY AND HUMAN GENETICS	Discussion – Participation
2	NUTRITIONAL AND DIETARY GENETICS: HOW DO OUR GENES REGULATE OUR NUTRITION AND NUTRITIONAL HEALTH?	Discussion - Participation
3.	FITNESS GENETICS AND GENETICS OF CHRONIC DISEASES: HOW DO OUR GENES REGULATE OUR RESPONSE TO EXERCISE AND HOW DO GENES REGULATE THE RISK OF CHRONIC METABOLIC DISORDERS?	Discussion - Participation
4.	DETOXIFICATION GENETICS: HOW DO OUR GENES REGULATE DETOXIFICATION WHICH INDIRECTLY IMPACTS OVERALL HEALTH AND DISEASE RISK?	Discussion - Participation
5.	NEUROGENETICS: HOW DO OUR GENES REGULATE THE SYNTHESIS AND BREAKDOWN OF NEUROTRANSMITTERS AND ITS IMPACT ON OUR HEALTH ?	Take home exam on sections 1-5, due during session 6
6.	GENETICS OF ENDOCANNABINOID PATHWAYS: HOW DO OUR GENES REGULATE THE RESPONSE TO CANNABIS?	Discussion - Participation
7.	SKIN GENETICS: HOW DO OUR GENES REGULATE OUR SKIN HEALTH ?	Take home assignment – due during session 8
8.	DISCUSSION AND PRACTICAL APPLICATIONS OF GENETIC TESTS DISCUSSED IN SESSIONS 2-7	Discussion - Participation

SESSION 1: INTRODUCTION TO MOLECULAR GENETICS, MOLECULAR BIOLOGY AND HUMAN GENETICS

Dr. Rahul Kushwah

March 27, 2019

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MOLECULAR BIOLOGY: THE BASIS OF GENETICS

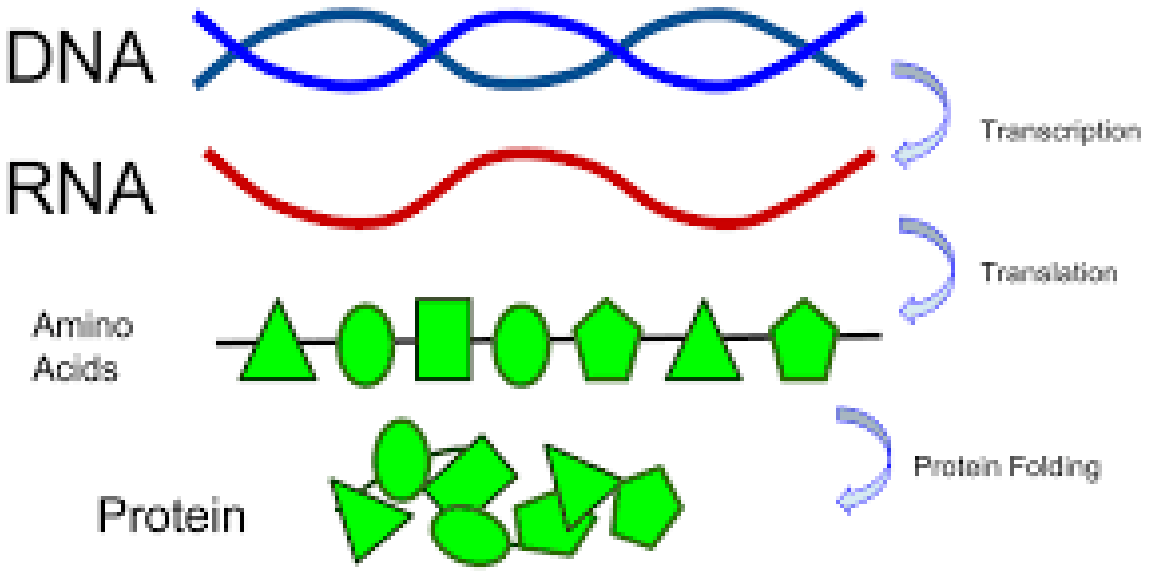
WHAT IS MOLECULAR BIOLOGY?

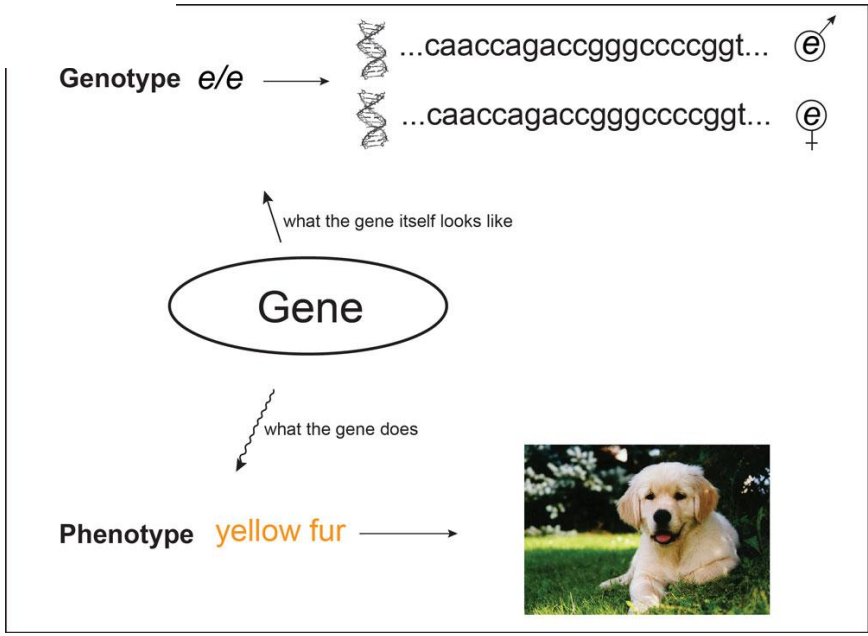
Molecular biology; the branch of biology that study gene structure and function at the molecular level.

The Molecular biology field overlaps with other areas, particularly genetics and biochemistry.

The Molecular biology allows the laboratory to be predictive in nature; events that occur in the future.

CENTRAL DOGMA OF MOLECULAR BIOLOGY

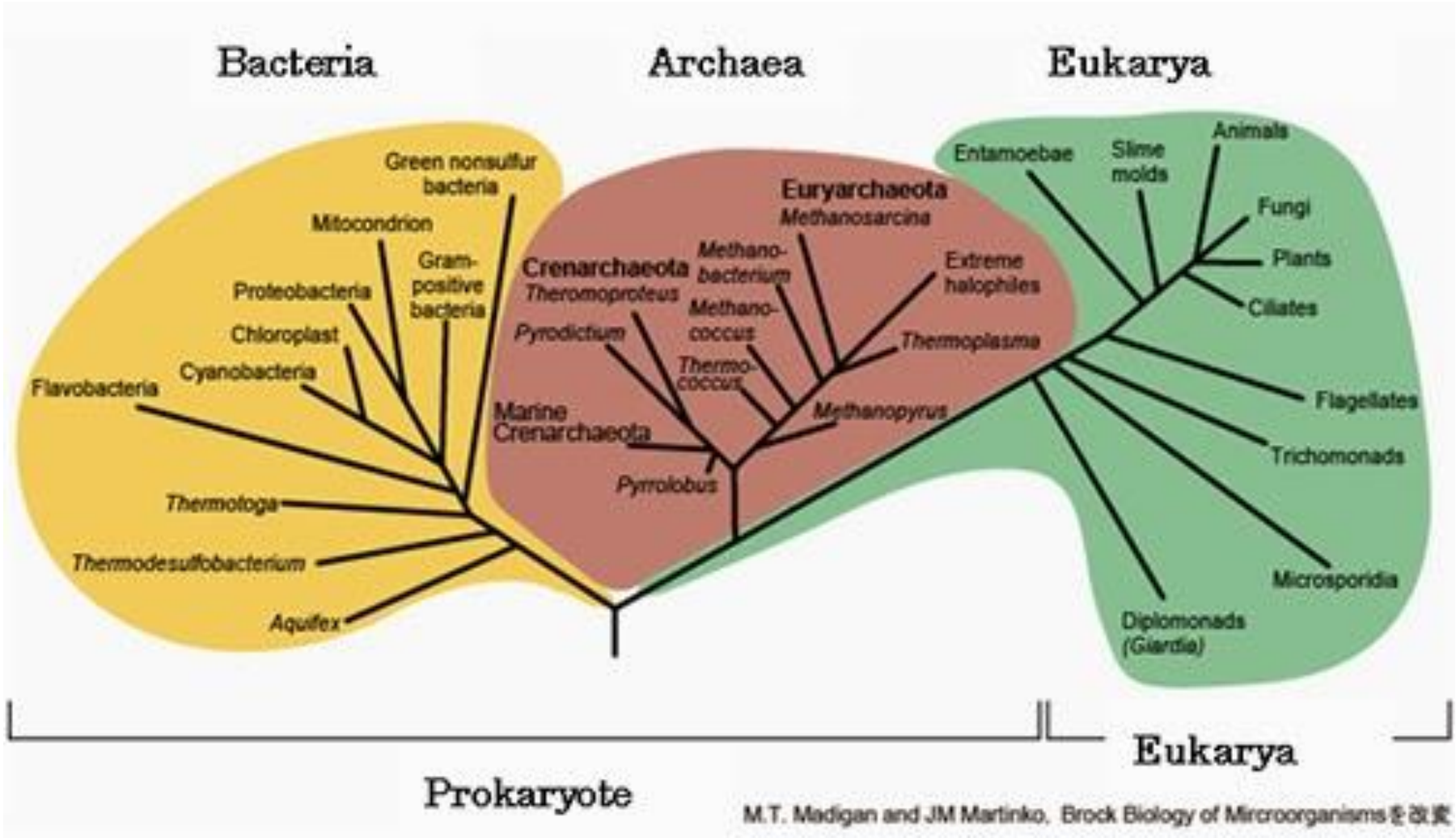




MOLECULAR BIOLOGY: THE BASIS OF GENETICS

- The Genome
- The Cell
  - Eukaryotic Cell
  - Prokaryotic Cell
- Three Domain of Life
- The General Structure of Nucleic Acid
  - DNA
  - RNA
- The Central Dogma of Molecular Biology
  - DNA Replication
  - From DNA to Protein
- Genetic Mutation
- Human genome project
- Functional Genomics/Transcriptomics /Proteomics

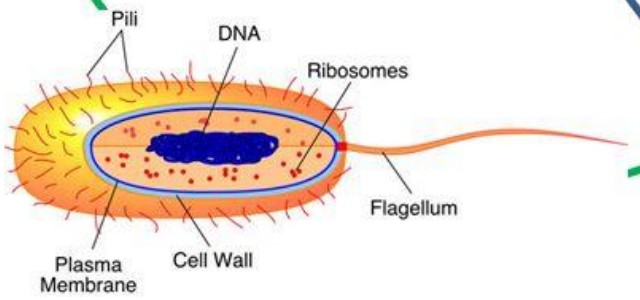
THE THREE DOMAINS OF LIFE



TWO DIFFERENT CELL TYPES

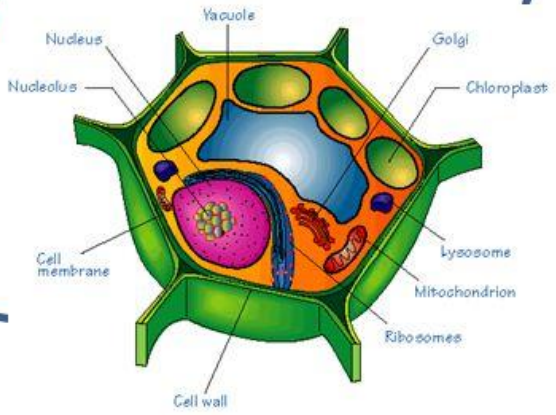
Prokaryotic Cells

- domains Bacteria & Archaea
- 1-10 µm
- DNA located in nucleoid region
- cell wall
- capsule
- pili
- flagella



Eukaryotic Cells

- domain Eukarya (protists, fungi, plants, & animals)
- 10-100 µm
- DNA located in nucleus
- organelles

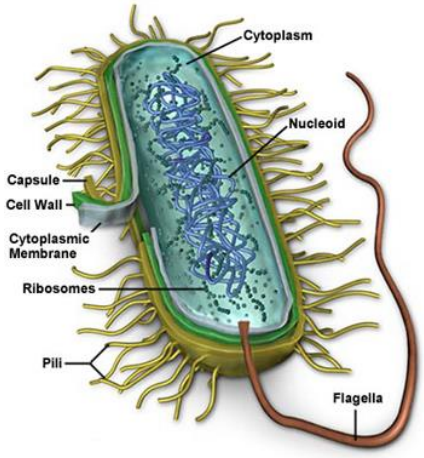


Both

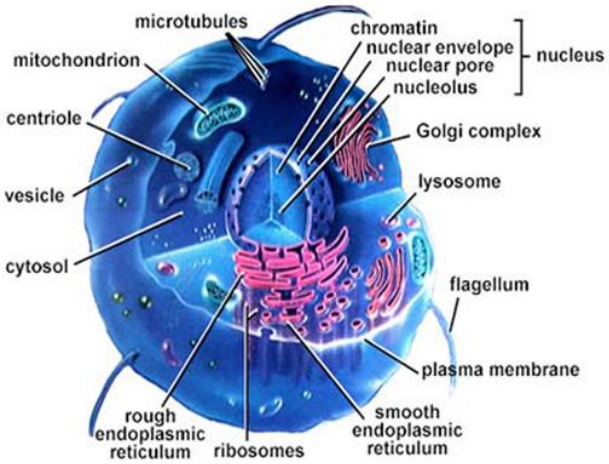
- plasma membrane
- cytosol/cytoplasm
- ribosomes
- chromosomes

EUKARYOTIC CELLS

- Eukaryotic cells are found in animals, plants, fungi and protists cell;
- Cell with a true nucleus, where the genetic material is surrounded by a membrane;
- Eukaryotic genome is more complex than that of prokaryotes and distributed among multiple chromosomes;
- Eukaryotic DNA is linear and Eukaryotic DNA is complexed with proteins called histones;
- Numerous membrane-bound organelles;
- Complex internal structure;
- Cell division by mitosis.

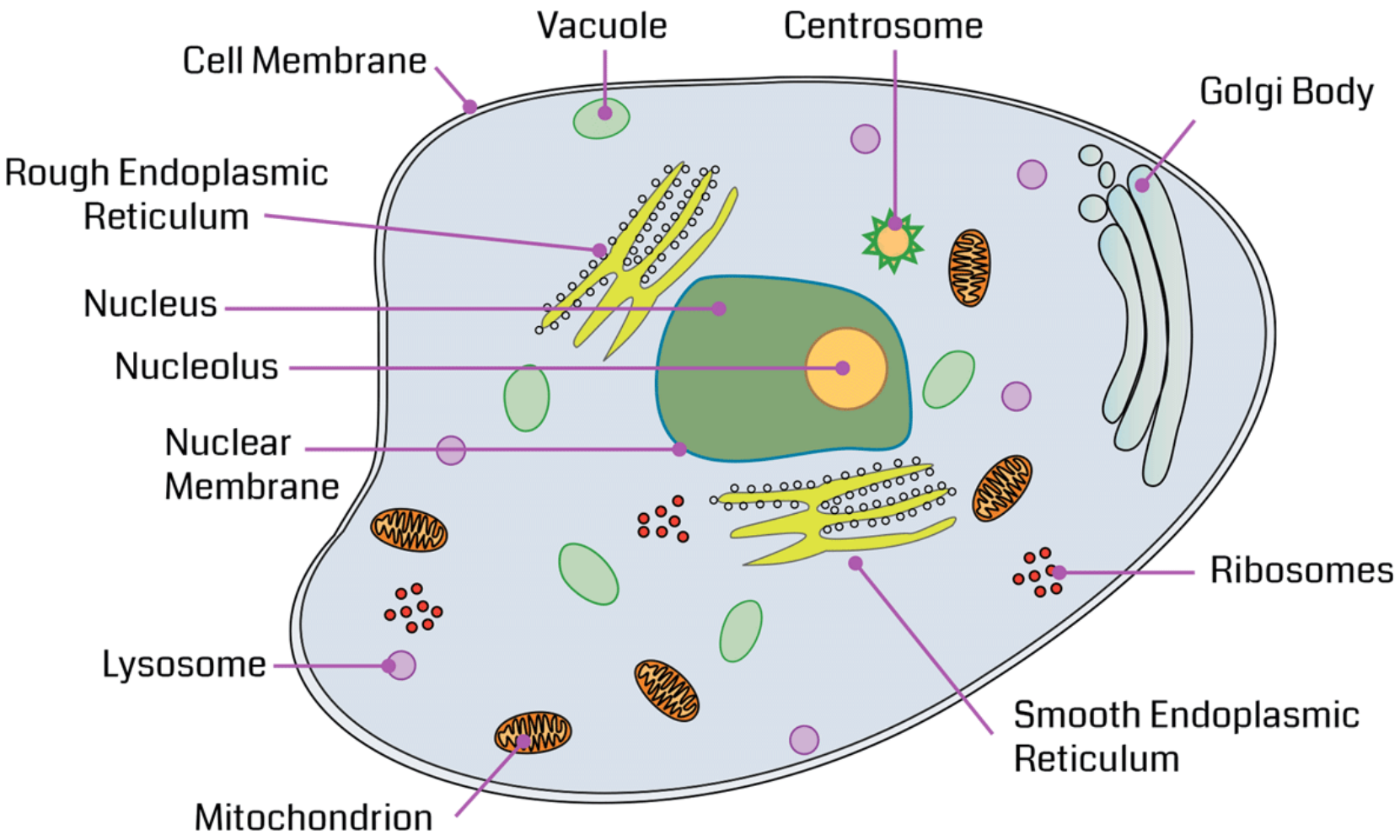


prokaryotic cell
(bacteria)

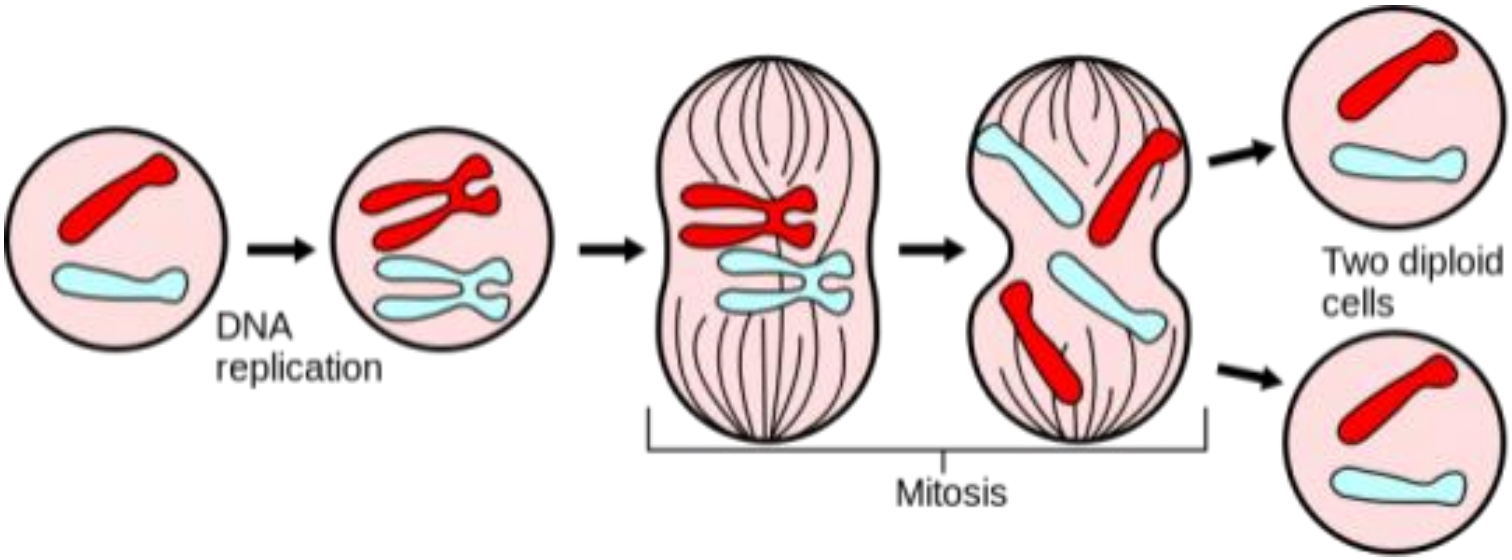


eukaryotic cell
(protists, fungi, animals, plants)

CELL ORGANELLES AND THEIR FUNCTION

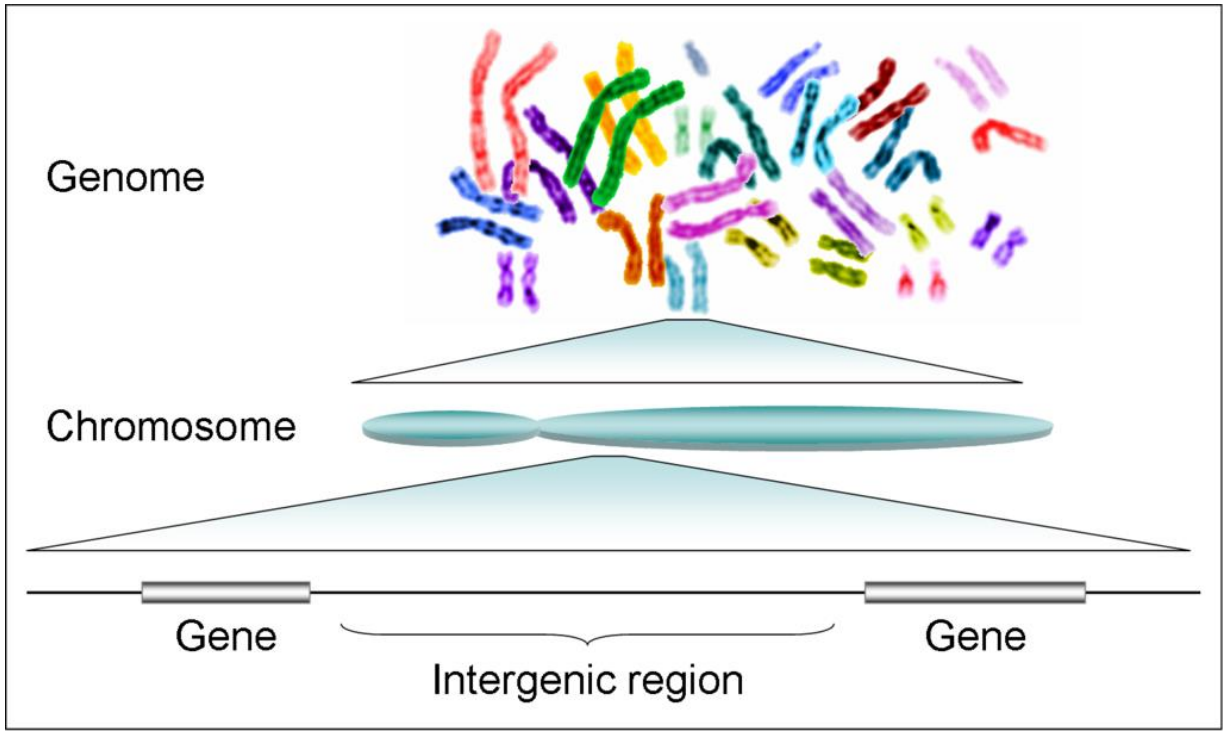


ALL CELLS SHARE THE SAME CYCLE

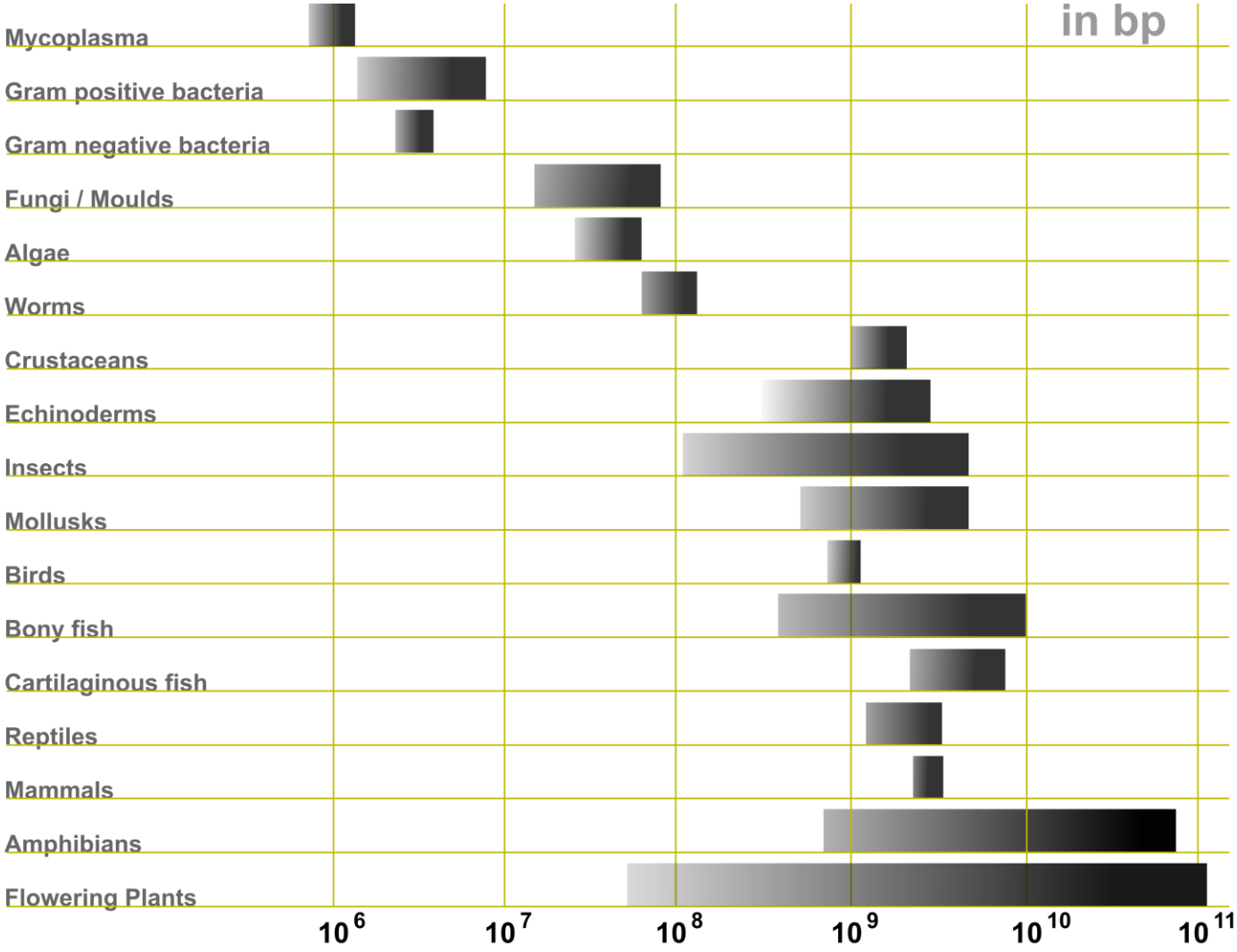


WHAT IS A GENOME?

- Genome is the complete collection of an organism's hereditary information.
- Encoded either in DNA or RNA (as in viruses)
- The genome includes the entire sequence of DNA (eg humans) or RNA (viruses)

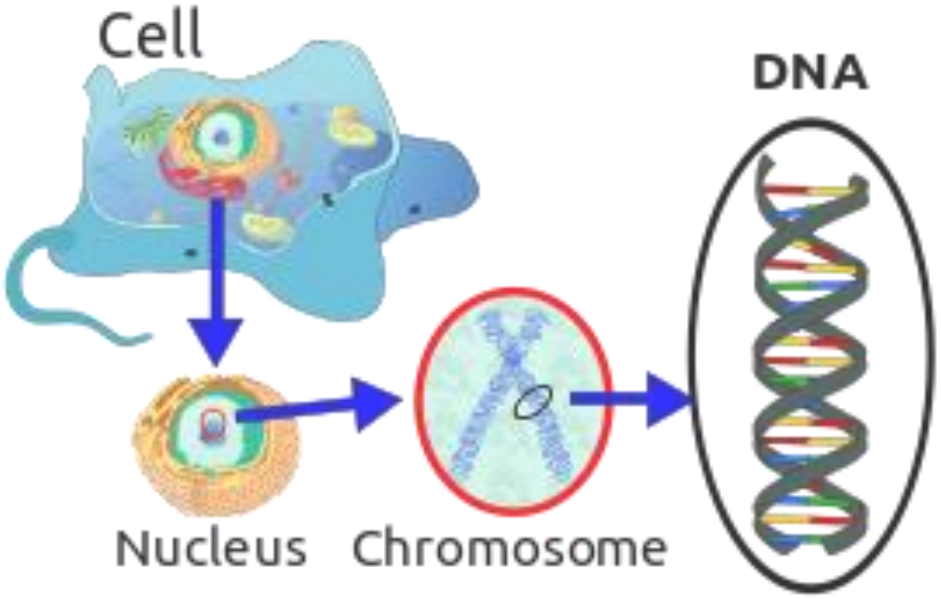


GENOME SIZE COMPARISON



WHAT IS A CHROMOSOME?

- DNA in eukaryotes in stored in structures called chromosomes
- Chromosomes are located inside the nucleus
- Chromosomes are highly condensed form of DNA
- DNA is wrapped around special proteins called histones which produces a compact structure
- Chromosomes are found in every cell and are passed from parents to progeny



SPECIES AND CHROMOSOMES

Chromosome number varies across species and specie size has no correlation to chromosome number

One set of chromosome comes from each parent

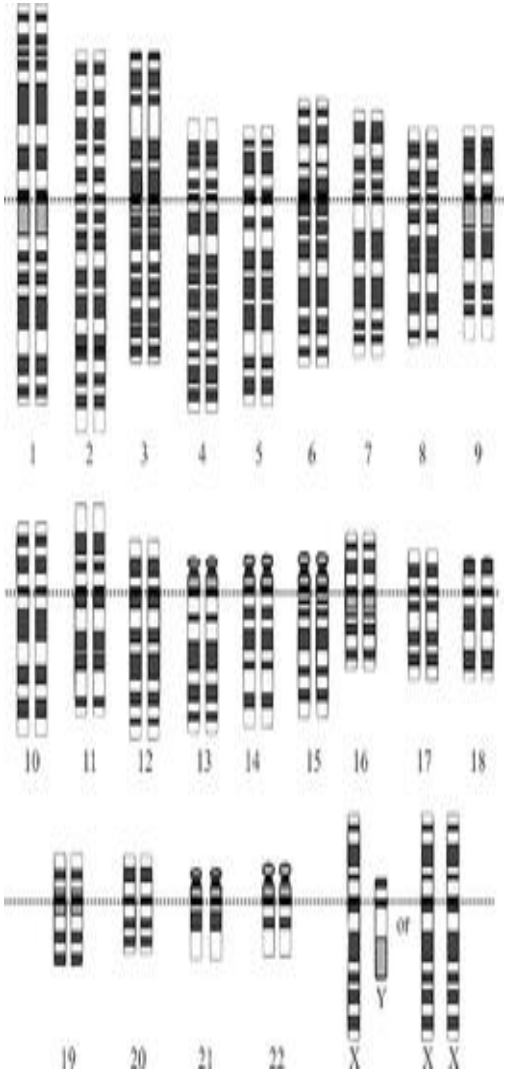
Common Name	Species	Diploid number	Common Name	Species	Diploid number
Animals (2n)			Plants (2n)		
Human	*Homo sapiens*	46	Corn	*Zea mays*	20
Monkey	*Macaca mulatta*	42	Potato	*S. tuberosum*	48
Dog	*Canis familiaris*	78	Green algae	*A. mediterranea*	20
Cat	*Felis domesticus*	38			
Mouse	*Mus musculus*	40	Fungi (2n)		
Frog	*Rana pipiens*	26	Yeast	*S. cerevisiae*	32
Fruit fly	*Drosophila melanogaster*	8			
Flatworm	*Planaria torva*	16			

http://plantcellbiology.masters.grkraj.org/html/Plant_Cellular_Structures13-The_Nucleus.htm

HUMAN GENOME

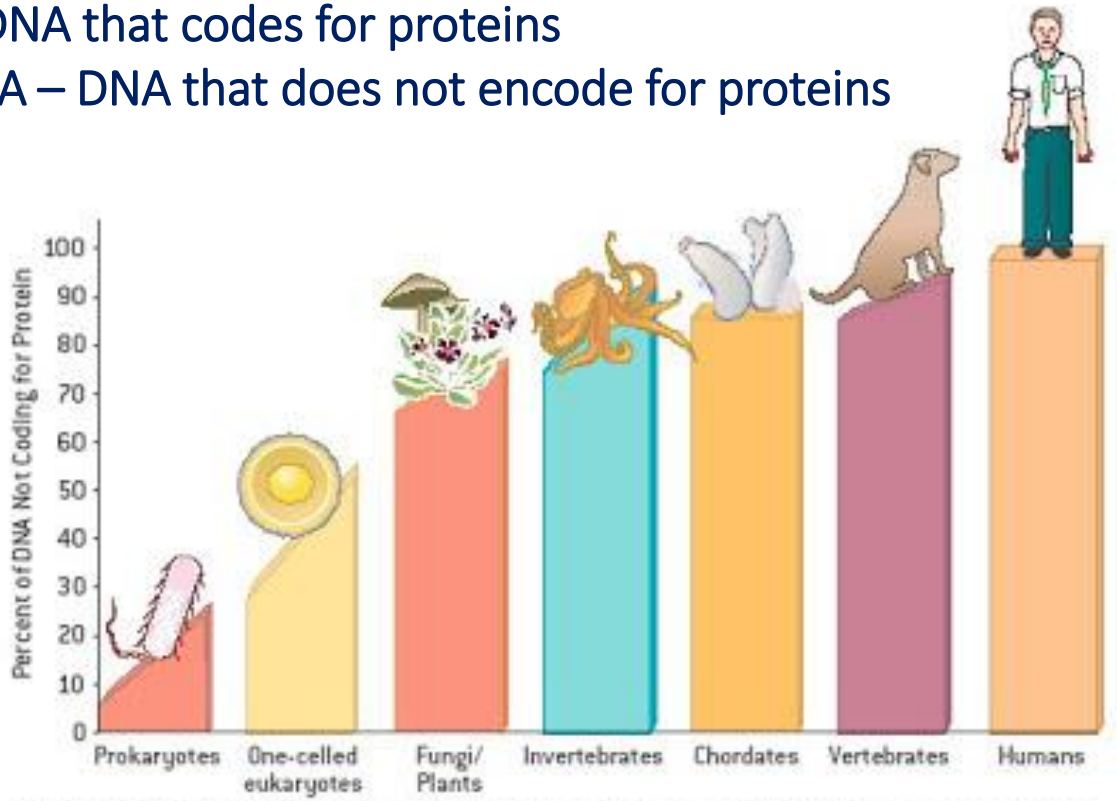
- Human Genome; Arranged on multiple chromosomes; twenty three pairs of chromosomes;
- Twenty two pairs (autosomes).
- One pair (sex chromosome) (xx) (female) or (xy) (male).

- Humans have 23 pairs of chromosome in every cell (except mature red blood cells..); Gametes or sex cells (sperm and eggs) have half the normal complement of chromosomes.



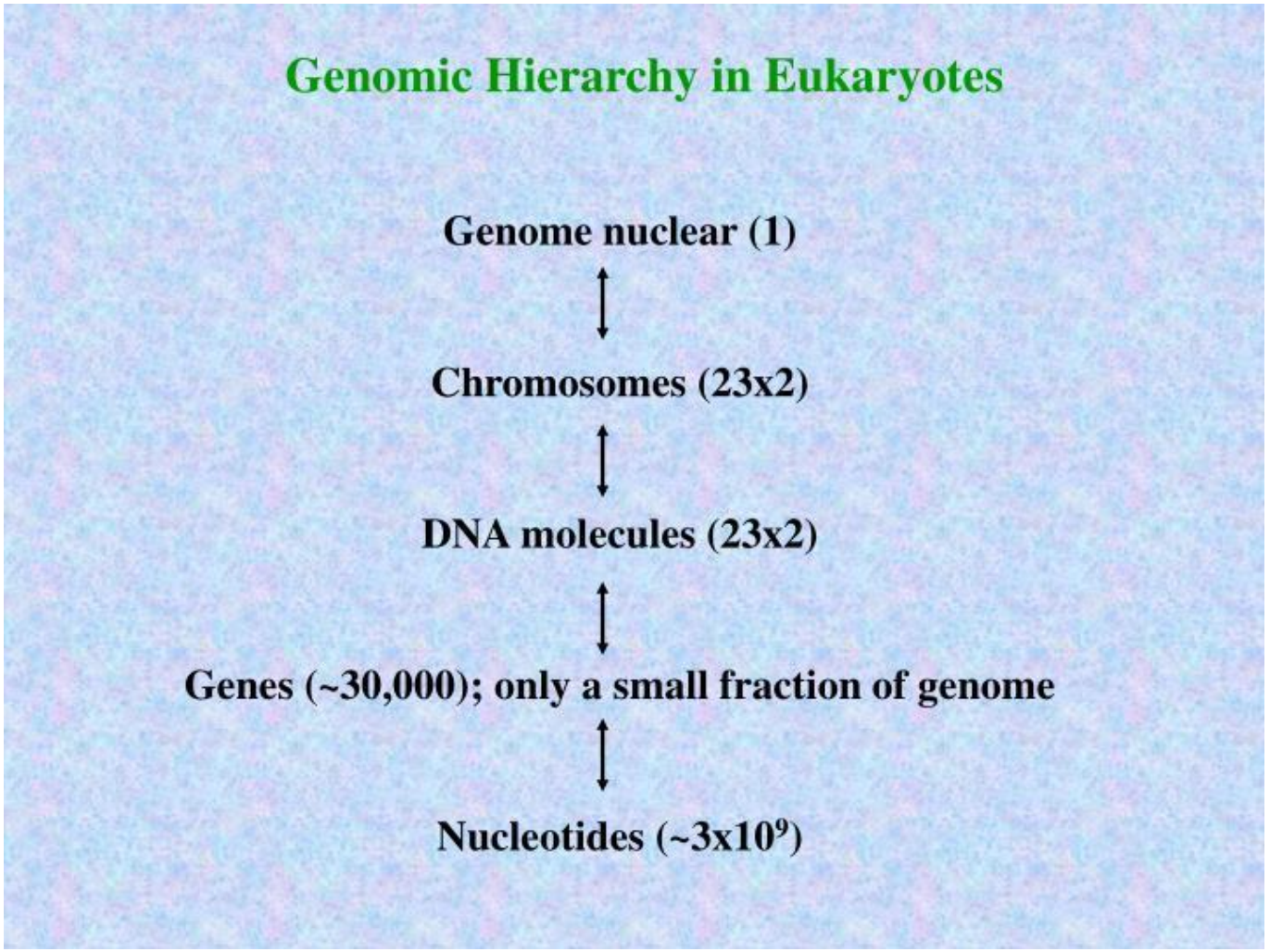
MOST OF THE HUMAN DNA IS NON CODING

- Coding DNA – DNA that codes for proteins
- Non Coding DNA – DNA that does not encode for proteins



NONPROTEIN-CODING SEQUENCES make up only a small fraction of the DNA of prokaryotes. Among eukaryotes, as their complexity increases, generally so, too, does the proportion of their DNA that does not code for protein. The noncoding sequences have been considered junk, but perhaps it actually helps to explain organisms' complexity.

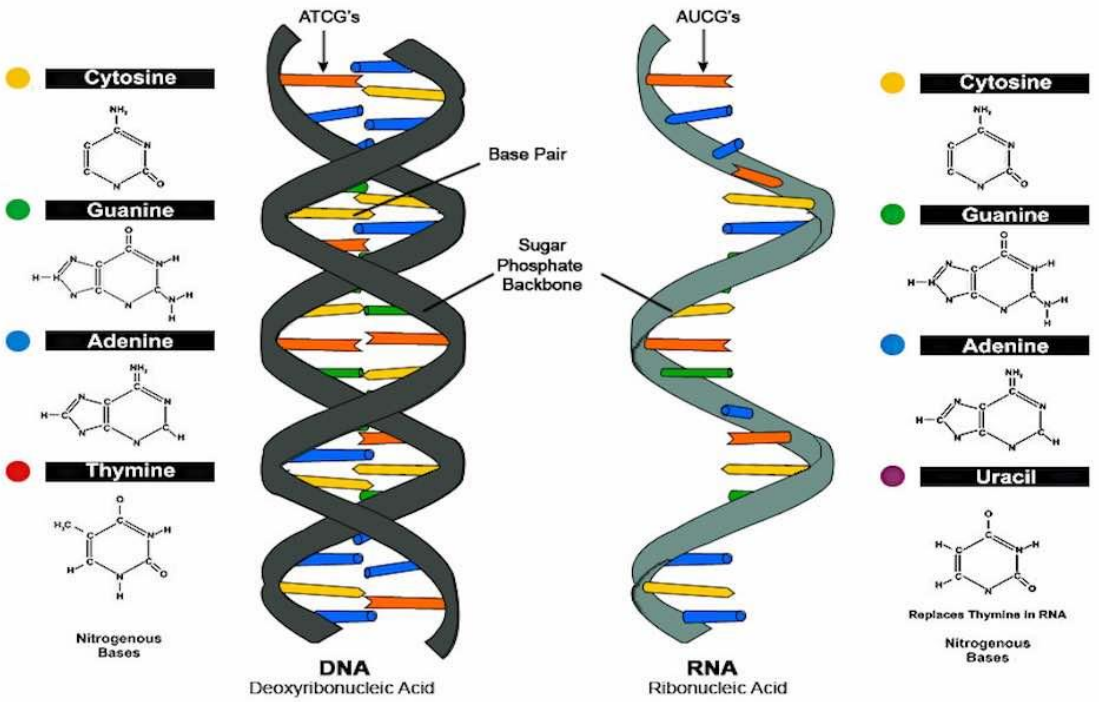
GENOMIC HIERARCHY - HUMANS



GENERAL STRUCTURE OF NUCLEIC ACIDS

Two major classes – DNA and RNA

DNA and RNA are long chain polymers of small chemical compound called nucleotides.



WHAT ARE NUCLEOTIDES?

Nucleotides; ring shaped structures composed of:

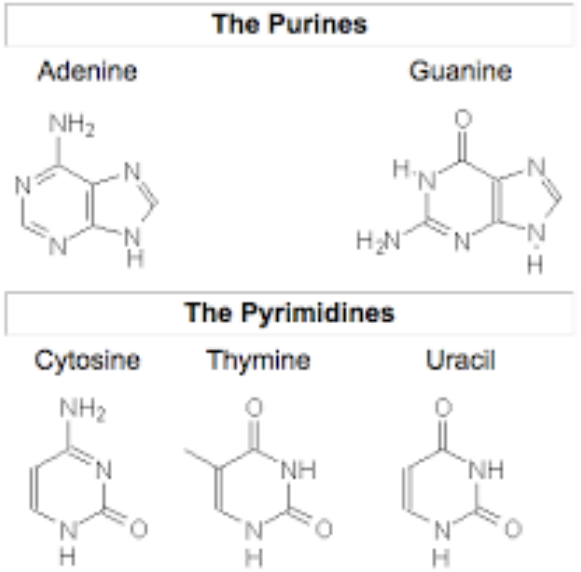
Nitrogenous base; these bases are classified based on their chemical structures into two groups:

Purine; double ringed structure (Adenine and Guanine).

Pyrimidine; single ring structures (cytosine and thymine).

Sugar

Phosphate group



NUCLEOTIDES

DNA: Four different types of nucleotides differ in nitrogenous base:

A is for adenine;

G is for guanine;

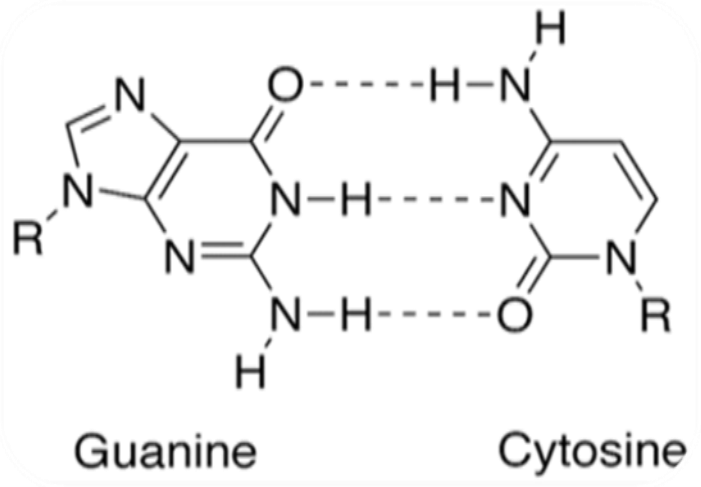
C is for cytosine and

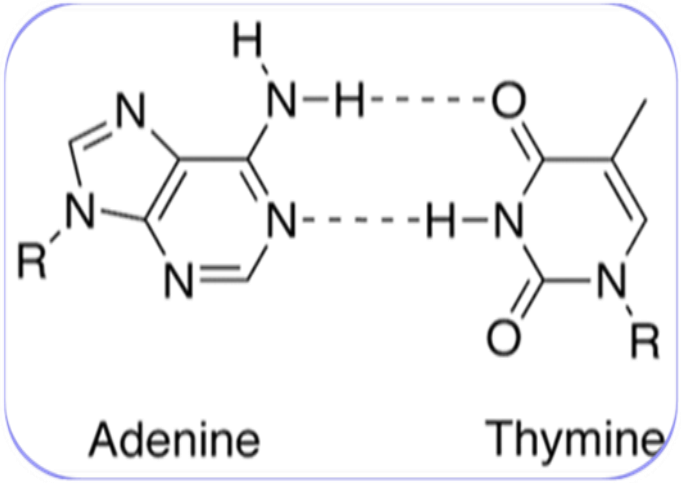
T is for thymine.

RNA: thymine base replaced by uracil base.

NUCLEOTIDES

A binds to T and G binds to C - DNA





WHAT IS DNA ?

Deoxyribonucleic Acid (DNA); the genetic material of all cellular organisms and most viruses.

DNA; the gigantic molecule which is used to encode genetic information for all life on Earth.

A human cell contains about 2 meters of DNA. DNA in the body could stretch to the sun and back almost 100 times. So it is tightly packed.

DNA responsible for preserving, copying and transmitting information within cells and from generation to generation.

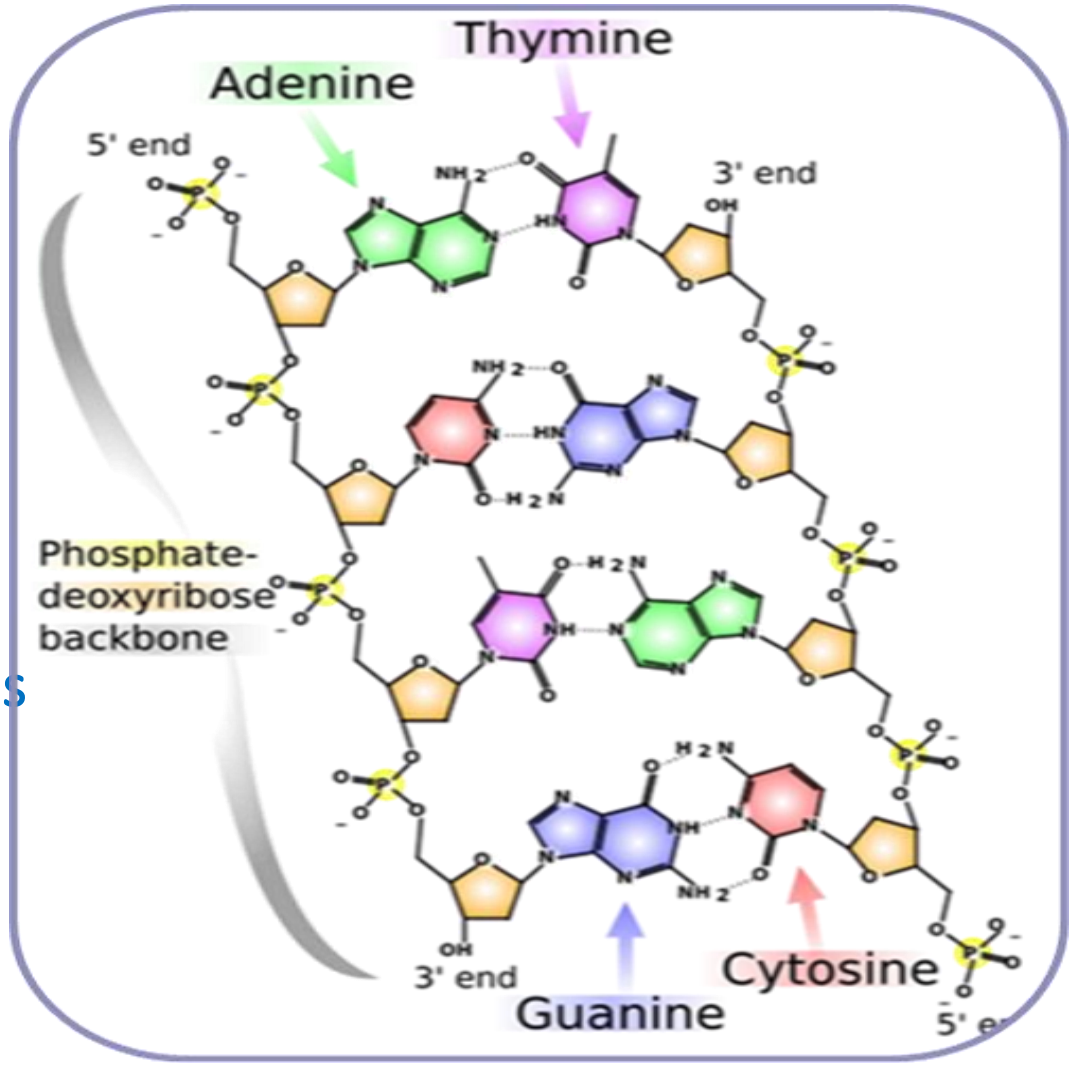
DNA – DOUBLE HELIX

Linked as a twisted ladder.

The curving sides of the ladder represent the sugar-phosphate backbone of the two DNA strands; the rungs are the base pairs.

Possess antiparallel polarity.

Stabilized by hydrogen bonds between the bases.



WHAT IS A GENE?

The gene; it is a segment within a very long strand of DNA

A segment of DNA that codes for an entire protein.

Genes are the basic units of hereditary.

Genes located on chromosome on its place or locus.

Allele; a variant of the DNA sequence at a given locus. Each allele inherited from a different parent.

WHAT IS AN ALLELE?

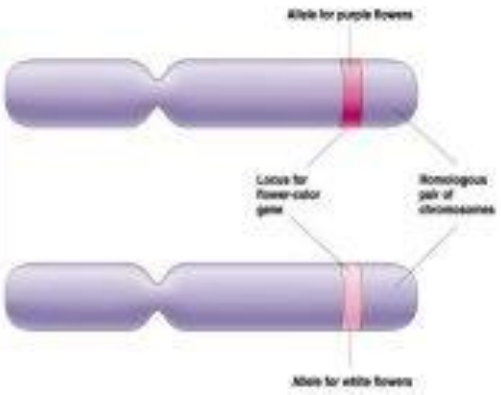
Every human has two copies of each gene with each being inherited from one parent

Single copy of each gene is referred to as allele

One allele of a gene is inherited from one parent

3.1 U.3 The various specific forms of a gene are alleles.

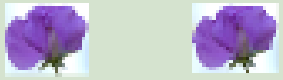
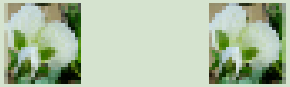
Allele is one specific form of a gene, differing from other alleles by one or a few bases only and occupying the same gene locus as other alleles of the gene.



FORMS OF ALLELES: DOMINANT & RECESSIVE

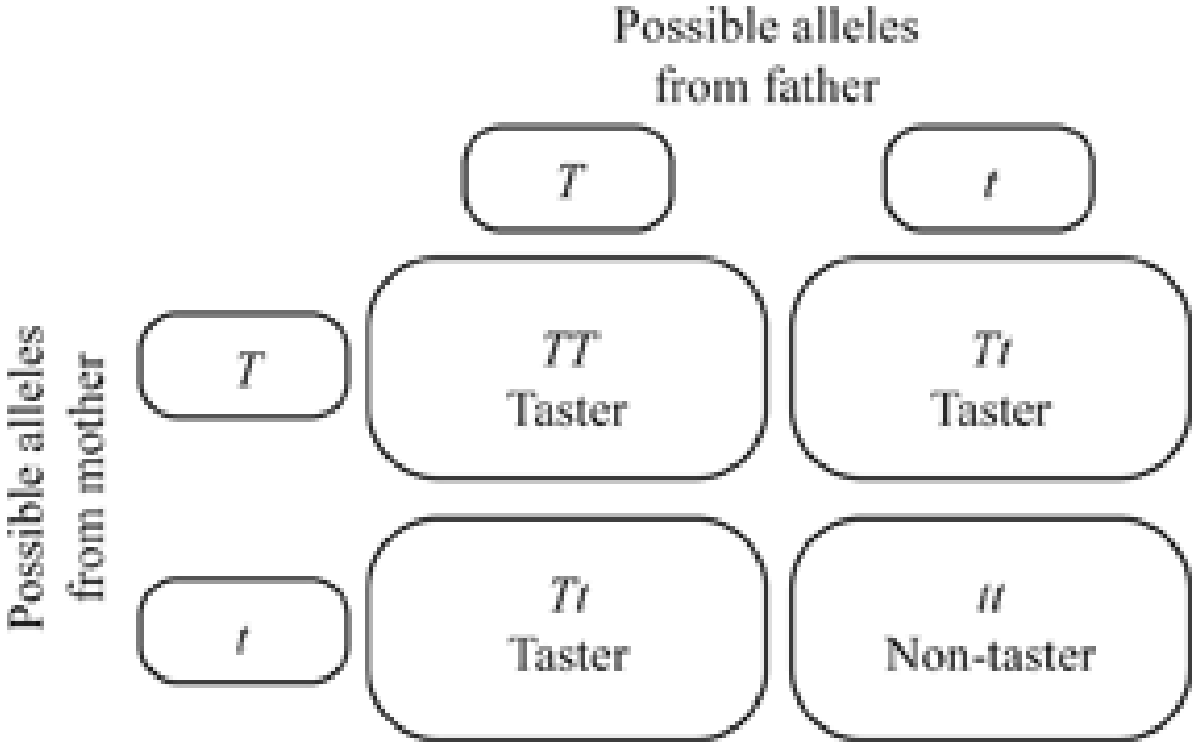
Dominant
The one pair of allele that masks the effect of the other when present in the same cell.
Recessive
The one pair of allele that is masked by the other when present in the same cell and capable of producing its characteristics phenotype in the organism only when two alleles is present and identical.

Homozygous Dominant
Heterozygous
Homozygous Recessive

Alleles	Alleles Expressed
Dominant, Dominant 	
Dominant, Recessive 	
Recessive, Recessive 	

DOMINANT vs RECESSIVE ALLELES

T – allele to taste potatoes – Dominant Allele
t – allele due to which one cannot taste potatoes – Recessive allele
Each parent is heterozygous and has Tt genotype

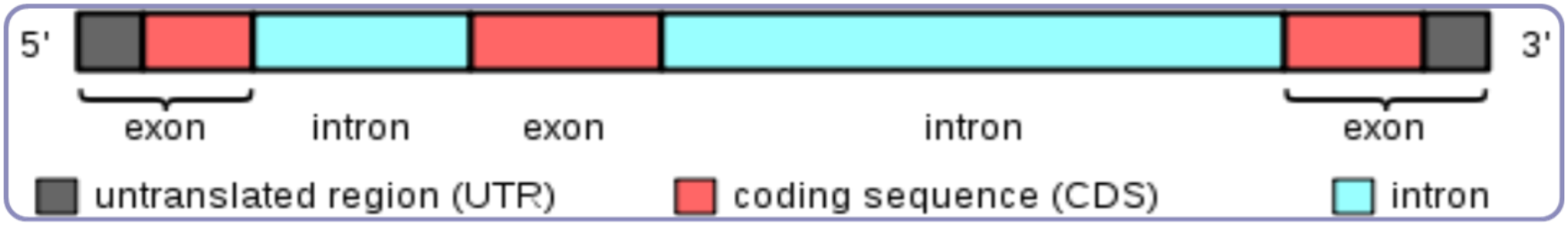


GENE STRUCTURE

Most of the genes consist of; short coding sequences or exons are interrupted by a longer intervening noncoding sequence or introns; although a few genes in the human genome have no introns.

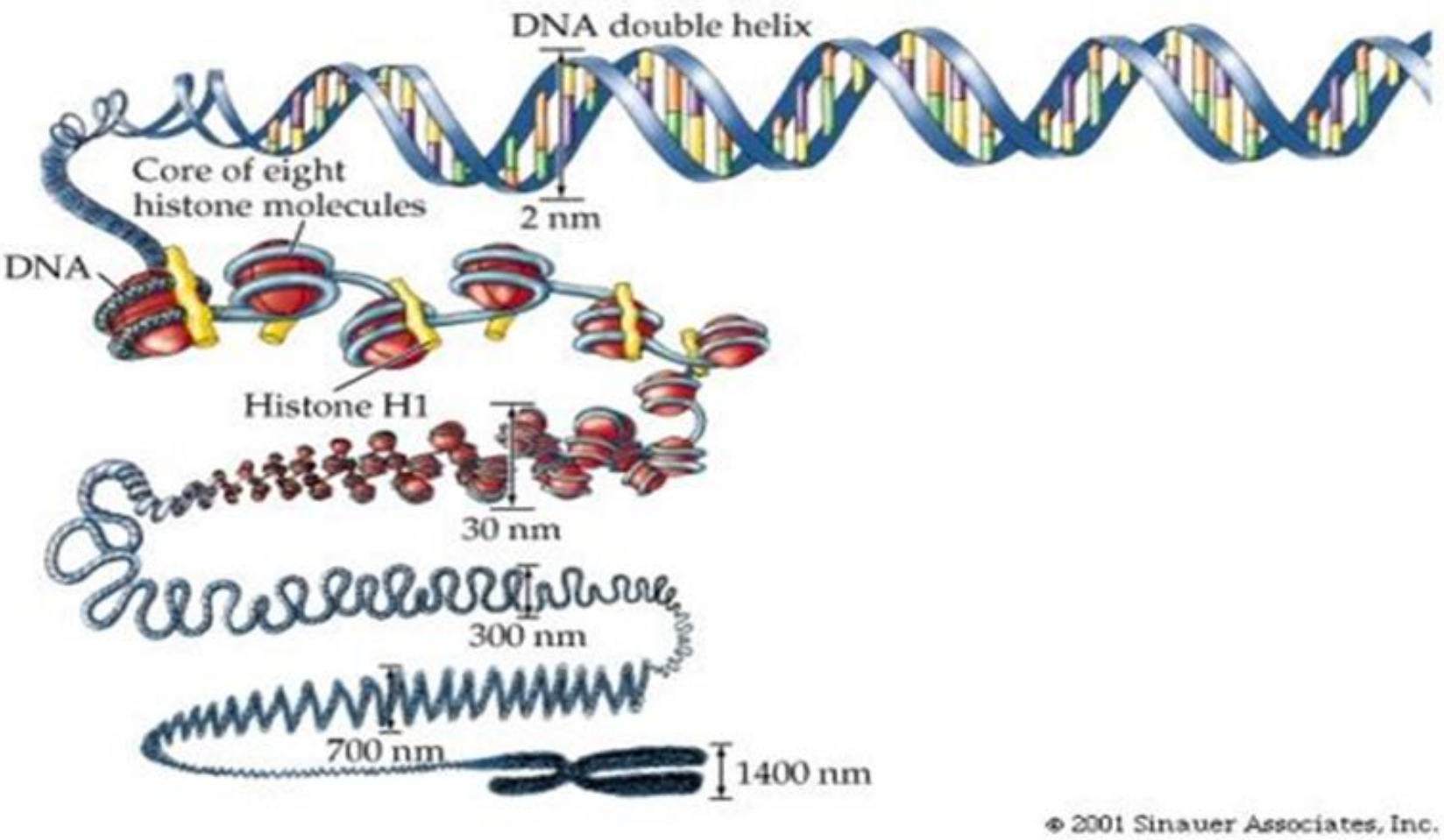
Introns – can allow for multiple proteins to be generated from same gene by splicing.

Jntrons can also encode for regulatory elements.



DNA ORGANIZATION

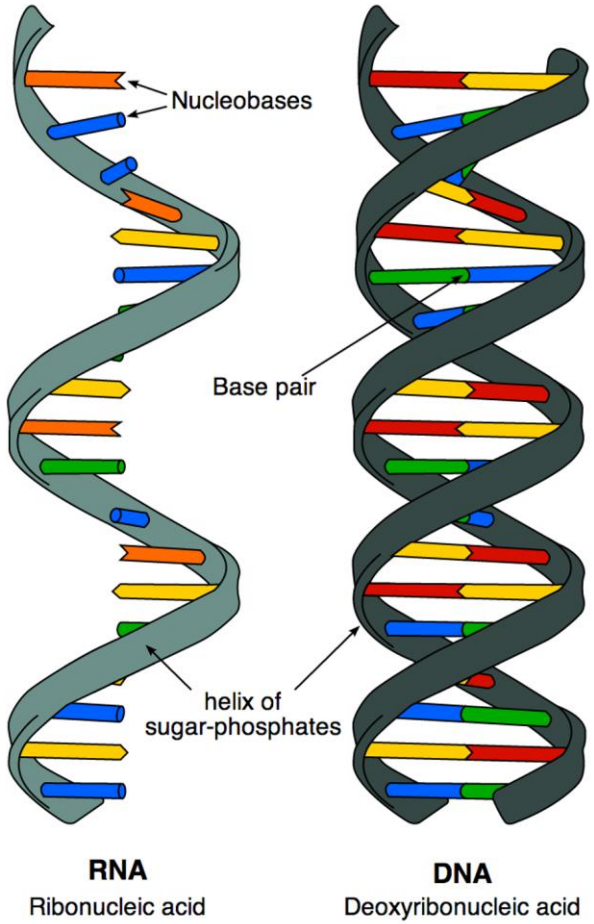
DNA is organized in the form of chromosomes



REVIEW/DISCUSSION POINTS

- 3 living domains
- Cell types
- Organization of DNA in cells
- Genome, Gene, Allele
- Nucleotides

RNA – RIBONUCLEIC ACID

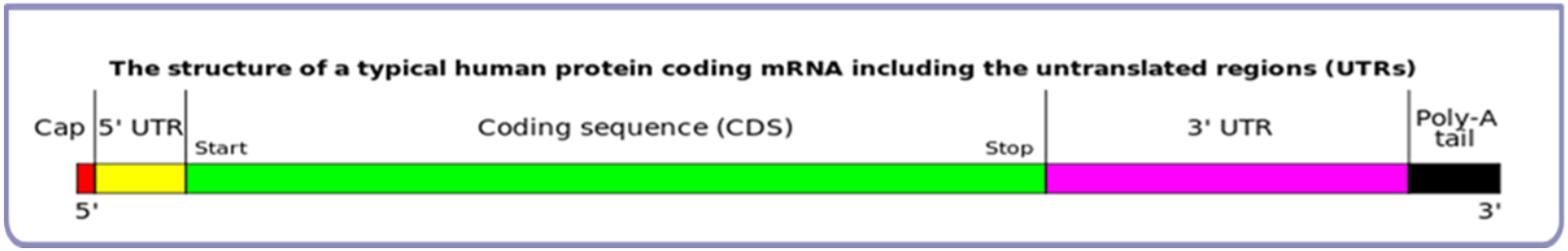


RNA is a single stranded; the pyrimidine base uracil (U) replaces thymine and ribose sugar replaces deoxyribose.

Three major classes of RNA: messenger (mRNA), transfer (tRNA) and ribosomal (rRNA). Minor classes of RNA include small nuclear RNA; small nucleolar RNA

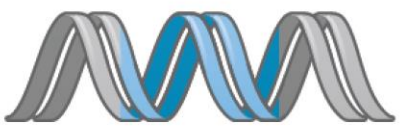
mRNA – Messenger RNA

- Transcripts of structural genes.
- Encode all the information necessary for the synthesis of a polypeptide of protein.
- The 5' terminus is capped by 7 methyguanosine triphosphate.
- Synthesis of the poly (A) tail involves cleavage of its 3' end and then the addition of about 200 adenine residues.
- Intermediate carrier of genetic information; deliver genetic information to the cytoplasm.



mRNA – THE SOFTWARE

DNA → mRNA → Protein



STORAGE

DNA stores instructions for proteins in the nucleus

SOFTWARE

mRNA is a temporary set of instructions for cells to make a protein; mRNA is made using DNA

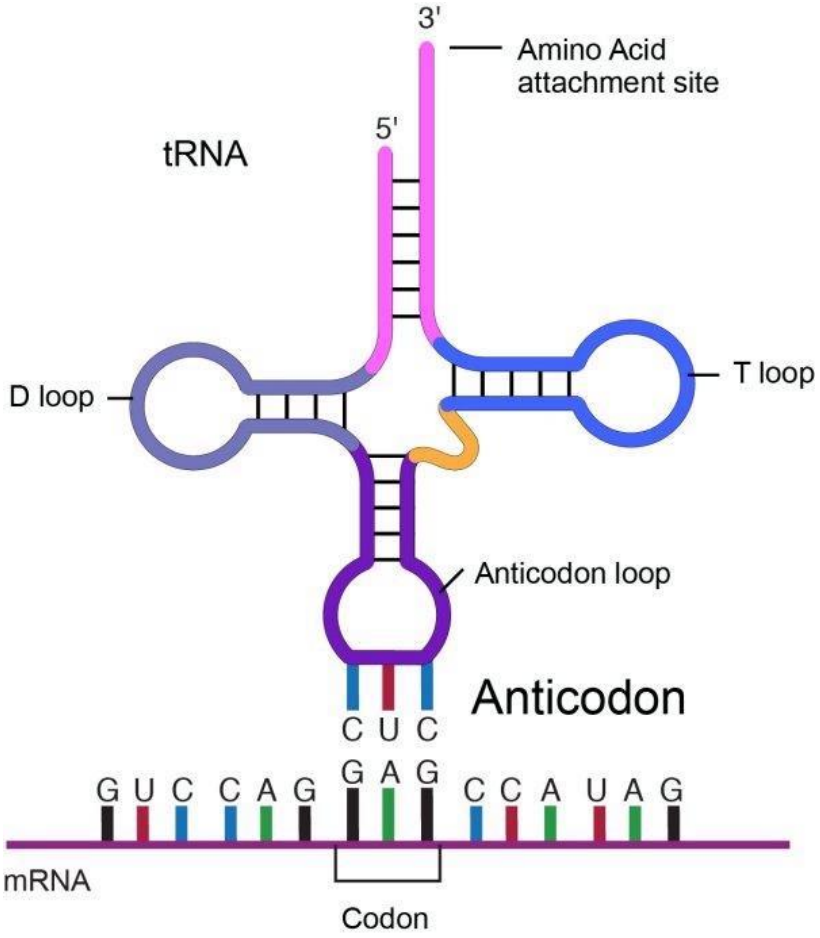
APPLICATIONS

Proteins form the basis of life by performing the functions required by every cell; proteins are made using mRNA

tRNA – TRANSFER RNA

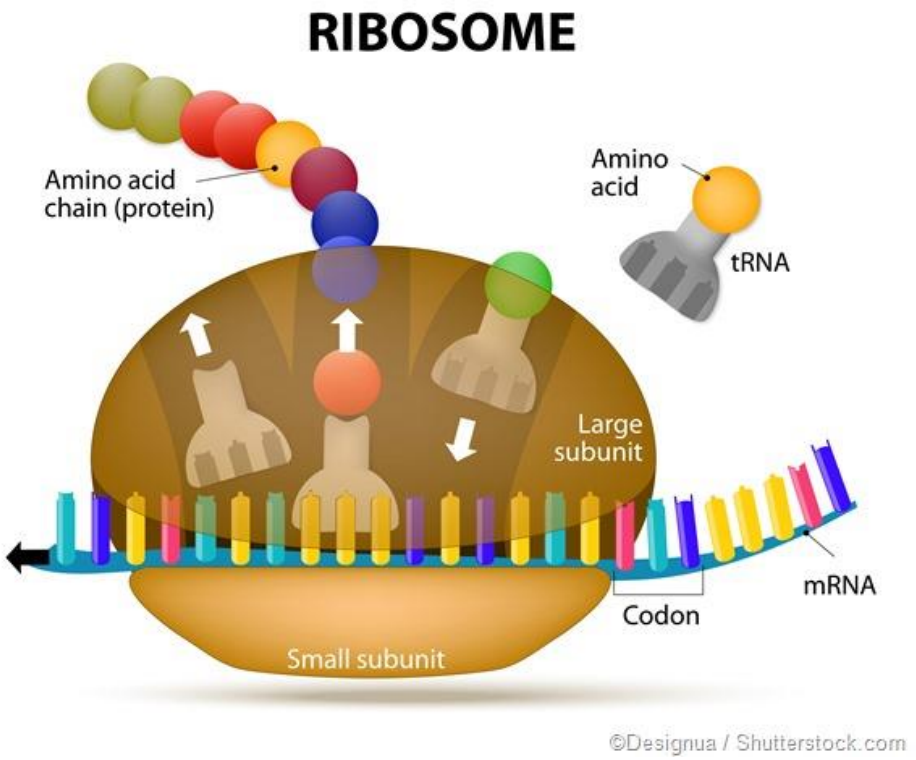
- All the tRNAs share a common secondary structure resembles a cloverleaf: They have four base-paired stems defining three stem-loops (the D loop, anticodon loop, and T loop) and the acceptor stem.

- tRNA carry correct amino acids to their position along the mRNA template to be added to the growing polypeptide chain.



rRNA – RIBOSOMAL RNA

- The central component of the ribosome.
- Ribosome; factory for protein synthesis; composed of ribosomal RNA and ribosomal proteins (known as a Ribonucleoproteinor RNP).
- rRNA provides a mechanism for decoding mRNA into amino acids.



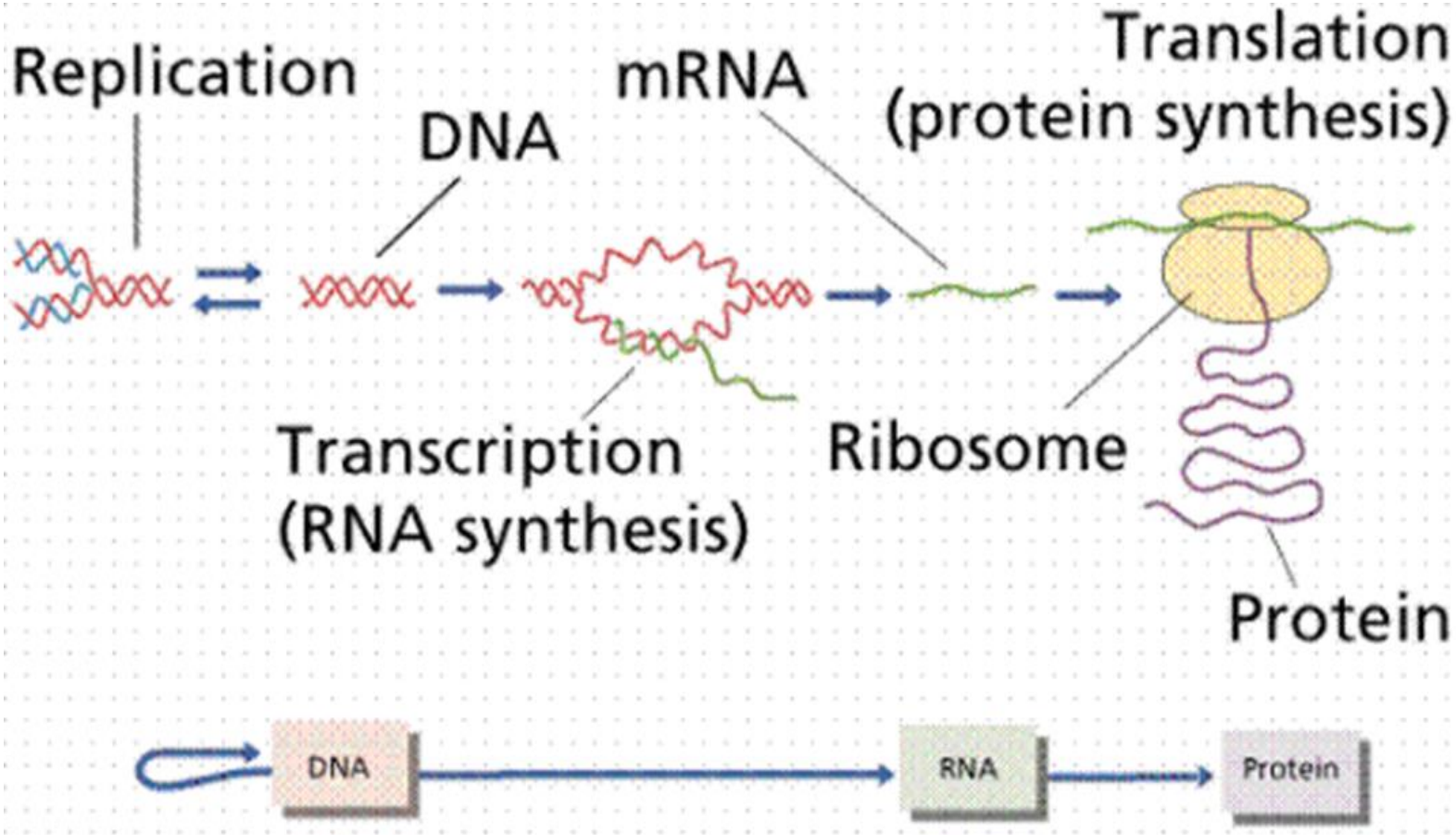
THE BASIC TENET OF MOLECULAR BIOLOGY

- DNA molecules serve as templates for either complementary DNA strands during the process of replication or complementary RNA during the process of transcription.

- RNA molecules serve as a template for ordering amino acids by ribosomes during protein synthesis.

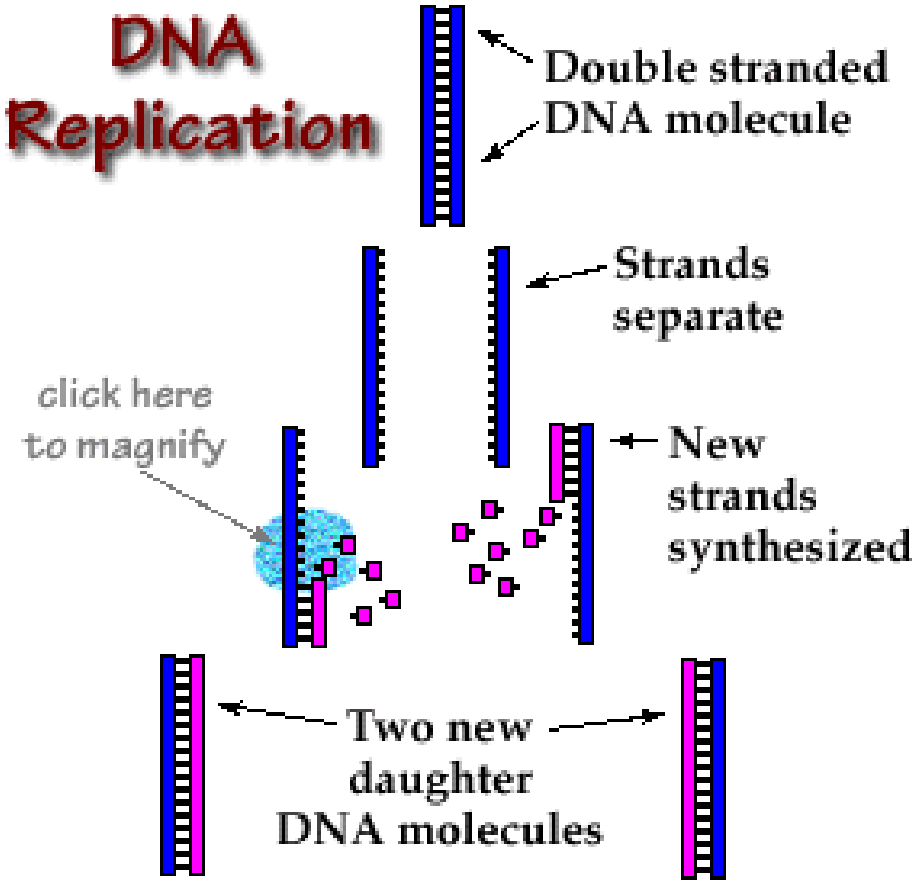
- REPLICATION – DNA MAKES MORE COPIES OF ITSELF WHEN CELLS DIVIDE
- TRANSCRIPTION – MRNA IS MADE FROM DNA
- TRANSLATION – PROTEIN IS MADE FROM MRNA

BASIS OF MOLECULAR BIOLOGY

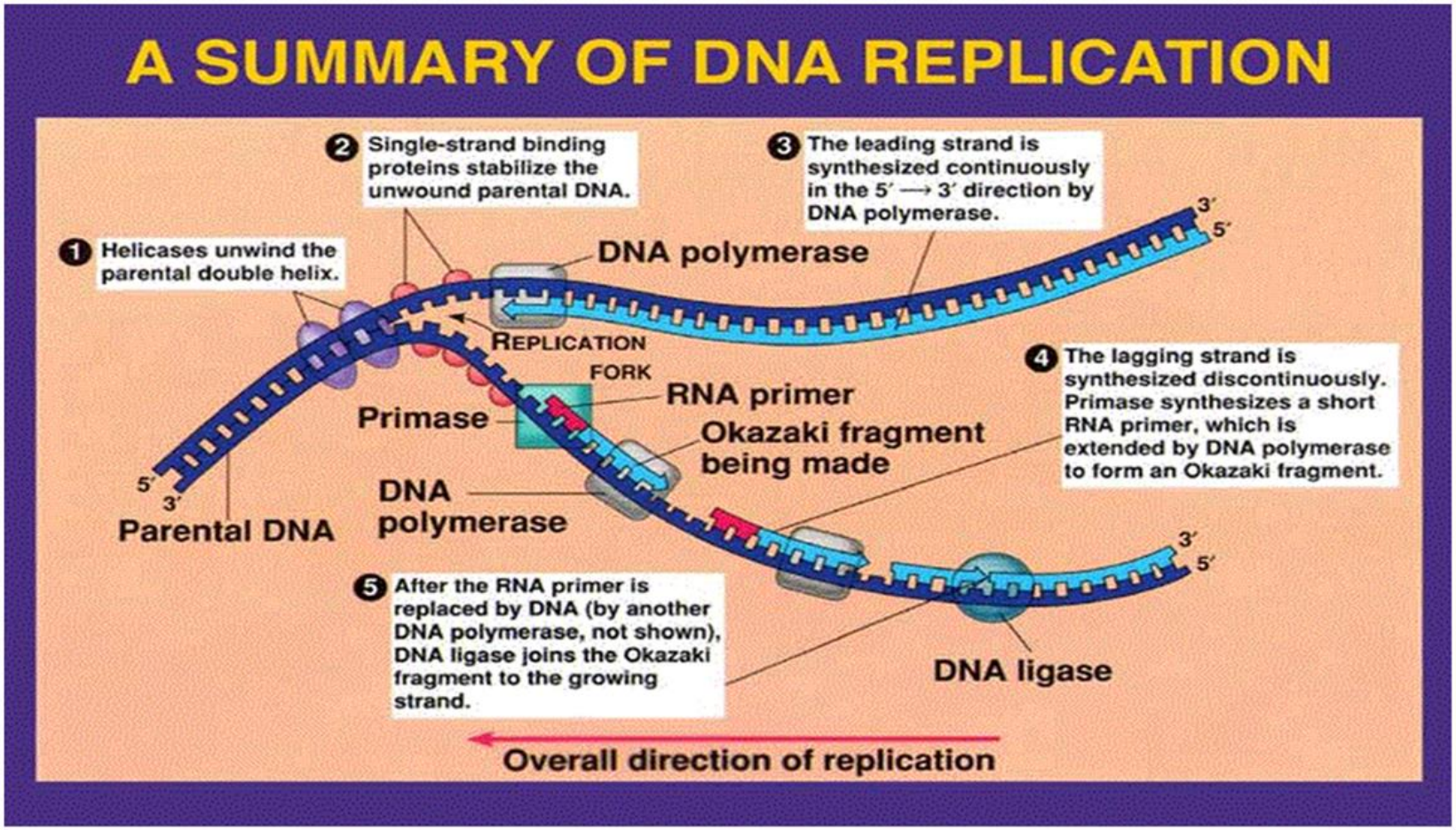


DNA REPLICATION

- The DNA duplication.
- The transfer the genetic information from a parent to a daughter cell.
- The DNA base sequences are precisely copied



DNA REPLICATION

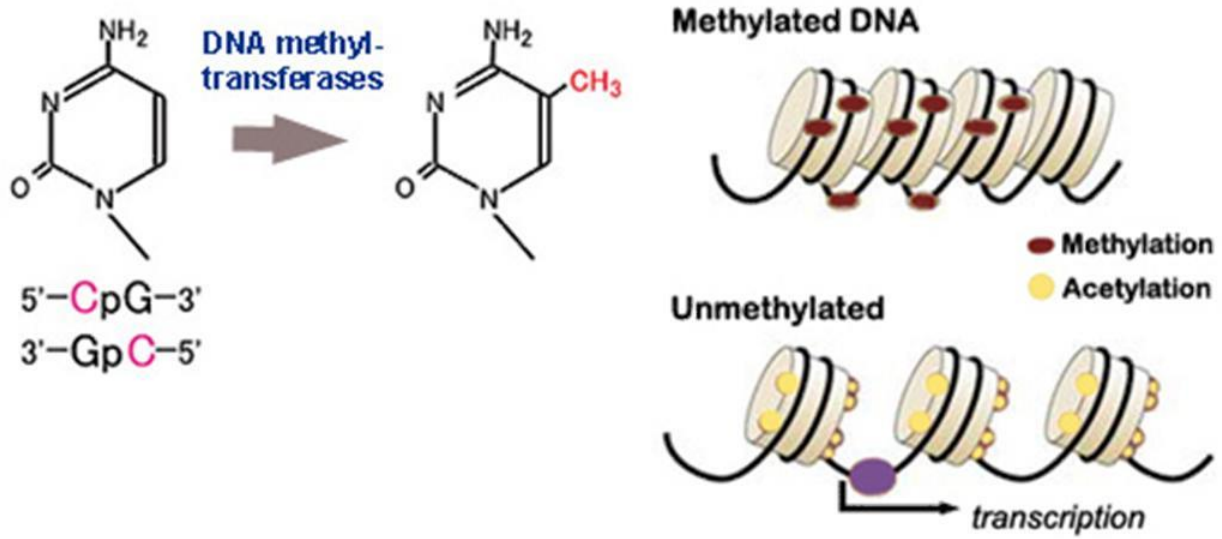


IDENTICAL TWINS

- Identical twins share the same DNA but still they are different. Why??
- Example – Different Hair Color
- POST REPLICATION DNA MODIFICATION

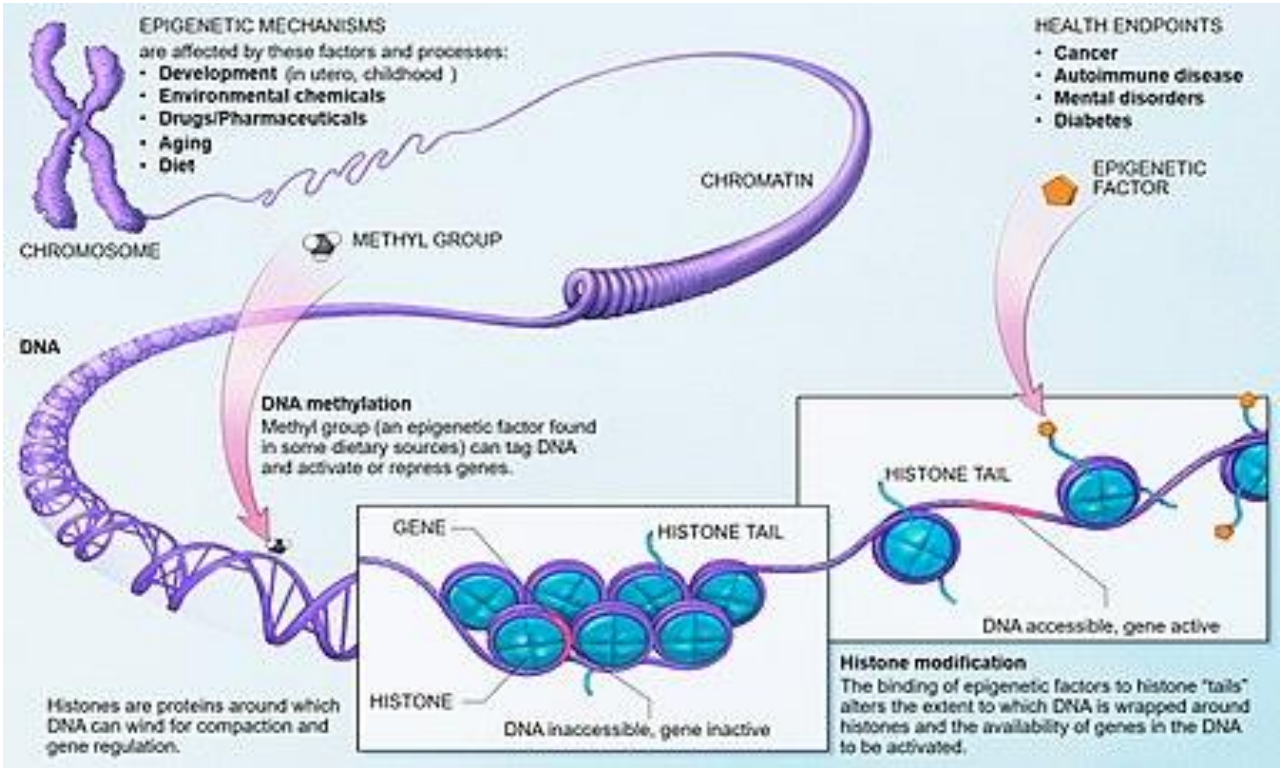
POST REPLICATION DNA MODIFICATION - METHYLATION

- Methylation; one of the major post- replicative reactions.
- Site of methylation of eukaryotic DNA is always on cytosine residues in CG dinucleotide.
- DNA methylation plays an important role for epigenetic gene regulation in development and disease.



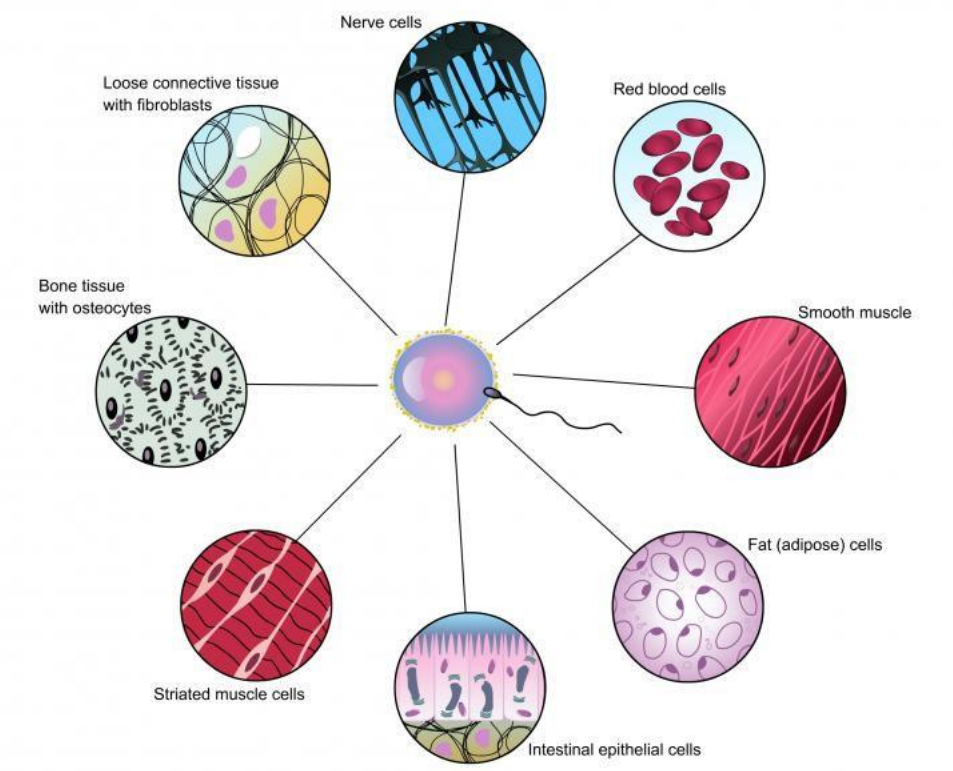
EPIGENETICS

- "Epi" – over, above, outer
- Epigenetics – stably heritable phenotype changes in a chromosome without alterations in the DNA sequence
- Histone modifications
- DNA methylation



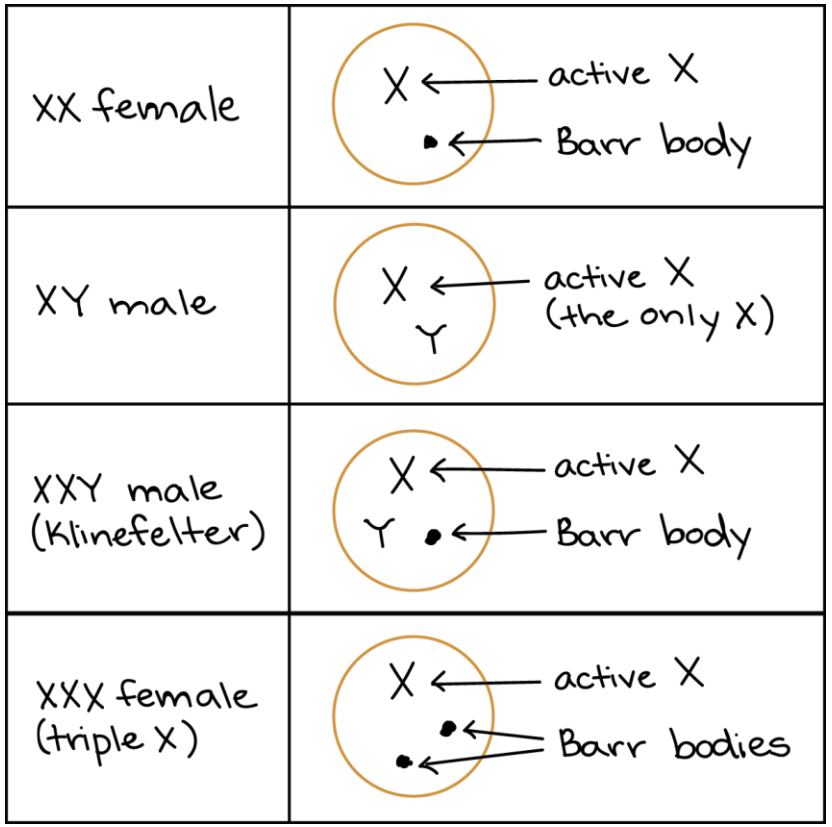
CELLULAR DIFFERENTIATION - EPIGENETICS

- Different types of cells share same DNA but yet are different cells
- What makes a liver cell vs neuronal cell ?
- Epigenetic control of cell fate
- Neuronal genes are turned on in neuronal environment promoting neuronal differentiation.

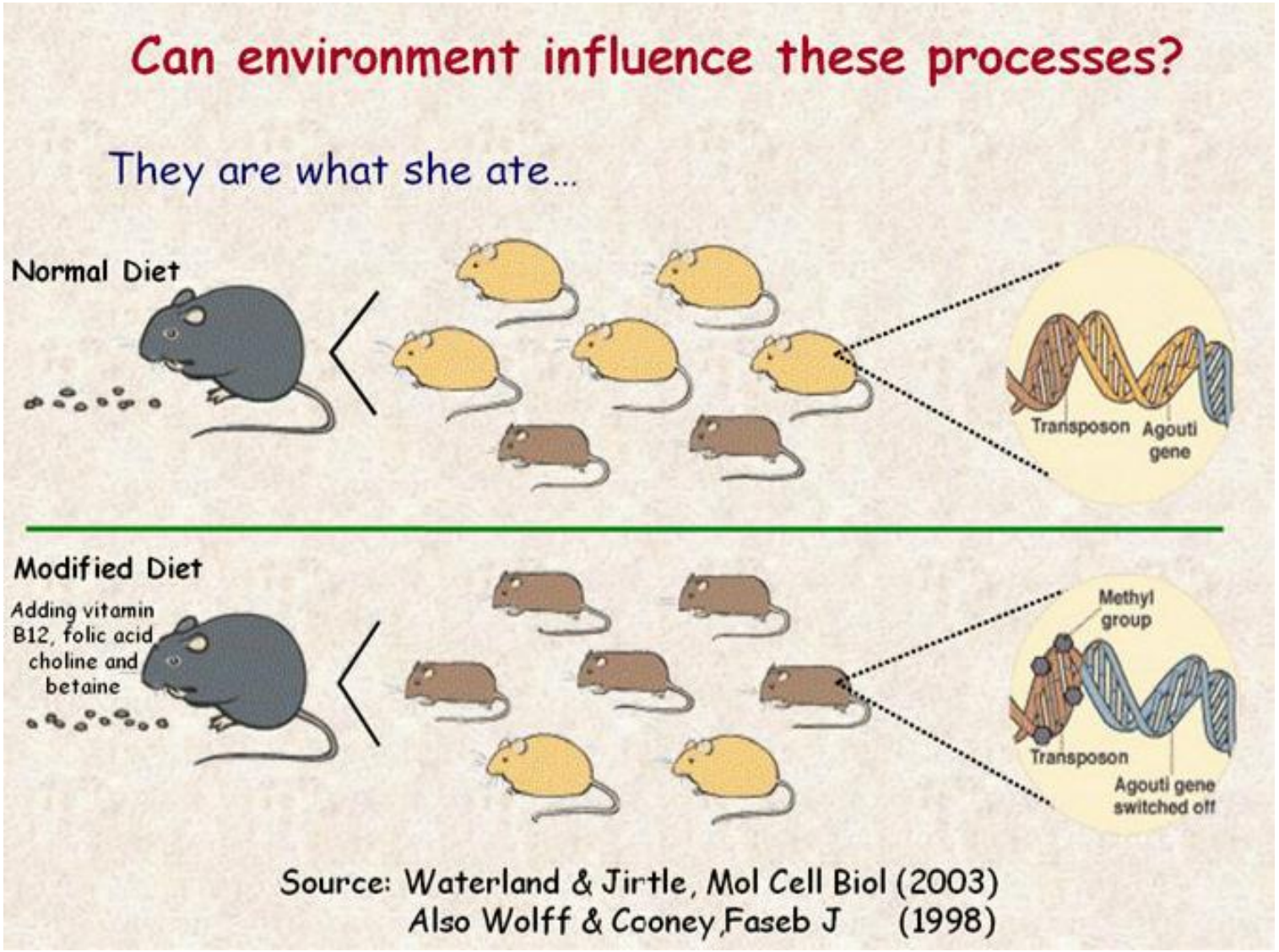


X INACTIVATION - EPIGENETICS

- Male: XY, Female: XX
- As nearly all female mammals have two X chromosomes, X-inactivation prevents them from having twice as many X chromosome gene products as males, who only possess a single copy of the X chromosome
- Inactive X chromosome also referred to as Barr Body



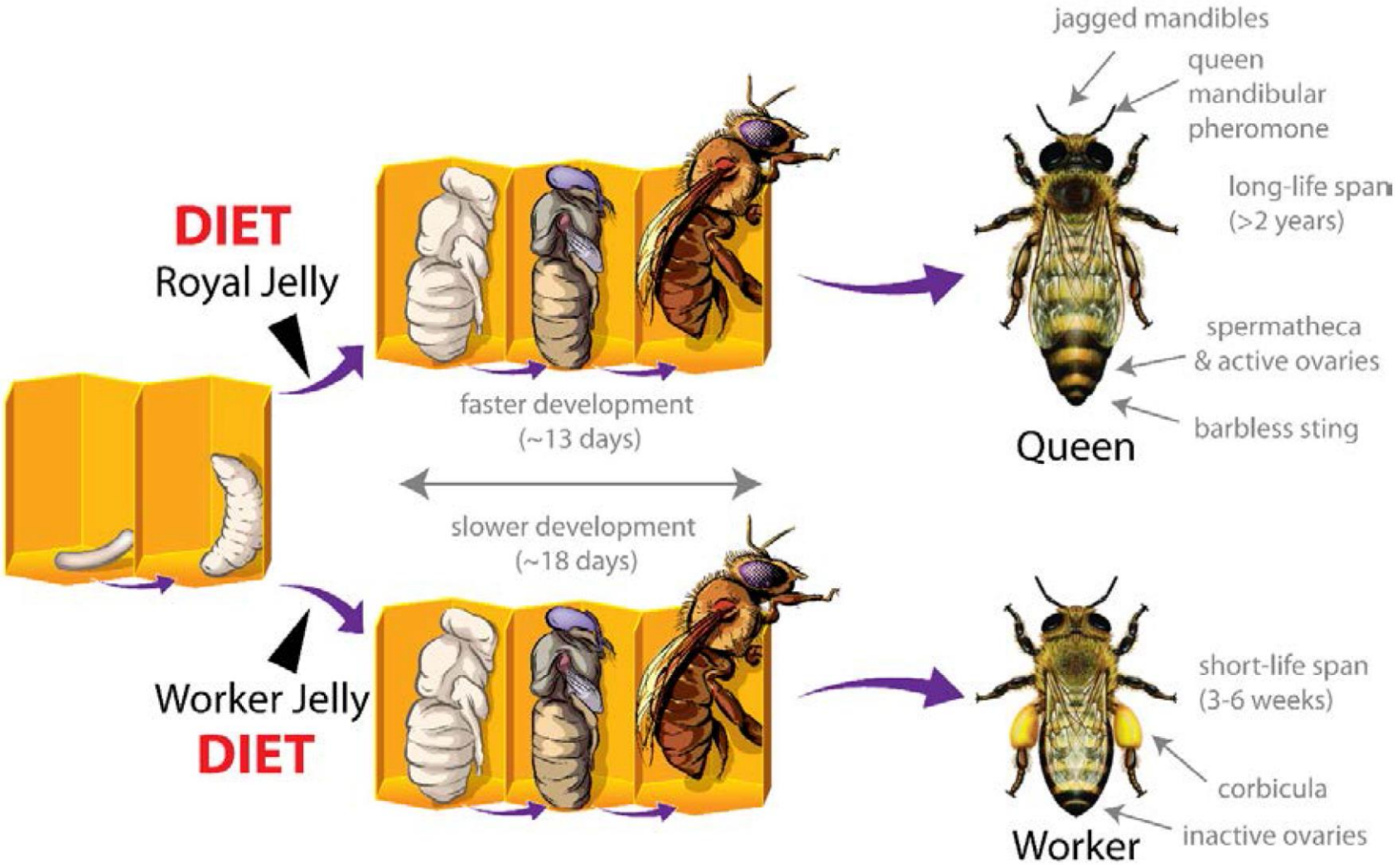
EPIGENETICS: DIETARY REGULATION



EPIGENETICS: DIETARY REGULATION

- Queen bee shares the same DNA as the bee workers – then what makes her the queen with the ability to reproduce ?
- ROYAL JELLY
- Royal jelly is a complex, protein-rich substance secreted from glands on the heads of worker bees. A larva destined to become a queen is fed large amounts of royal jelly inside a compartment called a queen cup.
- Royal jelly silences a key gene (Dnmt3), which codes for an enzyme that silences a group of queen genes. When Dnmt3 is turned "on," the queen genes are epigenetically silenced, and the larvae develop into the default "worker" variety. But when royal jelly turns Dnmt3 "off," the queen genes jump into action, turning the larvae into queens.

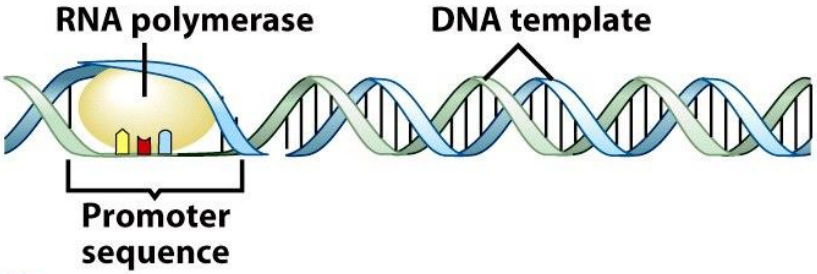
EPIGENETICS: DIETARY REGULATION



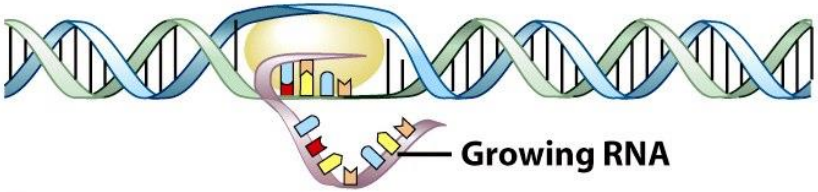
EPIGENETICS: DIETARY REGULATION

- German's blocked food to the Dutch in the winter of 1944.
- Calorie consumption dropped from 2,000 to 500 per day for 4.5 million.
- Children born or raised in this time were small, short in stature and had many diseases including, edema, anemia, diabetes and depression.
- The Dutch Famine Birth Cohort study showed that women living during this time had children 20-30 years later with the same problems despite being conceived and born during a normal dietary state.

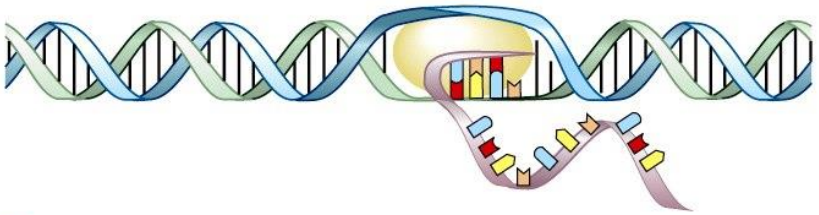
TRANSCRIPTION – DNA TO mRNA



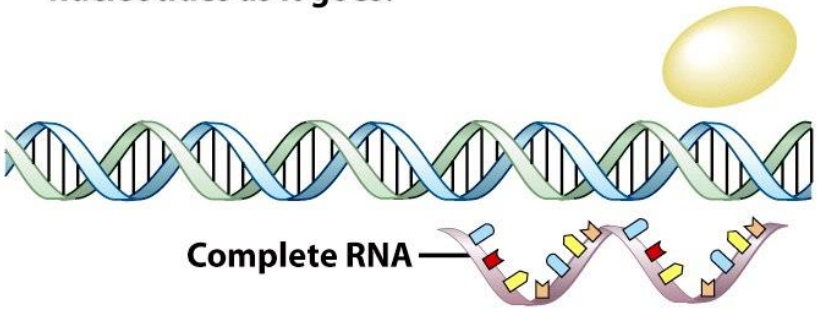
1. The RNA polymerase binds to the DNA at specific promoter sequences. The two strands of DNA are separated at a site near the promoter.



2. Complementary RNA nucleotides bind to only one of the exposed DNA strands. RNA polymerase joins the RNA nucleotides together to form a chain.



3. The RNA polymerase moves along the DNA, separating the strands and joining matched RNA nucleotides as it goes.

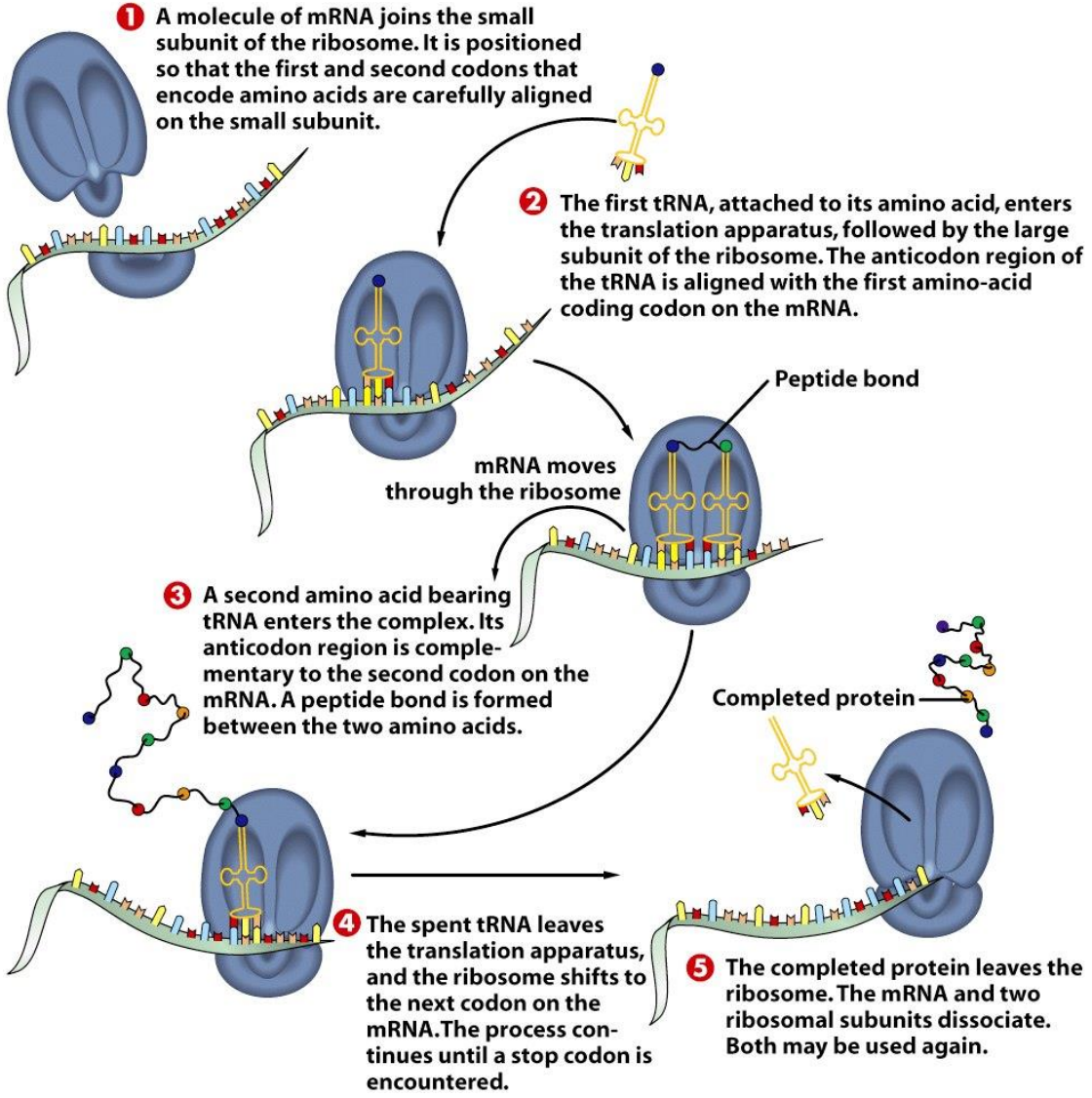


4. At the end of the coding sequence, the RNA polymerase and the newly synthesized RNA leave the DNA. The double helix closes again.

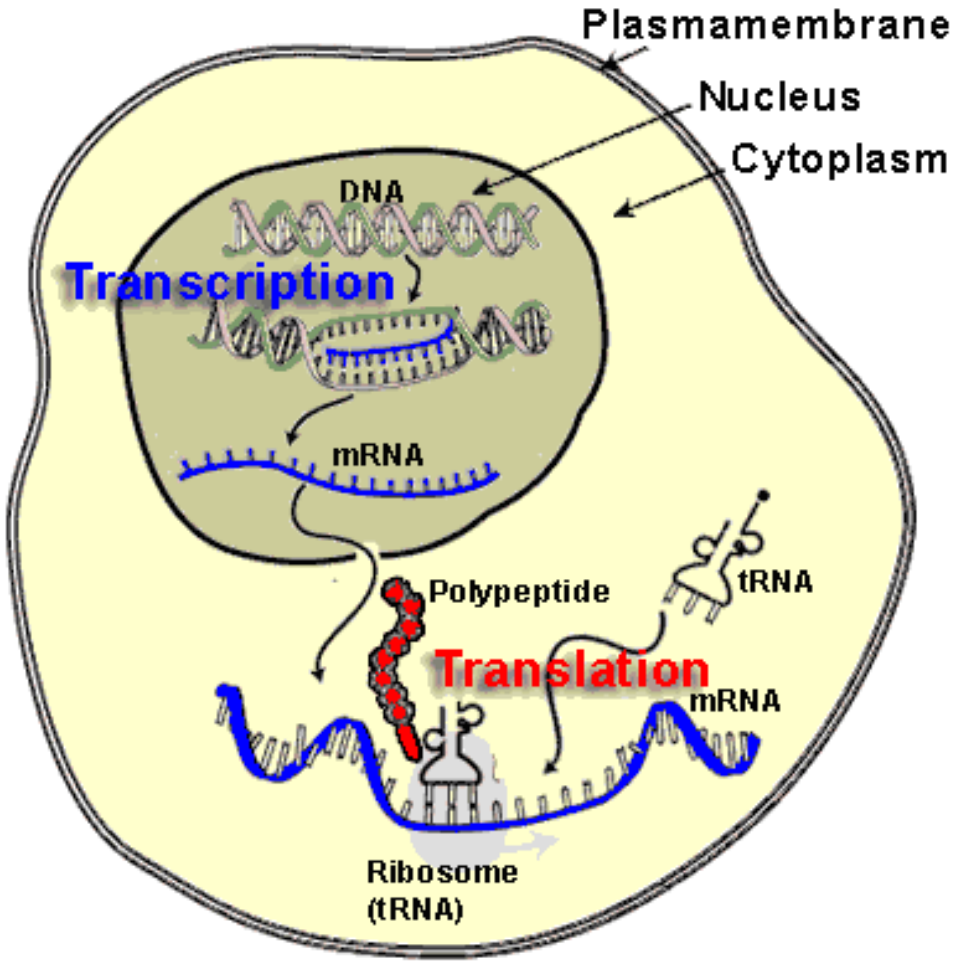
TRANSLATION – MRNA TO PROTEIN

- Occurs on ribosomes, mRNA translated to determine the sequence of amino acid in the protein being synthesized.
- Ribosomes are factory for protein synthesis.
- Ribosomes are composed of ribosomal RNA and ribosomal proteins (known as a Ribonucleoprotein or RNP).
- Translate (mRNA) to build polypeptide chains using amino acids delivered by (tRNA).
- Codon (mRNA) must be complementary to the anticodon (tRNA).
- Translation continues until ribosome encounters a stop codon.

TRANSLATION



FROM DNA TO PROTEIN



WHAT IS A PROTEIN ?

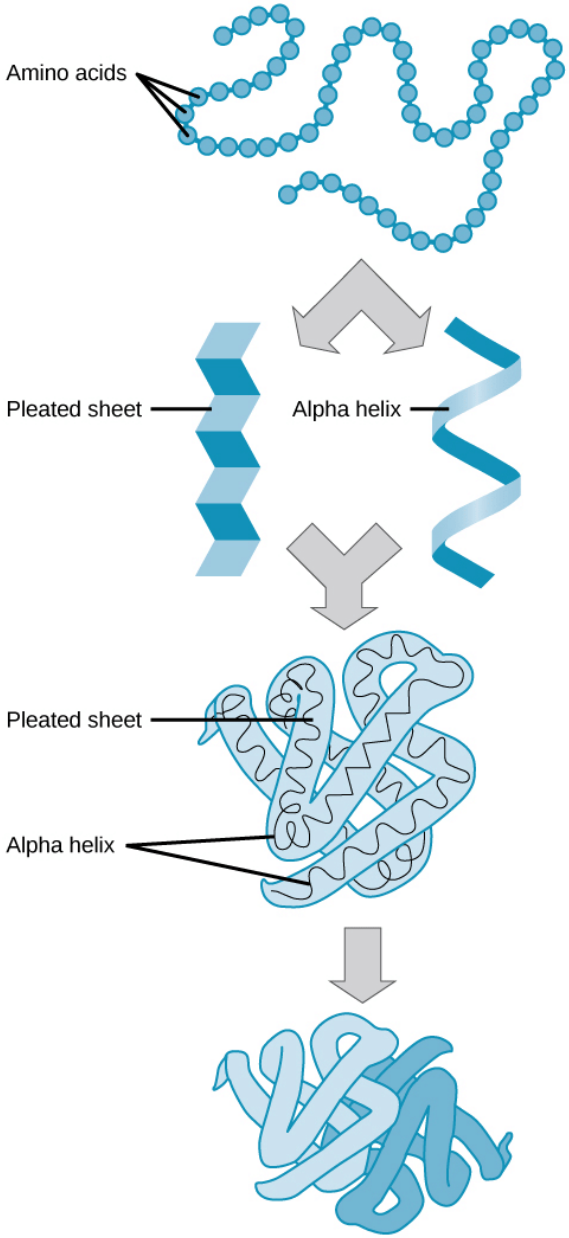
- Proteins are chain like polymers of a few or many thousands of amino acids.
- Amino acids: (Encoded by 3-nucleotide RNA sequences) (codon).
- 20 AMINO ACIDS

First Letter	Second Letter				Third Letter
	U	C	A	G	
U	phenylalanine	serine	tyrosine	cysteine	U
	phenylalanine	serine	tyrosine	cysteine	C
	leucine	serine	stop	stop	A
	leucine	serine	stop	tryptophan	G
C	leucine	proline	histidine	arginine	U
	leucine	proline	histidine	arginine	C
	leucine	proline	glutamine	arginine	A
	leucine	proline	glutamine	arginine	G
A	isoleucine	threonine	asparagine	serine	U
	isoleucine	threonine	asparagine	serine	C
	isoleucine	threonine	lysine	arginine	A
	(start) methionine	threonine	lysine	arginine	G
G	valine	alanine	aspartate	glycine	U
	valine	alanine	aspartate	glycine	C
	valine	alanine	glutamate	glycine	A
	valine	alanine	glutamate	glycine	G

PROTEIN STRUCTURE

- Primary protein structure: Sequence of a chain of amino acid.
- Secondary protein structure: A chain of amino acids linked by hydrogen bonds.
- Tertiary protein structure: It occurs when certain attraction occurs between alpha helices and pleated sheets.
- Quaternary protein structure: Protein containing more than one amino acid chains.

PROTEIN STRUCTURE



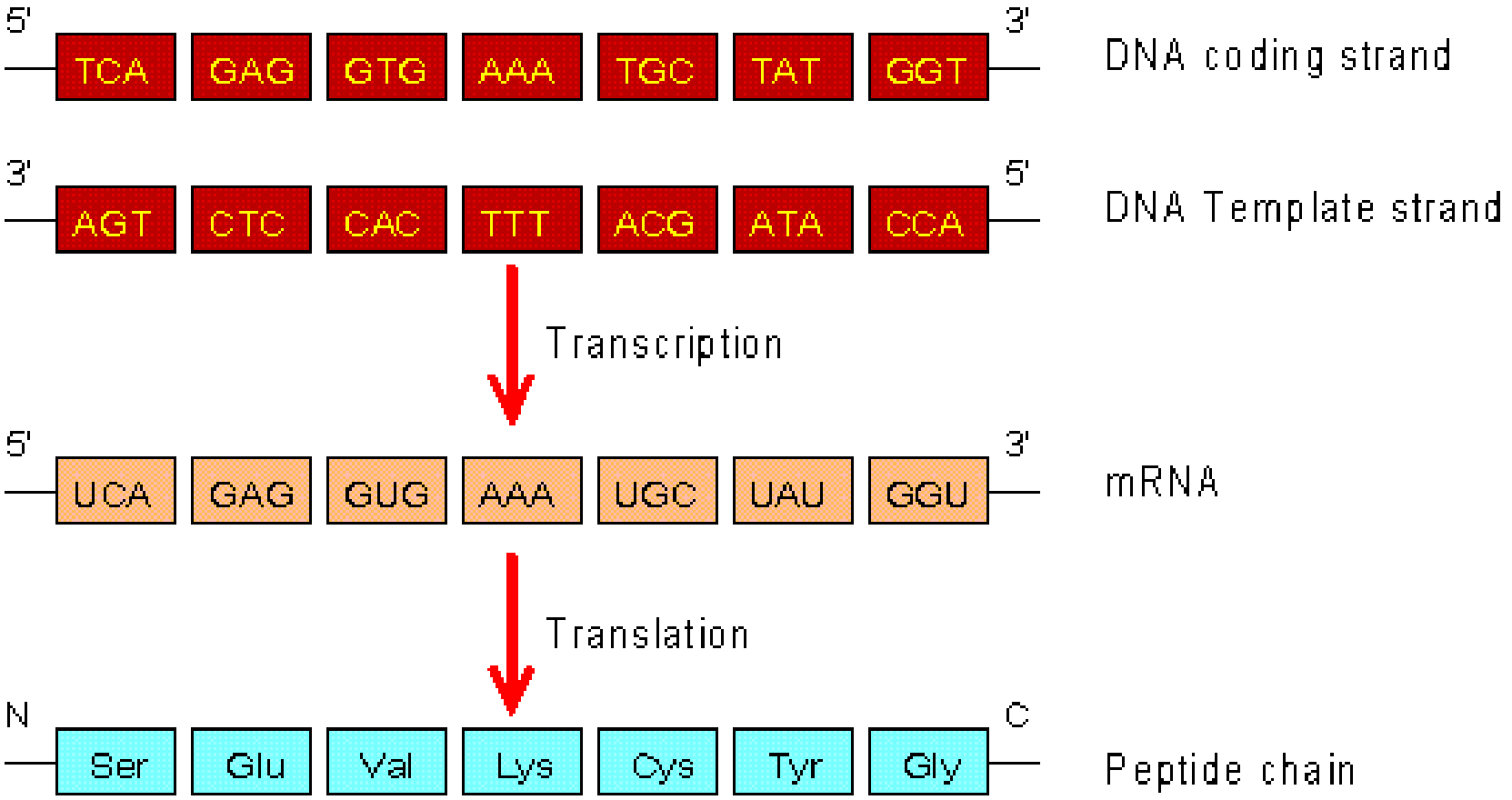
Primary Protein structure
sequence of a chain of animo acids

Secondary Protein structure
hydrogen bonding of the peptide backbone causes the amino acids to fold into a repeating pattern

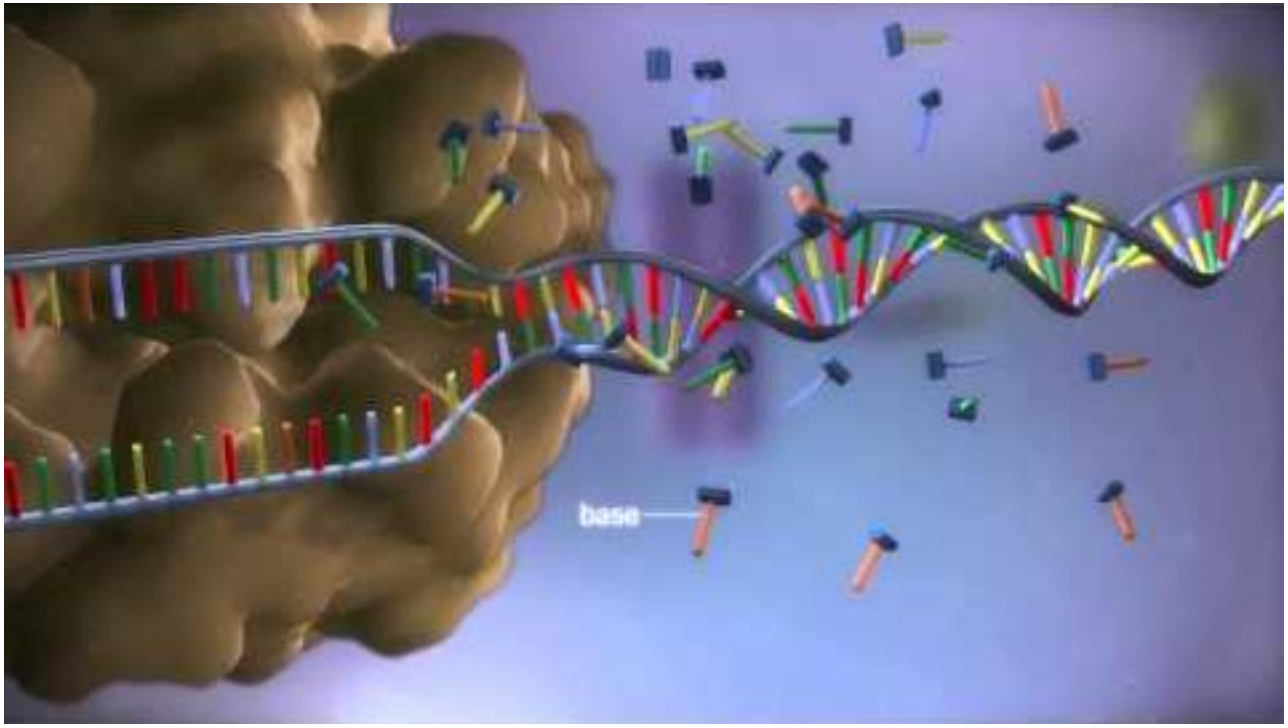
Tertiary protein structure
three-dimensional folding pattern of a protein due to side chain interactions

Quaternary protein structure
protein consisting of more than one amino acid chain

DNA TO PROTEIN



DNA TO PROTEIN



https://www.youtube.com/watch?v=gG7uCskUOrA

REVIEW/DISCUSSION POINTS

- Difference between RNA and DNA
- Types of RNA
- Replication
- Transcription
- Translation
- Epigenetics & DNA methylation

HOW DOES DNA CHANGE OVER TIME ?

- Mutations: a permanent change in the genetic material of a cell or organism.
- Can be inherited.
- Can involve whole chromosomes or changes in DNA sequences.
- Mutations are the basis to evolution.
- Not all mutations will have an effect.
- Some mutations may have a deleterious effect.
- Some mutations may have advantageous effect.

WHOLE CHROMOSOME MUTATIONS

- Polyploid: organism or cell containing three or more sets of chromosomes.
- Occurs due to a cell division error.
- Frequently seen in plants, rare in animals.
- Can have advantageous results.

- Nondisjunction: instances when paired chromosomes fail to separate during mitosis or meiosis
- Can result in an aneuploid: individual whose chromosome number is greater or less than normal

WHOLE CHROMOSOME MUTATIONS

- Down's Syndrome
- Due to nondisjunction with chromosome 21.
- Characterized by an impact on brain development, distinctive facial features.

WHOLE CHROMOSOME MUTATIONS

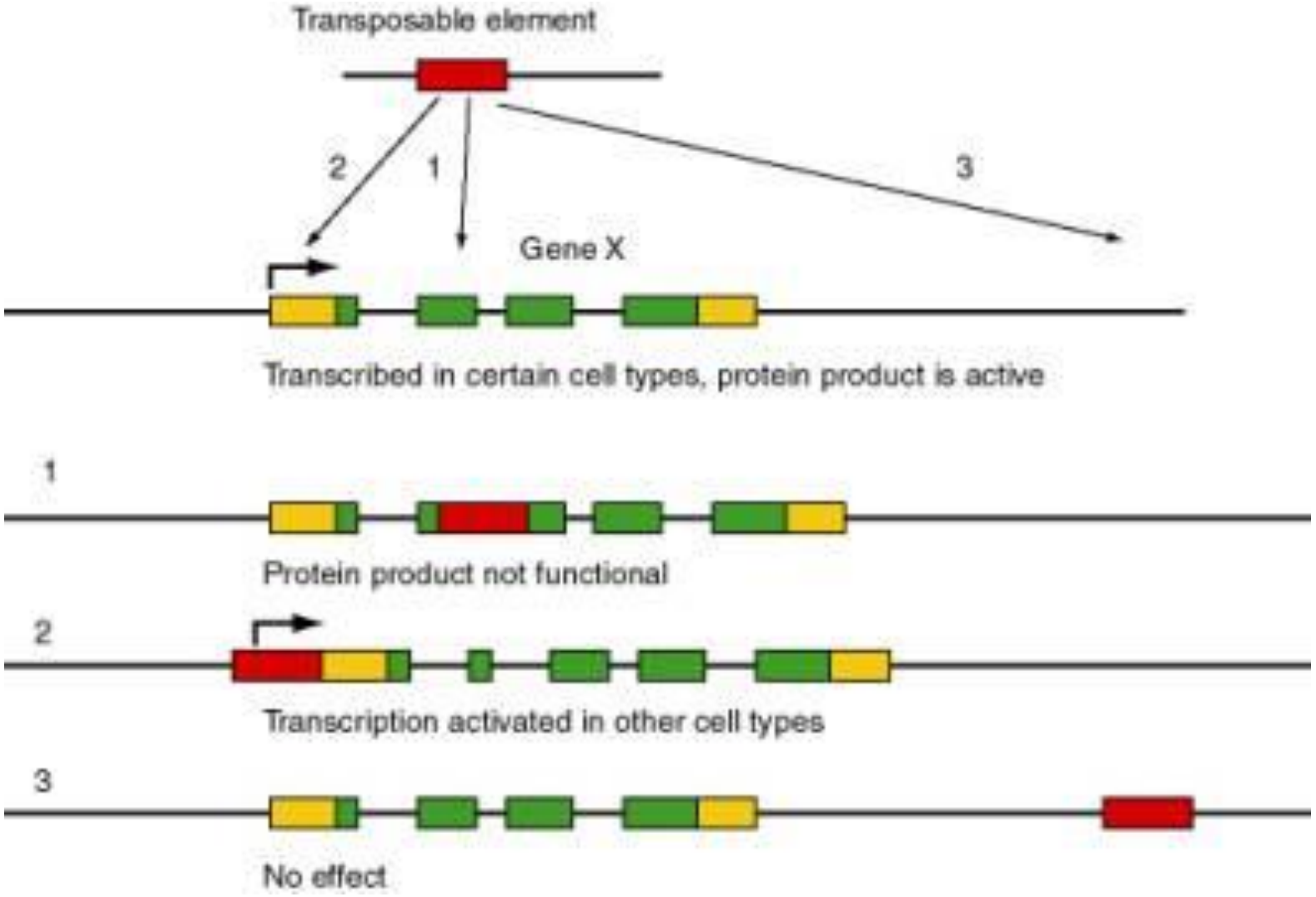
- Transposons:
- Variety of DNA sequences that can randomly insert themselves by transposition in various non-homologous regions on chromosomes and other DNA.

- Can generate new gene combinations

- Can also induce genetic errors

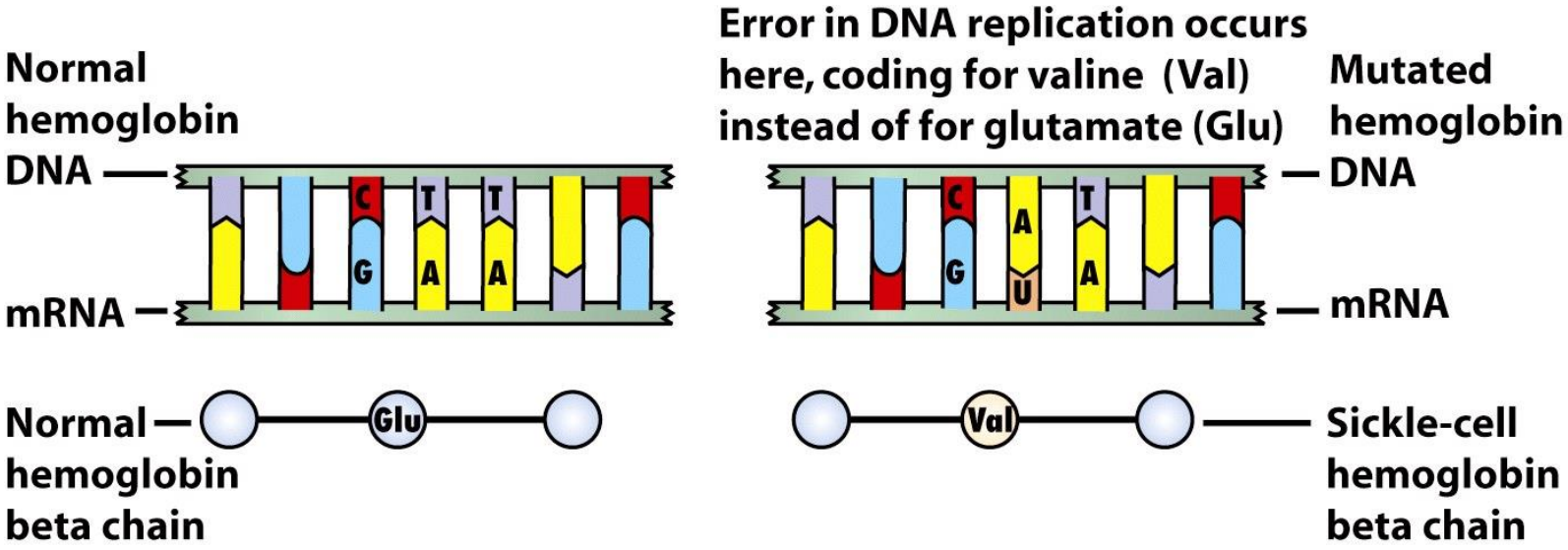
- Diseases often caused by TEs include hemophilia A and B, severe combined immunodeficiency, porphyria, predisposition to cancer, and Duchenne muscular dystrophy.

HOW DO TRANSPOSONS IMPACT GENE FUNCTION?

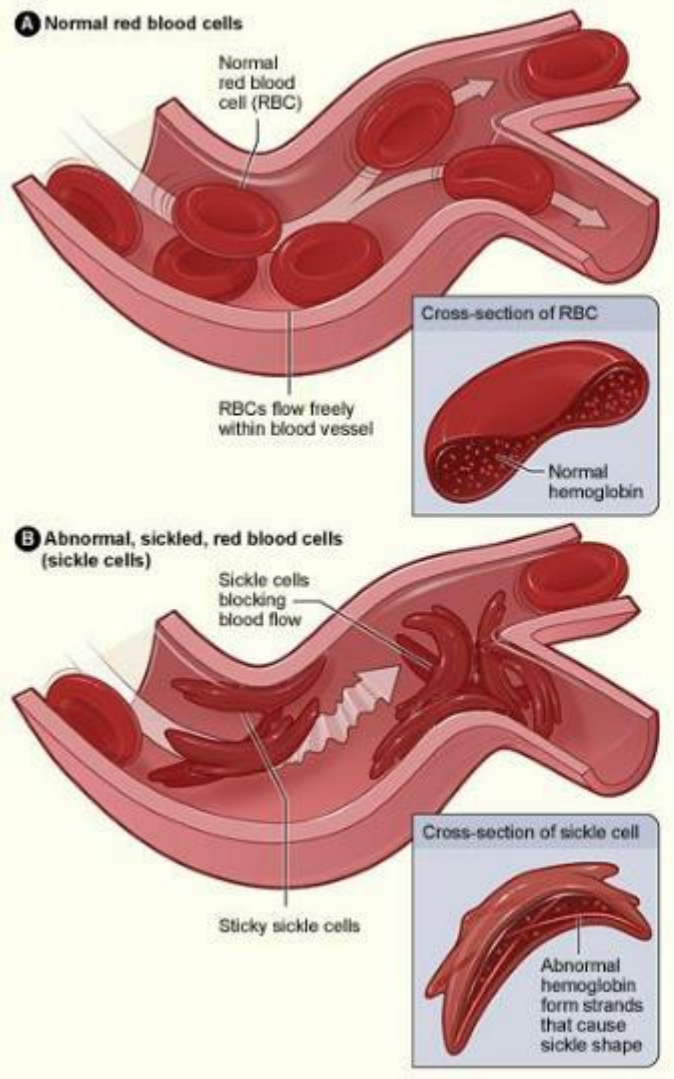


MUTATIONS: SINGLE DNA NUCLEOTIDES

- Point Mutations:
- Change in a single nucleotide base pair.
- Example: sickle cell anemia.



SICKLE CELL ANEMIA



Sickle cell anemia is an inherited disease where normal red blood cells sickle in shape.

They sickle because HgbS, (abnormal form of hemoglobin protein), is produced instead of normal HgbA.

The sickle shape of the cell ↓'s the ability of the RBC's to carry O_2 & move through the capillary. They often clump together forming clots.

MUTATIONS: SINGLE DNA NUCLEOTIDES

- Frame-shift mutation:
- A change in the reading frame resulting from an insertion or deletion of nucleotides in the DNA sequence for a protein.
- Extremely harmful.
- Can completely change the amino acid sequence of the protein following the point where mutation is introduced.

Normal:

JOE ATE THE HOT DOG

After deletion:

JEA THE OTD OG

Examples of Mutations in the DNA Sequence…

aaaccatctaggctatattcggatatcgatctatcggatctatctactagagctactacgatcagggactactacgagcatcgactacgag
gcttctagaggctatattctaggctactacgatcgatctacgtagctacgagatcgtgtgtgggggggacacagcgatctaatataaatc
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Normal

Examples of Mutations in the DNA Sequence…

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aaaccatctaggctatattcggatatcgatctatcggatctatctactagagctactacgatcagggactactacgagcatcgactacgag
gcttctagaggctatattctaggctactacgatcgatctacgtagctacgagatcgtgtgtgggggggacacagcgatctaatataaatc
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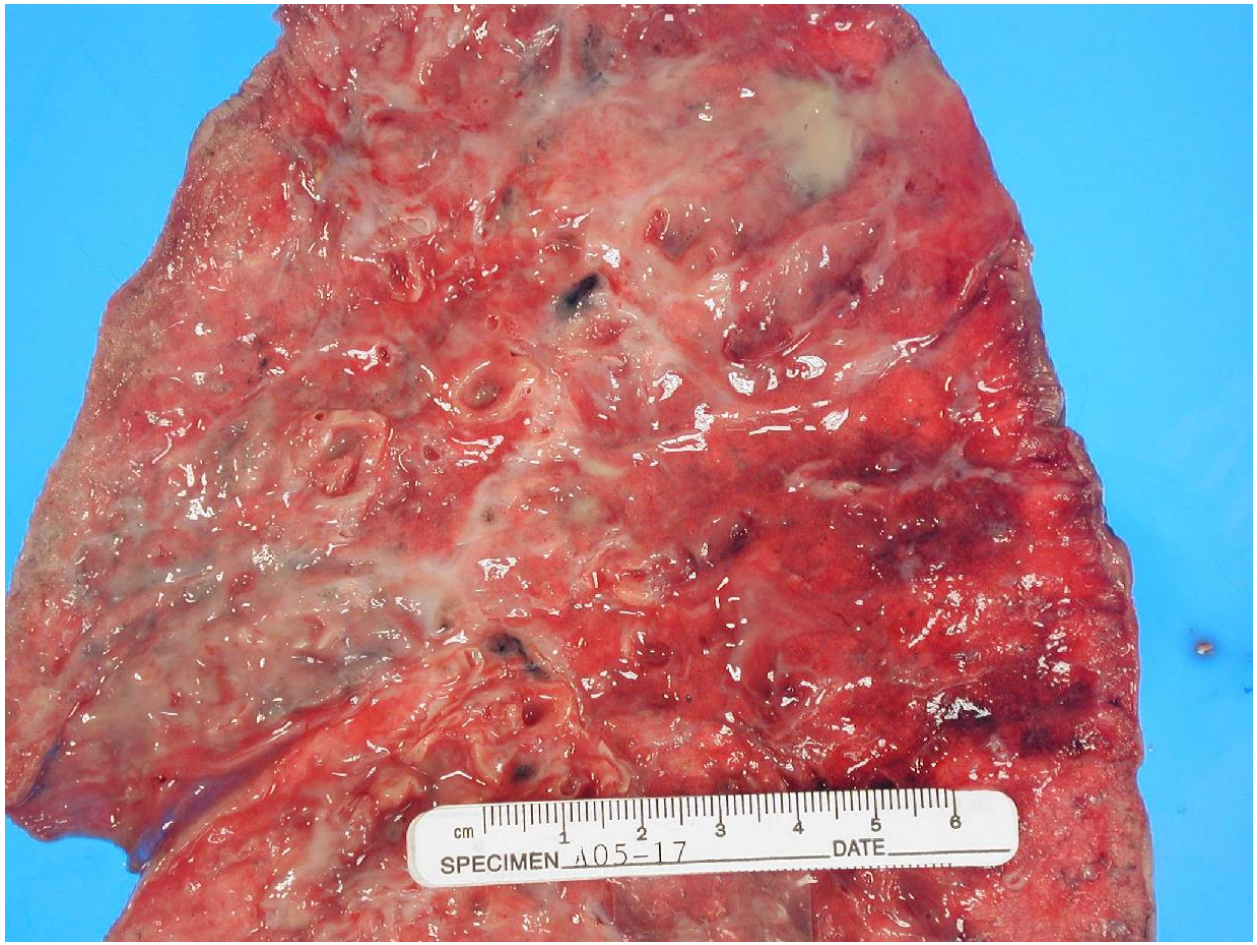
Deletion

(Cystic fibrosis)

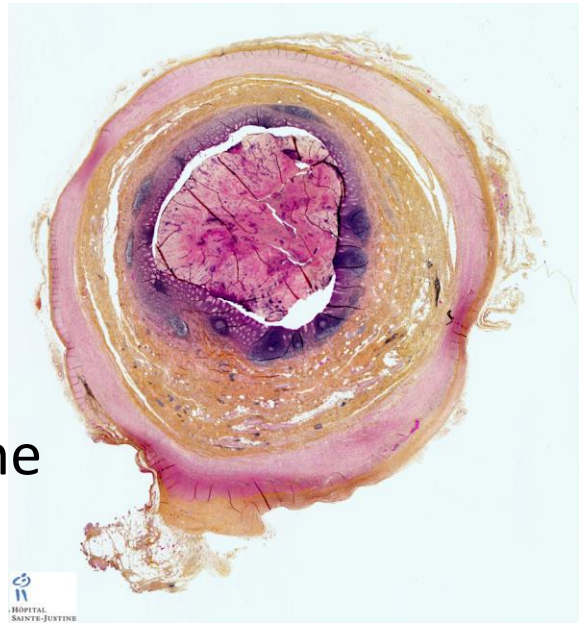
Presentation of Disease – CYSTIC FIBROSIS



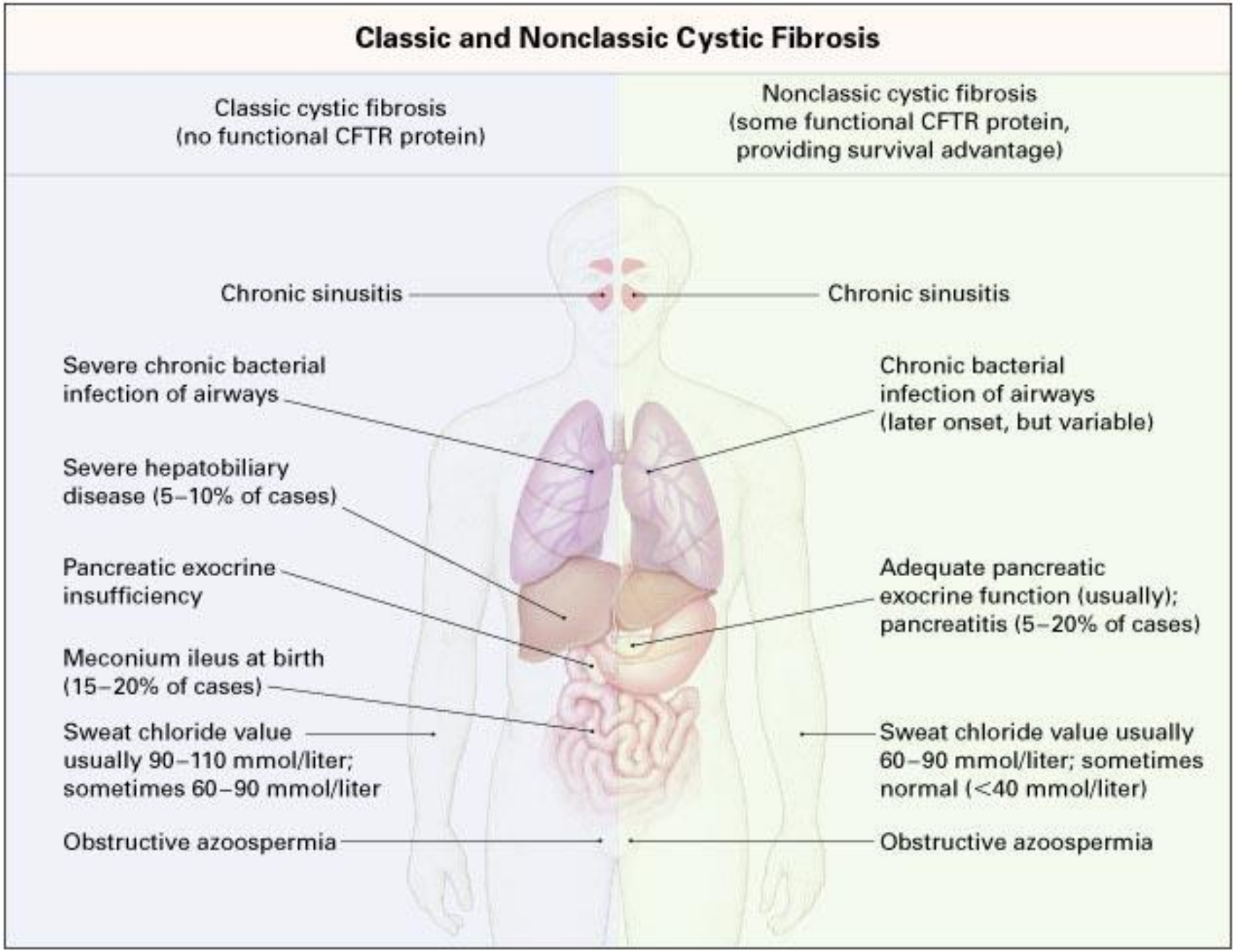
Mucous in the airways cannot be easily cleared from the lungs.

Clinical Features

- Cystic fibrosis is a heterogeneous recessive genetic disorder with features that reflect mutations in the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene.
- Classic cystic fibrosis is characterized by chronic bacterial infection of the airways and sinuses, fat maldigestion due to pancreatic exocrine insufficiency, infertility in males due to obstructive azoospermia, and elevated concentrations of chloride in sweat.
- Patients with nonclassic cystic fibrosis have at least one copy of a mutant gene that confers partial function of the CFTR protein, and such patients usually have no overt signs of maldigestion because some pancreatic exocrine function is preserved.



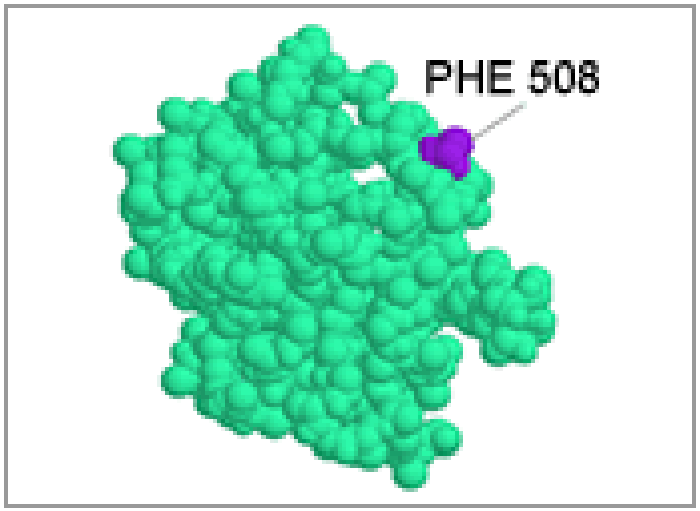
Classic and Nonclassic Cystic Fibrosis



Genotype and Phenotype

- Cystic fibrosis (CF) is caused by mutations in the CF transmembrane conductance regulator (CFTR) gene which encodes a protein expressed in the apical membrane of exocrine epithelial cells.
- This genotypic variation provides a rationale for phenotypic effects of the specific mutations. The extent to which various CFTR alleles contribute to clinical variation in CF is evaluated by genotype-phenotype studies.
- The poor correlation between CFTR genotype and severity of lung disease strongly suggests an influence of environmental and secondary genetic factors (CF modifiers).
- Several candidate genes related to innate and adaptive immune response have been implicated as pulmonary CF modifiers. In addition, the presence of a genetic CF modifier for meconium ileus has been demonstrated on human chromosome 19q13.2.
- The phenotypic spectrum associated with mutations in the CFTR gene extends beyond the classically defined CF. Besides patients with atypical CF, there are large numbers of so-called monosymptomatic diseases such as various forms of obstructive azoospermia, idiopathic pancreatitis or disseminated bronchiectasis associated with CFTR mutations uncharacteristic for CF.

Delta F508 – CFTR mutation



CFTR Sequence:

Nucleotide	A	T	C	A	T	C	T	T	T	G	G	T	G	T	T
Amino Acid	Ile			Ile			Phe			Gly			Val		
	506						508						510		

Deleted in ΔF508

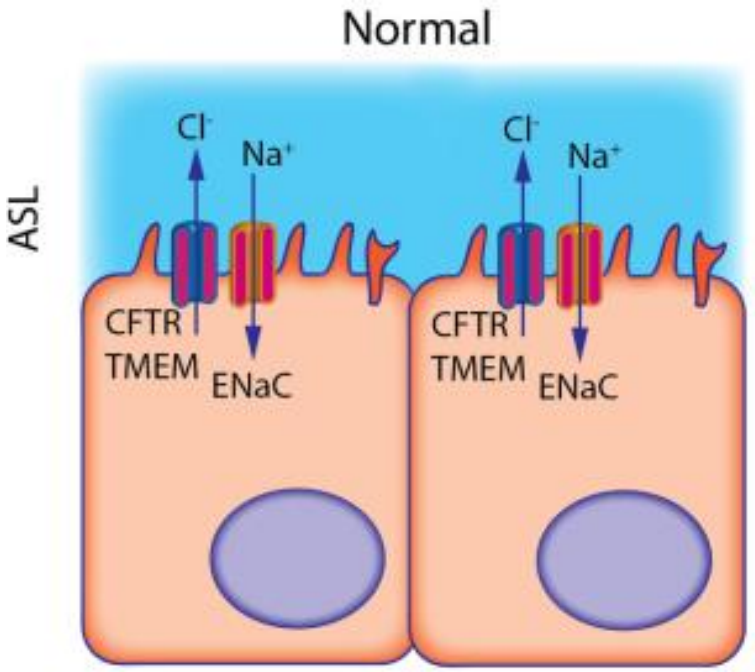
ΔF508 CFTR Sequence:

Nucleotide	A	T	C	A	T	T	G	G	T	G	T	T
Amino Acid	Ile			Ile			Gly			Val		
	506											

- When a CFTR protein with the delta F508 mutation reaches the ER, the quality-control mechanism of this cellular component recognizes that the protein is folded incorrectly and marks the defective protein for degradation. As a result, delta F508 never reaches the cell membrane.
- People who are homozygous for delta F508 mutation tend to have the most severe symptoms of cystic fibrosis due to critical loss of chloride ion transport.
- This upsets the sodium and chloride ion balance needed to maintain the normal, thin mucus layer that is easily removed by cilia lining the lungs and other organs. The sodium and chloride ion imbalance creates a thick, sticky mucus layer that cannot be removed by cilia and traps bacteria, resulting in chronic infections.

Protein Function and Biochemistry

- CFTR controls chloride ion movement in and out of the cell.
- Keeps airways hydrated so muco-ciliary action can remove debris and bacteria.



CF – GENE MUTATION

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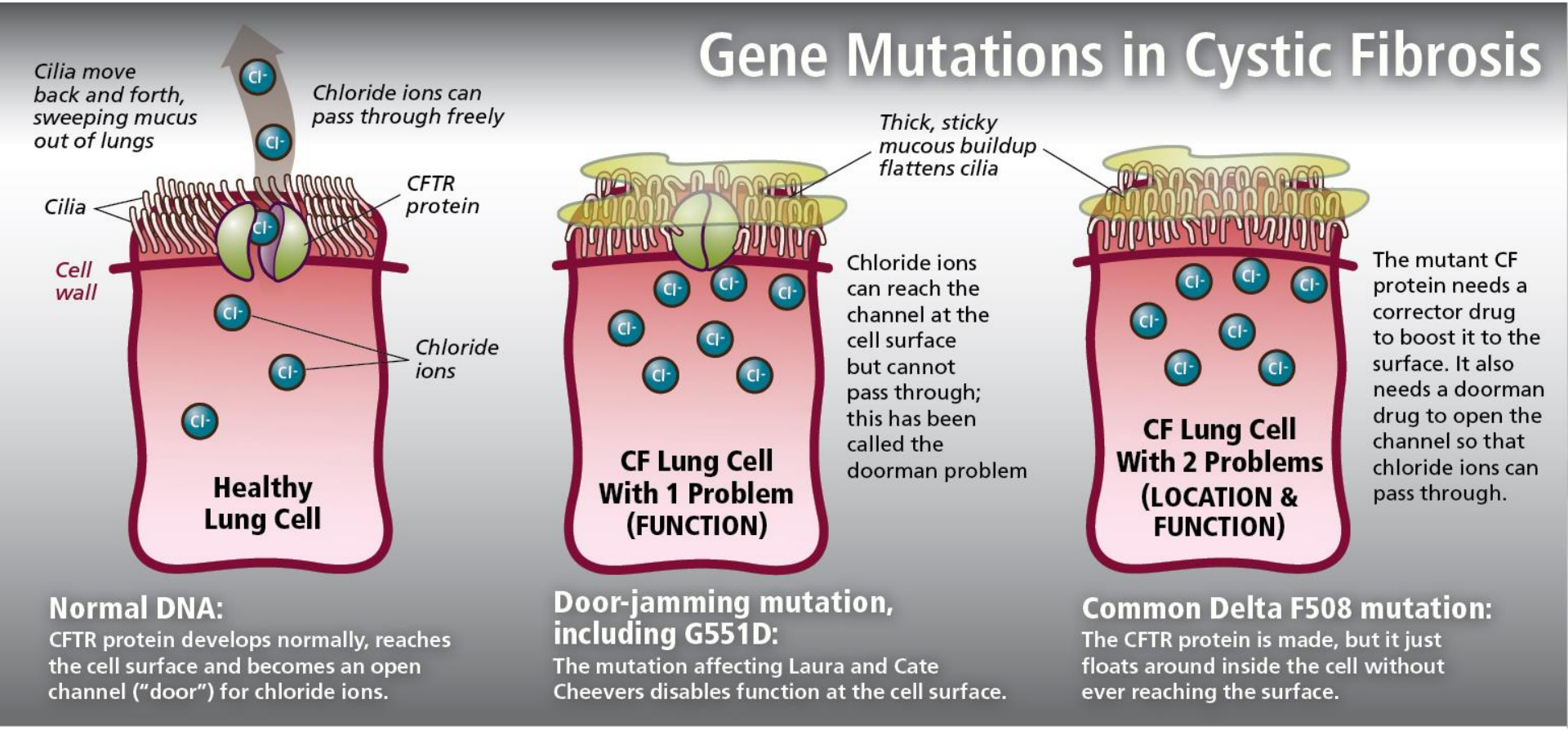
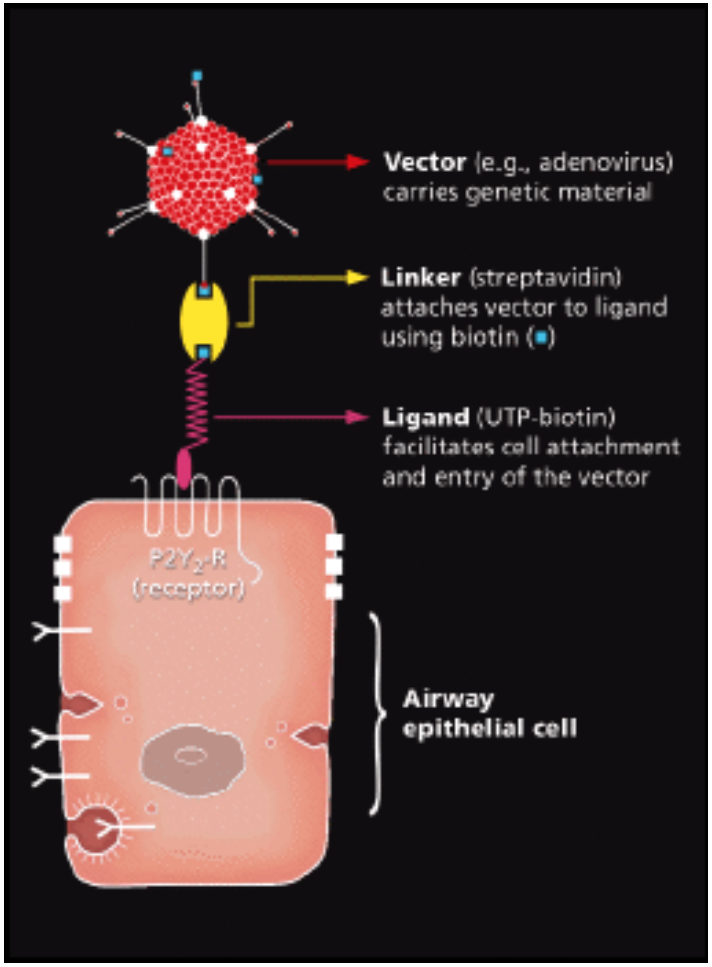


Gene Therapy

- Gene therapy is the use of normal DNA to "correct" for the damaged genes that cause disease.
- In the case of CF, gene therapy involves inhaling a spray that delivers normal DNA to the lungs.
- The goal is to replace the defective CF gene in the lungs to cure CF or slow the progression of the disease.



Examples of Mutations in the DNA Sequence…

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aaaccatctaggctatattcggatatcgatctatcggatctatctactagagctactacgatcagggactactacgagcatcgactacgag
gcttctagaggctatattctaggctactacgatcgatctacgtagctacgagatcgtgtgtgggggggacacagcgatctaatataaatc
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Frameshift or Insertion

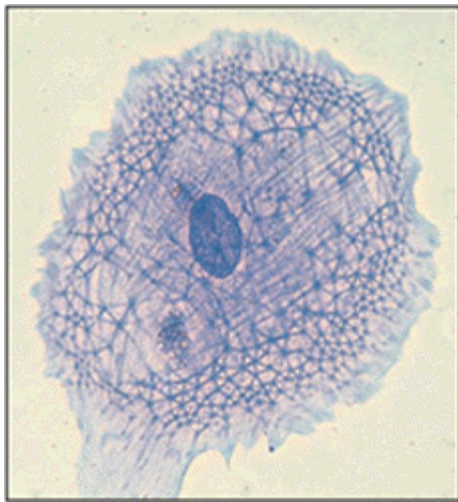
(Duchenne muscular dystrophy)

Duchenne Muscular Dystrophy

- DMD affects mostly males at a rate of 1 in 3,500 births.
- There are over 200 types of mutations that can cause any one of the forms of muscular dystrophy.
    - There are also mutations that occur within the same gene that cause other disease types.
- DMD is the most severe and common type of muscular dystrophy.
- DMD is characterized by the wasting away of muscles.
- DMD is the most aggressive form of muscular dystrophy.
- Diagnosis in boys usually occurs between 16 months and 8 years.
    - Parents are usually the first to notice problem.
- Death from DMD usually occurs by age of 30.

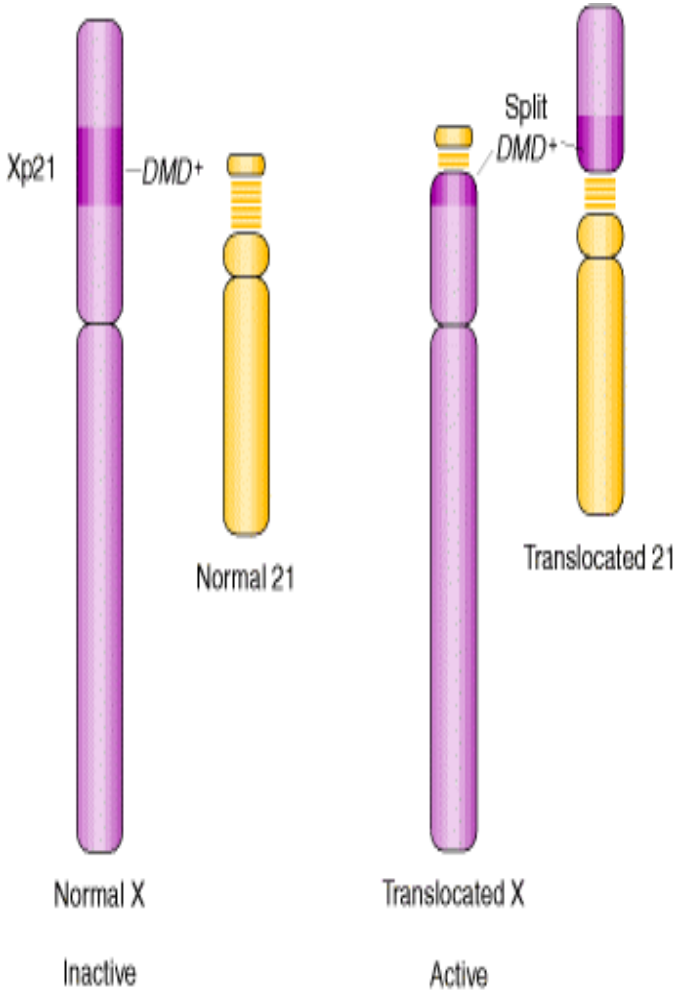
DMD Gene and Dystrophin Function

- The DMD gene encodes for the protein dystrophin, found in muscle cells and some neurons.
    - Dystrophin provides strength to muscle cells by linking the internal cytoskeleton to the surface membrane.
    - Without this structural support, the cell membrane becomes permeable. As components from outside the cell are allowed to enter the internal pressure of the cell increases until the cell bursts and dies.
        - Under normal wear and tear stem cells within the muscle regenerate new muscle cells and repair the damage.
        - In DMD the damage to muscle cells is so extreme that the supply of stem cells are exhausted and repair can no longer occur.



Clinical Features
Genotype of DMD

- Females carry the DMD gene on the X chromosome.
    - **Females are carriers and have a 50% chance of transmitting the disease in each pregnancy.**
        - **Sons who inherit the mutation will have the disease.**
        - **Daughters that inherit the mutation will be carriers.**
- The DMD gene is located on the Xp 21 band of the X chromosome.
- Mutations which affect the DMD gene.
    - **96% are frameshift mutations**
    - **30% are new mutations**
    - **10-20% of new mutations occur in the gametocyte (sex cell, will be pass on to the next generation).**
- **The most common mutation are repeats of the CAG nucleotides.**



Examples of Mutations in the DNA Sequence…

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ggctatattctaggctactacgatcgatctacgtagctacgagatcgtgtgtgggggggacacagcgatctaatataaatctgatgatca
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Insertion

(Huntington’s disease)

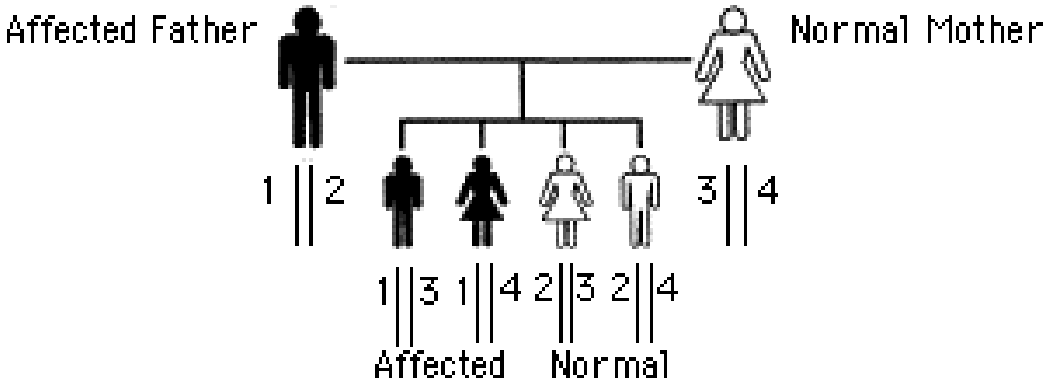
Clinical Features

- Huntington's disease is a rapidly progressive neurodegenerative disease that leads to dementia.
- Typically presents with alterations in mood as well as a change in character, defects in memory and attention.
- Progresses to a movement disorder consisting of involuntary, rapid motions.
- Usually not recognized until the patient is in their early 30's.

Genotype/Phenotype

- Huntington's is autosomal dominant.
  - This means that anyone with ONE abnormal copy of the gene will clinically have the disease.
  - There are no carriers for Huntington's.

- A parent with Huntington's will have a 50% chance of passing it on to their child.



Genomic Information

- The HD gene is located on chromosome 4, in the p-arm (4p16.3)
- It is a very large gene, consisting

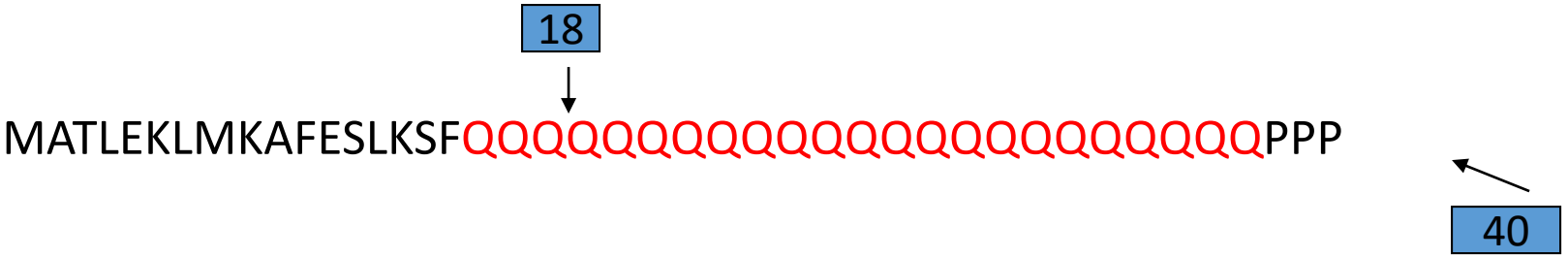
  of 169,280 base pairs.
- Contains 67 exons, which are spliced together the mRNA template.

Pathology

- The exact function of the normal Huntingtin protein is unknown.
- However, the accumulation of the abnormal protein is believed to be what causes neurological changes.
- The excess of the mutated protein interferes with neurotransmitters.

Protein Information

- There are 23 Q's (glutamine) in a repeating sequence in the normal protein. This is referred to as the 23Q repeat.



- The 23Q's in a normal Huntingtin protein are located as amino acids 18…40.

Pathological Protein Changes

- Expansion of this repeat region is what causes Huntington's disease. It translates to an unstable polyglutamine repeat in the protein product. Repeats in excess of 40 are considered to be pathological.

- The expanded ploy glutamine region of the pathological form of the protein causes impairment of the ubiquitin-proteasome system.

- This means that the dysfunctional protein is not removed and destroyed as it should be.

Examples of Mutations in the DNA Sequence…

aaaccatctaggctatattcggatatcgatctatcggatctatctactagagctactacgatcagggactactacgagcatcgactacgag
gcttctagaggctatattctaggctactacgatcgatctacgtagctacgagatcgtgtgtgggggggacacagcgatctaatataaatc
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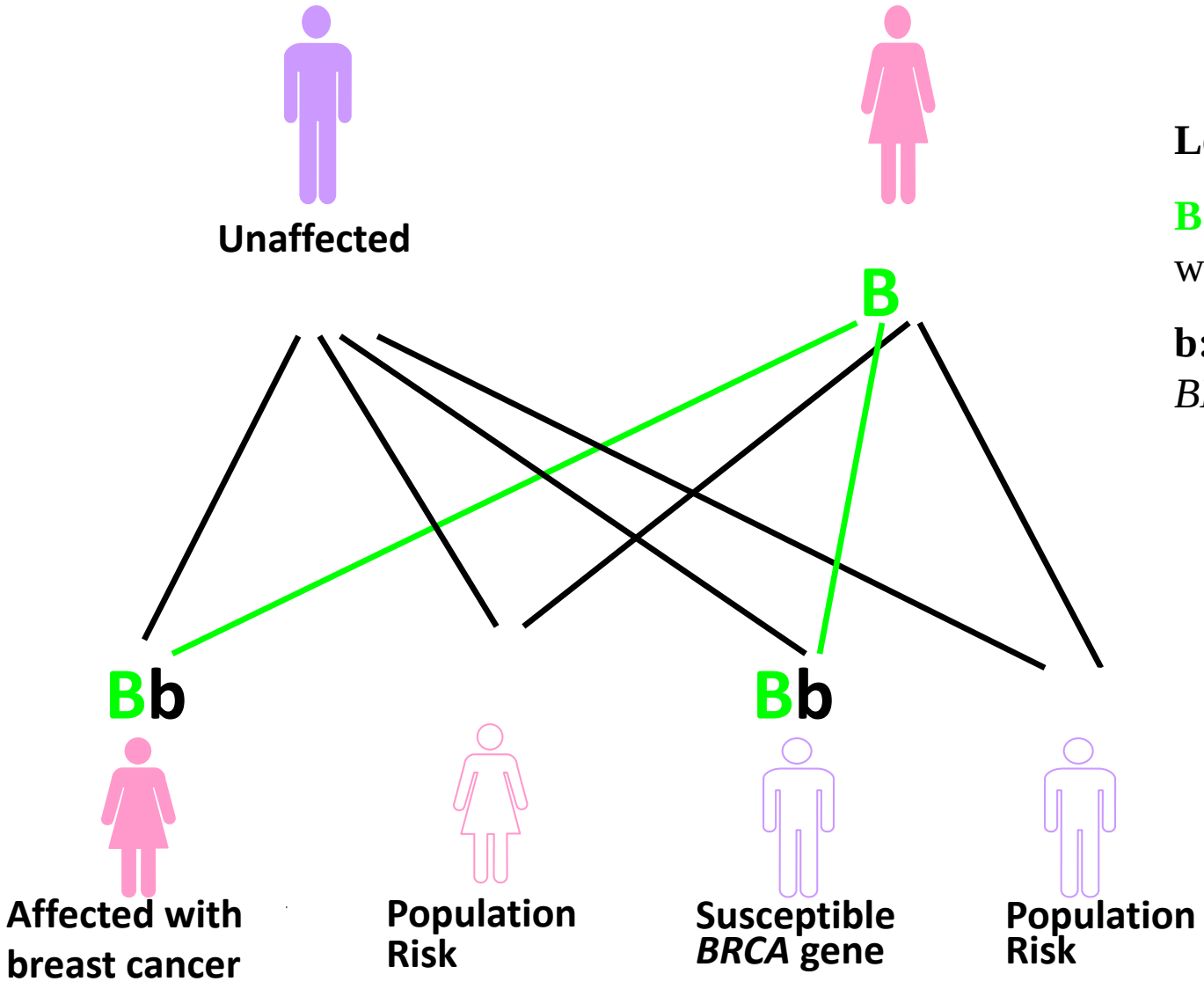
Multiple mutations

(Diabetes, susceptibility to breast cancer)

Genes involved in hereditary breast/ovarian cancer

- > 2,600 mutations in:
  - *BRCA1*- chromosome 17
  - *BRCA2* - chromosome 13
- Autosomal dominant transmission
- Over 12 other genes in addition to BRCA1 and BRCA2

Autosomal Dominant Inheritance



Legend

B: *BRCA* gene with mutation

b: normal *BRCA* gene

BRCA1 and *BRCA2*
What happens when their function is compromised ?

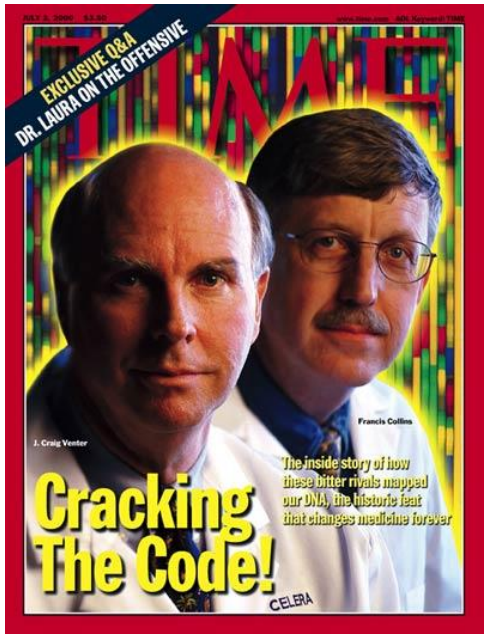
- Both genes are tumor suppressors:
  - Regulation of cell growth
  - Maintenance of cell cycle
- Mutation leads to:
  - Inability to regulate cell death
  - Uncontrolled growth, cancer

Consequences of having a *BRCA* mutation

	Estimated Risk in BRCA Mutation Carriers – by Age 70	In General Population
Breast Cancer ♀ *BRCA1* & *BRCA2*	50 - 85%	11%
Ovarian Cancer *BRCA1*	40-60%	1-2%
Ovarian Cancer *BRCA2*	10-20%	1-2%
Breast Cancer ♂ *BRCA2*	6%	<1%

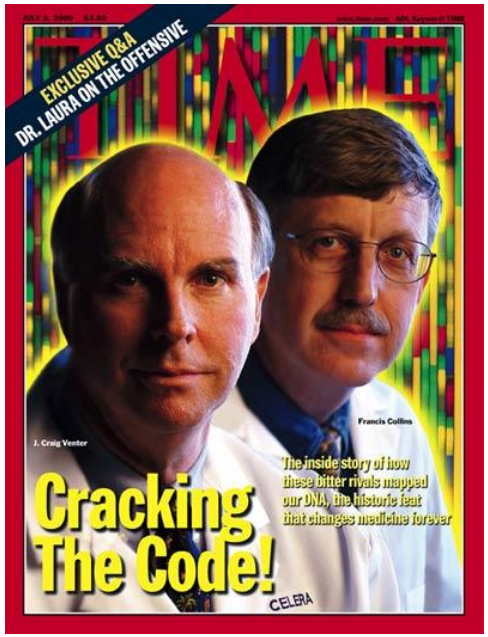
HUMAN GENOME PROJECT

- Initiated in 1990 and first draft published in Feb 2001
- Identification of human genes and sequencing of entire genome
- 1990, American geneticists started an ambitious quest to map and sequence the entire human genome.
- 1999, the final draft of human chromosome 22.
- 2000, the final draft of human chromosome 21.
- 2001, working draft of the whole human genome.
- 2004, the finished sequence of the euchromatic part of human genome.



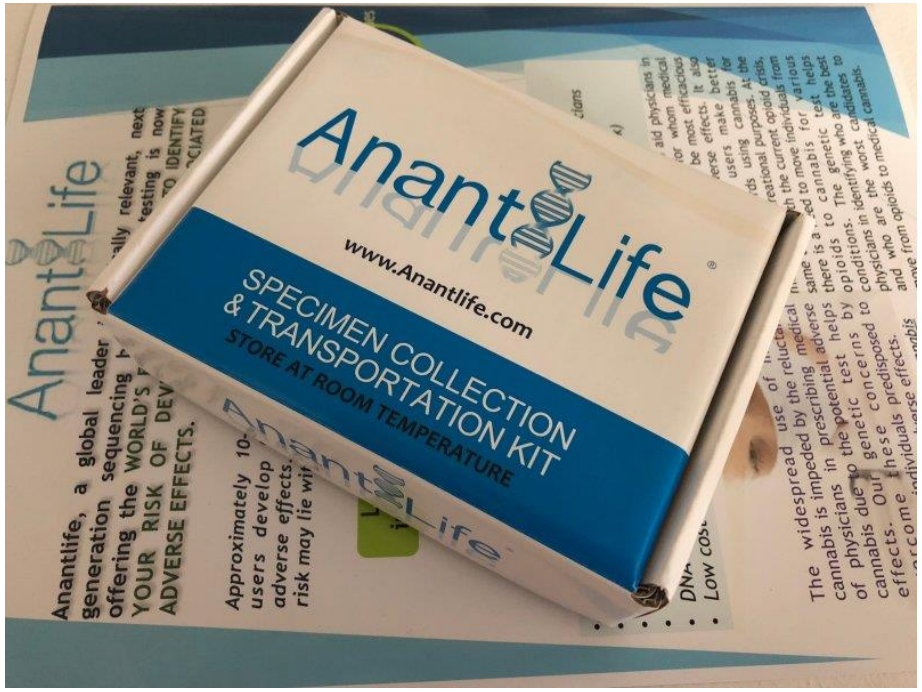
PERSONALIZED HEALTHCARE

- Nutrition based on Genetics
- Dietary Intake based on Genetics
- Fitness based on Genetics
- Detoxification based on Genetics
- Hormone metabolism based on Genetics
- Brain health and Genetics
- Diagnosis based on Genetics
- Predisposition screening and Genetics
- Pharmacogenetics



GENETIC TESTING

- Genetic testing is a type of medical test that identifies changes in chromosomes, genes, or proteins. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder.



TYPES OF GENETIC TESTING

- Diagnostic- establishes the basis of an existing disorder
- Predictive- determines the presence of genetic condition when there are no obvious symptoms
- Carrier- identifies heterozygotes
- Prenatal- assesses a fetus for abnormalities

DIAGNOSTIC GENETIC TESTING

- Based on physical signs and symptoms
- Can be performed on any age
- Not available for all genes or genetic conditions
- Examples:
  - Genetic testing for Celiac for a patient with prolonged diarrhea in response to gluten
  - Genetic testing for Cystic Fibrosis in a child failing to thrive and with recurrent lung infections

CARRIER GENETIC TESTING

- Identifies heterozygotes for X-linked and autosomal recessive disorders that occur with high frequencies in certain populations
- Knowledge of carrier status gives couples the opportunity to make reproductive choices
- Examples:
  - Genetic testing of a healthy child whose sibling has Cystic Fibrosis
  - Genetic testing of a healthy child with parents suffering from Sickle Cell Anemia

PRENATAL GENETIC TESTING

- Performed during pregnancy to determine whether there is a significant risk of having a child with a serious disorder
- Counseling should precede a prenatal test
- Examples:
  - Down Syndrome - Detected by Prenatal Testing
  - Accomplished with amniocentesis- an invasive procedure that involves taking amniotic from the mother and identifying fetal cells

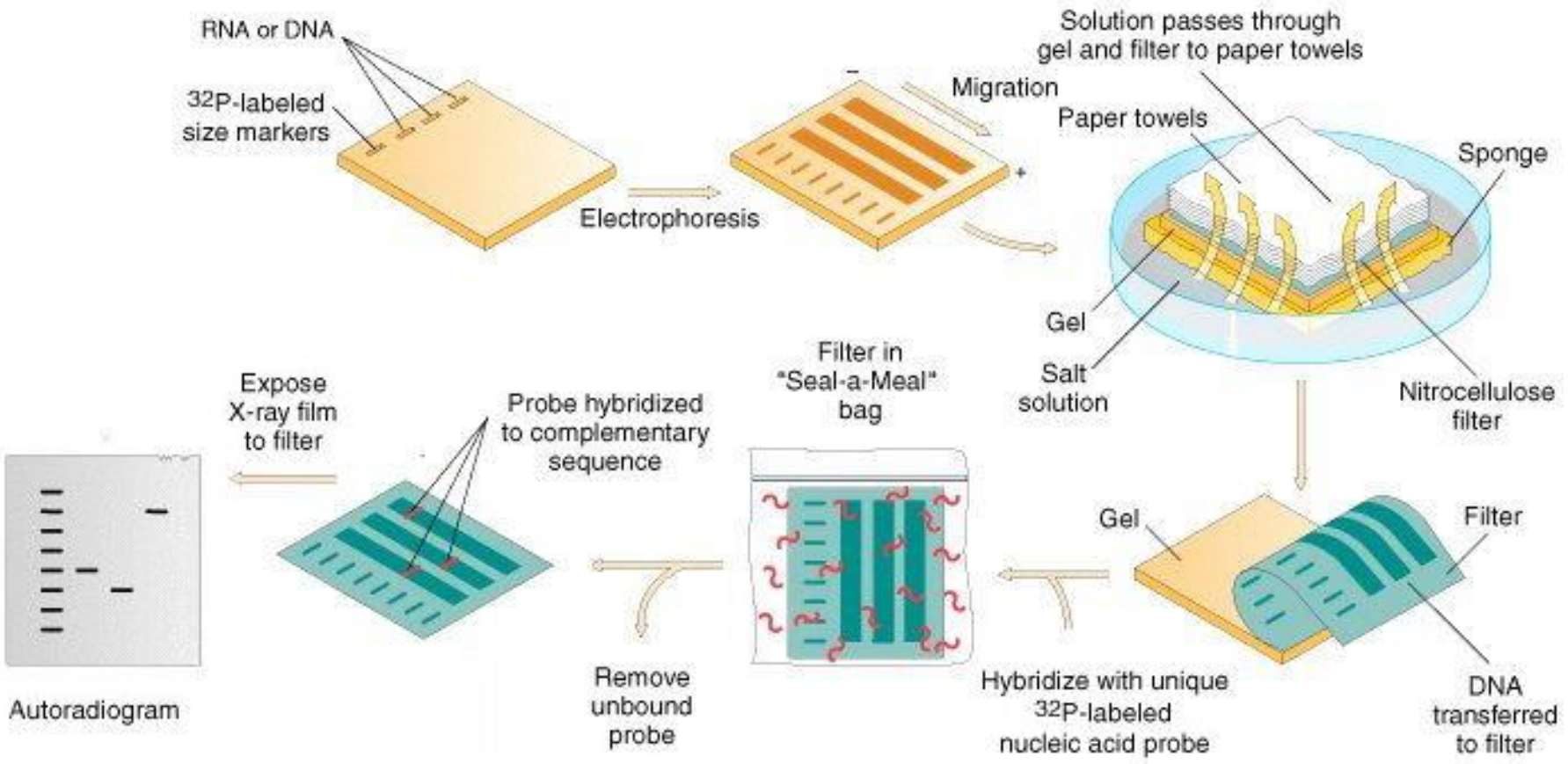
PREDICTIVE GENETIC TESTING

- Performed to predict risk of diseases, nutritional deficiencies, brain disorders
- Examples:
  - Breast Cancer Testing
  - Alzheimers disease testing

HOW IS DNA ANALYSIS PERFORMED?

- SOUTHERN BLOTS
- PCR – POLYMERASE CHAIN REACTION
- DNA MICROARRAYS
- NEXT GENERATION DNA SEQUENCING

SOUTHERN BLOT



SOUTHERN BLOT: LIMITATIONS

- Time consuming
- Use of radioactive probes
- Can only look at a single mutation at once
- Labour intensive
- Costly

PCR – POLYMERASE CHASE REACTION

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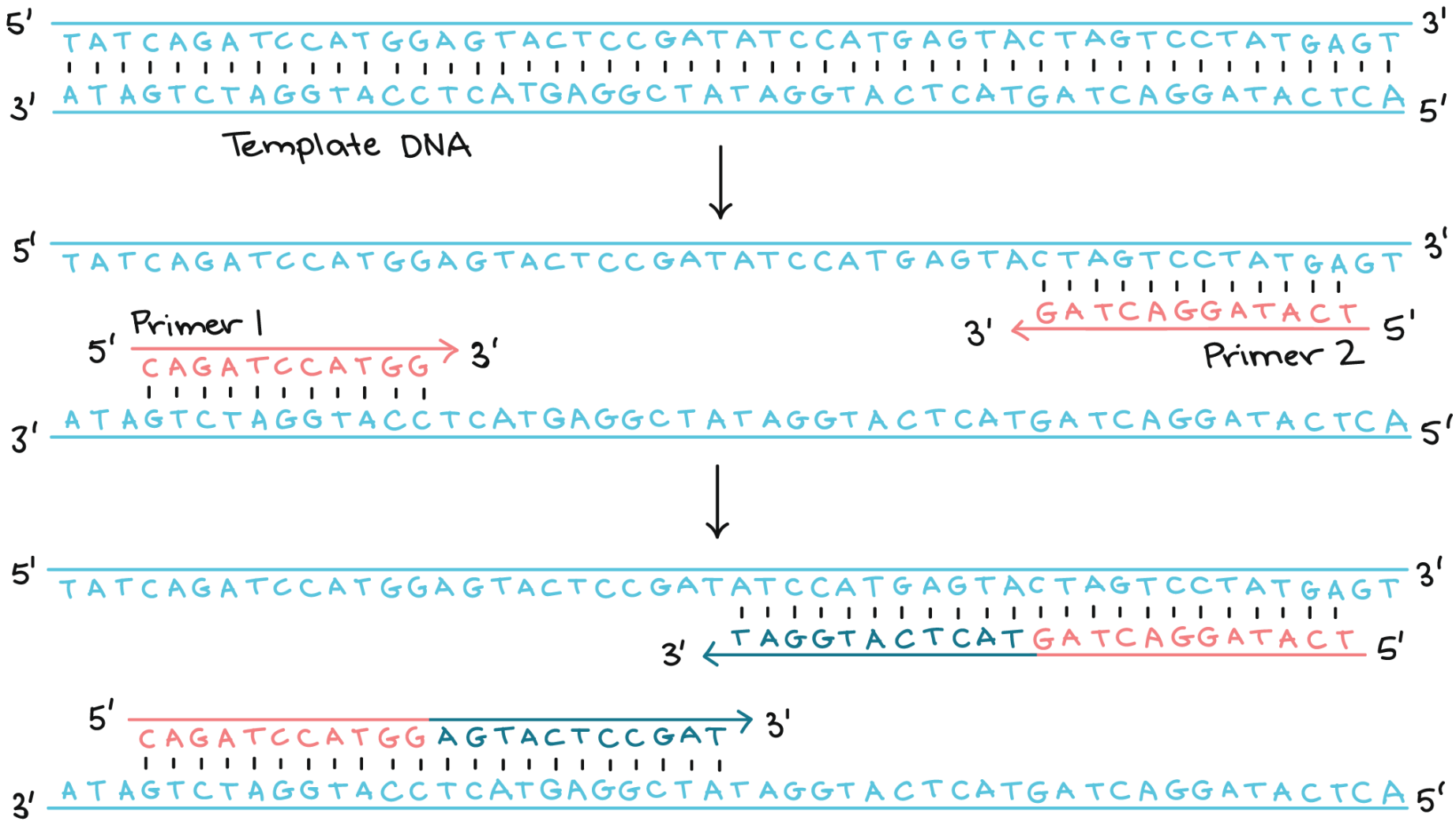
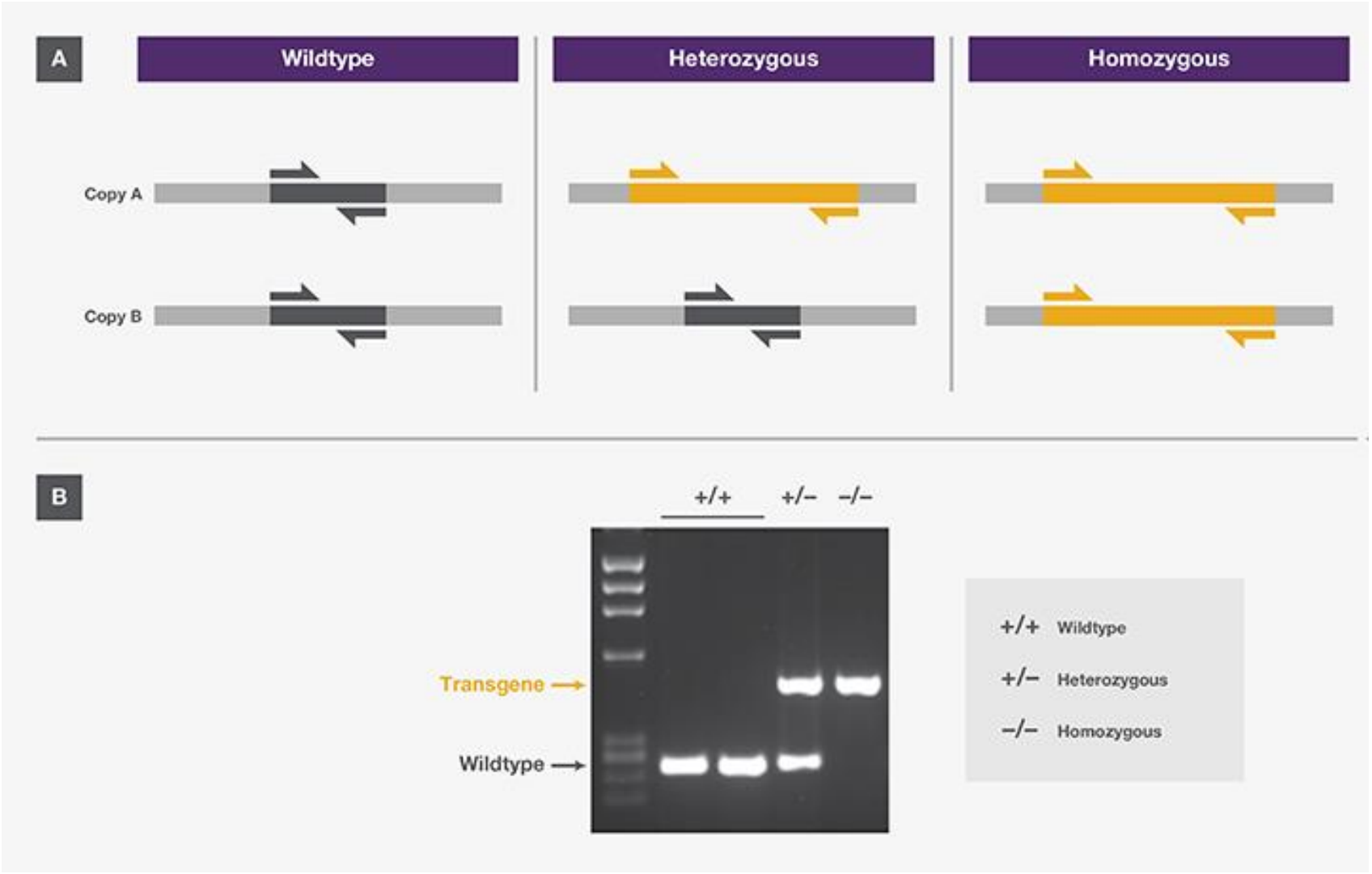
- Basis – artificial replication of the piece of DNA to be studied



PCR – POLYMERASE CHASE REACTION



DNA PCR: LIMITATIONS

- Prior information about the target sequence is necessary in order to generate the primers that will allow its selective amplification. This means that, typically, PCR users must know the precise sequence upstream of the target region on each of the two single-stranded templates in order to ensure that the DNA polymerase properly binds to the primer-template hybrids and subsequently generates the entire target region during DNA synthesis.

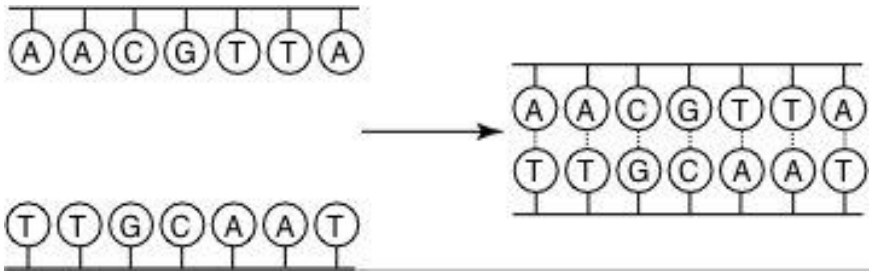
- Like all enzymes, DNA polymerases are also prone to error, which in turn causes mutations in the PCR fragments that are generated.

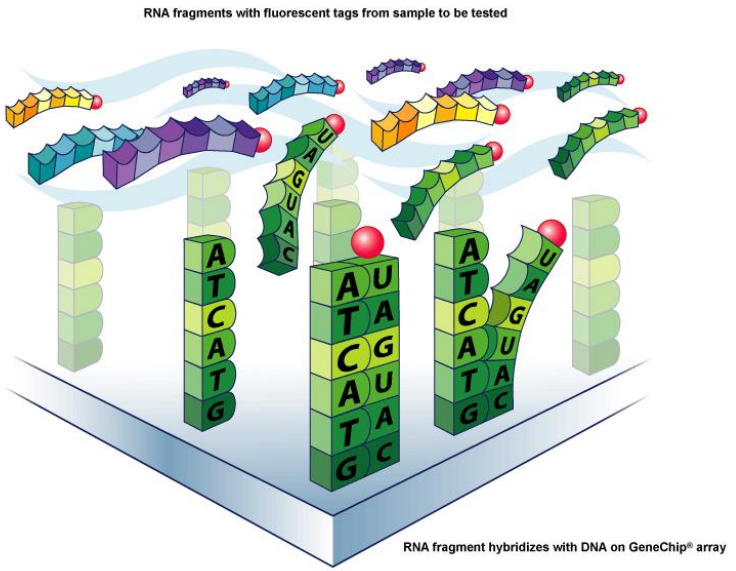
- Another limitation of PCR is that even the smallest amount of contaminating DNA can be amplified, resulting in misleading or ambiguous results.

DNA MICROARRAYS

- DNA Microarrays consist of 100 - 1 million DNA probes attached to a surface of 1 cm by 1 cm (chip).

- By hybridisation, they can detect DNA (DNA binding to a complementary probe)

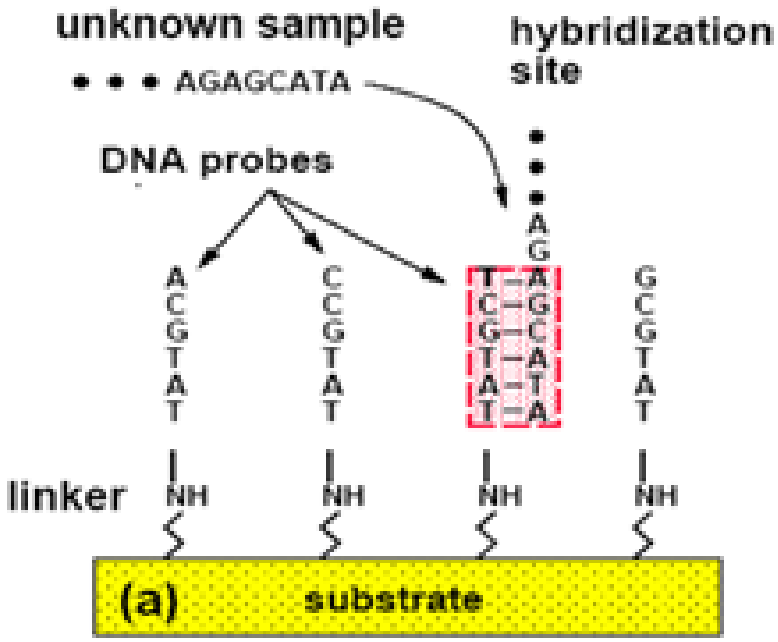


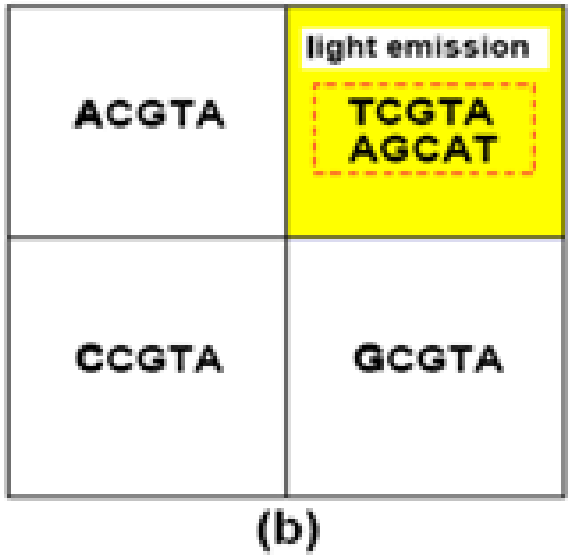


If the hybridised DNA is labelled fluorescently it can be quantified by scanning of the chip.

DNA MICROARRAY

Simple Example of DNA Microarrays





(a) Example immobilized DNA probes showing hybridization of unknown(target) to specific probe.

(b) Probes are arranged a planar arrays. The hybridized regions can be detected by the fluorescence of the duplex.

DNA MICROARRAY – ADVANTAGES AND DISADVANTAGES

Microarray

Advantages

- Well-defined protocols for hybridization
- Well-defined analysis pipelines
- Standardised approaches for data submission
- Relatively low cost

Disadvantages

- Analysis only for pre-defined sequences
- Dynamic range limited by scanner
- Relies on hybridisation
- Hybridisation potentially non-specific
- Might not give paralogue information
- High variance for low expressed genes
- Will generally not identify splice variants

NEXT GENERATION SEQUENCING

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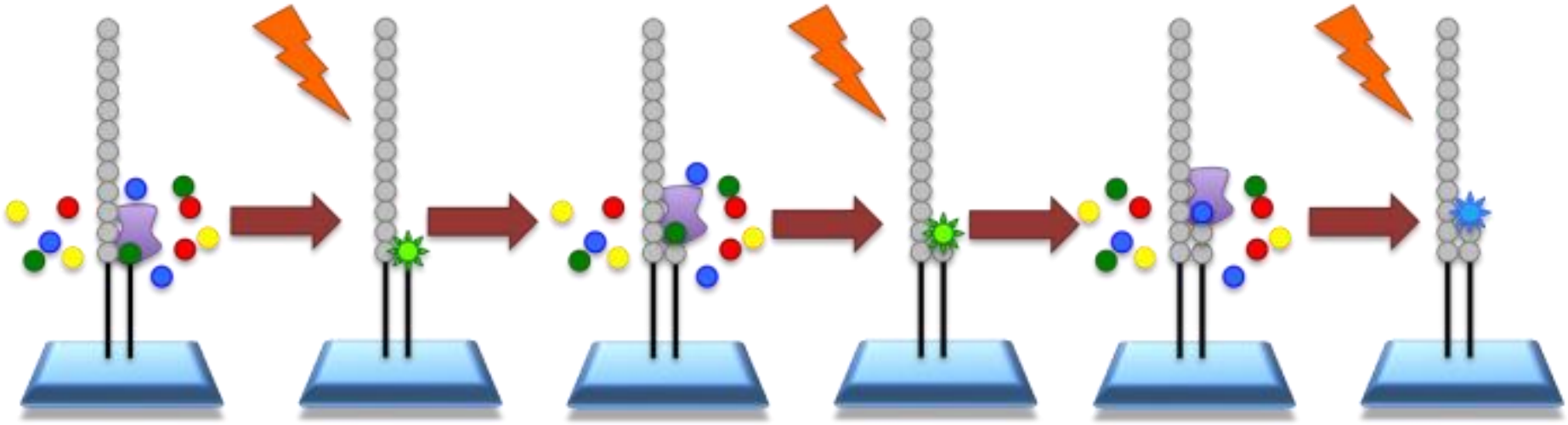
- The gold standard in DNA analysis
- Determines the entire sequence of the DNA segment/gene being studied



NEXT GENERATION SEQUENCING - ADVANTAGES

In contrast to microarray methods, NGS-based approaches have several advantages including:

- a prior knowledge of the genome or genomic features is not required;
- it offers single-nucleotide resolution, making it possible to detect related genes (or features), alternatively spliced transcripts, allelic gene variants and single nucleotide polymorphisms;
- higher dynamic range of signal;
- requires less DNA/RNA as input (nanograms of materials are sufficient);
- higher reproducibility
- No need to reanalyze the gene segment again! Ever again!

HOW DOES ANANTLIFE'S GENETIC TESTING WORKS?

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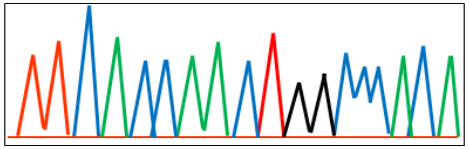
Patient Sample: Saliva

Extract DNA from Cells

NEXT GENERATION SEQUENCING

Sequence DNA



...TTCACCAACAGGCCCACA...

Compare Patient DNA Sequence to Reference Sequence

```
Reference  ...TTCACCAACATGCCCACA...
                  F  T  N  M  P  T
Patient    ...TTCACCAACAGGCCCACA...
               F  T  N  R  P  T
```

BIOINFORMATICS ANALYSIS

Bioinformatic analysis to Determine whether Patient Mutation is Associated with Disease

Our proprietary algorithm carries out risk stratification analysis

BIOINFORMATICS – DNA ANALYSIS

- Can be defined as the body of tools, algorithms needed to handle large and complex biological information.
- The NCBI defines bioinformatics as: "Bioinformatics is the field of science in which biology, computer science, and information technology merge into a single discipline”
- Requires large computing power as Gigabytes of data is being analyzed.
- Data is analyzed in reference to existing databases to identify at risk variants.

REVIEW/DISCUSSION POINTS

- Mutation, mutation types and examples
- Genetic testing, types of genetic testing
- Methods for DNA analysis
- Methods comparatives
- Bioinformatics