

#### Molecular Genetics Booklet 1





## Learner outcomes... What you need to know!

- summarize the historical events that led to the discovery of the structure of the DNA molecule, including the work of Franklin and Watson and Crick
- describe, in general, how genetic information is contained in the sequence of bases in DNA molecules in chromosomes and how the DNA molecules replicate themselves
- explain, in general, how restriction enzymes cut DNA molecules into smaller fragments and how ligases reassemble them
- explain how a random change (mutation) in the sequence of bases results in abnormalities or provides a source of genetic variability
- explain how base sequences in nucleic acids contained in the nucleus, mitochondrion and chloroplast give evidence <sup>2</sup> for the relationships among organisms of different species.

## Terms you need to know

Genome Deoxyribose Nucleic Acid **Phosphates** Nitrogen Bases Double helix **Rosalind Franklin** Watson and Crick **Complementary Base** Nucleotide Hydrogen Bond Gene Chargaff Rule Helicase **DNA Polymerase** Semi-conservative replication Human Genome Project

## **INTRODUCTION TO DNA**

- Genome of human cell has ~50,000-100,000 more genes (segments of DNA that encode for traits) than a bacterial cell and contains far more DNA per gene.
- The genetic information of a cell is contained in its DNA in the nucleus
- DNA <u>unique</u> to each individual





## **INTRODUCTION TO DNA**

- DNA stands for DeoxyriboNucleic Acid
- **Deoxyribose** is the sugar that makes up the backbone of DNA
- Named nucleic because it was initially found in the nucleus
- Acid the phosphates in the backbone cause DNA to be acidic
- DNA is universal it is the genetic code for all living cells whether they be plant, animal, bacteria or fungus



## **DNA Location**

#### IN THE NUCLEUS

#### **OTHER DNA OUTSIDE** THE NUCLEUS



### **Nuclear DNA**

In The Nucleus. Long strands of chromatin... then chromosomes



#### **Mitochondrial DNA (mtDNA)**

- a circular chromosome
- In plants and animals



Chloroplast DNA (cpDNA) – a circular chromosome -in plants

Inherit their genome from either paternal or maternal side -allows tracing back of ancestry

## **DNA** Location

- A <u>SON or DAUGTERS</u> mtDNA is identical (passed down from) their mother's but only the girls can pass it on.
- (unlike nuclear DNA which is from both mother and father)



This enables us to trace ancestry because you inherited your mitochondrial DNA from your mom who inherited it from her mom and so forth...

## Follow the Mitochondrial DNA...



Which children of the top two parents have passed down mitochondrial DNA?

## The Structure of DNA

- Rosalind Franklin was using X-Ray Diffraction to study DNA
- DNA is a helix, it is double stranded and the distance between DNA strands is constant.
- Her work allowed Watson and Crick to come up with model of DNA
  - Findings presented in 1953
  - Visually confirmed in 1969

NITROGEN BASES



SUGARS + PHOSPHATE BONDS

# The History of DNA

- Watson and Crick determined the 3-D structure of DNA
  - Watson entered university at 15 .....in Chicago
    - He ended up at Cambridge University with Crick, a physicist
- Scientists don't work alone, Watson and Crick relied on work done by Rosalind Franklin





## **DNA looks like a twisted ladde**

**<u>Structure</u>**: 2 complimentary strands

**Backbone** = phosphate(outside) + deoxyribose (sugar) - (inside the phosphate)

**R<u>ungs</u> = 2 nitrogen bases** 

•<u>A</u>denine pairs with <u>Thymine</u>

• <u>Guanine pairs with</u> <u>Cytosine</u>

Bases held together by <u>hydrogen</u> bonds (between A & T or C &G)





Straight letters go together A-T Curvy letters go together C-G

#### The Genetic Code

You and a worm are not that different...its true!!... ACORN WORM VIDEO

WHY?

You both use the same bases A,C,T, and G to make up your DNA

THE GENETIC CODE IS UNIVERSAL -all living things use the same code -all living things have the same bases -Codons code for the same amino acids

This is why genes from one organism can be spliced into a totally different one like a jellyfish into a cat.....to make a glowing cat Genetically speaking, how much do you think you have in common with a... <u>DNA SHARE VIDEO</u> An unrelated person next to you... 99.9%

A chimpanzee... 96%A cat... 90% A mouse... 85%A cow... 80% A chicken... 60%



A banana... 60% No wonder you are bananas!

# **Deoxyribonucleic acid**



If one strand of DNA has the following nucleotide bases, what **complementary bases** would the other strand have?



## Nucleotide



# Nucleotide

1 base(A,T,C,G) + 1 phosphate + 1 sugar(deoxyribose)



## Label this diagram with these words:



Hydrogen bond

Nucleotide

**Phosphate** 

Nitrogen Base- A, T, C, G



#### DNA Structure



#### **DNA Structure**





#### Each protein carries out a unique function



breaks down a sugar found in milk



National Human Genome Research Institute

## **DNA** Bases

#### These are on your DATA sheet.

Purines: Big ones!!

"<u>Pur</u>e <u>A</u>s <u>G</u>old" (Adenine and Guanine are Purines)

### Pyrimidines: Smaller

"<u>C</u>u<u>T</u> <u>P</u>y" (Cytosine and Thymine are Pyrimidines)



## Chargaff's Rule: Base Pairs

- Remember that adenine and thymine are complementary base pairs. This means that A = T
- Also, guanine and cytosine pair together, so

 $\mathbf{G} = \mathbf{C}$ 

#### When added together, all the base pairs = 100%.

- <u>Question:</u> If a DNA molecule were comprised of 30% adenine, how much guanine would be present?
- Solution: If A = 30%, then T = 30%. Added together, A + T = 60% of the DNA molecule.
- That means that the other 40% of the molecule is made up of G and C. Since G = C, then each is ½ of 40, or 40÷2 = 20.

## **DNA Base Pairs**



**Practice:** If a DNA molecule is 40% Guanine, then how much Thymine will there be in the molecule?

# **DNA and Uniqueness**

- Our DNA is like a cookbook of instructions
- The sequence of DNA bases determine the type of proteins produced
- Proteins are unique for each life form
- Offspring look similar to parents but not exactly the same...why?



- 1. New gene combinations (when sperm and egg fuse to form zygote)
- 2. Mutations (changes in DNA)
- 3. Crossing over

If all the DNA in your body were uncoiled and stretched out, it would reach to the sun and back approximately 3000 times!!

TED talk (4:26) The Twisting Tale of DNA

- DNA was the only known molecule capable of **replicating** itself
  - prions (special proteins) now known to do this too!
- Replication necessary for 1 cell to become 2 identical cells
- DNA replication occurs when a cell is going to divide



## **Semiconservative** Replication



Requirements

## DNA Unzips for 2 reasons!

- A. For replication prior to cell division
- B. To make mRNA for protein synthesis



# 1. DNA molecule untwists (straightens)

initiated by the enzyme
<u>helicase</u>



2. DNA 'unzips' (the hydrogen bonds break) with the help of helicase

 Helicase acts like a pair of scissors to break the weak H-bonds between nitrogen bases





- The exposed nucleotides attract their 'partner/complementary' nucleotides.
  - <u>DNA polymerases</u> are involved in the fusing of the free nucleotides.
    - -nucleotides start to attach
    - from 3' end of parent strands



#### \*\*FREE FLOATING NUCLEOTIDES IN THE NUCLEOPLASM COME FROM FOOD WE EAT!\*\*

 The sugar and phosphate molecules of adjacent nucleotides join to form
2 ladder-like structures each identical to the original.



**DNA Replication Fork Animation** 

 Semi-conservative replication - because each new molecule is composed of ½ the original strand and ½ the new strand.



Parental molecule of DNA Both parental strands serve as templates Two identical daughter molecules of DNA

Bozeman video (10:10)-skip 1:25- 2:48 Crash Course: DNA structure and replication

http://www.youtube.com/watch?v=FBmO\_rmXxIw&safe=active&safety\_mod e=true WORKBOOK PAGES 4, 6

# Mistakes in Replication – cause of protein mutations!

Mistakes in replication can be caused by environmental factors such as hazardous chemicals and radiation

One change in DNA sequence (mutation) can have a significant effect!

Mosquitoes Sprayed with DDT (insecticide)



# AFTER sprayed with roundup

BEFORE

This weed survived a herbicide spray (its DNA mutated) in a corn field. If it produces seed, what effect will that have on the field?

#### The <u>Human Genome Project</u>

Goals of the project:

- to **identify all genes** in human DNA.
- To determine the sequences of 30,000 genes which is the <u>3 billion base pairs</u> that make up human DNA (3 billion is 90 years of seconds)
- Took from 1990 2003 to complete

Nobel Prize Winner(1980) Walter Gilbert described the human genome as the Holy Grail of biology -helped develop method of identifying base pairs (1976)



## Human Genome

- Estimated to contain approx. 3 billion bases
- Mistakes (faulty base pairing) are infrequent and permanent damage prevented by proofreaders

– Enzymes

- These enzymes run along DNA looking for mismatched pairs
- Some enzymes snip out errors and replace them with correct nucleotide sequences



# Human Genome

#### **Benefits**

- Diagnose and treat genetic disorders (like BRCA gene – the gene for breast cancer)
- Develop new medications
- Prevent disorders
- Study evolutionary relationships
- DNA identification

#### <u>Issues</u>

- Who should have access to the information (employers, insurance companies, 'geneism')?
- Who owns the information?
- Reproduction issues how are we going to use the data?
- Cost to society?



Angelina Jolie has the mutated BRCA1

gene.

Angelina Jolie Reveals Plans For More Surgery After Double Mastectomy

After having a double mastectomy in 2013, that next stage would likely be to have an oophorectomy, a.k.a. ovary removal surgery. The procedure would combat Angelina's 50 percent risk (the number varies based on the individual) of developing ovarian cancer, the disease her mother, Marchelina Bertrand, died from at the relatively young age of 56. Angelina's risk of developing breast cancer and ovarian cancer was and is so high because she inherited the "faulty gene" known as BRCA1 from her mother.



#### BRCA1 and BRCA 2 Genes

BRCA1 and BRCA2 are human genes that produce tumor suppressor proteins. These proteins help repair damaged DNA and, therefore, play a role in ensuring the stability of the cell's genetic material. When either of these genes is mutated, or altered, such that its protein product is not made or does not function correctly, DNA damage may not be repaired properly. As a result, cells are more likely to develop additional genetic alterations that can lead to cancer.

#### **DNA REVIEW**

DNA stands for...

Describe Parts of DNA molecule:

Nitrogen Bases(4+1): Purine: Pyrimidine:

Nucleotide:

Gene:

Helicase:

Polymerase:

Semi-conservative replication: