READ, HIGHLIGHT, + ANSWER QUESTIONS #2

GENETICS: SOME OF THE BASICS...

The use of genetic technology in forensic science wasn't introduced until the 1980's - however it has proven to be a powerful tool in solving crimes, identifying unrecognizable human remains, and paternity testing.

Inside Your Cells

Every biological organism is composed of many individual microscopic cells. It is estimated that an average human is made up of over 300 trillion cells! A dog looks much different than a cat, while a human looks much different than a cow... however all the cells within each of these creatures contains the same type of organelles in their cytoplasm. For example, all biological cells contain a nucleus, mitochondria, lysosomes, ribosomes, and endoplasmic reticulum. All of these unique organelles are made up of various protein molecules.

The nucleus of every cell contains chromosomes. The number of chromosomes in an individual nucleus within EVERY normal body cell varies from organism to organism (ie. human = 46 , horse = 66, pig = 38, dog = 84). Every normal human body cell contains 46 chromosomes, except for sex cells -> ie. egg and sperm (which contain only 1/2 the number of chromosomes). Chromosomes contain the 'genetic blueprint' for the thousands of different proteins found within the body, and in order to create these proteins the information inside the chromosomes provides these important directions. Without your chromosomes, your body would no longer be able to function !

What is DNA ?

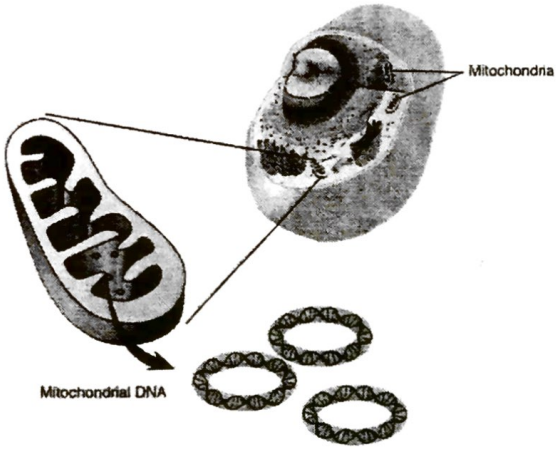
Every chromosome is composed of a molecule called DNA, which like other biological substances is just a molecule. In fact, its molecular name is *deoxyribonucleic acid*. DNA is literally 'crammed' inside each of the 46 chromosomes within the human nucleus.

DNA has a very unique molecular arrangement. It looks like a 'twisted ladder' (or double helix) and it is only a few microns wide, but would stretch up to two meters long if it was uncoiled! The DNA 'twisted ladder' consists of two matching strands that are fused together with hydrogen bonds. The precious interior of this 'twisted ladder' is composed of four nucleic acid bases. Each DNA strand has some 3 billion repeating nucleic acid bases within it.

Illustration of a single chromosome

The names of the 4 nucleic acid bases are adenine, thymine, cytosine and guanine. Nucleic acid bases are found within pairs in the interior of the DNA strand, but because of the unique shapes of each of the nucleic acid bases only certain bases will pair up/bond with each other. More specifically, adenine will only pair with thymine (A - T), while cytosine will only bond with guanine (C - G). The distinct sequence of these nucleic acids

creates genes. Each gene contains the instructions for a specific type of protein in the body. Because the human body is comprised of thousands of different proteins, there are thousands of different genes. Using a powerful microscope, some genes can even be seen on a chromosome in the form of gene bands, while others cannot (*see diagram*).



For a very long time scientists thought that DNA in was only found in the nucleus of all cells. This idea changed in the late 1960's when it was discovered that there was also DNA found with in the cellular organelles called **mitochondria**.

Cellular respiration occurs in the mitochondria of cells – this process produces adenosine triphosphate (ATP). ATP is an energy molecule required for all cell activities – every cell needs ATP to survive.

The DNA in the mitochondria of the cell is known as **mtDNA**. There are some differences between the DNA with in the mitochondria and nucleus:

-> The <u>shape</u> of mtDNA molecules differs from the double-helix/twisted ladder structure of nuclear DNA; mtDNA is smaller and tends to be looping and circular in shape.

➔ There is far less mtDNA than there is nuclear DNA within human cells.

➔ Of the 3 billion nitrogen-base pairs of human DNA, only 16,569 are mtDNA, while the remaining DNA is found within the chromosomes of the nucleus.

Human ovum have many mitochondria within them, while sperm cells do not; this is because sperm cells need just enough energy to power them on their journey to the ovum. Immediately after a sperm penetrates an ovum (conception), the mitochondria within the sperm cell are destroyed. Thus, we receive 50% of our nuclear DNA from our mother and 50% of our nuclear DNA from our father, but 100% of our mtDNA from our mother. Therefore, a child's mtDNA is identical to his/her mother, biological brothers & sisters, maternal grandmother, maternal aunts, uncles & cousins - anyone from the mother's side of the family. This fact is used by forensic scientists to identify:

- unrecognizable human remains *(ie. plane crash, murder, soldiers missing-in-action).*

- the maternity of adopted or missing children.

If the mtDNA pattern from an individual matches the mtDNA of another individual, it can prove his/her identity or simply that these two individuals are related.

<u>Every Person's DNA is Unique</u>

The sequence of the four basic nucleic acid bases makes up your genes and your genes determine what type of proteins your body will create. A specific gene creates every protein. Your body is made up of thousands of different proteins and they determine your unique appearance &

structure. Interestingly, only about 5% of your DNA is considered useful as it creates the 30,000 genes that your body uses. Also, 99% of this useful DNA is similar from person to person; therefore analysis of this similar type of DNA is NOT at all helpful in forensic science.

The function of about 95% of your DNA is unknown and often referred to as being 'junk' DNA! But what's interesting about this 'junk' DNA is that much of it is unique from person to person. Within the 'unknown/junk' sections of DNA, a nucleic acid base sequence often will repeat itself over and over again. These repeated base patterns are called VNTR, or *variable number tandem repeats.*

The VNTR's within a person's DNA therefore become the focal point within forensic DNA analysis. A total of <u>five</u> VNTR patterns have to be identical to be considered a 'match'. Again, the reason that VNTR patterns are checked is because of the uniqueness of VNTR sequences from person to person. A particular VNTR pattern may be repeated 14 times at a specific location in your DNA, 20 times at the same site of your sister's DNA, and maybe 52 times on your Uncle Henry's DNA. The odds against someone else have this EXACT same VNTR pattern repeated the EXACT same number of times on their DNA on at <u>five</u> sites is highly improbable. But the reality is that some VNTR patterns can be found in other people, while other VNTR patterns are very rare from person to person.

WORKSHEET: GENETICS - SOME OF THE BASICS...

Name: _____ Date: _____

1. Define each of the following important terms:

chromosomes:

_____

DNA:

_____

genes:

_____

nucleic acids:

_____

nucleus:

_____

mitochondria:

_____

2. If some unrecognizable flesh was found at a crime scene and investigators were uncertain whether it was human, what type of structure in the nucleus of the cells could help them determine its biological origin?

3. What is found <u>inside</u> a chromosome and what purpose does this substance serve?

4. Why can <u>mitochondrial DNA</u> (mtDNA) be useful to forensic scientists? List three scenarios where mtDNA would be specifically used & analyzed.

5. State <u>five people</u> in your family that would have the exact same mtDNA as you.

6. After the terrorist attack upon the World Trade Center on September 11th, 2001, family members of the thousands of victims were asked to give blood samples to the NYPD. Why do you think this was done? Which family members would not be able to give blood?

7. How much of your '<u>useful DNA</u>' is similar to that of other people?

8. What <u>type of DNA</u> would prove to be the most useful to forensic scientists in a criminal investigation? Explain why.

9. When a forensic DNA expert presents a <u>DNA print match</u>, he/she will also identify the <u>probability of certainty</u> for that match. Why will they do this?

10. As it stands today, is there <u>100% certainty</u> that a DNA print is a match?

DNA PRINT ANALYSIS TECHNIQUES: RFLP vs. PCR

In the late 1980's, Dr. Alec Jefferys founded the first two private DNA analysis labs in the world. One of these labs was based in England, while the other was situated in the United States. Dr. Jefferys called his private DNA analysis companies *Cellmark Diagnostics*. *Cellmark Diagnostics* is considered to be the finest private DNA analysis lab in the world today. In fact, *Cellmark Diagnostics* analyzed blood samples from the O.J. Simpson murder trial – the highly publicized criminal case that propelled forensic DNA analysis to the forefront.

RFLP vs. PCR

When Dr. Alec Jeffreys was at the University of Leichester in England, he developed the DNA analysis technique know as **RFLP** or *Restrictive Fragment Length Polymorphism*. In the RFLP method, DNA is extracted from a biological source such as blood, semen or skin and is tagged with a radioactive probe that is exposed onto a piece of X-ray film. The resulting illuminated pattern of gene bands looks somewhat like a 'bar code' you find on most store products. The gene bands patterns are then analyzed by experts and used for comparison. One disadvantage to this method is that many cells are required for the RFLP method: 5 000 to 50 000 cells.

A newer method of DNA analysis is called **PCR**, which stands for *Polymerase Chain Reaction*. In the PCR method, a single strand of DNA is quickly replicated and re-created over and over again. This method takes place inside a laboratory instrument called a thermal cycler. One major advantage to this method is that PCR results can yield results in a fraction of the time of the RFLP method. Also, PCR needs only around 50 cells rather than thousands required by RFLP.

Probability

The theory of DNA print 'matches' is based on the theory that segments of genetic material occur randomly across the population, so the odds against a match at five distinct VNTR sites is highly unlikely - but just how unlikely is the subject of much controversy. The reason that matches are based on probability rather than fact arises from the reality that an extensive worldwide data bank of human DNA does not exist. Some individual nations around the world have started DNA data banks for their respective countries, however only DNA taken from convicted criminals has been submitted.

The probability range for RFLP data is considered to be fairly accurate: one in hundreds of thousands or even one in a million or more. PCR data is less specific, as the probability range falls between 1 in 100 to one in 2000. This is due to the smaller DNA fragments that are used. These probabilities, rather than the facts, keep criminal defense lawyers busy and often confuse jurors. Defense lawyers will often argue and try to convince jurors that the chance of the DNA sample matching someone else in the population is not good enough to "*prove guilt beyond reasonable doubt*". Also, PCR results in particular offer more grounds for doubt.

DNA PRINT ANALYSIS TECHNIQUES:
TRUE & FALSE Challenge

Name: _____ Date: _____

Circle whether the following statements are TRUE or FALSE - correct all false statements.

a) DNA is extracted from red blood cells found in a blood sample: **TRUE** **FALSE**

b) Sections of DNA are extracted using a microscopic laser: **TRUE** **FALSE**

c) A DNA print is created using a process called gel electrophoresis: **TRUE** **FALSE**

d) DNA fragments are visible right after being placed on nylon membrane: **TRUE** **FALSE**

e) Nuclear DNA is used for unrecognizable remains identification: **TRUE** **FALSE**

f) DNA is placed at the (-) pole of the gel electrophoresis device: **TRUE** **FALSE**

g) Negatively-charged DNA moves towards the (+) pole: **TRUE** **FALSE**

h) DNA print analysis has been around since the 1950's: **TRUE** **FALSE**

i) Dr. Alec Jefferys developed the PCR technique: **TRUE** **FALSE**

j) A disadvantage to the RFLP method is that it requires 1000 cells: **TRUE** **FALSE**

k) The RFLP technique is faster than the PCR method: **TRUE** **FALSE**

l) Two VNTR sites have to be in the same order to be a 'match': **TRUE** **FALSE**

m) VNTR pattern similarities occur randomly across the population: **TRUE** **FALSE**

n) Every child born today has his/her DNA placed in a DNA data bank: **TRUE** **FALSE**

o) The probability range for RFLP is higher than PCR: **TRUE** **FALSE**

p) RFLP stands for Retroactive Fragment Length Polymorphism: **TRUE** **FALSE**

q) PCR stands for Polymerase Chain Reaction: **TRUE** **FALSE**

r) In a thermal cycler DNA is replicated & re-created over and over again: **TRUE** **FALSE**

s) Defense lawyers often argue DNA evidence always proves guilt beyond any doubt: **TRUE** **FALSE**

t) Most nations around the world have DNA data banks for convicted criminals: **TRUE** **FALSE**

STEP-BY-STEP DESCRIPTION
OF
DNA PRINT ANALYSIS

*The following is a description of the **RFLP** method, which is the oldest type and most common form of DNA print analysis used in forensics.*

-> A small sample of biological evidence is collected (e.g. blood, hair, semen, skin).

-> Using a powerful microscope, a nuclei or mitochondria from a cell is extracted using a micropipette.

(*Nuclear DNA is used for criminal comparison, while mitochondrial DNA is used for the identification of unrecognizable remains).*

-> The chromosomes from this nuclei are isolated. Then sections of DNA from within a chromosome are removed using a restrictive enzyme.

-> A fragment of DNA is then placed within a device called gel electrophoresis. The DNA fragment is then embedded within a thin layer of gel. One end gel has an overall negative charge thus it is known as the negative (-) pole.

-> Opposite to the imbedded DNA fragment is the positive (+) pole, which when activated emits a positive charge.

-> The positive charge emitted by the (+) pole attracts the negatively-charged DNA fragment, and as result the DNA slowly migrates and stretches through the gel to the opposite pole.

-> Once the DNA fragment has been stretched out accordingly, a 'DNA print' is transferred and preserved onto a nylon membrane or X-ray film.

-> Radioactive probes are applied to the DNA print - this makes distinct gene bands appear - becoming visible to the naked eye.

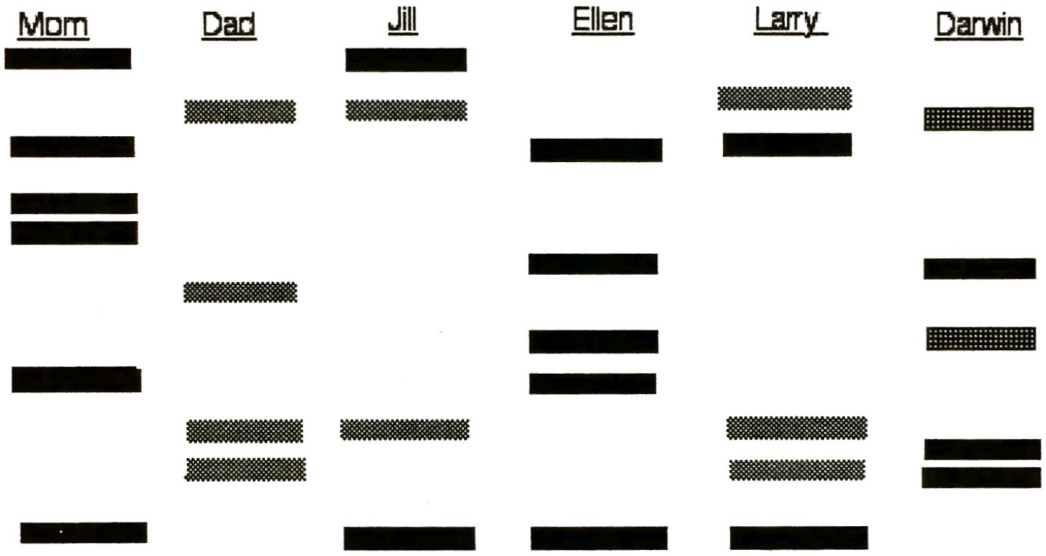
-> Once the DNA print is complete, the analysis and comparison of the distinct gene band pattern occurs.

Worksheet: DNA Print Analysis Exercises

Name: _____ Date: _____

Analysis I: Determination of Maternity & Paternity

The illustration below represents DNA print sequences from six different people. Shown are the DNA prints for Mr. and Mrs. Smith (Mom and Dad) and their four children: two daughters - Jill and Ellen and their two sons - Larry and Darwin.



Analyze the above illustration of a DNA Print in order to answer the questions below...

1. Predict which children are Mr. & Mrs. Smith's **biological** offspring. _____

2. Which of the Smith's children is adopted? _____

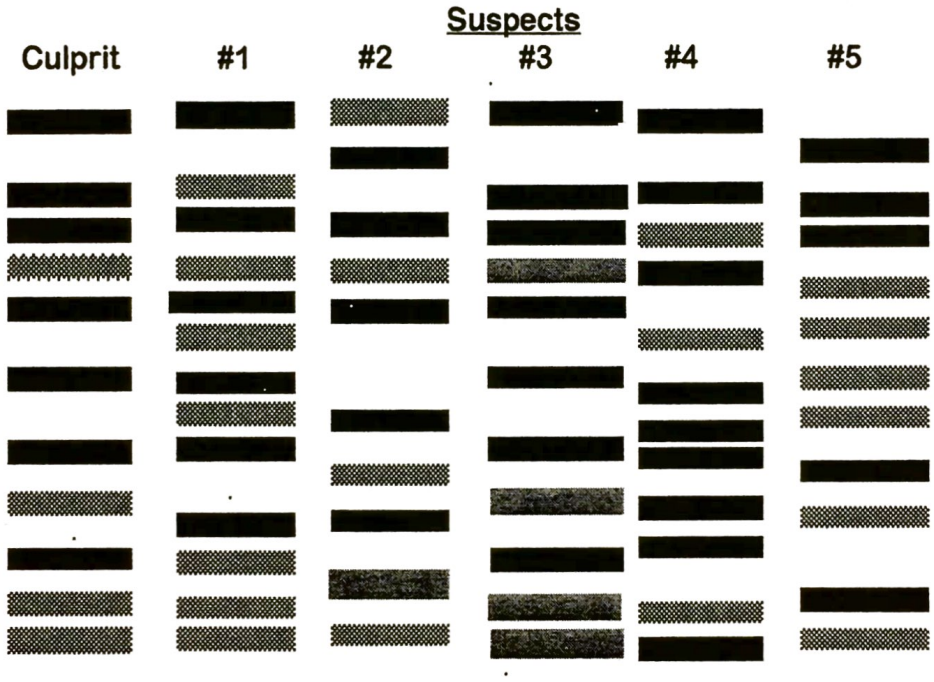
3. One of the four children is Mrs. Smith's child from a **previous marriage**. Predict which child this likely is? _____

Analysis II: DNA Left behind during a Sexual Assault

A sexual assault has been committed and a semen sample was collected from the body of the female victim. Fortunately for investigators, diploid cells are found and isolated within this semen sample. DNA is then extracted from these cells and an RFLP DNA analysis is completed.

Police apprehend five male suspects in this case and a blood sample is taken from each individual. White blood cells within the blood of each sample are isolated and are also subjected to an RFLP DNA analysis.

The illustration below shows a view of all the RFLP DNA analysis test results in this particular case. The DNA pattern from the semen sample left at the crime scene appears in the column labeled as culprit. Columns #1 thru #5 are DNA patterns obtained from various male suspects.



4. Which of the five suspect(s) do NOT match the crime scene evidence ? _____

5. Which of the five suspect(s) DOES match the crime scene evidence ? _____

#4

Name:_____ Catching Killers: DNA Profiling

1. How did police know the photos of Vicki Wegerle were not taken by them?

2. What did BTK stand for?

3. When did BTK first appear? Describe his first crime.

4. Describe how BTK communicated with the public. What did police try to determine from this?

5. What evidence was BTK careful not to leave behind at crime scenes?

6. What evidence to BTK leave at the Wegerle and Otero crime scenes?

7. What changed between when BTK was first active and when he reappeared?

8. What evidence was found on Lynda Mann? How did the police use it?

9. How long was it between the murders of Lynda Mann and Dawn Ashworth?

10. How did police determine that both girls were murdered by the same person?

11. Describe Richard Buckland's involvement in the case.

12. Describe the process of preparing the samples of analysis.

13. What two things was the DNA analysis able to determine?

14. What changed in DNA analysis between the two cases presented?

15. What does CODIS stand for? Was there a match for BTK in CODIS?

16. What lucky break did the police get in the Mann and Ashworth case?

17. Describe Colin Pitchfork.

18. Did Colin Pitchfork kill Lynda Mann and Dawn Ashworth?

19. How did the Pitchfork case change Forensic Science?

20. How did police get DNA evidence from people who didn't want to give a sample in the BTK case?

21. What piece of evidence broke open the BTK case?

22. Describe Dennis Rader.

23. How did police get Rader's DNA?

24. Describe Rader's arrest and confession.

25. What sentence did Rader receive?

26. How long does DNA analysis currently take? What is the goal?

27. What are some other goals for DNA in the future?