Homo sapiens 
Complete Mitochondrial Genome CRS 
16,569 bp

Instructor- Dr. Jeffery R. Hughey
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Front cover image is the genetic map of the mitochondrial genome of the CRS
# Introduction to Forensic DNA Analysis - Biology 13

## Lecture Syllabus

<table>
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<tr>
<th>Date</th>
<th>Lecture Topic</th>
<th>Reading</th>
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<tr>
<td>August 28</td>
<td>Evidence collection and packaging</td>
<td>30-34</td>
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<td>September 4</td>
<td>Cytology and Histology, “Killer in the county” Video</td>
<td>8-9, 29, 40-43</td>
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<td>September 11</td>
<td>DNA + “Secret of Photo 51” Video</td>
<td>8-16</td>
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<td>September 18</td>
<td>Mitosis and Meiosis, Human Genetics</td>
<td>150-156</td>
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<tr>
<td>September 25</td>
<td>Molecular Biology Lab Techniques 1</td>
<td>51-58, 69-70, 78-80, 101-104</td>
</tr>
<tr>
<td>October 2</td>
<td>Midterm Examination</td>
<td><strong>All of the above</strong></td>
</tr>
<tr>
<td>October 9</td>
<td>Molecular Biology Lab Techniques 2</td>
<td>55-64, 70-78, 81-87</td>
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<tr>
<td>October 16</td>
<td>Gender and mtDNA</td>
<td>118-122, 158-159</td>
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<td>October 23</td>
<td>“Journey of Man” Video</td>
<td>118-122, 158-159</td>
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<td>October 30</td>
<td>CODIS and Y-chromosome</td>
<td>48-50, 90-117, 163-166, 184-188</td>
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<td>November 6</td>
<td>DNA from other biological sources</td>
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<td>November 13</td>
<td>DNA limitations</td>
<td>105-113, 128-135, 209-210</td>
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<td>November 20</td>
<td>“Zodiac” Video</td>
<td>---</td>
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<td>November 27</td>
<td><strong>Thanksgiving Holiday, College Closed</strong></td>
<td>---</td>
</tr>
<tr>
<td>December 4</td>
<td>Probability and DNA Databases</td>
<td>Chapters 4 &amp; 7 <strong>Bring a calculator</strong></td>
</tr>
<tr>
<td>December 11</td>
<td>DNA in the Courtroom</td>
<td>Chapter 5</td>
</tr>
<tr>
<td>December 18</td>
<td>Comprehensive Final Examination (3:00-6:00 PM)</td>
<td><strong>STUDENT REPORTS DUE</strong></td>
</tr>
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</table>

**STUDENT REPORTS DUE**

**Bring a calculator**
COURSE DESCRIPTION
An introduction to forensic DNA analysis with emphasis on biology and case studies. Students will develop an appreciation for forensic DNA methodologies and an understanding of their limitations in solving crimes. Designed for all students interested in learning the basic principles of molecular human identification.

COURSE STUDENT LEARNING OUTCOMES
1. Given the DNA molecule, the student will be able to demonstrate a knowledge of its biochemistry.
2. Given a tissue sample, the student will be able to outline the methods of DNA extraction, amplification, and typing.
3. Given allele frequencies, the student will be able to calculate the probability of a genetic match.
4. Given a forensic article, the student will be able to summarize the key findings.

COURSE OBJECTIVES:
The student will
1. apply the scientific method to forensic investigations.
2. arrange the principles of crime scene investigation into their correct order.
3. propose a search pattern given a crime scene.
4. compare and contrast presumptive versus confirmatory tests.
5. outline and label the components of a basic chemical reaction.
6. demonstrate proper evidence collection and packaging techniques.
7. identify the features of all human cells.
8. illustrate the differences between prokaryotic and eukaryotic cells.
9. construct a list of human tissues and fluids from which DNA can be isolated.
10. distinguish between covalent, ionic, and hydrogen bonds.
11. analyze the structure of DNA.
12. assemble the components of the DNA molecule.
13. investigate mitosis and meiosis.
14. examine the chromosomal basis for Mendelian genetics.
15. compare and contrast DNA isolation procedures.
16. describe how gender, mitochondrial, and Y-chromosomal DNA analyses are performed.
17. analyze genetic diversity in human populations.
18. list types of organisms from which DNA are commonly analyzed for forensic purposes.
19. interpret DNA results from forensic case studies.
20. identify behavior that contributes to field and laboratory contamination of samples.
21. assess the strength of genetic evidence using basic statistics.
22. compare and contrast continuous and discrete allele systems.
23. perform frequency estimate calculations on DNA typing data.
24. investigate admissibility standards for using DNA in the courtroom.
25. examine governmental requirements for gaining DNA typing certification and accreditation.
26. evaluate landmark criminal cases where the results from DNA analyses were not admitted.
27. debate the current political and legal issues for using genetic evidence in the courtroom.
INSTRUCTOR INFORMATION
Instructor- Dr. Jeffery R. Hughey
Office- Building N, Room 26C
Office Telephone- (831) 770-7054
Email- jhughey@hartnell.edu
Lecture and Laboratory Material- http://www.hartnell.cc.ca.us/faculty/jhughey/course_doc.htm
Office Hours- Monday 12:30-1:30 PM, Tuesday 5:00-6:00 PM, Thursday 5:00-6:00 PM

REQUIRED TEXTBOOKS

POSSIBLE POINTS AND GRADE DETERMINATION

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<th>Total Points Earned (Percentage)</th>
<th>Definition</th>
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<tr>
<td>A</td>
<td>315-350 pts. (90-100%)</td>
<td>Excellent</td>
<td>4</td>
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<tr>
<td>B</td>
<td>280-314 pts. (80-89%)</td>
<td>Good</td>
<td>3</td>
</tr>
<tr>
<td>C</td>
<td>245-279 pts. (70-79%)</td>
<td>Satisfactory</td>
<td>2</td>
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<tr>
<td>D</td>
<td>210-244 pts. (60-69%)</td>
<td>Barely passing</td>
<td>1</td>
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<tr>
<td>F</td>
<td>209 or fewer (59% or less)</td>
<td>Failing</td>
<td>0</td>
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</tbody>
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LECTURE EXAMINATIONS
Lecture examinations will consist of a combination of multiple choice, true or false, matching, short answer, and essay questions. Questions will come from lecture material, but may also be derived from assigned readings. Supply your own Scantron 882-E form and a number 2 pencil for all examinations.

STUDENT REPORT
Students will select a research paper from the Journal of Forensic Sciences and write a 2-3 page summary. Reports must be typed, single spaced, and printed using “Times New Roman” font size 11. They are to be handed directly in print form to Dr. Hughey at the beginning of the class on December 11th, 2014, not emailed. If you fail to hand the report in on time you will receive a zero. The report is 50 points.

MAKE UP EXAMINATION POLICY
If you are unable to attend an examination please notify me by telephone, email, or in person prior to the test. If you fail to contact me in advance and you miss the examination, submit a written letter signed by the authority involved (doctor, policeman, judge) that includes their phone number and an explanation. If you have a valid excuse, I will schedule an intellectually comparable make up oral or essay examination. The final exam will be given on December 18th at 3:00 pm, no exceptions. No make ups will be granted for the final examination.
ATTENDANCE POLICY
Regular attendance and consistent study are your responsibility and the two factors that contribute most to a successful college experience. I expect you to attend all of the class sessions. Any lack of attendance, which leads in my judgment to unsatisfactory progress, or absence in excess of two weeks (consecutive or non-consecutive), will result in being dropped from the course.

DROPPING THE COURSE
It is your responsibility to drop the course. Do not assume that I will submit the drop for you if you decide to stop coming to class. The last day to drop for a “W” is November 21st. Students that do not officially drop the course by November 21st will receive a letter grade based on their Total Points Earned.

COMMUNICATION WITH YOUR INSTRUCTOR
When communicating by email, phone, or in person, do so in a professional manner. Namely, when emailing, include your complete name, course name, and use complete sentences. Do not abbreviate words or use slang.

CLASSROOM VISITORS
No one is permitted to attend this class unless he or she is a registered student.

CONDUCT
Please show respect for your peers and your instructor. If I observe any student performing or aiding in any of the types of misconduct listed under “Codes of Student Conduct” on page 31 of the Hartnell Catalog, that student will be dropped from the course. Disruptive behavior will not be tolerated (this includes text messaging during class).

INSTRUCTIONAL SUGGESTIONS
Course suggestions and requests that you feel will improve comprehension, retention, and cognition are warmly welcomed. Requests for less information, postponement of examinations, or easier examinations are not welcomed.

SUGGESTIONS FOR DOING WELL IN BIOLOGY 13
Read the textbook, complete and learn the study guides, review your notes before coming to class, study for the examinations with a partner or in a group, attend class and take complete notes, participate in classroom exercises and discussions.
Introduction to Forensic DNA Analysis

Forensics

Aristotle

The Scientific Method

What are the characteristics/traits of a good forensic DNA scientist?

The Scientific Method

• A procedure used to ____________ ____________ or ______________ ______________.
• A way of thinking and looking at the ______________.
• A technique used to gather information and reach ________________.

Steps to the Scientific Method

Principles of Crime Scene Investigation

____ Document the scene
____ Protect the scene
____ Help the victim
____ Search the scene
____ Photograph and draw location of items of evidence
____ Transport evidence to the laboratory
____ Package evidence

Search the Scene

• Goal- “no ______________________________.”
• Patterns and search strategies
  – ______________ and ______________
  – _______________ _______________
  – ________________ ______________
  – ________________ configuration
Presumptive vs. Confirmatory Tests

• Presumptive
  —____________________
  —____________________ specific
  —____________________ specific 
  =____________________ false positives

• Confirmatory
  —Less____________________
  —Specific
  =____________________ to _____________ false positives

Blood

• Presumptive
  —____________________
  —____________________
  —____________________

• Confirmatory
  —____________________crystal test
  —ABAcard_____________________________

Luminol

Hemastix

ABAcard Hematrace

Diffusion

• Diffusion- the __________________________ tendency of a __________________________ (solute or solvent) to move from a _______________ concentrated to a _______________ concentrated area.
  —T or F. Diffusion results in the uniform distribution of a substance.

Double Diffusion=

Spermatozoa

• Presumptive
  —____________________ light
  —____________________ reaction
• Confirmatory
  – __________________________
  – __________________________ or P30
  – __________________________

Male reproductive system and vasectomized individuals

P30 Analysis- ABAcard

Saliva
• Presumptive
  – __________________________ reaction
• Confirmatory
  – __________________________

Fecal Material
• Presumptive
  – __________________________ for undigested materials
  – __________________________ reagent UV
    – __________________________-green fluorescence

Urine
• Presumptive
  – __________________________ light
  – __________________________
    – Litmus paper
  – __________________________
    – __________________________ test

Cold Case Files, A & E- “Frozen in time”
Evidence Collection and Packaging

Evidence Packaging: A How-To Guide Terry Spear, John Rush, Jerry Massetti, Jim Weigand and Mark Traughber, California Department of Justice, Bureau of Forensic Services

Goals of Evidence Packaging
• ___________________________ from possible hazards associated with evidence.
• Correctly __________________ samples.
• Protect evidence against:
  – ___________________________
  – ___________________________
  – ___________________________
  – ___________________________  
  • Suspect to victim  
  • Victim to suspect  
  • Scene to scene  
  • Item to item

Packaging Information
• __________________________: What is it?
• __________________________ of collection.
• __________________________.
• __________________________ of collector.
• __________________________.
  – __________________________ that correlates with a specimen labeled in the schematic drawing or photographic documentation of evidence.
• Custodial agency.
• What processing is needed.
  – “_____________ processing required”
• __________________________ (e.g. sharp knife).
• Required or preferred __________________________.
  – “Store at room temperature” or “Store __________________________”
  – __________________________ and __________________________ over the seal.
• Chain of custody.

What type of packaging is optimal?
• __________________________ of container
• __________________________ of container
  – __________________________ - envelopes and bags.
  • Allows samples that are not completely dry to finish drying.
  – Cardboard __________________________.
  – Plastic safety __________________________.

Processing of Biological Evidence
• Allow samples to __________________________.
  – Do not heat.
• Package evidence __________________________.
• __________________________ evidence when necessary.
  – Firearms and knives.
• Seal, initial, and date evidence.
Does evidence need to be prepared for packaging?
• _________! Certain kinds of evidence may need to be dried before it can be packaged:
  – ________________________ biological samples.
  – Fresh ______________________ material.
• Silica gel desiccant.

What Can Happen When Biological Evidence is Packaged Wet?
• It “leaks” through to the exterior of the container, resulting in
  – Sample ______________________
  – Sample ______________________
  – ______________________

Do Not Package a Wet Bloodstain
• Do not use a packaging device that limits air exchange.
  – Biological samples ______________________ if it takes too long for them to dry.

How to Package Bloodstains
• ______________________ samples.
• Package the samples and the ______________________ into separate envelopes.

Packaging of Clothing
• Minimize the chance of cross transfer by placing clothing items in ______________________ containers.

Packaging Bloody Clothing #1
• Allow bloodstains to ______________________ as much as possible.
• Place clothing onto a piece of ______________________ ______________________.

Packaging Bloody Clothing #2
• Place paper between stained areas to ______________________ ______________________.

Packaging Bloody Clothing #3
• ______________________ and ______________________ paper package.
• Insert paper package into ______________________ ______________________ bag.

This is Not the proper way to package a bloody knife

Why?
• Knives can ______________________ the paper envelope and endanger anyone who handles the evidence.
  – Human immunodeficiency virus (HIV).
  – B virus (HBV).
  – Hepatitis C virus (HCV).
• Blood from the blade can be easily ______________________.

How to Package a Bloody Knife
• Use a ______________________ ______________________ (not airtight).
• ______________________ knife to protect both personnel and the blood sample.

How Not to Handle Firearms Evidence
• Do Not insert anything into the barrel.
EXTREMELY ______________________!
May remove ______________________ or ______________________ evidence.

Packaging Firearms Evidence
• Immobilized with a ______________________ _________________.
• Close, label, and seal box.
• Write “______________________” notification on outside of container.

Drug Evidence: Syringes
• Syringes may contain genetic evidence.
• They may also contain blood borne ______________________ _________________.

Packaging Syringes
• Package syringes in ______________________ - ______________________ containers.

Victim Physical Evidence Recovery Kits (Rape Kit)
1. Sexual assault information form.
2. Outer clothing and debris collection.
3. Lips/Lip area swabs and smear.
4. Oral rinse.
5. Hair contaminated with semen.
6. Head hair standard.
7. Underpants and tampons/sanitary napkin collection.
8. Pubic hair combings.
10. Thighs/external genitalia swabs and smear.
11. Vaginal/cervical swabs and smear.
15. Foreign material collection.
Packaging Trace Evidence
• Trace evidence is ______ evidence and can be easily lost.
• Use ________, ________ blades, and _______ ________ to collect evidence.
• Examples of trace evidence:
  — __________
  — Epidermal ____________ from prints.
  — __________

What do you do once the Evidence is Packaged?
• Seal it with ________.
• Initial and date along the seal to indicate the person responsible for packaging the evidence.
• Why seal?
  — Guard evidence against ________________.
  — Guard evidence ________________.
  — Protect from the ________________ or ________________ of material.

How Not to Seal a Bag
How Not to Seal Envelopes
How to Correctly Seal Evidence
• Tape seal across entire flap of envelope.
• ________________ and ________________ the seal.
Cytology and Histology

Levels of Organization
_____ Tissue Level
_____ Chemical Level
_____ Individual Level
_____ System Level
_____ Cellular Level
_____ Population Level
_____ Organ Level

Matching. Match the definitions on the right to the levels of organization on the left.
_____ Chemical a. group of similar cells that perform a common function
_____ Cellular b. a single organism
_____ Organ c. the chemical components of cells
_____ System d. a group of related organs that have a common function
_____ Individual e. all individuals of the same species in a given area
_____ Population f. two or more different tissues that perform a common function
_____ Tissue g. the basic unit of life

__________________ (1635-1703)
• and ___________ cells in 1665.
• Using a microscope with ____ X magnification, he observed __________ cells from an oak tree.

Cell Theory
• CELL THEORY- proposed in 1839 by Schleiden and Schwann.
  – All living things are _______________ of cells.
  – The cell is the _______________ _____________ of life.
  – ___________ cells arise only from __________________________ cells.
  – Cells contain __________________ ________________________ which is passed from cell
to cell during cell division.
  – All cells are similar in chemical ______________________.
  – All of the energy flow of life occurs within cells.

Prokaryotes and Eukaryotes

Prokaryotic Cell

Eukaryotic Cell
  – Organelles-__________________________________

Types of Biological Evidence
<table>
<thead>
<tr>
<th>Evidence</th>
<th>Possible Location of DNA on the Evidence</th>
<th>Source of DNA</th>
</tr>
</thead>
<tbody>
<tr>
<td>baseball bat or similar weapon</td>
<td>Handle, end</td>
<td>sweat, skin, blood, tissue</td>
</tr>
<tr>
<td>hat, bandanna, or mask</td>
<td></td>
<td>________, ________, dandruff</td>
</tr>
<tr>
<td>eyeglasses</td>
<td></td>
<td>sweat, skin</td>
</tr>
<tr>
<td>facial tissue, cotton swab</td>
<td>Surface area</td>
<td>mucus, blood, _____, semen, ___ _____</td>
</tr>
<tr>
<td>dirty laundry</td>
<td>Surface area</td>
<td>_____, sweat, _____</td>
</tr>
<tr>
<td>toothpick</td>
<td>Tips</td>
<td></td>
</tr>
<tr>
<td>used cigarette</td>
<td>Cigarette butt</td>
<td></td>
</tr>
<tr>
<td>_____ or</td>
<td>Licked area</td>
<td>saliva</td>
</tr>
<tr>
<td>tape or ligature</td>
<td>Inside/outside surface</td>
<td>skin, sweat</td>
</tr>
<tr>
<td>bottle, can, or glass</td>
<td>Sides, mouthpiece</td>
<td>saliva, sweat</td>
</tr>
<tr>
<td>used condom</td>
<td>Inside/_______ surface</td>
<td>_____, vaginal or rectal cells</td>
</tr>
<tr>
<td>condom wrapper</td>
<td>_______ surface</td>
<td>_______ cells</td>
</tr>
<tr>
<td>blanket, pillow, ____</td>
<td>surface area</td>
<td>sweat, hair, semen, urine, saliva</td>
</tr>
<tr>
<td>&quot;through and through&quot; bullet</td>
<td>outside surface</td>
<td>blood, tissue</td>
</tr>
<tr>
<td>bite mark</td>
<td>person's _____ or clothing</td>
<td>saliva</td>
</tr>
<tr>
<td>fingernail, partial fingernail</td>
<td>Scrapings</td>
<td>blood, sweat, tissue</td>
</tr>
</tbody>
</table>
**Tissue** - a group of similar cells that perform a common function.

- **4 Types of Tissues**
  - __________________________
  - __________________________
  - __________________________
  - __________________________

**Epithelial Tissue**
- **Functions** - protects, secretes, and absorbs.
- __________________________ and __________________________ epithelium
  - Epidermis of skin.
  - Lining of blood vessels, respiratory, reproductive, urinary, and GI tract.
- __________________________ epithelium
  - Thyroid, adrenal, sweat glands.

**Layers of the Epidermis**
- Stratum __________________________
- Stratum __________________________
- Stratum __________________________
- Stratum __________________________
- Stratum __________________________ = Stratum germinativum

**Epidermal Ridges**
- Epidermal ridges form in fetus as epidermis conforms to __________________________ __________________________.
  - Increase grip of hand.
  - Fingerprints are left by __________________________ __________________________ that open on the tops of ridges.
  - Stratified squamous epithelial cells are deposited with the print.

**Accessory Structures of Skin**
- **Epidermal derivatives**
  - __________________________
  - Skin __________________________
  - __________________________

**Hair (Pili) Structure**
- Projects from the surface.
  - Medulla, cortex, & cuticle.
- Medulla, cortex, & cuticle.
- Surrounds the root.
  - External and internal root sheaths.
  - **Bulb** - base of the follicle
- Papilla of the hair.
- Matrix.

**Glandular Epithelium**
- **Function:**
- **Gland:** one cell or a group of highly specialized epithelial cells that secrete _______________________ into ________________, onto a ________________, or into the blood.
- **Classified into two categories**
  - Endocrine glands
  - _______________ glands
    - Sebaceous (__________) glands.
    - Sudoriferous (__________) glands.
    - Ceruminous (__________) glands.
    - Mammary (__________) glands.

**Connective Tissue**
- **Functions:** binds organs together, provides support & protection, fills spaces, produces blood cells, & stores fat.
- **Composed of two basic elements**
  - Cells and extracellular matrix
- **Mature Connective Tissue**
  - ________________ connective tissue
  - ________________ connective tissue
  - ________________ Tissue
  - ________________ Tissue
  - ________________ Tissue

**Teeth- dentes**
- **Numbers**
  - 8 ________________
  - 4 ________________
  - 8 ________________
  - 12 ________________
- Accessory digestive structures used to macerate organic materials.
- ________________ cavity and root ________________ contain the most ________.

**Blood**
- **ology:** the study of blood, blood forming tissues, and the disorders associated with them.
- **Blood consists of components**
  - ________________ (55%)
  - ________________ (45%)
    - Erythrocytes (99%)- ________________ blood cells.
    - Leukocytes (<1%)- ________________ blood cells.
    - Platelets (<1%)- fragment of ________________ enclosed in a cell membrane.

**Characteristics of Blood**
• **Description**- tissue with a liquid matrix (plasma) and red blood cells, white blood cells, and platelets.
• ________________ of blood contains ____ million blood cells, about 400,000 of which are ____________.
• Blood is denser than water.
• Blood temperature is about 100.4 °F.
• Blood is about ________% of total body weight.
• Blood volume in
  – ________________ is 5-6 liters (1.5 gallons).
  – ________________ is 4-5 liters (1.2 gallons).

**Muscular System**
• **Functions**- generate force and produce __________________________.
• **Composed of** ________________________________.
• There are 3 types of muscle tissue.
  – ________________
  – ________________
  – ________________

**Neurons**
• **Neuron**- the functional unit of the nervous system.
• **Numbers**
  – 100 billion in the ________________.
  – 1 billion in the ________________.

**Nervous Tissue**
• **Description**- consist of two kinds of cells, neurons and __________________________.
  – Neurons (nerve cells)
    • ________________
    • ________________ Body
    • ________________
  – Neuroglia

**Male Reproductive System**

**Semen and Sperm**
• **Semen**- mixture of ________________ and ________________________________ (60% seminal vesicles, 25% prostate, and about 15% from the bulbourethral gland).
  – Slightly alkaline (7.2-7.7ph).
• Typical ejaculate is ____________ to ____________ ml in volume.
• Normal sperm count is 50 to 150 _____________________/ml.
Cold Case Files, A & E- “Killer in the County”
DNA- Deoxyribonucleic Acid
• Johann Friedrich ___________________________
  — “__________________”
  — White blood cells from ________________.
  — Salmon spermatozoa, nucleus accounts for more than 90% of mass of sperm cells.
    • ____________________ contains ___________________________ information.
    • Rejected this idea after analysis of egg cells.

Phoebus Levene- _____________________________________________________________
Linus Pauling- ______________________________________________________________

Avery, MacLeod, McCarty 1944- __________________________________________________

Erwin Chargaff
• Chargaff’s Rules- ____________________.
  — The number of adenines = _____________________. A=T
  — The number of guanines = _____________________. G=C

The Structure of DNA
• James ______________________ and Francis ______________________ published in 1953.
• Maurice Wilkins and ______________________ ________________________.
  X-ray diffraction pattern

NOVA- “Secret of Photo 51”
Nucleic Acids
• Nucleic acids- _______________________ of nucleotides.
  – Nucleotide- ___________ group, a five-carbon ___________, and a nitrogenous ___________.
• There are _________ types of nucleic acids.
  – ______________- ribonucleic acid.
    • Sugar is ribose.
    • Single stranded.
    • Nitrogenous bases- adenine (A), uracil (______), guanine (G), cytosine (C).
  – ______________- deoxyribonucleic acid.
    • Sugar is __________________________.
    • ______________________ stranded.
    • Nitrogenous bases- adenine (A), thymine (_______), guanine (G), cytosine (C).

DNA Structure
Central Dogma of Molecular Biology
• 1957- James Watson and Francis Crick.
  • Three processes involved in information transfer.
    – __________________- the synthesis of DNA.
    – __________________- the synthesis of RNA.
    – __________________- the synthesis of a protein.

Periodic Table of the Elements
• Elements- substances composed of only one type of atom.
  – ___________ naturally occurring.
    – Carbon, Nitrogen, ______________________, and _______________________ account for 90% of the elements in the human body.

Atoms
• Atom- the smallest unit of an element.
• Atoms contain a ___________ and a _______________________ or shells.
• Atoms consist of 3 subatomic particles.
  – ___________________ = + charge
  – ___________________ = no charge
  – ___________________ = - charge

Atom Stability
• The inner shell can hold _________ electrons.
• The outer shell can hold _________ electrons.
  • Atoms are more stable when their shells are full, 2 for the inner and 8 for each additional outer shell.

Covalent Bond
• Covalent Bond- 2 or more atoms _______________________ electrons in their outer shells.

Ionic Bonds
• Ionic Bond- atoms _______________________ or take on an electron to stabilize their outer shells.
Hydrogen Bond
• Hydrogen Bond- an _______________________ between a slightly positively charged _______________________ atom and a slightly _______________________ charged atom.

DNA Damage
• DNA is susceptible to degradation.
  – Depurination
    – _______________________ Damage
    – _______________________ Damage

DNA Packaging
Chromatin- ____________________________________________.

Fully Condensed Chromosome
• Sister Chromatids- two identical chromosomes.
  • _______________________ - constricted region that joins two sister chromatids.
  • p- ____________ arm.
  • q- ____________ arm.

Matching. Match the definitions on the right to the correct level of organization on the left.
Molecular  a. two or more different tissues that perform a common function
Cell      b. a single organism
Organ     c. the basic unit of life
System    d. a group of related organs that have a common function
Individual e. all individuals of the same species that occupy a given area
Population f. a group of similar cells that perform a common function
Tissue    g. the chemical components of cells
**Mitosis and Meiosis**

**The Human Life Cycle Involves**

• 1) ______________________ – ________________________- a nuclear division of a parental cell that produces ____ daughter cells with the ________ chromosome number as the parental cell. (2N= _______)

• 2) ______________________ – ________________________- two nuclear divisions of a parental cell that produces ___________ daughter cells with ______________ the chromosome number as the parental cell. (N= ________)

Spermatogenesis- _____________________________________________________________________.

Oogenesis- __________________________________________________________________________.

**Cell Cycle**

• Cell cycle consists of:

  – ______________________

  – ______________________ - nuclear division.

  – ______________________ - cytoplasmic division.

**Mitosis**

________________ Stages to Mitosis- PMAT.

– P____________________

– M____________________

– A____________________

– T____________________

Interphase- DNA ______________________________.

Late Interphase- chromatin ____________________________.

Prophase- nuclear envelope ___________________________ and __________________ begins to form.

Metaphase- chromosomes are __________________ at the equator.

Anaphase- daughter chromosomes _____________ to the ________________.

Telophase- two _________________ __________ begin to form and the spindle _____________.

Cytokinesis- ______________________________________.
Meiosis
• Meiosis consists of ________ successive nuclear divisions.
• Genetic material between homologous chromatids is ____________________.
• The product of meiosis is ________ ______________________ nuclei.

• Homologous chromosomes- chromosomes that associate in ___________ in the first stage of meiosis (Prophase ________); each member of the pair is derived from a different parent.

Genetic Exchange
• _____________________ - the pairing of homologous chromosomes.
• ____________________ - __________ - the exchange of genetic material between homologous chromatids.

Crossing Over- Meiosis I
Human Genetics

Gregor Mendel- father of genetics

Mendel’s Methods
• _______________ crosses- an experimental cross between individuals that differ by a ________ trait.
• First generation (____________).

Mendel’s F1 Observations
• One of the traits could be _______________ in the F1 generation= ________________
• One of the traits was _______________ in the F1 generation= ________________

Genetic Terminology #1
• _______________- an organisms traits.
• _______________- an organisms genetic makeup.
• _______________- identical alleles for a gene.
• _______________- two different alleles for a gene.

Genetic Terminology #2
• _______________- a sequence of DNA that codes for a protein.
• _______________- the position on a chromosome occupied by a gene.
• _______________- one of 2 or more alternative forms of the same gene.

Mendel’s F2 Observations
• Flower color
  —Purple:white
  705:224
  _____:_____

Mendel’s Dihybrid Cross
• 315:108:101:32
  = 9:____:____:1

Mendel’s Work Yielded These Genetic Rules
• Alternative versions of genes (different ______________) account for variations in inherited characters.
• For each ______________, an organism inherits two alleles, ___ from the mother and ___ from the father.
• If two alleles differ, then one, the dominant allele is fully ______________ in the organism’s appearance.
• The two alleles for each trait segregate during ________________ ________________.
• Alleles of a gene segregate ________________ of the alleles of other genes.

Punnett Square
• Punnett square- a diagram used to predict the result of a genetic cross.

Freckles (FF or Ff) vs. no Freckles (ff)
Short fingers (SS or Ss) vs. long fingers (ss)

Widow’s peak (WW or Ww) vs. straight hairline (ww)

Unattached earlobes (EE or Ee) vs. attached earlobes (ee)

Phenylthiocarbamide taster (TT or Tt) vs. non-Phenylthiocarbamide taster (tt)

Tongue rolling (TT or Tt) vs. unable to roll tongue (tt)

THO1 (Intron 1 of Tyrosine hydroxylase gene)

________________________ Dominance
• The _____________ allele produces a functional protein and the protein’s effects are ___________.
• The ___________ allele produces a less functional protein or none at all and the trait is _____ apparent.

Codominance- blood types
• Codominance- the effects of both alleles are apparent.

Incomplete Dominance
• Incomplete dominance- inheritance in which the F₁ is __________________ in phenotype between the parents.
• __________________ allele is dominant. Examples= _______________ and ________________.

Continuous Variation is the result of __________________________ Inheritance
• Continuous variation- a __________________________ in phenotype; indicates that a trait is controlled by _________ or ___________ genes. Examples= _______________ and _______________.

________________________
Linkage

- **Linkage**: the tendency for certain genes to be inherited together because they are in __________________________ on the __________________________ chromosome.

- **Linked genes**: genes that are __________________________.

Position of Forensic STR Markers on Human Chromosomes

Allelic Ladders

Paternity
DNA Methodologies Part I

DNA Methodologies

• Sterilization
  – Clean the workstation with ______________________ and ______________________.
  – ______________________ and ultraviolet light (______________ radiation).
• Consumables and reagents.

• Equipment
  – ______________________ - P______, P______, P______
  – Block heater
  – ______________________
  – Centrifuge
  – PCR machine
  – Electrophoresis box and power supply
  – ______________________ machine
  – Computer

DNA Protocol

• DNA
  – FTA Card
  – Chelex
  – Spin columns
  – Organic- simple and differential.

• DNA
  – Direct, non-blot
  – Direct, slot-blot
  – Real Time PCR

• ______________________
  – Polymerase Chain Reaction

• ______________________

Human Cell

How do we get the nuclear and mitochondrial DNA out of the cell?

_____________________________________

_____________________________________

_____________________________________

DNA Extraction Protocols #1

• ______________________ Card
  – ______________________ aliquot on card.
  – ______________________ on paper and trap DNA.
  – Use ______________________ to remove paper for DNA analysis.
  – ______________________
  – ______________________
FTA Protocol

DNA Extraction Protocols #2
• ______________________
  – Incubate blood sample in _______% Chelex at _______ °C for 30 min.
  – ______________________ for 8 min.
  – ______________________ to remove inhibitors and cellular debris.

DNA Extraction Protocols #3
• ______________________
  – ______________________ cells and ______________________ proteins.
    • Physical, heat, detergent
    • Proteinase ________
  – ______________________ DNA to ______________________ membrane by centrifugation.
  – ______________________ with 70% ethanol by centrifugation.
  – ______________________ DNA with TAE (Tris-Acetate - EDTA) or water by centrifugation.

DNA Extraction Protocols #4
• ______________________
  – Lyse cells and digest proteins.
    • Physical, heat, detergent
    • Proteinase K
  – Organic extraction.
    • ______________________
  – Centrifuge to remove supernatant.
  – ______________________ DNA.
    • ______________________ or ______________________
  – Centrifugation to ______________________ DNA.
  – ______________________ DNA with TAE or water.

Differential Extraction
• Technique used to ______________________ ______________________ cells from _______ -sperm cells (______________________ cells).
  1. Epithelial cells are lysed with a __________ extraction buffer using Sodium Dodecyl Sulfate (____).
  2. ______________________ to pellet sperm cells, DNA from epithelial cells is in the supernatant.
  3. ______________________ sperm cells using Dithiothreitol (______________________).
DNA Quantitation

• **DNA Quantitation**
  - Direct, ______________________ -blot
  - Direct, ______________________ -blot
  - Real Time PCR

• **Why quantitate?**
  - __________ Standards require it!
  - 1ng-2.5ng yields ________________ typing ______________________.
  - 1 cell = 6.1pg (________ cells needed for analysis)
  - Too ________ DNA leads to artifacts and too much signal.
  - Too ________ DNA leads to allelic dropout.

DNA Quantitation #1

• **Direct, non-blott**
  - ______________________ (Promega Corp.)
  - Human-specific probe that binds to ________ insertions (highly repetitious DNA).
  - Luciferin-__________ reaction and a luminometer to detect the amount of light.
  - Emission is compared ______________________ ______________________.
  - Standards of known concentration are compared against a sample.

DNA Quantitation #2

• **Direct, slot-blott**
  - ______________________ (Applied Biosystems)
  - Human-specific probe (D17Z1) binds to DNA.
  - ________________ is used to determine the DNA concentration against a set of standards.

DNA Quantitation #3

• **Real Time PCR**
  - ______________________ (Applied Biosystems)
  - Human genes are ______________________.
  - Gene number ______________________ after each ______________________.
  - Each cycle yields ______________________.
  - Fluorescence is recorded and compared against ______________________ to determine DNA concentration.
Study Guide for the Midterm Examination

General recommendations- study your notes, PowerPoint slides, and answer/study the questions in this guide.

Multiple Choice. Select the single best possible answer:
1. The scientific method is ____________________________________________________________________.
   a. a procedure used to solve problems or answer questions
   b. a way of thinking and looking at the world
   c. a technique used to gather information and reach conclusions
   d. applied to everyday situations as well as forensics
   e. all of the above
2. The scientific method involves four steps. Select from below these steps placed in their correct order.
   a. hypothesis, experiment, observation, conclusion
   b. conclusion, hypothesis, experiment, observation
   c. hypothesis, observation, experiment, conclusion
   d. observation, hypothesis, experiment, conclusion
   e. hypothesis, experiment, conclusion, observation
3. There are seven principles to crime scene investigation, which one includes making schematic diagrams,
   photography, and videotaping?
   a. help the victim
   b. protect the scene
   c. document the scene
   d. search the scene
   e. transport to the laboratory
4. Searching the scene involves geometric patterns, which of the following is not one of the patterns typically used?
   a. square
   b. triangular configuration
   c. pie configuration
   d. rectangle
   e. concentric circles
5. Which of the arrangements below lists the levels of organization in the correct order (from smallest to largest)?
   a. molecular, tissue, cellular, organ, system, individual, population
   b. molecular, tissue, organ, system, individual, cellular, population
   c. cellular, molecular, tissue, organ, system, individual, population
   d. molecular, cellular, tissue, organ, system, individual, population
   e. cellular, molecular, organ, tissue, system, individual, population
6. The Englishman Robert Hooke is best known to biologists for ______________________.
   a. developing DNA extraction techniques
   b. describing and naming cells (“cellula”)
   c. inventing the microscope
   d. proposing the cell theory
   e. eating pork and muttering “damn this stuff is good”
7. Which of the following is not one of the tenets of the cell theory?
   a. all living things are composed of cells
   b. the cell is the basic unit of life
   c. new cells arise only from preexisting cells
   d. cells contain hereditary information
   e. cells lyse when they reach maturity
8. Organelles _______________________________________________________________________________.
   a. are membrane-bound compartments
   b. have membranes
   c. are found in eukaryotic organisms
   d. include mitochondria, golgi, endoplasmic reticulum
   e. all of the above
9. Which of the following is not one of the four types of human tissue?
10. Which of the following is **not** an example of connective tissue?
   a. cartilage
   b. neuroglia
   c. lymph
   d. blood
   e. bone

11. The DNA sequencing machines used today
   a. are unable to separate 105 bp DNA fragments from 106 bp DNA fragments
   b. use an argon laser and a camera to capture fluorescence emitted by dyes
   c. run DNA through acrylamide for separation
   d. run DNA through agarose for separation
   e. none of the above

12. The epidermis
   a. contains bone
   b. contains epidermal ridges that form to the dermal papillae
   c. contains one type of cell, they are called keratinocytes
   d. accounts for about 80% of skin
   e. includes the dermis

13. The integument consists of two parts,
   a. the epidermis and dermis
   b. the epidermis and hypodermis
   c. the epidermis and hair
   d. the epidermis and skin
   e. the epidermis and glands

14. Blood
   a. contains plasma
   b. contains white and red blood cells
   c. contains platelets
   d. transports nutrients, gases, hormones, and waste
   e. all of the above

15. Hair
   a. is also called ‘pili’
   b. includes a shaft, root, and hair follicle
   c. contains cells in its medulla, cortex, and root sheaths that contain DNA
   d. is nourished by blood vessels
   e. all of the above

16. Exocrine glands
   a. secrete milk, sweat, oil, and wax
   b. are composed of nervous tissue
   c. secrete products directly into the bloodstream
   d. are composed of connective tissue
   e. secrete hormones, such as human growth hormone (hGh)

17. A typical PCR thermocycling profile for denaturation, annealing, and extension steps is ____________.
   a. 94°C for 30 sec, 55°C for 30 sec, and 72°C for 60 sec (x 32 cycles)
   b. 94°C for 2 min, 70°C for 30 sec, and 72°C for 60 sec (x 32 cycles)
   c. 94°C for 2 min, 70°C for 30 sec, and 72°C for 60 sec (x 100 cycles)
   d. 105°C for 30 sec, 35°C for 30 sec, and 72°C for 60 sec (x 32 cycles)
   e. none of the above

18. Leukocytes
   a. contain nuclei (and therefore DNA)
   b. are red blood cells
c. are found in higher percentages in blood than red blood cells
d. are the same as platelets
e. spend their life embedded in collagen

19. Semen
a. is a mixture of sperm and seminal fluid
b. may contain spermatozoa
c. is slightly alkaline
d. can be identified using ultraviolet light
e. all of the above

20. Evidence should be protected against
a. cross-transfer
b. loss
c. contamination
d. deterioration
e. all of the above

21. When packaging evidence, what are two important things to consider?
a. type and manufacturer of the container
b. type and shape of the container
c. size and type of the container
d. size and shape of the container
e. nothing, as long as its packaged

22. When processing biological evidence, which of the following is incorrect?
a. allow samples to air dry
b. package evidence separately
c. immobilize evidence when necessary
d. heat specimens so they will dry
e. seal, initial, and date evidence

23. When biological evidence is packaged wet, it
a. may leak
b. will deteriorate
c. may result in sample loss
d. may result in contamination
e. all of the above

24. Physical evidence recovery kits contain
a. combs for pubic hair sampling
b. swabs for various sampling
c. envelopes for specimen collection
d. a tube for a known blood sample
e. all of the above

25. DNA
a. is single stranded
b. contains the following nitrogenous bases- A, U, G and C
c. is a polymer of amino acids
d. is a double stranded polymer of nucleotides
e. contains the sugar ribose

26. DNA was first isolated
a. using saliva from a chimpanzee
b. from plants
c. by Johann Friedrich Miescher in 1869
d. by Gregor Mendel in 1858
e. from yeast

27. Humans contain
a. 46 chromosomes, 44 of which are autosomes, 2 are sex chromosomes
b. 46 chromosomes, 2 of which are autosomes, 44 are sex chromosomes
c. 45 chromosomes, 2 of which are autosomes, 43 are sex chromosomes
d. 44 chromosomes, 2 of which are autosomes, 42 are sex chromosomes
28. Chromosomes _________________________________________________________.
   a. carry genetic material
   b. have a short (p) and long arm (q)
   c. consist of histones and DNA
   d. contain centromeres
   e. all of the above

29. The cell cycle _________________________________________________________.
   a. occurs only in reproductive cells
   b. spends about 90% of its time going through mitosis
   c. includes G1, S, G2, mitosis, and cytokinesis
   d. consists entirely of interphase
   e. does not occur in human cells

30. Chargaff’s rules state that _____________________________________________.
   a. the # of adenines (A) = # of guanines (G) and # of thymines (T) = # of cytosines (C)
   b. the # of adenines (A) = # of thymines (T) and # of guanines (G) = # of cytosines (C)
   c. the # of adenines (A) = # of cytosines (C) and # of guanines (G) = # of thymines (T)
   d. the # of adenines (A) = # of thymines (T) and # of guanines (G) = # of thymines (T)
   e. the # of adenines (A) = # of cytosines (C) and # of guanines (G) = # of cytosines (C)

31. Which of the following statements about DNA is false?
   a. it is 2 nm wide
   b. it forms a helix by making one full turn every 10 nucleotides
   c. it is an acronym for “deoxyribonucleic acid”
   d. it has a phosphate/sugar backbone
   e. it is bonded together by one type of bond, that is the covalent bond

32. Nucleotides _________________________________________________________.
   a. are found only in DNA
   b. are biomolecules that wash up on the seashore
   c. consists of a phosphate group, a sugar, and a nitrogenous base
   d. occur in 20 different forms in the nucleus
   e. none of the above

33. Which of the following does not occur in high concentrations in the human body?
   a. oxygen
   b. nitrogen
   c. sodium
   d. carbon
   e. hydrogen

34. Atoms are most stable when _____________________________________________.
   a. the inner shell contains 2 electrons, and the outer shell 8 electrons
   b. the inner shell contains 2 protons, and the outer shell 8 protons
   c. the inner shell contains 2 electrons, and the outer shell 2 electrons
   d. the inner shell contains 2 protons, and the outer shell 2 protons
   e. the number of neutrons are not equal to the number protons in the nucleus

35. Covalent bonds occur when _____________________________________________.
   a. two or more atoms share electrons in their outer shells
   b. atoms give up or take on an electron to stabilize their outer shells
   c. a slightly positively charged hydrogen atom and a slightly negatively charged atom are attracted
   d. sodium atoms bond with chlorine atoms
   e. water bonds with oxygen

36. Which of the following is a false statement about the Central Dogma of Molecular Biology?
   a. DNA self-replicates
   b. transcription of DNA results in the synthesis of mRNA
   c. translation of mRNA results in the synthesis of a polypeptide (protein)
   d. it suggests that proteins are the inherited genetic material
   e. it was proposed by Watson and Crick in 1957

37. Which of the following is not true about Gregor Mendel?
a. he studied 7 characters (traits)
b. he crossed garden peas
c. he was an Austrian monk
d. he is considered the Father of Genetics
e. he concluded that traits in peas could not be predicted mathematically

38. Mendel learned from his F₁ observations that ____________________________________________.
   a. dominance in flower color does not exist in the pea
   b. homozygous recessive, in terms of flower color, was always purple
   c. one of his traits was always dominant and the other was recessive
   d. 50% were the purple phenotype, 50% were white phenotype
   e. none of the above

39. Assuming the freckle (F) form of the gene is dominant over the non freckle (f) form of the gene, what is the probability that a male with the genotype FF and a female with ff will produce a child with freckles?
   a. no freckles
   b. 25% of the time freckles
   c. 50% of the time freckles
   d. 75% of the time freckles
   e. 100% of the time freckles

40. Assuming the freckle (F) form of the gene is dominant over the non freckle (f) form of the gene, what is the probability that a heterozygous male and a heterozygous female will produce a child with freckles?
   a. no freckles
   b. 25% of the time freckles
   c. 50% of the time freckles
   d. 75% of the time freckles
   e. 100% of the time freckles

41. Assuming that the unattached earlobes (E) allele is dominant over attached earlobes (e) allele, what are the resulting genotypes when a heterozygous male and a homozygous recessive female are crossed?
   a. 2 Ee, 1 EE, and 1 ee
   b. 1 Ee, 2 EE, and 1 ee
   c. 2 Ee and 2 ee
   d. 2 Ee and 2 EE
   e. none of the above, your crazy Hughey

42. Which of the following is not an example of polygenic inheritance?
   a. height
   b. skin color
   c. eye color
   d. weight and intelligence
   e. hairline

43. The human life cycle involves ___________________________________.
   a. growth
   b. reproduction
   c. mitosis
   d. meiosis
   e. all of the above

44. Which of the following is not one of the stages of mitosis?
   a. telophase
   b. metaphase
   c. prophase
   d. mitophase
   e. anaphase

45. Meiosis _____________________________________.
   a. consists of two successive nuclear divisions
   b. allows for homologous chromatids is exchange genetic material
   c. takes place in the gonads (ovaries and testes)
   d. produces four haploid nuclei
46. Which of the following is not a necessary prerequisite to mitosis?
   a. organelle duplication
   b. cytoplasmic synthesis
   c. replication or synthesis of DNA
   d. synthesis of proteins
   e. interphase

47. Which of the following is not one of the protocols commonly used to extract DNA?
   a. FTA card
   b. chelex
   c. spin columns
   d. GeneAmp
   e. organic-phenol/chloroform

48. Which of the following is not one of the protocols commonly used to extract DNA?
   a. FTA card
   b. chelex
   c. spin columns
   d. GeneAmp
   e. organic-phenol/chloroform

49. Which of the following is not involved in a FTA card extraction?
   a. pipette aliquot on card
   b. centrifugation
   c. white blood cells lysing on paper
   d. hole punch
   e. wash

50. The purpose of a differential extraction is to ____________________________.
   a. combine sperm cells with non-sperm cells
   b. separate sperm cells from non-sperm cells
   c. lyse sperm cells so that non-sperm cells can be analyzed
   d. remove inhibitors
   e. none of the above

51. Why quantitate DNA extractions?
   a. U.S. FBI Standards require it
   b. 1ng-2.5ng yields consistent typing results
   c. too much DNA leads to artifacts
   d. too little DNA leads to allelic dropout
   e. all of the above

52. Which of the following is not one of the methods commonly used to quantitate DNA?
   a. non-blot with AluQuant
   b. non-blot with using a yield gel
   c. slot-blot with QuantiBlot
   d. real time PCR using the Quantifiler kit
   e. slot-blot with QiaQuant

53. Which of the following is not involved in a column extraction?
   a. lysing of cells and digestion of proteins
   b. binding DNA to silica membrane by centrifugation
   c. washing with 70% ethanol by centrifugation
   d. precipitation of DNA
   e. eluting of DNA with TAE (Tris-Acetate - EDTA) or water by centrifugation

True or False. If false, correct the statement.
   ______ Forensics deals with the application of science in solving legal problems.
   ______ Presumptive tests are less sensitive and more specific than confirmatory tests.
   ______ Presumptive tests sometimes give false positives.
   ______ The luminol reaction is a type of chemiluminescence.
   ______ Cytology is the study of bones.
   ______ Histology is the study of tissues.
The cell is the basic unit of life. An organ is a group of similar cells that perform a common function. A population is all individuals of the same species that occupy a given area. Prokaryotic cells contain organelles. Eukaryotic cells contain a “true kernel” or a nucleus. The plasma membrane is the thin and flexible outer boundary of the mitochondria. The plasma membrane is composed of a phospholipid bilayer. The nucleus and the golgi apparatus contain DNA.

In addition to sweat, stratified squamous epithelial cells are left with a fingerprint. Sweat from sudoriferous glands contains cells that can be used for DNA analysis. Connective tissue consists of cells and extracellular matrix. Muscle tissue does not contain cells, and therefore cannot be used for DNA analysis. The three types of muscle tissue are skeletal, striated, and cardiac. DNA in neurons comes from the nucleus, which is located in the axon of the cell. Protecting personnel from possible hazards is one of the goals of evidence packaging. Certain kinds of evidence may need to be dried before packaging. When packaging multiple clothing items, items should be placed in one bag. Packaging clothing items requires the use of paper to prevent cross-transfer from stained areas. Knives and firearms are immobilized with a plastic tie in a air-tight container prior to returning to the lab. Trace evidence is small evidence and can be easily lost. Trace evidence may require the use of tweezers, razor blades, and tape lifts for collection. DNA is packaged in the nucleus. Adenine and cytosine are purines.

The bonds that hold DNA together are not susceptible to degradation. Oxidative and hydrolytic damage results in fragmented and unanalyzable DNA. Monohybrid crosses are experimental crosses between individuals that differ by a single trait. A homozygous individual has identical alleles for a gene. The product of mitosis in humans is two identical cells with the same number of chromosomes (2N= 46). The genotype of an organism refers to its appearance, not its genes. A Punnett square is a diagram used to predict the result of a genetic cross. Codominance in genetics occurs when the effects of both alleles are apparent. Incomplete dominance is where the F1 is intermediate in phenotype between the parents. Continuous variation is due to polygenic inheritance. An individual that is homozygous for an STR locus will contain alleles with the same number of repeats. Walther Fleming described mitosis in 1882. Crossing-over is the exchange of genetic material between homologous chromatids. Mitosis and meiosis are types of nuclear division. The result of meiosis in humans is a reduction in the number of chromosomes from 46 (2N) to 23 (N). The Ouchterlony procedure is a double diffusion technique. The Ouchterlony procedure can be used to determine if a sample of biological fluid is human in origin.

Match the test type on the right to the method on the left.
1. Luminol a. presumptive
2. Phenolphthallin b. confirmatory
3. Takayama crystal test
4. Ultraviolet light
5. Microscopy
6. Lugol’s (I2KI) reaction
7. Brentamine
8. Christmas tree stain
9. Phadebas
10. Hemastix
11. ABAcard Hematrace
12. P30
13. Edelman’s reagent
14. Jaffe test
Match the tissue on the right to the method on the left.
1. Luminol   a. blood
2. Phenolphthalin   b. seminal fluid
3. Takayama crystal test   c. saliva
4. Ultraviolet light   d. spermatozoa
5. Microscopy   e. fecal material
6. Lugol’s (I₂KI) reaction   f. urine
7. Brentamine
8. Christmas tree stain
9. Phadebas
10. Hemastix
11. ABAcard Hematrace
12. P30
13. Edelman’s reagent
14. Jaffe test

Matching. Match the definitions on the right to the correct level of organization on the left.
_____ Molecular   a. two or more different tissues that perform a common function
_____ Cell   b. a single organism
_____ Organ   c. the basic unit of life
_____ System   d. a group of related organs that have a common function
_____ Individual   e. all individuals of the same species that occupy a given area
_____ Population   f. a group of similar cells that perform a common function
_____ Tissue   g. the chemical components of cells

Match the function on the right to the tissue type on the left.
1. Epithelial   a. binds and supports body parts
2. Connective   b. moves the body and its parts
3. Muscle   c. covers body surfaces and lines body cavities
4. Nervous   d. receives stimuli and conducts nerve impulses

Match the definition on the right to the physiological response on the left.
1. Gene   a. a sequence of DNA that codes for a protein
2. Locus   b. an organism’s traits
3. Allele   c. the position on a chromosome occupied by a gene
4. Genotype   d. one of 2 or more alternative forms of the same gene
5. Phenotype   e. an organism’s genetic makeup

List a dozen types of biological evidence that DNA can be extracted from.
_______________, _______________,_______________,_______________,_______________,_______________,_______________,_______________,_______________,_______________,_______________,_______________.

Place the following principles of crime scene investigation in the correct order:
_____ Document the scene
_____ Protect the scene
_____ Help the victim
_____ Search the scene
_____ Photograph and draw location of items of evidence
_____ Transport evidence to the laboratory
_____ Package evidence

When packaging evidence what type of information needs to be listed on the package?
_______________, _______________,_______________,_______________,_______________,_______________,_______________.
Describe the correct technique for sealing packaged evidence.

Summarize the video “Secret of Photo 51”.

Describe 4 differences between DNA and RNA.

Describe, illustrate, and label the components involved in and the events of mitosis.

Describe the events of meiosis.

**Match the pipette on the right to the appropriate volumes on the left.**

1. 200-1000 μl  
   - a. P20

2. 20-200 μl  
   - b. P200

3. 1-20 μl  
   - c. P1000
DNA Methodologies Part II

DNA Typing Methods

• ______________________ - restriction fragment length polymorphism.
• *AmpliType®* + ______________________
• ______________________
  – ______________________ DNA typing.
• ______________________ - short tandem repeats (PCR based).

DNA Typing and Criminal Investigations

• 1980s- Lynda Mann and Dawn Ashworth.
  – kitchen porter.
  – Confessed to the second murder.
• Scotland Yard.
  – Semen samples and blood from Buckland taken.
  – ______________________ at Leicester University.
  –DNA ______________________.
  – Buckland was __________________________________________ the rapist/murderer!
• Voluntary blood submission and DNA testing.
  – ______________________ and Ian Kelly- bakery workers.
  – Kelly bragged about the blood switch.
• Colin Pitchfork was arrested and subsequently confessed in 1987.
• DNA analysis ______________________ match.

Restriction Enzymes

• An enzyme that __________ DNA at __________________ internal sites in the nucleotide sequence.
• They recognize specific __________________-stranded sequences in DNA.
• Recognition sites are ______ base pairs (bp) long.
• Recognition sites are ______________________.
• Arrows indicate cut sites.

The First Human Genetic Fingerprint

Gel Electrophoresis

• Technique used to ______________ on the basis of _________ (number of base pairs).
• DNA samples (mixed with loading dye) are loaded into ______________________.
• Samples are pushed through a gel (____________________ or ______________________) while under the influence of an ____________________________ electrode (____________________).
• DNA is negatively charged due to ______________________ groups and thus migrates towards the ______________________ electrode (____________________).

  The ______________________ the DNA, the ______________________ it travels.

*AmpliType®*PM+DQA1
PCR-________________________________________________

• Nobel Prize in chemistry in 1993.

• PCR is used to target and replicate ________________________________.
  – Copy high numbers (1 trillion) of target in 2-3 hours.

• A technique that involves repeated cycles of 3 steps:
  – __________________________
  – __________________________
  – __________________________

**Thermal Cycling Temperatures**

**PCR Components**

• __________________________ - catalyzes the attachment of the nucleotides to the growing strand of DNA.
• __________________________ - deoxynucleotide triphosphates (A,T,G,C).
• __________________________ - required to activate Taq Polymerase.
• __________________________ - salt and pH balanced.
• __________________________ - a single-stranded short chain of nucleotides (10-30 nucleotides in length).
• __________________________

• Water

**PCR Reaction**

• 25 µL reactions
  – PCR tubes- ______________, for rapid heat transfer.
  – __________________________
  – An instrument that is programmed to repeatedly raise and lower the temperature of a heating block.

  – Cycling Parameters
  • 94°C for 3 minutes- initial denaturation.
  • __________________________ cycles of the following:
    – __________ °C for 30 seconds.
    – __________ °C for 30 seconds.
    – __________ °C for 60 seconds.
  • 72°C for 5 minutes- final extension.
  • 4°C until processing.

**PCR Products**

PCR products are separated on agarose gel.

**PCR products can be sequenced.**
ABI 377 DNA Sequencing Machines

Capillary Electrophoresis- AB 310

Capillary System

DNA Separation Mechanism

ABI 3100 Array Detection

Fluorescent Dyes Used in 4-Color Detection

Fluorescent Emission Spectra for ABI Dyes

Principles of Sample Separation and Detection

MALDI-TOF Mass Spectrometry
Short Tandem Repeats (______________)
• The ___________________________ is ____________________ between samples while the
  ___________________________ regions where primers anneal are ____________________.
    – ___________________________ - both alleles are the same length.
    – ___________________________ - alleles differ in length.

Multiplex PCR
• ___________________________ PCR reactions are run simultaneously in ________ tube.
  – Up to ________ reactions.
  – <=1 ng sensitivity.
• ___________________________ dyes used to distinguish STR alleles with overlapping
  size ranges.

Position of Forensic STR Markers on Human Chromosomes

STR Primers

Fluorescent Labeling of STR PCR Products
• ___________________________ are attached to one ___________________________ in a pair used to amplify a STR
  marker.
• Dye-labeled oligonucleotide is incorporated into PCR product during multiplex PCR amplification
  giving a specific color “____________________” to each PCR ___________________________.

Genome Sequencing- Next Generation Sequencing
Gender ID and Mitochondrial DNA

Sex Typing- Amelogenin
• ________________ - a gene that determines tooth bud formation in the fetus; gene for tooth pulp.
• Located on _______ and _______ chromosomes.
• The ______________ of the gene is _______ bp (AAAGTG) ______________ than the X.

Mitochondrial DNA Analysis
• Mitochondrial Genome= ________________ bp.
  – ______________ sites are variable in humans.
• D-Loop - ______________________
  – ______________ variable Sequence 1 (HVS 1)
    • 16024-16383 (360 bp).
    • __________ var sites occur in the HVS 1.
    • Variation ranges from _______ - _______ bp.
    • On ______________, humans vary by _______ bp.
  – ______________ variable Sequence 2 (HVS 2)
    • 73-340 (268 bp).
    • __________ var sites occur in the HVS 2.
    • Variation ranges from 0-4 bp.
    • On average, humans vary by 2 bp.
• Why analyze the HVS 1 and 2 regions?
  – Shows a lot of ______________.
  – There are ______________ copies/cell (200-10,000).
  – Good for ______________ or ______________ materials.
  – Large ______________ of the world’s variation for comparison and analysis.

Haplogroup Designation
• ________________ - the paternal or maternal genetic constitution of an individual.
  – __________ haplogroups have been defined.
    • Many additional ______________ have been described.
• Designation- based on ______________ observed in the HVS 1 sequence of the d-loop, and also other SNPs throughout the mitochondrial genome.
  – Cambridge Reference Sequence (_____________).

Haplogroup Designation
CRS  GTATTTCTGATATTCTGGCAGCCACCCTGAATATTGTACGGTACCATAAAATACTTGACCACC
F5   GTATTTCTGATATTCTGGCAGCCACCCTGAATATTGTACGGTACCATAAAATACTTGACCACC

CRS  GTATTTCTGATATTCTGGCAGCCACCCTGAATATTGTACGGTACCATAAAATACTTGACCACC
F5   ..........................................................C. ........................................
Haplogroup Origins

- _______________: H, T, U, V, X, K, N, I, J
- _______________: L, L1, L2, L3, L3*
- _______________: A, B, C, D, E, F, G (note: C, D, E, & G belong to macro-haplogroup M)
- _______________: A, B, C, D, and X

The Romanov Family

- Ruled ____________ for years, overthrown in ____________. 
- The family was ________________ by firing squad in 1918. 
- ________________ was discovered in 1991 with four males and five females, supposedly six females were executed. 
- ____________________ - Polish peasant girl?, claimed to be Anastasia.

Anastasia and Anna Anderson

Heteroplasmy-

Mass Disasters and mtDNA

- ________________ Mass Graves
- 9/11
- ________________ Flight 111
- 2004 Indonesian ________________
- ________________ investigations
CODIS and Y-STRs

FBI’s CODIS DNA Database

• Launched October ___________
• Links to all _________ states.
• Used for linking serial crimes and unsolved cases to repeat offenders.
• Current backlog of > ______________________ samples.
• Requires >4 RFLP markers and/or ________________________________

Position of Forensic STR Markers on Autosomes

Information on 13 CODIS STRs

Short Tandem Repeats (STRs)

Commercial STR Kits

Commercially Available STR Kits
Promega Corporation-
Applied Biosystems-

Why STRs are Preferred Genetic Markers
• ______________________ processing.
• Abundant throughout the genome.
• __________________________ within populations.
• __________________________ allows multiplex development.
• Discrete alleles allow digital record of data.
• Allelic ladders __________________________.
• PCR based which __________________________ material.
• Small product size __________________________ with __________________________ DNA.
Microsatellite Mutations
•Polymerase _____________________
•_________________ _____________________ during meiosis

Paternity Testing with Codis STRs

Bill Clinton

Separating the Men from the Sample

Why the Y Chromosome?
•Applications
  – ___________% of violent crime by ____________.
  – __________________ purposes.
  – __________________ studies.
•Advantages to Y Identity Testing
  –Male component isolated without ____________________________.
  –Paternal lineages.

Haplogroups and geographic origins of all individuals in the Y Chromosome Consortium (2002) collection- What is this similar to?
Y-Chromosomal STRs
• ________,000,000 base pairs (only chromosome 22 is smaller).
• ____________ genes.
• 95% is ____________ (nonrecombining Y).
• 5% of it at the tips (telomeric) recombines.
• _________ Y-STR loci have been identified.
• __________________ (Scientific Working Group on DNA Analysis) endorses the use of _____ loci.

Promega Corporation- ________________________________

Applied Biosystems- ________________________________

Reliagene- Y-Plex 6, ________________________________

DNA From Other Sources

DNA From Other Sources
•  
• Archaea
•  

STRs in Grapes

STRs in _________ and _________

Cannabis Typing Using AFLP= ____________________________

Bryophytes

Insect Mitochondrial DNA- Maggots
• Data can be used to identify:
  — The human ____________ that the maggot was eating.
  — The ____________ of maggot.

PBS, Scientific American Frontiers- “Palo Verde Tree”
DNA Limitations

DNA Analysis Outcomes
   1. Match   2. Exclusion   3. Inconclusive

   a. The genotype comparison shows profile differences that can only be explained by _______ _______ _______ _______ from ____________ sources.

   b. Peaks between the compared STR profiles have the ____________________  ____________________ and no unexplainable differences exist between the samples.

   c. The data does not support a conclusion as to whether the profiles match. This finding might be reported if ___________ are in ____________ after review and discussion of the data. Insufficient information exists to support any conclusion.

DNA and Its Problems/Limitations

Blobs and STRs
*Free dye (________________________________________) can be injected into the capillary and will interfere with the typing of STR alleles.

*Dye blobs are ________________ and usually of ________________ intensity than true STR alleles.

Dye Blobs and ___________________________

Stutter Products
*Stutter Products- peaks that show up primarily ____________________ than the true allele as a result of strand slippage during DNA synthesis.

*Stutter is less pronounced with larger repeat unit sizes, dinucleotides > tri- > tetra- > penta-.

*Stutter peaks make _________________  _______________________ more difficult.

Non-template Addition
*Some Taq polymerases will add an ____________________ “A” to the end of the PCR product.

*Addition is enhanced with ____________________ at the end of the PCR cycle (e.g., 15 min @ 72 °C).

*Addition can be eliminated using _____________ polymerases.

*For analysis it is best if there is NOT a mixture of “+- A” peaks.
DNA Sequence Variation in the PCR Primer Binding Site May Cause _________________________

Null Alleles
*Allele is present in the DNA sample but __________________ to __________________ due to a nucleotide change in a primer binding site.
*Allelic dropout is a problem because a heterozygous sample appears falsely as a homozygote.
*Two PCR primer sets can yield different results on samples originating from the same source.
*This phenomenon impacts DNA databases.
*Large concordance studies are typically performed prior to use of new STR kits.

Microvariants in STRs
*Microvariant- __________________ that are
    —not __________________ of the basic repeat motif.
    —________________ of the repeat motif.
    —both of the above.

Mutations can complicate the analysis

Measured Mutation Rates

Multiple Contributors- mixed samples.
Probability and Databanks

Product Rule
• What are the odds of a flipped coin landing on heads?
  \(-50:50 = \frac{1}{2} (1 \text{ in } 2) = \text{________________________ frequency}\)
• The odds of flipping 2 coins simultaneously and landing 2 heads?
  \(-\frac{1}{2} \times \frac{1}{2} = \frac{1}{4} (1 \text{ in } 4) = \text{________________________ frequency}\)
• The odds of flipping 3 coins simultaneously and landing 3 heads?
  \(-\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{8} (1 \text{ in } 8) = \text{________________________ frequency}\)

Allele Combination- 6 alleles
• How many alleles for this locus?
• How many combinations are possible?
• What is the probability of rolling 1 and 3?

Identifiler Allelic Ladders

Allele Frequencies CSF1PO

Hardy-Weinberg
• Allele frequencies at any particular locus can be determined by _________________ a number of _________________ from that _________________.
• The combined frequencies of all possible alleles must account for 100%.

The Probability of Identity (__________)
• Probability of Identity (\(P_i\))- the probability that _________________ selected at _________________ will have an _________________ ________________.

AmpFISTR Identifiler

Allele Frequencies CSF1PO

Identifiler Allelic Ladders
Allele Combinations- 2 alleles

• Heads= ____________________
• Tails= ____________________
• How many combinations are possible?
  – Homozygous 1= ______________
  – Homozygous 2= ______________
  – Heterozygous= ______________

Determining the Combined Probability of Identity (CPi)

• Combined $P_i= (P_i1) (P_i2) (P_i3) (P_i4) (P_i5) (P_i6) (P_i7) (P_i8) (P_i9) (P_{10}) (P_{11}) (P_{12}) (P_{13}) (P_{14})$

• Homozygous loci- $pp$ or ________ ($freq \times freq$) (Actually its $p^2 + (p \times (1-p) \times \theta) = frequency$)
• Heterozygous loci- ________ ($2 \times freq \times freq$)

Determining the Combined Probability of Identity (CPi)

• Combined $P_i= (0.0534) (0.0168) (0.0856) (0.0738) (0.0039) (0.0632) (0.0246) (0.0696) (0.0364) (0.0394) (0.0536) (0.0506) (0.0518) (0.0720) (0.0796) = 2.76 \times 10^{-21}$

\[
\frac{1 \times 10^{21}}{2.76} = 3.62 \times 10^{20}
\]

$3.62 \times 10^{20}= 1 \text{ in } 362,000,000,000,000,000,000$

Power of Discrimination
• Power of Discrimination= ______________
  – The power to ________________ people at a given locus/loci.

\[P_D = 1 - P_I\]

Combined $P_I= 2.76 \times 10^{-21}$

Commercially Available STR Kits
Probability and mtDNA

• Mitochondrial DNA sequences are referred to as ________________ (HVS 1 and HVS 2).
• If the database consists of 100,000 profiles and the mitotype in question has not been reported previously, then the odds are:
  – 1 in ____________________.

Power of Exclusion (_____) - the probability, averaged over all possible mother-child pairs, that a ________________ _____________ __________________ will be __________ from paternity.

Cumulative Power of Exclusion (CPE)= 1- (1-P1) (1-P2) (1-P3) (1-P4) (1-P5) (1-P6) (1-P7) (1-P8) (1-P9) (1-P10) (1-P11) (1-P12) (1-P13) (1-P14) (1-P15)

The Probability of Paternity

• The existence of non-matching markers:
  – Probability of Paternity= ___________% that the alleged father is the biological father.
• The existence of matching markers:
  – Probability of Paternity= ___________% that the alleged father is the biological father.

STR and mtDNA Databases

• Computers and DNA Databases.
• As of October 2008 the STR profile composition of the ____________________________________ (NDIS) is:
  – Total number of profiles:
    • 6,384,379
  – Total forensic profiles:
    • 241,685
  – Total convicted offender profiles:
    • 6,626,064
DNA in the Courtroom

\[ \text{____________} \text{ v. United States. 1923. The Frye Standard} \]

• Lie-detector (_________________________ test).
• Sufficiently established to have gained _______________ ________________ by scientific community.
• The Frye court bestowed on judges a gatekeeping function.

\[ \text{____________} \text{ v. Merrell Dow Pharmaceuticals, Inc. 1993. The Daubert Standard.} \]

• Has the scientific technique been tested? Validity?
• Peer review or publication?
• Is the rate of error known?
• What is the degree of acceptance?

TWGDAM and SWGDAM

• TWGDAM- _________________ Working Group on DNA Analysis Methods.
  – Established to assure that DNA testing was performed reliably.
• SWGDAM- _________________ Working Group on DNA Analysis Methods.
  – Recommend revisions to quality assurance standards.
  – Serve as a forum to discuss, share, evaluate forensic biology methods.
  – Recommend and conduct research to develop and validate methods.

Admissibility of RFLP Data


STR and mtDNA Decisions

• PCR-STR DNA Evidence- 9 cases in California.
  – not disputed, but there are exceptions.
• Mitochondrial DNA- 1 case in California.
  – Admissible in at least 18 states.
Exonerating the Innocent

• The _______________________  _______________________
  —Barry __________________ and Peter _____________________ 1992
  —Rape and homicide
  —______________ inmates exonerated as of 5/8/2007
  —Most cases received go unresolved
  —________ states allow for postconviction DNA testing

• DNA Exonerations By Year In US
• Factors Leading To Wrongful Convictions

Roger Keith Coleman

• Roger waiting for an interview on death row in the Greensville Correctional Center, Jarratt, VA, May 20,1992.
  New DNA tests confirmed the _________________ of Coleman, who went to his death in Virginia’s electric chair in 1992.

Defending DNA Evidence

• Use an ___________________
  —Admission of DNA ______________  _________________
  —Admission of ______________________
  —Admission of an ______________________ of the DNA results

Attacking DNA Evidence

• Use an expert/s
  —Admission of DNA test results
  —Admission of statistics
  —Admission of an explanation of the DNA results

Admission of DNA test results

• New type of DNA test
• Expert not qualified to testify to DNA results
• Laboratory not accredited
• Testing not performed by certified technicians
• Lack of discovery material or notice with respect to the admission
• Improperly obtained DNA evidence
• DNA profile should have been purged from database
Admission of Statistics
• Expert not qualified to testify to statistics
• Statistics do not conform to standards accepted by the scientific community
• Improper database used

Admission of an explanation of the DNA results
• Expert not qualified to testify to statistics for specific context
• Attacking laboratory techniques and conditions
  — Use of accepted techniques
  — Quality control and assurance
  — Use of proficiency testing and audits
  — Laboratory error

General Routes of Attack
• DNA test that is used
• Chain of custody
• Expert Witness
• Contamination
• Choice not to employ several different DNA tests, sequencing
• Use of PCR
• Preventing testimony regarding the issue being decided
BIO 13- Study Guide for the Final Examination

Multiple choice and other miscellaneous practice questions.

1. Different sized DNA molecules can be separated _____________________________________________.
   a. using capillary based electrophoresis
   b. using agarose gel electrophoresis
   c. using acrylamide gel electrophoresis
   d. because DNA is negatively charged
   e. all of the above

2. Which of the following is not one of the techniques used for DNA typing?
   a. RFLP
   b. AmpliType®/PM+DQA1
   c. NMR
   d. STR
   e. DNA sequencing

3. Dr. Alec Jeffreys from Leicester University in the UK is known for _____________________________.
   a. inventing the polymerase chain reaction
   b. sequencing human DNA
   c. being the first to use DNA in forensics
   d. being the first to extract DNA from buccal swabs
   e. none of the above

4. Which of the following is not true of restriction enzymes?
   a. they cut DNA at specific internal sites in the nucleotide sequence
   b. they recognize specific double stranded sequences in DNA
   c. the recognition sites are palindromes
   d. they can only be used to cleave (cut) mitochondrial DNA
   e. recognition sites are 4-8 base pairs (bp) long

5. The polymerase chain reaction _____________________________________________________________.
   a. makes trillions of copies of any DNA target in 2-3 hours
   b. requires buffer and enzyme
   c. involves denaturation, annealing, and extension steps
   d. requires a thermocycler
   e. all of the above

6. The amelogenin gene _____________________________________________________________________.
   a. is used to determine the sex of an individual
   b. determines tooth bud formation in the fetus
   c. is found on the X and Y chromosomes
   d. on the Y chromosome is 6 bp longer than the X chromosome
   e. all of the above

7. Which of the following is not true about the mitochondrial genome?
   a. it contains about 16,569 nucleotides
   b. about 530 of its nucleotide sites are variable in humans
   c. it contains 2 Hypervariable Sequence regions (HVS 1 and HVS 2)
   d. it occurs in low copy numbers per cell (10-40 copies/cell)
   e. it is ideal for analyzing trace or degraded materials

8. Mitochondrial haplogroups are determined by _______________________________________________.
   a. using STR technology
   b. identifying differences observed in the HVS 1 sequence and/or SNPs throughout the genome
   c. sequencing the Y chromosome
   d. quantifying the genomic DNA of an individual and then comparing that data to their mother
   e. using a molecular-weight size marker

9. Heteroplasmy ____________________________________________________________________________.
   a. is an individual that has 2 distinct mitochondrial genomes in their cells
   b. is when differentiated cells that makeup an individuals body tissues begin to leak protoplasm
   c. is when two individuals have the same mtDNA
d. is having a different type of cytoplasm from one’s mother

10. The use of mtDNA techniques for the purpose of human identification after mass disasters is common practice. List four disasters where DNA was employed: _______________________________, _______________________________, _______________________________, _______________________________.

11. DNA analysis is useful __________________________________________.

   a. in determining paternity
   b. in identifying human remains following mass disasters
   c. in missing persons investigations
   d. for matching the suspect with the evidence found at a crime scene
   e. all of the above

True or False. If false, correct the statement.

_____ Gel electrophoresis is a method used to separate DNA.

_____ DNA migrates towards the negative electrode (anode).

_____ AmpliType® PM+DQA1 uses a paper-like strip with probes to analyze PCR products.

_____ The 310 Genetic Analyzer from Applied Biosystems uses agarose gel to separate DNA fragments.

_____ Multiplex PCR is when multiple PCR reactions are run simultaneously in 1 tube.

_____ The acronym STR is an abbreviation for “short tandem repeat.”

_____ In STR typing one of the two primers is tagged with a fluorochrome.

_____ On an electropherogram a female individual will show 1 amelogenin peak.

_____ A haplogroup is defined as the paternal or maternal genetic constitution of an individual.

_____ Using mitochondrial and Y-chromosomal DNA the ethnic background of a male can be predicted.

_____ Mitochondrial and Y-chromosomal sequences can sometimes resolve an individual’s country of origin.

_____ Mitochondrial DNA sequences are referred to as mitotypes.

Describe the Polymerase Chain Reaction (Yes you will need to do this on the test, guaranteed!).

Describe how the capillary electrophoresis sequencing machine works.

12. CODIS is an acronym for the ______________________________________________________________.
   STR is an acronym for _____________________________________________________________________.

13. The “Core set” or CODIS STRs, includes _______ loci.
   a. 9   
   b. 10  
   c. 11  
   d. 12  
   e. 13  

14. Which of the following best illustrates a tetranucleotide motif?
   a. AAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAA
   b. ATATATATATATATATATATATATATATAT
   c. ATTGATTGATTGATTGATTGATTGATTGATTG
   d. ATATGATATGATATGATATGATATGATATGATAG
   e. ATTATTATTATTATTATTATTATTATTATTATTATTATT

15. What is the name of the company that produces the “AmpFISTR® Identifiler™” kit? ______________________

16. What is the name of the company that produces the “PowerPlex™ 16” kit? ______________________________

Given the following:

17. Circle the loci that are heterozygous.

18. Which locus produces the largest DNA fragment? (Hint- the sizes in base pairs are listed along the top)

19. How do the two fragments shown for D8S1179 differ? What do the 12 and 14 stand for?

20. In “D7S820”, what does the “D” stand for? What does the “7” indicate? What does the “S” indicate? What does the “820” stand for?

21. Allelic ladders are ________________________________________________________________.
   a. DNA standards that are used to determine the sizes of unknown alleles from sample data
   b. DNA mixes that contain one heterozygous and one homozygous individual
   c. physical agents that induce DNA mutations
   d. short segments of single-stranded DNA that are tagged with radioactivity
   e. used by vertically challenged forensic DNA scientists to load the DNA sequencing machine

22. Which of the following is not a reason why STRs are preferred genetic markers?
   a. rapid processing
b. abundant throughout the genome
c. highly variable within populations
d. small size ranges allow for multiplex PCR
e. 1,000-5,000 copies/marker per cell

23. Based on the STR data below, which one of the following alleged fathers is the biological father? Why?

<table>
<thead>
<tr>
<th></th>
<th>Mother</th>
<th>Child</th>
<th>Alleged #1</th>
<th>Alleged #2</th>
<th>Alleged #3</th>
</tr>
</thead>
<tbody>
<tr>
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<td>12, 13</td>
<td>11, 12</td>
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<td>6, 7</td>
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<td>8, 9</td>
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<td>8, 9</td>
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<td>13, 14</td>
<td>18, 18</td>
<td>13, 18</td>
<td>11, 12</td>
</tr>
<tr>
<td>D21S11</td>
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<td>29, 32</td>
<td>28, 29</td>
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</tr>
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<td>11, 13</td>
<td>10, 13</td>
<td>14, 16</td>
<td>12, 12</td>
<td>17, 18</td>
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<td>22, 25</td>
<td>20, 24</td>
<td>16, 30</td>
<td>17, 18</td>
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<td>15, 16</td>
<td>15, 19</td>
<td>14, 17</td>
<td>14, 14</td>
<td>20, 21</td>
</tr>
<tr>
<td>D3S1358</td>
<td>15, 17</td>
<td>17, 17</td>
<td>16, 18</td>
<td>16, 20</td>
<td>16, 16</td>
</tr>
</tbody>
</table>

This is a trick. In the column above labeled “Child”, circle the allele that was contributed by the biological father.

24. Why analyze the Y chromosome?

25. What are YFiler, Y-PLEX 12, and the MegaPlex used for?

26. List 5 “other” sources from which DNA could be extracted, PCR amplified, and used for forensic purposes:
   1) ________________, 2) ________________, 3) ________________, 4) ________________, 5) ________________.

27. List 6 problems/limitations with using DNA for forensic purposes:
   1) ________________, 2) ________________, 3) ________________, 4) ________________, 5) ________________, 6) ________________.
Matching. Match the images with the technical problem below.

28. Microvariant
29. Bad capillary
30. Dye blobs
31. Allelic dropout
32. Stutter Products

A.  
B.  
C.  
D.  
E.  

33. Based on a review of the image below, what is your conclusion? Why?
34. Based on a review of the image below, what is your conclusion? Why?

![Image of DNA profiling bands for D6S818, D13S317, and D7S820 loci.

**35.** True or False. IF FALSE, CORRECT THE STATEMENT!
- **D6S818**
  - True or False: The FBI launched CODIS in 1989.

- **D13S317**
  - True or False: CODIS can be used for linking serial crimes and unsolved cases to repeat offenders.
  - Reason: CODIS can be used for linking serial crimes and unsolved cases to repeat offenders.

- **D7S820**
  - True or False: STRs targeted by forensic scientists are mostly pentanucleotide repeats.
  - Reason: STRs targeted by forensic scientists are mostly pentanucleotide repeats.

- **36.** True or False: If both alleles for the STR D7S820 contain 8 repeats, then that individual is heterozygous for D7S820.
  - Reason: If both alleles for the STR D7S820 contain 8 repeats, then that individual is heterozygous for D7S820.

- **37.** True or False: Commercial STR kits are commonly used to genotype biological evidence.
  - Reason: Commercial STR kits are commonly used to genotype biological evidence.

- **38.** True or False: Forward STR primers are labeled with fluorochromes (dyes) that when excited emit light.
  - Reason: Forward STR primers are labeled with fluorochromes (dyes) that when excited emit light.

- **39.** True or False: Microsatellite mutations are due to polymerase slippage and unequal crossing-over during meiosis.
  - Reason: Microsatellite mutations are due to polymerase slippage and unequal crossing-over during meiosis.

- **40.** True or False: Dye blobs are wider and usually of less intensity than true STR alleles.
  - Reason: Dye blobs are wider and usually of less intensity than true STR alleles.

- **41.** True or False: Human error is a possibility in all DNA analyses.
  - Reason: Human error is a possibility in all DNA analyses.

- **42.** True or False: Some Taq polymerases will add a guanine (“G”) to the end of the PCR product.
  - Reason: Some Taq polymerases will add a guanine (“G”) to the end of the PCR product.

- **43.** True or False: Null alleles are present in the DNA sample but fail to amplify.
  - Reason: Null alleles are present in the DNA sample but fail to amplify.

- **44.** True or False: Allele frequencies at a locus can be determined by sampling individuals from that population.
  - Reason: Allele frequencies at a locus can be determined by sampling individuals from that population.

- **45.** True or False: The PI is the probability that 2 individuals selected at random will have different genotypes.
  - Reason: The PI is the probability that 2 individuals selected at random will have different genotypes.

- **46.** True or False: The P0 is the power to differentiate two individuals at a given locus.
  - Reason: The P0 is the power to differentiate two individuals at a given locus.

- **47.** True or False: The existence of matching markers in a paternity case indicates a probability of paternity of 99.99%.
  - Reason: The existence of matching markers in a paternity case indicates a probability of paternity of 99.99%.

- **48.** True or False: Currently, about 11 million STR profiles are deposited in the National DNA Index System database.
  - Reason: Currently, about 11 million STR profiles are deposited in the National DNA Index System database.

- **49.** True or False: PCR-STR DNA evidence has been rendered inadmissible due to the use of different technology.
  - Reason: PCR-STR DNA evidence has been rendered inadmissible due to the use of different technology.

- **50.** True or False: The Combined Probability of Identity (CP) is calculated using the product rule.
  - Reason: The Combined Probability of Identity (CP) is calculated using the product rule.

- **51.** True or False: The PE is the probability, averaged over all possible mother-child pairs, that a random alleged father will be excluded from paternity.
  - Reason: The PE is the probability, averaged over all possible mother-child pairs, that a random alleged father will be excluded from paternity.

**Multiple choice and other miscellaneous practice questions continued...**

55. According to the **Frye Standard**,
   - a. data since 1923 from the polygraph machine is admissible in a court of law
   - b. scientific evidence is admissible if the thing from which the deduction is made is sufficiently established to have gained general acceptance by the scientific community
   - c. judges are permitted to allow expert testimony if they should see fit
   - d. judges can exclude relevant evidence if they determine its value to be deemed potentially confusing
   - e. the court is allowed to determine the qualifications of a person to be a witness

56. According to the **Daubert Standard**, which of the following is not a guideline that can be used by the trial courts to determine admissibility of scientific evidence?
   - a. whether the scientific technique can be tested
   - b. if an expert from the F.B.I. vouches for the scientific technique
   - c. if there is widespread acceptance in the scientific community of the technique
   - d. if the rate of error is known
   - e. if the scientific technique has been peer reviewed or published

57. SWGDAM is an acronym for ________________________________________________________________.

58. TWGDAM is an acronym for ________________________________________________________________.
Given the following for the D3S1358 locus:

<table>
<thead>
<tr>
<th>Allele</th>
<th>Population Frequencies</th>
<th>Allele Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>12</td>
<td>8</td>
<td>8/714 = 0.011</td>
</tr>
<tr>
<td>13</td>
<td>4</td>
<td>4/714 = 0.006</td>
</tr>
<tr>
<td>14</td>
<td>86</td>
<td>86/ =</td>
</tr>
<tr>
<td>15</td>
<td>220</td>
<td>204/714 = 0.286</td>
</tr>
<tr>
<td>16</td>
<td>204</td>
<td>138/714 = 0.193</td>
</tr>
<tr>
<td>17</td>
<td>138</td>
<td></td>
</tr>
<tr>
<td>18</td>
<td>48</td>
<td></td>
</tr>
<tr>
<td>19</td>
<td>6</td>
<td></td>
</tr>
</tbody>
</table>

Do your allele frequencies make sense? \( p + q + r + s + t + u + v + w = 1 \)
\( \text{_____ + _____ + _____ + _____ + _____ + _____ + _____ + _____ = 1} \)

61. What percentage of your population has allele 17? Hint- move the decimal point. Show you work.

62. What is the probability that an individual in the above population will carry allele 12 for D3S1358?

\[
\text{Probability} = \frac{1}{0.011} = 90.9
\]

Answer = 1 in 90.9

63. What is the probability that an individual in the above population will have the 12, 16 genotype?

\[
\text{Probability} = 2pq
\]
\[
\text{Probability} = (2) (0.011) (0.286) = 0.00629
\]
\[
\text{Probability} = \frac{1}{0.00629} = 159
\]

Answer = 1 in 159

64. What is the probability that an individual in the above population will have a 14, 14 genotype? Show you work.

\[
\text{Probability} = p^2 \text{ or you can use the official formula which is: } p^2 + (p \times (1-p) \times \theta), \text{ where } \theta \text{ is equal to 0.01 for large populations.}
\]
Given the following frequencies for 6 loci in an African-American population, use the product rule to determine the Combined Probability of Identity (CPI) for these genotypes:

<table>
<thead>
<tr>
<th>Locus</th>
<th>Allele 1</th>
<th>Allele 2</th>
</tr>
</thead>
<tbody>
<tr>
<td>D7S820</td>
<td>8, 8</td>
<td></td>
</tr>
<tr>
<td>D16S539</td>
<td>8, 9</td>
<td></td>
</tr>
<tr>
<td>D8S1179</td>
<td>14, 15</td>
<td></td>
</tr>
<tr>
<td>CSF1PO</td>
<td>10, 10</td>
<td></td>
</tr>
<tr>
<td>D13S317</td>
<td>10, 12</td>
<td></td>
</tr>
<tr>
<td>D5S818</td>
<td>9, 9</td>
<td></td>
</tr>
</tbody>
</table>

Combined PI= \( (P_1)(P_2)(P_3)(P_4)(P_5)(P_6) \)

\( P_i \) for Heterozygotes= \( 2pq \)

Homozygotes= \( p^2 \) or \( p^2 + (p \times (1-p) \times \theta) \)

where \( \theta \) is equal to 0.01 for large populations.

<table>
<thead>
<tr>
<th>Allele</th>
<th>%</th>
<th>Allele</th>
<th>%</th>
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<tbody>
<tr>
<td>D7S820</td>
<td>6</td>
<td>5</td>
<td>0</td>
</tr>
<tr>
<td></td>
<td>7</td>
<td>8</td>
<td>0.0042</td>
</tr>
<tr>
<td></td>
<td>8</td>
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<td>0.1877</td>
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<tr>
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<td>15</td>
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</table>

<table>
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<tbody>
<tr>
<td>D8S1179</td>
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<td>8</td>
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<td>14</td>
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<td></td>
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<table>
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<tbody>
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<td>D13S317</td>
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<td>7</td>
<td>0.0308</td>
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<table>
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<tr>
<td>D5S818</td>
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<tr>
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<td>14</td>
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</table>

Convert the CPI into a statistic (1 in ________________).