# Forensic Molecular Biology and Population Genetics
# CHE 348
**(4 credits)**

# Cedar Crest College
# Department of Chemical and Physical Sciences
# Spring, 2008

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## Course Syllabus

## Forensic Molecular Biology and Population Genetics
## Introduction

The extraordinary growth of the application of forensic science in the criminal justice system in recent years is primarily due to advances in the area of biological testing. Since Sir Alex Jeffreys introduced DNA fingerprinting in the mid 1980s, many advances in the area of forensic biological testing have been developed that now enable forensic scientists to uniquely link forensic biological samples to a particular individual. The development of CODIS (combined DNA index system) gave crime laboratories the potential to link an unknown DNA profile with convicted suspects or crime scene samples from other cases. This has not only helped in the solving of many crimes but has helped develop links between cases with the same perpetrator (e.g. serial rapists). Prior to DNA testing, forensic biological testing was accomplished by the identification of polymorphs of protein and enzymes and blood group antigens. These methods were useful in differentiating between individuals, but had only limited individualization potential.

In such a rapidly changing field, case laboratories have been forced to change the physical structure of their facility as well as change case management philosophy. Through this period of change, however, the goals have remained the same as before the forensic molecular biology revolution. Although the methods for individualization have changed, the steps prior to individualization, namely the examination of physical evidence and the identification of particular physiological fluids have not.

Forensic biology will always be part of the area of forensic science known as criminalistics. Criminalistics involves the recognition, identification, and individualization of physical evidence from criminal investigations. The attempt at individualization (in other words, trying to determine an unique or particular source of an item of physical evidence) is what separates criminalistics from all other scientific endeavors. Through individualization, criminalistics attempts to link victims with suspects and people with crime scenes that subsequently can lead to the reconstruction of crimes.

The purpose of this course is to acquaint the student with the history of forensic biological testing but will concentrate on current methods. Emphasis will be placed on PCR technology and STR fragment analysis. The meaning of DNA profile matches utilizing population statistics based on population genetics will also be emphasized.

The laboratory aspect of this course will consist mainly of work on unknowns designed to simulate physical evidence problems and to stimulate thinking about them. The lecture part of the course will provide much of the theoretical knowledge required to complete the exercises.

Good laboratory procedure should be practiced as an integral part of each and every experiment. Care should be exercised to avoid contamination problems. For example, gloves should be worn when handling biological materials and pipette tips must be repeatedly changed when handling multiple sample tubes. Prior to each exercise, the instructor will discuss preventative contamination measures.

**Course Objectives:**

1. To familiarize the student with the history and current state of forensic biological testing and the role of a forensic biologist in a forensic investigation.

2. To develop competency in the use of equipment and techniques typically employed in a forensic biology laboratory.

3. To discuss the different types of biological evidence encountered in a forensic investigation and the analyses of each.

4. To introduce the student to the proper documentation and handling of physical evidence containing biological evidence.

5. To develop good documentation and note-taking skills.

6. To develop competency in the utilization of population statistics in DNA testing.

7. To introduce the student to forensic science literature and the role that research has played in the development of forensic biological techniques.

**Course Outcomes:**

1. The student will understand the history and current state of forensic biological testing and the role of a forensic biologist in a forensic investigation. The student will also understand the role that the scientific method plays in a forensic biological investigation.

2. The student will develop competency in the use of equipment and techniques typically employed in a forensic biology laboratory

3. The student will know the different types of biological evidence encountered in a forensic investigation and the analyses of each.

4. The student will demonstrate good documentation skills in the description of physical evidence and their analysis.

5. The student will learn the proper methods for the handling of biological evidence.

6. The student will develop competency in the application and understanding of population statistics in biological testing.

7. The student will become familiar with the various peer-reviewed journals in forensic science and various important journal articles dealing with forensic biological testing.

**Course Assessment**

Student progress in laboratory exercises will be assessed after each exercise by reviewing the analytical data and conclusions for each exercise and by reviewing the laboratory notebook to ensure that documentation guidelines are followed.

Progress in lecture will be monitored through 4 written in-class examinations (3 of which will be given during the semester and 1 during the final examination period).

Each student will orally present a journal article (from a designated list) in the area of forensic biological testing (see page 7-11)

**Required Text:** Butler, John M.
*Forensic DNA Typing: Biology and Technology behind STR Markers,* second edition, Elsevier Press, 2005
ISBN:0-12-147952-8

**Required Reading:** Lee, H.C.
*The Identification and Grouping of Bloodstains*
Forensic Science Handbook, Volume I, first edition
Richard Saferstein, editor.
Pearson Education (Prentice-Hall), 1982.

Jones, E.L.
*The Identification of Semen and Other Body Fluids*
Forensic Science Handbook, Volume II, second edition
Richard Saferstein, editor.
Pearson Education (Prentice-Hall), 2005

Baird, M.L
*Analysis of Forensic DNA Samples by Single Locus VNTR Probes*
Forensic DNA Technology
Mark Farley, James Harrington, editors
Lewis, 1991

**Lecture Time:** **12:00-12:50 M, W, F**

# Lecture Outline

| Topic | | Reading Assignment |
|---|---|---|
| *January 14 - 30* | | |
| I. | Introduction of Course | |
| II. | Body Fluid Stain Identification | Lee, Jones, Butler, 39-42 |
| III. | Species of Origin Determination | Lee |
| IV. | Recognition, Documentation, and Collection of Biological Evidence at The Crime Scene: The Role of the Forensic Biologist at the Crime Scene | Butler, 33-39 |
| V. | Examination of Biological Evidence in the laboratory<br>A. Homicide and Assault Evidence<br>B. Sexual Assault Evidence<br>i. Rape kits | |
| VI. | Overview of DNA | Butler, 1-31 |
| *February 1-8* | | |
| VII. | RFLP | Baird |
| VIII. | PCR Theory | Butler, 63-75, 79-81 |
| *February 11- 15* | | |
| IX. | DNA Extraction | Butler, 42-50 |
| X. | DNA Quantitation | Butler, 5-56, 75-79 |

*February 18* – **Exam #1**.

*February 20, 22* – Class Presentations (8 students)

| **Topic** | | **Reading Assignment** |
|---|---|---|
| *February 25-29, March 10* | | |
| XI. | STRs | Butler, 85-113, 115-117 |
| XII. | CODIS | Butler, 435-449 |
| XIII. | Y Chromosome DNA Testing | Butler, 113-115, 200-232 |
| XIV. | Mitochondrial DNA | Butler, 241-288 |
| *March 12-26* | | |
| XV. | DNA Separation Methods | Butler, 313-323, 345-360 |
| XVI. | DNA Detection Methods | Butler 325-344 |
| XVII. | Biology of STRs,<br>STR Genotyping Issues,<br>Forensic Issues | Butler, 373-387<br>123-174 |
| *March 28* – **Exam #2** | | |
| *March 31, April 2* – Student Presentations (8 students) | | |
| *April 4-14* | | |
| XVIII. | Basic Genetic Principles | Butler 455-471 |
| XIX. | Population Databases | Butler 473-494 |
| XX. | Profile Frequency Estimates | Butler 497-515 |
| XXI. | Statistical Approaches to Mixtures | Butler 519- 527 |
| *April 16* – Student Presentations (4 students) | | |
| *April 18-21* | | |
| XXII. | Paternity Testing | Butler 140-142<br>529-534 |

XXIII. New Technologies Butler 413-430

*April 23, 25* – Student Presentations (6 students)

*April 28* – **Exam #3**

*April 29* – Student Presentations (3 students)

There will be a **cumulative** final examination that will be held during the final exam week.

Grading
Your final grade will be determined as follows:

| | |
|---|---|
| Exam #1 | 20% |
| Exam #2 | 20% |
| Exam #3 | 20% |
| Final Examination | 30% |
| Presentation | 10% |

Letter grades will be assigned as follows:

| | |
|---|---|
| 91-100% | A |
| 89-90% | A- |
| 87-88% | B+ |
| 81-86% | B |
| 79-80% | B- |
| 77-78% | C+ |
| 71-76% | C |
| 69-70% | C- |
| 60-68 | D |
| <59% | F |

Prior to each lecture, students may be asked questions about the previous lecture. Students are expected to give intelligent and thoughtful answers to questions. Completely off-base or "I don't know" answers will result in a 1-point subtraction from the student's final grade. Good answers will result in a 1-point addition to the student's final grade.

**Presentation**

Each student will give a 12-minute presentation on an important journal article in forensic biology. In the presentation, each student will discuss the objectives of the article, the techniques and research design used, the methods used for data evaluation (e.g. statistical tests), and results and conclusions. Other papers that augment, support, or dispute the claims of the authors should also be discussed. The impact that the paper had

on the field of forensic biology should also be discussed. All students are required to read each journal article. Material on the article is fair game on exams.

**Journal Articles**

1. Juusola JJ, Ballantyne J,
*mRNA profiling for body fluid identification by multiplex quantitative RT-PCR.*
J Forensic Sci 52 (2007): 1252-1262.

2. Gross AM, Harris KA, Kaldun GL,
*The effect of luminol on presumptive tests and DNA analysis using the polymerase chain reaction.*
J Forensic Sci 44 (1999): 837-840.

3. Karlsson AO, Holmlund G,
*Identification of mammal species using species-specific DNA pyrosequencing*.
Foren Sci Int 173 (2007); *16-20.*

4. Strasser S, Zink A, et al.,
*Age determination of blood spots in forensic medicine by force spectroscopy.*
Foren Sci Int 170 (2007); 8-14.

5. Tobe SS, Watson N, Nic Daeid NN,
*Evaluation of six presumptive tests for blood, their specificity, sensitivity, and effect on high molecular-weight DNA*.
J Forensic Sci 52 (2007); 102-109.

6. Jeffreys AJ, Wilson V, Thein SL,
*Individual specific "fingerprints" of human DNA.*
Nature, 316 (1985): 76-79.

7. Elliott K, et al.,
*Use of laser microdissection greatly improves the recovery of DNA from sperm on microscope slides.*
Foren Sci. Int., 137 (2003); 28-36.

8. Nicklas JA, Buel E,
*Development of an Alu-based, real-time PCR method for quantitation of human DNA in forensic samples.*
J Forensic Sci 48 (2003); 936-944.

9. Saiki RK, et al, *Analysis of enzymatically amplified β-globin and HLA-DQα with allele specific oligonucleotide probes.*
Nature 324 (1986); 163-166.

10. Kimpton C, et al,
*Evaluation of an automated DNA profiling system employing multiplex amplification of four tetrameric STR loci.*
Int J Legal Med 106 (1994); 302-311.

11. Ruitberg CM, et al,
*STRBase: a short tandem repeat DNA database for the human identity testing community.*
Nucleic Acids Research 29 (2001); 320-322.

12. Divine A-M, et al,
*Forensic casework analysis using the HVI/HVII mtDNA linear array assay.*
J Forensic Sci 50 (2005); 548-554.

13. Calloway C, et al,
*The Frequency of heteroplasmy in the HVII region of mtDNA differs across tissue types and increases with age.*
Am. J Hum Genet 66 (2000); 1384-1397.

14. Buel E, et al,
*Capillary electrophoresis STR analysis: comparison to gel based systems.*
J Forensic Sci 43 (1998); 164-170.

15. Cadenas AM, Regueiro T, et al,
*Male amelogenin dropouts: phylogenetic context, origins and implications*.
Foren Sci Int 166 (2007); 155-163.

16. Lukka M, Tasa G,
*Triallelic patterns in STR loci used for paternity analysis: Evidence for a duplication in chromosome 2 containing the TPOX STR locus.*
Foren Sci Int 164 (2006); 3-9.

17. Smith JAL, Budowle B,
*Source Identification of Body Fluid Stains Using DNA Profiling.*
FBI Newsletter (1998); 89-90.

18. Butler JM, et al.,
*The development of reduced size STR amplicons as tools for analysis of degraded DNA.*
J Forensic Sci 48 (2003); 1054-1064.

19. Moeller A, et al,
*Different types of structural variation in STRs: HumFES/FPS, HumVWA and HumD21S11.*
Int J Leg Med 106 (1994): 319-323.

20. Grubwieser P, Thaler A, et al,
*Systematic study of STR profiling on blood and saliva traces after visualization of fingerprint marks.*
J Forensic Sci 48 (2003); 733-741.

21. Phipps M, Petricevic S,
*The tendency of individuals to transfer DNA to handled items.*
Foren Sci Int 168 (2007); 162-168.

22. Smith PJ, Ballantyne J,
*Simplified low-copy-number DNA analysis by post-PCR purification.*
J Forensic Sci 52 (2007); 820-829.

23. Ballantyne KN, Roland AH, et al,
*Comparison of two whole genome amplification methods for STR genotyping of LCN and degraded DNA samples.*
Foren Sci Int 166 (2007); 35-41.

24. Craft KJ, Owens JD, Ashley MV,
*Application of plant DNA markers in forensic botany: Genetic comparison of Quercus evidence leaves to crime scene trees using microsatellites.*
Foren Sci Int 165 (2007); 64-70

25. Butler JM, et al,
*Reliable genotyping of short tandem repeat loci without an allelic ladder using time-of-flight mass spectrometry.*
Int J Legal Med 112 (1998); 45-49.

*26.* Kidd KK, Pakstis AJ, et al,
*Developing a SNP panel for forensic identification of individuals.*
Forensic Sci Int 164 (2006); 20-32.

27. Mitnik L, Carey L, et al,
*High-speed analysis of multiplexed short tandem repeats with an electrophoretic microdevice.*
Electrophoresis 23 (2002); 719-726.

28. Kuste CR, Barnes SB, et al,
*Environmental survey for four pathogenic bacteria and closely related species using phylogenetic and functional genes.*
J Forensic Sci 51 (2006); 548-558.

29. Hellman AP, Rohleder U, et al,
*A proposal for standardization in forensic canine DNA typing: allele nomenclature of six canine-specific STR loci.*
J Forensic Sci 51 (2006); 274-281.

**Community Standards for Academic Conduct**

Academic integrity and ethics remain steadfast, withstanding technological change. Cedar Crest College academic standards therefore apply to all academic work, including, but not limited to, handwritten or computer-generated documents, video or audio recordings, and telecommunications.

As a student at Cedar Crest College, each student shall:
• Only submit work which is his/her own.
• Adhere to the rules of acknowledging outside sources, as defined by the instructor, never plagiarizing or misrepresenting intellectual property.
• Neither seek nor receive aid from another student, converse with one another when inappropriate, nor use materials not authorized by the instructor.
• Follow the instructions of the professor in any academic situation or environment, including taking of examinations, laboratory procedures, the preparation of papers, properly and respectfully using College facilities and resources, including library and computing resources to ensure that these resources may be effectively shared by all members of the College community.
• Abide by the Cedar Crest Computer Use Policy.
• If a student perceives a violation of the Academic Standards, he/she will go to their instructor.
• If you are unable to resolve the problem with the instructor, you should go to the chair of the department. If you need further assistance after consultation with the instructor and the chair, you should see the Provost.

**Classroom Protocol**

Appropriate classroom behavior is defined and guided by complete protection for the rights of all students and faculty to a courteous, respectful classroom environment. That environment is free from distractions such as late arrivals (students will be deducted one point from each late arrival after the second time), early departures, inappropriate conversations and any other behaviors that might disrupt instruction and/or compromise students' access to the Cedar Crest College education.

Attendance in lecture is mandatory. It is understood that students may need to miss class or laboratory due to illness or personal obligations. Students needing to be absent from class must contact the instructor prior to class or laboratory. Students with valid reasons will not be penalized. In all cases, students will be responsible for all material covered in the missed class. Make-up exams will be given only in the event of illness or compelling personal matter. If a make-up exam is not granted, a zero will be given. If the instructor

is not notified prior to the exam, documentation explaining the reason for the absence may be required.

Students will not be allowed **any** unexcused absences. For each unexcused absence, students will be deducted 1% from their final grade.

**Honor Philosophy**

The Cedar Crest College Honor Philosophy states that students should uphold community standards for academic and social behavior in order to preserve a learning environment dedicated to personal and academic excellence. Upholding community standards is a matter of personal integrity and honor. Individuals who accept the honor or membership in the Cedar Crest College community of scholars pledge to accept responsibility for their actions in all academic and social situations and for the effect their actions may have on other members of the College community.