

Oakland University
Biomedical Diagnostic and Therapeutic Sciences

MLS 4020: Molecular Diagnostics
(Previously MLS 402)
Winter 2018

Professor:	Rebekah Martin, Ph.D., MLS(ASCP) ^{CM}	Lecture Day/Time:	MW 8:00-8:55am
Office:	3163 HHB	Lecture Location:	HHB 5036
Phone:	248-364-8674		
Email:	rmartin2@oakland.edu		
Office Hours:	Tu/Th, 1:00-2:00pm, or by appointment	Lab (10509) Day/Time:	M 9:00-11:55am; HHB 5023
		Lab (14756) Day/Time:	M 12:00-2:55pm; HHB 5023

Course Description: Discussion of diagnosis of disease on a molecular level including current molecular diagnostic techniques and procedures, and correlation with clinical conditions.

Required Materials:

- **Text:** Molecular Diagnostics: Fundamentals, Methods and Clinical Applications, Lela Buckingham, 2nd ed, ISBN: 0-8036-2677-0
- **Gloves:** Nitrile gloves (no latex, please)

Course Objectives: The student will achieve upon completion of this course the ability to:

1. Describe and explain the molecular techniques related to:
 - a. Separation and Detection
 - b. Nucleic Acid Amplification
 - c. DNA Sequence Analysis
 - d. Other techniques (RFLP, array technology, mass spectrophotometry, etc.)
 2. Explain medical laboratory operations related to:
 - a. Contamination (biological, amplified, and non-amplified nucleic acid)
 - b. Quality assurance
 - c. Guidelines and regulations
 - d. Safety
 3. Explain and apply current uses of medical molecular testing
 - a. Infectious Disease
 - b. Oncology
 - c. Genetics
 - d. Other (histocompatibility, parentage, pharmacogenomics, etc.)
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Class Policies:

1. **Academic conduct policy:** All members of the academic community at Oakland University are expected to practice and uphold standards of academic integrity and honesty. Academic integrity means representing oneself and one's work honestly. Misrepresentation is cheating since it means students are claiming credit for ideas or work not actually theirs and are thereby seeking a grade that is not actually earned. For further information on academic dishonesty at Oakland University, visit <https://www.oakland.edu/pace/policies-procedures>. Following are some examples of academic dishonesty:
 - a. **Cheating on examinations.** This includes using materials such as books and/or notes when not authorized by the instructor, copying from someone else's paper, helping someone else copy work, substituting another's work as one's own, theft of exam copies, or other forms of misconduct on exams.
 - b. **Plagiarism** is using someone else's work or ideas without giving that person credit; by doing this, students are, in effect, claiming credit for someone else's thinking. Whether students have read or heard the information used, they must document the source of information. When dealing with written sources, a clear distinction should be made between quotations (which reproduce information from the source word-for-word within quotation marks) and paraphrases (which digest the source of information and produce it in the student's own words). Both direct quotations and paraphrases must be documented. Even if students rephrase, condense or select from another

person's work, the ideas are still the other person's, and failure to give credit constitutes misrepresentation of the student's actual work and plagiarism of another's ideas. Buying a paper or using information from the Internet without attribution and handing it in as one's own work is plagiarism.

- c. **Unauthorized collaboration** on computer assignments and unauthorized access to and use of computer programs, including modifying computer files created by others and representing that work as one's own.
2. **You are expected to prepare for and take exams on the date and time scheduled.** Additional time for exams will not be allowed if you are tardy. You will receive a zero for missed exams due to unexplained absences.
 - a. You will have **55 minutes** to complete your exams.
 - b. If you arrive in class on exam day after the first person has left the room, you may not be allowed to take the exam.
3. Assignments must be turned in at the beginning of class to receive credit. **Assignments turned at any other time during class will not be scored.**
4. **Attendance:** In accordance with professional behavior, it is expected that you will attend course lectures/labs and be punctual. If you are unable to come to a class, professional courtesy asks that you send an email to me explaining your absence, however, attendance for this class is not mandatory. **Attendance will be part of your grade for the laboratory segment of this course.**
5. **Excused Absence Policy:** This policy for university excused absences applies to participation as an athlete, manager or student trainer in NCAA intercollegiate competitions, or participation as a representative of Oakland University at academic events and artistic performances approved by the Provost or designee. Students shall inform their instructors of dates they will miss class due to an excused absence prior to the date of that anticipated absence. For activities such as athletic competitions who schedules are known prior to the start of a term, students must provide their instructors during the first week of each term a written schedule showing days they expect to miss classes. For other university-excused absences, students must provide each instructor at the earliest possible time the dates that they will miss.
 - a. Make-up work
 - i. It is the responsibility of the student to request from the instructor an opportunity to complete missed assignments, activities, labs, examinations or other course requirements in a timely manner.
 - ii. Students are responsible for all material covered in classes that they miss, even when their absences are excused, as defined above.
 - iii. Missed classroom activities will be rescheduled at the discretion of the instructor.
6. **Add/Drops:** The university policy will be explicitly followed. It is the student's responsibility to be aware of deadline dates for dropping courses.
7. **Special Considerations:** Students with disabilities who may require special considerations should make an appointment with campus Disability Support Services, 106 North Foundation Hall, phone 248 370-3266. Students should also bring their needs to the attention of the instructor as soon as possible. For this class, you are required to bring me a Letter of Accommodation from Disability Support Services if you need any special considerations. For academic help, such as study and reading skills, contact the Academic Skills/Tutoring Center, 103 North Foundation Hall, phone 248 370-4215.
8. **Religious Observance Policy:** Oakland University policy to be developed. If you have a conflict with class due to a Religious Observance, please alert me ahead of time to make up assignments or exams. You will receive a zero for missed assignments or exams due to unexplained absences.
9. **Student Sexual Misconduct Policy: Sexual Misconduct** by a student can occur in any University sponsored program, both on- campus and off-campus or at an off campus location if the effects of the misconduct adversely affects or creates a hostile environment on campus, endangers or threatens the health or safety of any person, and/or is detrimental to the University's interests and/or educational mission. Sexual misconduct is unwelcome conduct of a sexual nature without consent and includes *sexual harassment, sexually hostile environments* and *sexual violence*. Review sexual misconduct policies, reporting procedures, and resources at <https://www.oakland.edu/policies/health-and-safety/625/>

10. **Diversity:** At Oakland University, actions defines our unwavering commitment to diversity. We accept and support our differences and commonalities, whether in race, sex, gender identity, gender expression, sexual orientation, age, height, weight, disability, color, religion, creed, national origin or ancestry, marital status familial status or veteran status, class, geography, language, socioeconomic status, or other aspects of the human condition. Review information on diversity, equity and inclusion at <https://www.oakland.edu/diversity>
11. **Emergency Preparedness:** In the event of an emergency arising on campus, the instructor will notify you of actions that may be required to ensure your safety. It is the responsibility of each student to understand the evacuation and “lockdown” guidelines to follow when an emergency is declared. These simple steps are a good place to start:
 - a. OU uses an emergency notification system through text, email, and landline. These notifications include campus closures, evacuation, lockdowns and other emergencies. Register for these notifications at oupolice.com/em/alerts.
 - b. Ensure that one cellphone is on in order to receive and share emergency notifications with the instructor in class.
 - c. If an emergency arises on campus, call the OUPD at 248-370-3331. Save this number in your phone, and put it in an easy-to-find spot in your contacts.
12. **Email Communication:** During the business week (Mon-Fri 8-5pm) I will typically respond to emails within 24 hours. I may not respond to emails right away during the evening or over the weekend. Before you send me an email, make sure the information you require is not available elsewhere (OU website, Moodle, syllabus etc.) and make sure you include a greeting, your name, class section, brief message, and a salutation (this is good practice and a good habit to get into; once we have established contact we can be less formal).
13. **Electronic Devices Policy:** You may use cell phones, laptops, and other electronic devices for learning purposes or urgent matters **in lecture**. Please keep phones on silent or vibrate. If your use of these devices becomes disruptive to me or your colleagues, I reserve the right to dismiss you from the classroom and/or give you a zero for that week’s laboratory attendance. See below for laboratory electronics policy.
14. **Laboratory Safety:** The laboratory component of this course requires you to work with specimens of human origin. Following Standard Precautions is mandatory and includes the wearing of a lab coat, shoes (not sandals), and gloves. You must wash your hands before leaving the lab. Sharps must be placed in a sharps container and waste disposed of properly. Workstation must be cleaned and properly decontaminated with a 5% bleach solution made daily before leaving lab.

NO CELL PHONES ARE ALLOWED AT WORKSTATIONS DURING THE LABORATORY EXERCISES. You must take off your gloves and wash your hands before you use a phone.

Course Grading:

4 exams	50 pts each	200 points
Case Studies	25 pts each	50 points
Group Presentation	100 pts	100 points
Lab Homework/Reports		
/Attendance	15 pts per lab	150 points
Cumulative Final Exam	125 pts	125 points

TOTAL		625 points
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Group Presentation: You will prepare a presentation on a genetic disease that is NOT covered in class. Details are attached.

Lab Homework/Reports: Homework assignments are due at the beginning of lecture/lab exercises. Assignments turned in late will be given a zero. Lab reports (when applicable) must be turned in before leaving the lab. Reports turned in late will be given a zero. If more than 3 labs are missed, you will need to drop the course and repeat it for credit.

Approximate Grading Scale: Please note that this grading scale is approximate.

Grade	Grade Point	%	Grade	Grade Point	%	Grade	Grade Point	%	Grade	Grade Point	%
A	4.0	98-100	B	3.0	80	C	2.0	70	D	1.0	60
A	3.9	97	C	2.9	79	D	1.9	69	No Credit	0.0-0.9	0-59
A	3.8	95-96	C	2.8	78	D	1.8	68			
A	3.7	93-94	C	2.7	77	D	1.7	67			
A	3.6	90-92	C	2.6	76	D	1.6	66			
B	3.5	89	C	2.5	75	D	1.5	65			
B	3.4	87-88	C	2.4	74	D	1.4	64			
B	3.3	85-86	C	2.3	73	D	1.3	63			
B	3.2	83-84	C	2.2	72	D	1.2	62			
B	3.1	81-82	C	2.1	71	D	1.1	61			

Course Schedule: Dates are subject to change

Week of:	Day: Topic #	Chapter:	Lab:
1/1	M: NO CLASS W: PANOPTO LECTURE Topic 1. QA and QC in Molecular labs	16	No Lab
1/8	Topic 2. Nucleic Acids: Isolation, Purification	4 and 5	Lab 1: Micropipetting / Lab math
1/15	M: NO CLASS Topic 3. Nucleic Acids: Quality and Quantity	5 and 7	No Lab
1/22	Topic 3. Nucleic Acids: Quality and Quantity Topic 4. Nucleic Acid Amplification	7	Lab 2: Whole blood DNA isolation "Salting out"
1/29	M: Exam 1 (Topics 1-3) Topic 4. Nucleic Acid Amplification		Lab 3: Whole blood DNA isolation column extraction
2/5	Topic 4. Nucleic Acid Amplification Case Melanoma	6	Lab 4: DNA extraction and PV92 PCR
2/12	Topic 5. Hybridization and Arrays W: Exam 2 (topics 4)	8/9	Lab 5: Finish PV92 Lab
2/19	Winter Break		No Lab
2/26	Topic 5. Hybridization and Arrays	9/10	Lab 6: Western Blot & Radial Immunodiffusion
3/5	Topic 6. Detection of chromo and gene mutations	1 and 11	Lab 7: Finish Western Blot & Finish Radial Immunodiffusion
3/12	Topic 8. Polymorphisms W: Exam 3 (topics 5-6)		Lab 8: Forensic DNA Fingerprinting
3/19	Topic 7. DNA sequencing Case Oligodendroglioma	12 and 13	Lab 9: DNA repair & HIV detection by ELISA
3/26	Topic 10. Molecular detection of inherited diseases Topic 11. Molecular methods in oncology	14	No Lab
4/2	Topic 9. Microorganisms W: Exam 4 (Topics 8 - 10)	15	Lab 10: Familial Cancer
4/9	M: Student presentations W: Student presentations		No Lab
4/16	M: Student presentations / Case studies / Review		
4/23	Cumulative FINAL (Lecture/lab) EXAM Mon 4/23 8:00-11:00am		

Group Presentation

As a laboratory professional you will interact with peers, physicians, nurses, and even patients. Your ability to effectively communicate your thoughts and ideas to these diverse groups of people is essential to providing the utmost quality in patient care. This assignment will allow you to practice your oral communication skills, work closely and cooperatively with colleagues, and enhance your skills in educating others using visual media.

Assignment:

1. Presentation:

Using PowerPoint, your group will create a **8-10-minute presentation** on a genetic disease (see list of those that you may choose from, below). Your presentation should include the following information about the chosen disease (not necessarily in this order):

- Background (brief)
 - Allele frequency
 - Signs and symptoms
 - Traditional diagnosis
 - Prognosis
 - Treatments or therapies

 - Molecular Diagnostics
 - Chromosomal location of each known gene mutation involved
 - Name of each gene that is mutated (if there are many, include at least 3 locations)
 - Type of mutations that are associated with the disease
 - Current testing or theoretical testing for the mutations
 - Explanation of functional defects that the gene mutation contributes to the disorder.
 - What is the normal function of the gene and of the gene product?
 - What pathway is the gene product involved in?
 - If the gene product is an enzyme, how does accumulation of the substrate contribute to the pathology?
 - How does the mutated gene product lead to the pathology?

 - References (Include **at least one peer reviewed publication**, later than 2012)
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Conditions that you may choose for your presentation:

- | | | |
|---|-------------------------------|------------------------|
| • Bloom syndrome | • Gorlin syndrome | • Parks Weber syndrome |
| • CHARGE syndrome | • Hurler syndrome | • Pendred syndrome |
| • Ectrodactyly-Ectodermal
Dysplasia-Cleft Lip/Palate (EEC
syndrome) | • Hypertrophic cardiomyopathy | • Tangier disease |
| • Familial Mediterranean fever | • Lesch Nyhan syndrome | • Tay-Sachs Disease |
| • Gaucher Disease | • Lynch syndrome | • Werner syndrome |
| | • Marfan syndrome | |
| | • Mowat-Wilson syndrome | |
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An excellent resource to help you get started: <http://ghr.nlm.nih.gov/>

Other useful resources include:

- OMIM
- Gene Cards
- Gene Reviews
- 1000 Genome's Browser
- Journal of Molecular Diagnostics
- Journal of Clinical Chemistry

***Tip:** Your presentation should flow. While each student should contribute a particular set of information to the presentation, the delivery of your topic should NOT be broken into two or three obviously unique pieces. This is not an exercise in 3 separate presentations, but one, cohesive, informational activity.

2. Multiple Choice Questions

You must supply 4 multiple-choice questions with your final presentation. Each question must contain 4 choices (A-D). You must map each question to a learning outcome (below). Your presentation must include information that directly relates to these learning outcomes:

- I. Identify normal activity of the gene product (that is dysfunctional in the disease).
- II. Define the molecular defect of the disease (type of mutation).
- III. Explain how the functional defect contributes to the pathophysiology of the disease.
- IV. Propose molecular diagnostic testing for the molecular defect of this disease.

Deadlines:

You will **email** me your presentation topic by the **beginning** of class on **Wednesday, January 17** on a first come, first served basis. If you choose a topic that another group has already chosen, then you must choose a different topic. It would benefit you to choose early and to have a backup choice.

You will turn in an outline of your presentation by the **beginning** of class on **Wednesday, February 14**. It should encompass the order with which you choose to present your topic as well as the preliminary details of your research into the topic. You **must include** your **references (APA formatted)** with your outline.

You must email a final draft of your presentation along with your multiple choice questions to me by the **beginning** of class on **Monday, April 2**. You will then have a week to look over, practice, and make **MINOR** changes to your presentation.

Your groups will present on, **April 9, 11, and 16**.

Deadline Overview: Each deadline must be met by the **beginning** of class or will be considered late.

Topic Due:	Wednesday, January 17
Outline and first Peer Evaluation Due:	Wednesday, February 14
Presentation and Multiple choice questions Due:	Monday, April 2
Presentation and second Peer Evaluation Due:	April 9, 11, and 16

Grading:

You will be graded on completeness of the assignment and your ability to effectively communicate your ideas. You will also fill out evaluations on your group mate(s) throughout the semester. Your peers will assess your effort on this assignment and your grade will be adversely affected by lack of participation. The average score from each evaluation will be included in your grade for this assignment.

Peer Evaluations		
Average score of evaluations		_____/ 15
Asking questions during presentations		_____/ 5
Assignment Guidelines		
Deadlines		_____/ 10
Time-Limit		_____/ 10
Quality of Presentation		_____/ 10
Organization/Flow		_____/ 10
Preparedness		_____/ 10
Speech		_____/ 10
Quality of Sources		_____/ 10
Multiple Choice Questions		_____/ 10
TOTAL		_____/ 100

Peer Evaluation Form for Group Presentation

Write the name of each group member in a separate column. For each group member, indicate the degree to which you agree with the statements on the left, using a scale of 1-3 (1=strongly disagree; 2=neutral; 3=strongly agree). Total the numbers in each column.

YOUR NAME:

Evaluation Criteria	Group member:	Group member:
Is punctual in attending scheduled group sessions		
Contributes meaningfully to group discussions		
Prepares work in a quality manner		
Demonstrates cooperative and supportive attitude		
Contributes overall to the success of the project		
TOTAL:		

1. Indicate each group member's assignment/contribution for the project.
2. Provide specific comments about any group members.
3. Identify any problems or disputes that occurred during your interactions.
4. How could disputes have been avoided and/or how were they alleviated or resolved?
5. Did the group process have a positive effect on your learning? Please explain.

MLS 4020 Presentation Rubric

	- 10 -	9 8 7	6 5 4	3 2 1
Deadlines	Topic, outline, presentation, MC questions/answers/grades were turned in on time and required no or minor revisions. MC questions are mapped to objectives.	Topic, outline, presentation were turned in on time and required no or minor revisions. MC questions on time, but answers, and mapping to objectives late.	Presentation was turned in on time but required significant revisions. Only some MC questions. Grades late. Mapping to objectives incomplete.	Presentation was turned in late or required major revisions. No or only some MC questions. Grades late. Mapping to objectives incomplete.
Time-Limit	Presentation is 8-10 minutes long.	Presentation is too long or too short by 1-2 minutes.	Presentation is too long or too short by 3-4 minutes.	Presentation is too long or too short by 5 or more min.
Quality of Presentation	Followed guidelines for a quality appearance to the presentation on all slides	Most slides follow guidelines for a quality appearance	Some slides follow guidelines for a quality appearance	Presentation does not follow guidelines for a quality appearance
Content	Includes all criteria, explains each one completely. High degree of creativity with visuals and explanations.	Includes all criteria and explains each one. Some creativity noted.	Misses one criterion, explanation. Limited creativity demonstrated.	Misses more than one criterion, explanation. Limited/no creativity demonstrated.
Organization/Flow	Information is presented in a logical sequence	Information is presented in a logical sequence. More or less information is needed	Information is inconsistently organized (i.e. jumping around)	There is no sequence of information and/or so much is missing that the presentation makes little sense.
Preparedness	Provides expert analysis and insight regarding topic. Answers anticipated questions. Limited reliance on notes/slides. No typos.	Presenter is somewhat comfortable with topic. Some reliance on notes/slides. One typo.	Relied heavily on notes/slides. Not comfortable with all material. Two typos.	Not comfortable with the material. Only read from notes/slides. More than two typos.
Speech	Speaks clearly and distinctly all (100-95%) the time and mispronounces no words. Volume is appropriate. Defines all new words.	Speaks clearly and distinctly all (100-95%) the time, but mispronounces one word. Volume is appropriate. Defines most new words.	Speaks clearly and distinctly most (94-85%) of the time. Mispronounces more than one word. Volume is loud enough to be heard by audience at least 75% of the time.	Difficult to understand diction. Mispronounces more than one word. Does not define new words. Volume often too soft to be heard by audience most of the time.
Quality of Sources	3 professional sources (may include textbook), one peer reviewed pub., later than 2012. Formatted correctly.	2 professional sources (may include textbook), one peer reviewed pub., later than 2012. One – two ref. formatted incorrectly.	1 professional source, one peer reviewed pub., later than 2012. More than 2 ref. formatted incorrectly.	1 or more unprofessional or inappropriate source(s), no peer reviewed source. Multiple ref. with wrong formatting.
Multiple Choice Questions	All questions are clearly written, encourage critical thinking, and represent 4 objectives. All questions clearly mapped to obj.	All questions are clearly written or encourage critical thinking. Represent 3 objectives and are clearly mapped.	Some questions are unclear, poorly written, or too easy/difficult. Some questions not multiple choice. Questions cover less than 3 objectives.	Questions do not relate to topic or do not follow objectives. None are multiple choice.