

BMB527 – Medical Genetics

Fall – 2016

(updated 8/31/16)

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Notice to Students: Although course syllabi at MSUCOM have a consistent format, vitally important details differ by course. For this reason, you must read the syllabus thoroughly at the onset of each course to know what the course will provide and what is expected of you.

Section 1 – Course Information

Course Description

BMB 527 is a 1 credit hour course that provides students with knowledge of basic principles of human medical genetics, storage and expression of genetic information, and transmission of genetic information to progeny.

Course Goals

There are two main objectives in this course: (1) to instruct students in the basic concepts, principles, and language of human genetics; and (2) to equip students to analyze diseases and the responses to diseases in molecular and genetic terms. The concepts and principles will be illustrated using examples from clinical practice. We anticipate that this training will enable students to recognize and solve genetic problems encountered in subsequent clinical training and experience.

Specific instructional objectives are provided within each lecture or other learning activity of this course. These objectives represent a subset of the core competencies in genetics that are expected of all health professionals, as put forth in guidelines from the American Society for Human Genetics and the National Coalition for Health Professional Education in Genetics.

Prerequisites

This course assumes that each student has fulfilled the admission requirement of having completed a 3-semester hour course in genetics at the college level. In addition, mastery of concepts introduced in prior first-year medical school courses (e.g., BMB 515) will be expected.

Section Codes for the Course:

- 301 for COM-EL
- 302 for COM-DMC
- 303 for COM-MUC

Course Coordinator

(Note - Preferred method of contact is shown in italics)

Name:	Martha Faner
Phone:	313-578-9669
Email:	<i>fanermar@msu.edu</i>
Address:	CG-90, DMC

Course Faculty

(Note - Preferred method of contact is in italics)

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Site:	A-322F East Fee Hall, EL	402C Biochemistry, EL	502B Biochemistry, EL

Curriculum Assistants

	EL	DMC - Lead	MUC
Name:	Cheryl Luick	Katelyn Johnston	Charity Troutt
Phone:	517-884-3880	313-578-9628	586-226-4788
Email:	cheryl.luick@hc.msu.edu	katelyn.johnston@hc.msu.edu	charity.troutt@hc.msu.edu

Lines of Communication

- For administrative aspects of the Course: contact the course coordinator.
- For content questions relating to a specific lecture or topic: contact the faculty presenter for that specific portion of the course or your SE MI on-site instructor.
- For absences/missed exams (see excused absence information below)

Office Hours

Office hours for each campus are announced in site-specific folders under the Content tab of the D2L course website. Students are also encouraged: (a) to address questions to instructors via the E-mail system; (b) to seek individual consultation with the lecturer or the on-site instructor by appointment throughout the semester; and (c) to use the discussion board on D2L course website.

Course Web Site

The URL for the Course web site is <https://d2l.msu.edu/>

You should pay attention to these MAIN items at this website:

- **News** – Course-related communication to the class will be made here. You should check for announcements on a daily basis.
- **Syllabus** - Information about textbooks, exam dates, grading system, rules and regulations, etc. as well as information on the instructional team. This file is under the **Content** tab of the Course website.
- **Content** – Lecture recordings, tutorials (TT), self-study modules (SSM), and all other scientific material will be deposited here.
- **Discussion Board** – List of student “asked” questions organized by week and by lecture. The discussion board will be primarily student led. It is intended for students to use as a way to communicate with each other and teach each other. The Course faculty will monitor the discussion board periodically. It is highly recommended that you check the discussion board prior to sending a question to faculty as it is very likely that you are not the only student with a given question and it may already have been answered.

Although each visit to any section of D2L by an individual student is “tracked” by the computer and the instructors of the course will have access to such information, we do not intend to use it.

Textbooks and Reference Materials

Required	Recommended	Optional
<ul style="list-style-type: none"> ▪ BMB 527 2016 Course Pack ▪ Turnpenny and Ellard, <u>Emery's Elements of Medical Genetics</u>. 14th ed. Elsevier: Churchill 		<ul style="list-style-type: none"> ▪ Jorde, Carey, Bamshad and White, (2015) <u>Medical Genetics</u>. 5th Ed. [978-0323188357]

<p>Livingstone. [978-0702040436]</p> <ul style="list-style-type: none"> ▪ Ferrier, <u>Lippincott's Illustrated Reviews: Biochemistry</u>. 6th ed. Wolters Kluwer: Lippincott, Williams, and Wilkins. [978-1451175622] 		<ul style="list-style-type: none"> ▪ Nussbaum, McInnes, and Willard, (2016) <u>Thompson and Thompson Genetics in Medicine</u>. 8th ed. [978-1437706963]
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Other Instructional Material

The Cytogenetics and Treacher Collins Tutorials can be found as a subfolder labeled "Tutorial" on the BMB 527 course Mediasite catalog. Additional instructional materials, including assigned readings, problem sets, and computer-based instructional aids, may be provided as links at the course website or as handouts at lecture sessions. These materials are intended to be an integral aspect of the course; instruction in some course objectives may be accomplished entirely through these exercises and experiences, and thus might not be explicitly addressed during lectures. Students are expected to make use of them.

Students enrolled in BMB 527, 2016 will have the unique opportunity to hear a presentation by Mr. Sam Drazin from 12-1 PM on 9/30/16 in DMC – G029, MUC – UC3 208 and EL – 102 Conrad Hall. **This presentation is optional but we expect you to take advantage of it and attend.** Mr. Drazin has Treacher Collins syndrome, a condition that has resulted in him looking different from others and also deal with hearing loss. Please see course pack page 176 for more details on this event.

i>Clicker Use in Course

i>Clickers will be used in this class. An i>Clicker 2 is required for this class. The mobile application, i>ClickerGO, will not be allowed.

In this course, i>Clicker input may be used in the following ways: to provide practice with concepts and principles, to stimulate discussion and/or to give mini-quizzes. Questions may be posed at any time during the class hour. No make-up experiences will be provided should you forget your i>Clicker. The i>Clicker will be the only mechanism to record attendance during large group lectures.

- If the i>Clicker is used to take attendance, you will be expected to arrive in class on time and to stay for the duration of the assigned activity.
- If you bring your i>Clicker and it fails during the lecture, please see the course representative immediately after the lecture to inform us of the problem. NO points for attendance will be given unless you notify us at the time of the lecture.

Please refer to additional i>Clicker policy information provided in Section 2 of this syllabus.

Course-based Academic Support

The value you derive from this course (and those that follow it) will depend on many factors, but most importantly the amount of time and effort you put into it. In undergraduate courses, students often concentrate on just getting through the next exam or individual courses. But medical education is different because it is cumulative. Study for understanding, not just short term memorization. This will allow you to understand concepts and carry them forward with you to the next step in your medical education.

You are strongly urged to:

- Consult the course D2L web site frequently to see announcements and to access various study aids (e.g., practice exam questions, and answers to frequently asked questions).
- Complete the preparatory work assigned for each session; this includes working through online modules, problem sets or any other advance study activities.
- Attend every lecture session.
- Actively annotate your Course Pack as you prepare for each class session, as each class session progresses, and also during your follow-up study.
- Complete the follow-up (supplemental) reading and self-study exercises as directed in the Course Pack and on the D2L web site.
- Use the practice exam questions (from Exam Soft) to help guide your review and preparation for exams. Do NOT wait until the day before an exam to look at these practice exam questions. Instead, start using them several days prior to each exam to help guide your review and exam preparation.
- Each member of the teaching team has a well-deserved reputation for being approachable and for helping students achieve success. Avail yourself of the opportunities for help provided by the course faculty -- in person, via e-mail, or call them to schedule an appointment time.
- The time immediately before or after a course lecture is often too hectic to provide a good opportunity to get help from course instructors. In contrast, scheduled office appointments provide an excellent time to ask questions of course faculty.
- Keep in mind that you can contact course faculty by e-mail with your questions. Note: Whenever you pose a question by e-mail, include what you THINK the answer is. This makes it much easier for the instructor to either confirm your understanding or to see where you are going off-track.
- Face-to-face contact with faculty at lecture sessions -- In addition to the faculty person giving a lecture, one or more of the course faculty regularly sit in on course lecture sessions at each site. This provides you with an opportunity to pose a quick question or to request a personal meeting with your local campus faculty. E-mail is also a good way to set up a personal meeting with a particular faculty member.
- Study groups - Many students find it beneficial to study with one or more partners, and we strongly encourage this activity. Studying together can be efficient (what one student doesn't understand, another one will), stimulating (personal interaction can help keep you focused and alert), and motivating (commitment to a partner supplements self-discipline). We encourage you to study with suitable partners. We caution you, however, to avoid study groups that turn into "gab fests" or where one or two students do all the talking. Remember, you may THINK you understand a concept when you hear someone else explain it, but you'll KNOW you understand the concept only when YOU can explain it to someone else. So, make sure you get to talk in your study group!
- Caution: Scribe note services are not sanctioned by MSUCOM and are not endorsed by the course faculty. Course faculty assume no responsibility whatsoever for errors in the "scribe notes". It is unwise to expect the "scribes" to substitute for your own attendance in lecture or lab, your own note taking, or your own studying.
- Additional academic support resources can be accessed at:

http://com.msu.edu/Students/Academic_Career_Guidance/index.htm

In summary, the course faculty members are here to facilitate your learning. The large number of students in this course (about 300) necessitates a degree of formality. Also, since your schedules are very full, we must adhere rigidly to the lecture, small group and lab times assigned to this course. However, within these constraints, the needs of individual students will be accommodated whenever possible. Please feel free to contact the Course Coordinator with any personal issues you may have involving this course.

Course Begin and End Dates

The first scheduled day of instruction for BMB 527 is Wednesday, September 14, 2016 and ends on October 28, 2016. See addendum for detailed daily course schedule.

Exams/Assessments

The educational objectives, defined in the Course Pack for each section of this course, will serve as the basis for evaluating student performance. Mastery of these objectives will be expected whether those topics have been discussed in lecture sessions or explored using other resources (tutorial recordings, assigned reading, computer-based instructional aids). Student attainment of these objectives will be evaluated using two quizzes, two exams, one homework exercise on retrieving information from online databases, and one "flipped classroom" session.

a) Each quiz will contain 7 questions, to be completed in the first 10 minutes of the class in the lecture venue on the dates stated (see Table below). The quizzes will be administered using your personal computer-based testing device. BMB 527 will follow the same procedure as BMB 515 for conducting the quizzes, as detailed in the document "Quiz Protocol" under the Content Tab of the D2L course website.

b) Unit exams 2 and 3, respectively, will contain 24 questions and 27 questions from BMB 527 (see Table below).

c) Following a lecture during which information regarding the use of online tools is presented, a homework exercise will provide students with the opportunity to gain experience in searching for clinically relevant genetics information using these resources. After reading the clinical scenario, the student is to search and consult the online resources to answer five multiple-choice questions, submitted through the D2L course website. Detailed information regarding the homework exercise will be provided in the Course Pack. This exercise will contribute 5 points to the total Course Grade.

d) The "flipped classroom" session requires students to do preparatory work at home and the classroom session will consist of student groups presenting to their peers what they have learned. The preparatory work at home will include: (i) watch a recording of a mini-lecture (~15 minutes); (ii) read an assigned article; and (iii) take notes on key points to contribute to a 1-page summary of what was learned. There will be individual readiness and post-exercise assessments, contributing a total of 7 points to the Course Grade. We will be using participation in i>Clicker questions as the mechanism to monitor attendance throughout the session. **You will not be eligible to earn points in the Post Exercise Quiz (PEQ) unless you participate in the i>Clicker questions during the session.**

Exam/Quiz	Date	Projected Points	Material to be Covered	Percentage of Grade
Quiz #1	Wed. 9/21/16	7	Sessions 1 through 4 [1 TT]	9.1
Unit Exam 2	Mon. 9/26/16	24	Sessions 1 through 8 [1 TT]	31.2
Quiz #2	Tue. 10/3/16	7	Sessions 9 through 14 + Treacher Collins	9.1
Homework exercise	Due by 5 pm Fri. 10/7/16	5	Retrieving information from online databases	6.5

Unit Exam 3	Mon. 10/10/16	27	Comprehensive for the Course but with emphasis on Sessions 9-17 + Treacher Collins	35.0
Flipped classroom exercise	Due by 11 pm Fri. 10/28/16	7	Approaches to correction of genetic conditions	9.1

Course Grades

A total of 77 points can be derived from the two quizzes, two exams, the homework exercise, and the "flipped classroom" session (see Table on page 6).

- **P-Pass**
This means that credit is granted and that the student achieved a level of performance judged to be satisfactory by the instructor. To obtain a "P" grade for this course, a student must accumulate 54 points (70% of 77 points).
- **N-No Grade**
This means that no credit is granted and that the student did not achieve a level of performance judged to be satisfactory by the instructor. A student who accumulates less than 54 points (or an accumulated score below 70%) will receive an "N" grade.
- **Remediation**
Since all of the core courses in the MSUCOM preclerkship curriculum are required, any student receiving an "N" grade must remediate, if eligible, or repeat the course when next offered. Please refer to the remediation policy information provided in Section 2 of this syllabus for information on College requirements and eligibility determination. Consistent with COM policy, the remediation opportunity for BMB 527 will be by examination. The Remediation Exam will consist of 50 questions, comprehensive for the course. Passing is 70%.

Student Evaluation of the Course

We want your feedback on how to make this course better for yourself and also for the students who come after you.

- The faculty of BMB 527 will be monitoring the effectiveness of the instruction throughout the semester and will be responsive to constructive student feedback. Three main mechanisms are available to assess the attainment of instructional objectives: (a) Direct student contact with the instructors; (b) the use of "focus groups" and class liaisons; and (c) the use of instructor/course evaluations.
- Over the long-term, student feedback via "focus groups" and instructor/course evaluations provides the instructors with invaluable information regarding student perspectives on the performance of the faculty and the quality of the course. The information gained from these evaluations will be used to develop future offerings of genetics.
- SIRS Evaluations – You are not required to complete the course evaluations available to you via SIRS. If you would like to opt out of the ability to evaluate this course using SIRS, login to sirsonline.msu.edu, click the link to the survey, then select button at the bottom that says "Decline to Participate."
- Informal Feedback: Feel free to approach the Course Coordinator, Dr. Martha Faner, or any of

the other course faculty with your reactions and suggestions. Or write out your comments and e-mail them to the Course Coordinator or Faculty. From time to time, we may also convene focus groups of students, as an additional way to elicit your suggestions.

- **Formal Evaluation:** As part of professional behavior, it is an expectation of the College that all students provide feedback on the course and instructors. Student feedback provides Course Coordinators with valuable information regarding their performance, the performance of their colleagues, and the quality of the course. The information gained from these evaluations is used to continuously improve future offerings of this course. Students can access the evaluation system at: http://kobiljak.msu.edu/Evaluation/UnitI_II.html. **The evaluation system will open on the following schedule:**
 - **Course Evaluation – October 5th**
 - **Dr. Amalfitano Faculty Evaluation – September 28th**
 - **Instructor Iacoboni Faculty Evaluation– October 5th**
 - **Site Specific Faculty Evaluations – October 5th**
 - **All Evaluations will be available until November 11th**

Section 2 – Policies

Academic Honesty and Professionalism

http://com.msu.edu/Students/Professional_Development/Statement_on_Professionalism.htm

Every student is responsible for their behavior and is expected to adhere to all MSU and MSUCOM policies of academic honesty and professionalism. If there is any instance of academic dishonesty or unprofessionalism discovered by a member of the faculty, administration or staff, it is his or her responsibility to take appropriate action.

Such action may include, but is not limited to: giving a failing grade, referring a student for judicial review, directing the student to the Associate Dean of Student Services, and any other actions outlined in the Medical Students' Rights and Responsibilities document.

<http://com.msu.edu/AP/Professionalism/MSRR-Offical-Document-Final.pdf>

Absences from Mandatory Class Sessions, Examinations, Assessments and Activities

MSUCOM students are expected to attend all mandatory class sessions (e.g., lectures, laboratories, group activities) and take all examinations/assessments during their originally scheduled times. If this is not possible an excused absence may be requested.

Excused absences will not be given to all who make a request. If a request for an excused absence is denied, the student will receive a zero (0) grade for the mandatory session or examination/assessment in question, which may result in the issuance of an "N" grade in the course.

An excused absence does not relieve the student from responsibilities for missed mandatory class sessions and examinations. It is highly possible that an excused absence will not be granted for a mandatory class session due to scheduling of equipment and faculty. If one of these mandatory sessions is missed, even though an excused absence is granted, and cannot be made up, the student will receive a zero(0) grade for the event and may be required to repeat the course at its next offering.

Personal Emergencies:

To obtain an excused absence, complete the Excused Absence Request form found on the student portal.

A personal emergency is typically defined as the death of an immediate family member, serious illness, automobile accident and/or hospitalization. Situations including, but not limited to: failure to be on time, conflicting appointments and failure to provide proper identification will not be considered a personal emergency, and requests based upon these situations may be denied.

If an examination/assessment or other mandatory experience is missed due to medical reasons, a medical provider's written confirmation may be required before the request is considered.

Advance Notice of Absence Available:

A student must submit his or her excused absence via the Excused Absence Form at least one week in advance of any scheduled mandatory class session or examination/assessment. Requests for excused absences regarding weddings, family celebrations and vacations will be denied.

Conferences, Conventions, Meetings, College Sponsored Activities:

If a student wishes to attend a conference, convention, meeting, or college sponsored activity which will cause him or her to miss a mandatory class [session(s) and/or examination(s)], he or she must complete and submit an [Excused Absence Request Form](#) at least two weeks prior to the expected absence period and provide a copy of the conference, convention, meeting or college sponsored activity announcement. Examination dates and mandatory class sessions will not routinely be changed for these activities.

Extended Absences:

MSUCOM will not excuse students for extended absences involving elective travel and medical mission work in another country. Approval of extended absences for unavoidable situations will be considered by Academic Programs osteomedap@hc.msu.edu on a case-by-case basis.

Computer-Based Testing (CBT)

http://www.com.msu.edu/AP/preclerkship_program/preclerkship_curriculum/preclerkship_curriculum%20.htm

It is the responsibility of each and every student to know and be in compliance with the MSUCOM policy regarding computer-based testing. It is possible that adjustments may need to be made to this policy, and students will be notified of those adjustments when necessary.

In addition, each and every student must possess his or her own electronic device that is compatible with the software program SoftTest, and ensure that it is fully functional and operational at the time of every computerized assessment.

If a student has difficulties with respect to their technology prior to an assessment, he or she can send

an email to OsteoMedAP@hc.msu.edu (which is monitored during normal business hours) for a response within 24 hours of viewing or call the lead curriculum assistant for more urgent matters.

i>Clicker Policy

http://www.com.msu.edu/Students/Policies_and_Programs/iCLICKER_Policy.htm

You are expected to have your i>Clicker registered prior to the beginning of this class. You are responsible for bringing your i>Clicker to every class with you. Class will proceed as planned, even if you have forgotten to bring your i>Clicker with you. Paper completion of i>Clicker activities will not be accepted as a substitute for the i>Clicker response. Please make sure that your i>Clicker is always in working order.

As a matter of professionalism, please note that under no circumstances should you loan your i>Clicker to another student. Nor should you ever be in the possession of an i>Clicker other than your own.

Answering questions or checking in for attendance on behalf of another student by using his or her i>Clicker is considered to be an act of dishonesty and may result in dismissal from the college.

Remediation Policy

Remediation of an "N" grade will be governed by the MSUCOM Policy for Retention, Promotion and Graduation (relevant content found under Remediation section),

<http://www.com.msu.edu/Students/Registrar/MSUCOM%20Remediation%20of%20N%20Grade%20Algorithm.pdf>) and by the remediation section of each course syllabus.

It is the responsibility of each student in the Michigan State University College of Osteopathic Medicine to verify his/her eligibility, with the Office of Student Services, prior to the administration of the remediation examination/experience. Also, it is the student's responsibility to ask the course coordinator about the format and expectations of the remediation experience.

Students deemed eligible for remediation by the registrar will be informed by the registrar's office.

Semester 2 Course Remediation Exams

Individual course written Comprehensive Remediation Exams for the following Semester 2 courses (**BMB 515, BMB 527, MMG 531, MMG 532, OST 566, PHM 564, PSL 536, PSL 537**) will be held either on Thursday, January 5, 2017 or Friday, January 6, 2017 between the hours of 8 am and 5 pm. Students will have 2 hours to complete an individual course remediation exam. Students affected will be notified of the exact date, time, and venue for their course specific exam.

Requests for Special Accommodations

Michigan State University is committed to providing equal opportunity for participation in all programs, services and activities. Requests for accommodations by persons with disabilities may be made by contacting the Resource Center for Persons with Disabilities at 517-884-RCPD or on the web at <http://www.rcpd.msu.edu/>. Once your eligibility for an accommodation has been determined, you may be issued a Verified Individualized Services and Accommodation (VISA) form. Please present the VISA to Cheryl Luick, luick@msu.edu A329 East Fee Hall at the start of the term and/or two weeks prior to the assessment event (test, project, labs, etc.). Requests received after this date will be honored whenever possible.

Please note: In the event of any unforeseen situations, the instructor(s) may make changes to any

portion of the syllabus, within reason and without notice. If such a situation arises, the instructor(s) will inform you as soon as possible of the necessary adjustments/updates. It is the student's responsibility to make note of these adjustments/updates.

Addendum: Course Calendar on the next page

Addendum: Course Schedule

BMB 527, Fall, 2016 Calendar

<u>Session</u>	<u>Day</u>	<u>Date</u>	<u>Time</u>	<u>Topic</u>	<u>Registration</u>	<u>Lecturer</u>
1	Wed.	9/14	10 am	Introduction to medical genetics	EL	Amalfitano
	Thurs.	9/15	online	Cytogenetics tutorial	online	Iacoboni
2	Mon.	9/19	11	Chromosome structural and numerical abnormalities I	EL	Iacoboni
3	Tues.	9/20	10	Common chromosomal disorders	EL	Amalfitano
4	Tues.	9/20	11	Macro- and micro-deletion disorders	EL	Amalfitano
5	Wed.	9/21	8	(quiz 1: sessions 1-4 + tutorial) Chromosome structural and numerical abnormalities II	EL	Iacoboni
6	Wed.	9/21	9	Modes of inheritance	EL	Iacoboni
7	Thurs.	9/22	10	Factors modulating inheritance patterns	DMC	Iacoboni
8	Thurs.	9/22	11	Population genetics (END of material for Unit Exam 2, Mon. 9/26)	DMC	Iacoboni
	Thurs.	9/22	online	Treacher Collins tutorial	online	Iacoboni
9	Tues.	9/27	10	NF and Marfan (full title of lecture: Common Multiple Anomaly syndromes due to Single Gene Mutations)	EL	Amalfitano
10	Tues.	9/27	11	Multifactorial (complex) disorders	EL	Amalfitano
11	Wed.	9/28	10	Genetic anticipation	MUC	Amalfitano
12	Wed.	9/28	11	Genetic imprinting	MUC	Amalfitano
13	Fri.	9/30	8	Cancer genetics	EL	Iacoboni

14	Fri.	9/30	9	Genetic testing	EL	Iacoboni
Optional	Fri.	9/30	noon	Sam Drazin Presentation (Treacher Collins) (HIT Needed)	EL	Sam Drazin
15	Mon.	10/3	11	(quiz 2: sessions 9-14 + Treacher Collins) Case Discussion (HIT needed)	Local	Local Faculty
16	Tues.	10/4	11	Prenatal screening	EL	Iacoboni
17	Wed.	10/5	8	Teratogens	EL	Iacoboni
				(Unit Exam 3, Mon. 10/10 comprehensive for the course but with emphasis on Sessions 9-17 + Treacher Collins)		
Clinical Application	Fri	10/28	1-2:50 or 3-4:50	Flipped Classroom: Current and future approaches to genetic disorders (HIT Needed)	Local	Local Faculty