

<p>Fall 2021 Course Number: GS11 1622 - Topics in Medical Genetics I</p> <p>Meeting Location (Building/Room # or WebEx/Zoom): Medical School Basement, B.612 (unless otherwise noted below)</p>	<p>Program Required Course: <input checked="" type="checkbox"/> Yes <input type="checkbox"/> No Approval Code <input checked="" type="checkbox"/> Yes <input type="checkbox"/> No (If yes, the Course Director or the Course Designee will provide the approval code.)</p> <p>Audit Permitted: <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No Classes Begin: 8/19/21 Classes End: 12/2/21 Final Exam Week: 12/9/21</p>
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Class Meeting Schedule:

Day	Time
Monday	12:30 pm – 2:00 pm*
Thursday	12:30 pm – 2:00 pm *
	*unless otherwise noted

<p>Course Director: Leslie Dunnington, MS, CGC Assistant Professor, Department of Pediatrics UT Houston Email Address: Leslie.A.Newman@uth.tmc.edu Contact Number: (713) 500-5667</p> <p>Course Co-Director/s: Kathryn Gunther, MS, CGC Clinical Instructor, Department of Pediatrics UT Houston Email Address: Kathryn.A.Gunther@uth.tmc.edu Contact Number: (713) 500-5969</p> <p>NOTE: Office hours are available on request. Please email us to arrange a time to meet.</p>	<p>Instructor/s: (Use additional page as needed)</p> <ol style="list-style-type: none"> 1. Leslie Dunnington, MS, CGC UT Houston Leslie.A.Newman@uth.tmc.edu 2. Kathryn (Katie) Gunther, MS, CGC UT Houston Kathryn.A.Gunther@uth.tmc.edu 3. Janice Smith, Ph.D. Baylor College of Medicine JS5@bcm.edu 4. James Hixson, Ph.D. UT Houston James.E.Hixson@uth.tmc.edu 5. Molly Daniels, MS, CGC MD Anderson Cancer Center MSDaniel@mdanderson.org 6. Myla Ashfaq, MS, CGC UT Houston Myla.Ashfaq@uth.tmc.edu 7. Paige Roberts, MS, RD UT Houston Paige.Roberts@uth.tmc.edu 8. Jaqueline Hecht, Ph.D., UT Houston Jacqueline.T.Hecht@uth.tmc.edu
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Textbook

Korf B and Irons MB (2013) Human Genetics and Genomics, ISBN #978-0-470-65447-7
 Gardner, Sutherland, and Schaffer (2012).

Chromosome Abnormalities and Genetic Counseling, ISBN# 978-0195375336

Thompson & Thompson (2016) Genetics in Medicine, ISBN-10: 1416030808

Supplemental Readings

These resources have general overviews of many of the syndromes discussed in this class. You may find it helpful to review certain syndromes.

<u>Pediatric/Specialty/General texts</u>	
Management of Genetic Syndromes	Cassidy & Allanson
The Metabolic and Molecular Basis Inherited Disease	Scriver
Atlas of Metabolic Diseases	Nyhan, Barshop, & Ozand
Human Malformation and Related Anomalies	Stevenson & Hall
Smith's Recognizable Patterns Of Human Malformation 7-ed	<u>Kenneth</u> Jones
Genetics in Primary Care & Clinical Medicine	Seashore & Wapner
Chromosome Abnormalities and Genetic Counseling	Gardner & Sutherland
Emery and Rimoin's Principles and Practice of Medical Genetics	Rimoin, Connor, Pyeritz, & Korf
Unbalanced Chromosome Aberrations in Man	Schinzel
Radiology of Syndromes, Metabolic Disorders & Skeletal Dysplasias	Lachman & Taybi
Syndromes of the Head and Neck	Gorlin
Introduction to Risk Calculation in Genetic Counseling	Ian Young
Practical Genetic Counseling	Harper
Hematology of Infancy and Childhood	Nathan & Orkin
Growth curves	GGC

http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM
www.geneclinics.org
http://www.acmg.net/resources/policy-list.asp
www.kumc.edu
http://www.nsgc.org/about/position.cfm
www.nsgc.org
www.abgc.net
http://www.marchofdimes.com/professionals/15829.asp
http://www.childrensheartinstitute.org/educate/heartwrk/heartse.htm
www.reprotox.org username: 20050001 password GENE11
http://www.possumcore.com username: uthealth password: possum321

Course Objective/s:

Upon successful completion of this course, students will have a knowledge base to understand diagnosis, etiology, natural history, prognosis and treatment/management of genetic conditions as well as be able to elicit targeted medical and family histories.

Specific Learning Objectives:

1. To build a medical genetics knowledge base, including but not limited to, diagnosis, etiology, natural history, prognosis, and treatment/management of genetic conditions and birth defects
2. To understand what information is necessary to elicit in a given clinical scenario in order to accurately assess the medical and family histories
3. To analyze and interpret information for use in determining differential diagnoses

Student responsibilities and expectations:

1. You are required to complete the assigned readings and attend lecture.
2. Slides are available on Google Drive, when available.
3. You are expected to participate during in-class discussions.
4. **Show respect to presenters.**
 - a. **DO NOT TEXT, CHAT, USE/CHECK SOCIAL MEDIA SITES, WORK ON OTHER ASSIGNMENTS, ETC. DURING A LECTURE.**
 - i. **In-Person:** Anyone seen doing so will receive a 0 for their participation grade in the class. You will be notified as soon as this behavior is witnessed, as well as receive notification in writing that will include the details of the event and the consequences.
 - ii. **WebEx/Virtual:** You are responsible for being an active participant in your education. This includes:
 1. Active listening during the presentation
 2. Asking clarifying questions
 3. Engaging with the presenter when opportunities arise
 4. Showing respect to presenters as if you were in-person as described above

Attendance

- Attendance is mandatory for all assigned classes.
- More than one unexcused absence will result in a reduction in the student's final letter grade by one full letter grade. Only extenuating circumstances, such as illness with a doctor's note, will be considered for exceptions to this rule. Make-up work will be required for any missed class.
- **Two tardies (5 minutes late or more) will be considered one unexcused absence.**
 - **WebEx Lectures:** Attendance will be taken 5 minutes after class starts by reviewing the names listed in the "Participants" section

Grading System: A-F

Student Assessment and Grading Criteria :

Vocabulary Quizzes (15 %)	There will be a total of five vocabulary quizzes that are each worth 3% of your final grade. Prior to each quiz, you will be given a list of 20 vocabulary words. The quiz will consist of 10 of these words. The quiz will be in a free answer format. You will be graded on spelling of the vocabulary word as well as definition.
Aneuploidy Presentations (10 %)	Students will give a 10 minute powerpoint presentation on their assigned topic. Assignments and grading rubric can be located in the google drive. Students are expected to identify primary literature on their assigned topic and have the literature approved prior to sharing with the class.
Block I (30 %)	The exam will be administered in-person and will be cumulative for block I. The format will be a combination of multiple choice, matching, fill in the blank and short essay.
Block II (30 %)	The exam will be administered in-person and will be cumulative for block II. The format will be a combination of multiple choice, matching, fill in the blank and short essay.
Clinical Documentation Assignment (10%)	Following your clinical documentation lecture, you will be given a clinical scenario and asked to write a clinic note. This will be graded on completion and is intended to give you practice writing notes prior to clinic.
Participation and/or Attendance (5%)	You are expected to attend class on time and participate in any discussion.

CLASS SCHEDULE

Day/Date	Duration (Hr)	Lecture Topic	Lecturer/s
8/19/2021	1.5	Cytogenetics I	Jan Smith Ph.D.
8/23/2021 Webex	1.5	Molecular Genetics	James Hixson, Ph.D.
8/26/2021	1.5	Cytogenetics II	Jan Smith Ph.D.
8/30/2021	1.5	Patterns of Inheritance I	Leslie Dunnington, MS, CGC
9/2/2021	1.5	Patterns of Inheritance II Vocabulary Quiz 1	Leslie Dunnington, MS, CGC
9/6/2021	NA	No Class	NA
9/9/2021	1.5	Genetic Testing Techniques*	Molly Daniels, MS, CGC
9/13/2021	1.5	Genetic Nomenclature	Myla Ashfaq, MS, CGC
9/16/2021	1.5	Biochemical Testing	Kathryn Gunther, MS, CGC

			Paige Roberts, MS, RD
9/20/2021	1.5	Chromosomes by the Number Vocabulary Quiz 2	Kathryn Gunther, MS, CGC
9/23/21	1.5	An Introduction to Autism Spectrum Disorder	Emily Hansen-Kiss, MS, CGC
9/27/21	2 12 pm to 2 pm	Cytogenetic Abnormalities Note: Required Reading Posted in Google Drive	Jaqueline Hecht, Ph.D., CGC
9/30/2021	1.5	Sequencing Techniques	Michelle Jackson, MS, CGC
10/4/2021	2 12:30 pm to 2:30 pm	Aneuploidy Student Presentations	Class of 2023
10/7/2021	1.5	Autism Spectrum Disorder: The Psychologist's Perspective	Michael Assel, Ph.D.
10/11/2021	1.5	Block I Exam	Leslie Dunnington, MS, CGC Kathryn Gunther, MS, CGC
10/14/2021	1.5	Database and Variant Interpretation	Alana Cecchi, MS, CGC
10/18/2021	1.5	Multiple Congenital Anomaly Syndromes Vocabulary Quiz 3	Kathryn Gunther, MS, CGC
10/21/2021	1.5	Imprinting Disorders	Paul Hillman, MD, Ph.D.
10/25/2021	1.5	Overgrowth Disorders	Kathryn Gunther, MS, CGC
10/28/2021	1.5	Craniofacial Genetics	Emily Hansen-Kiss, MS, CGC
11/1/2021	1.5	Epilepsy Genetics Vocabulary Quiz 4	Kate Mowrey, MS, CGC
11/4/2021	1.5	Bleeding Disorders: Hemophilia and More	Emma Li, MS
11/8/2021	1.5	Neuromuscular Disorders	Kate Mowrey, MS, CGC
11/11/2021	1.5	Normal Human Development	Leslie Dunnington, MS, CGC
11/15/2021	1.5	Dermatologic and Radial Ray Syndromes	Kathleen Shields, MS
11/18/2021	1.5	Approach to the Medical Genetics Evaluation	Myla Ashfaq, MS, CGC
11/22/2021	1.5	A Day in Medical Genetics Clinic: Common Indications Vocabulary Quiz 5	Leslie Dunnington, MS, CGC Kathryn Gunther, MS, CGC
11/25/2021	NA	No Class	NA
11/29/2021	1.5	Huntington's Disease	Leslie Dunnington, MS, CGC
12/2/2021	1.5	Clinical Documentation in Medical Genetics	Paul Hillman, MD, Ph.D.
12/6/2021	1.5	TBD	TBD
12/9/2021	1.5	Block II Exam	Leslie Dunnington, MS, CGC Kathryn Gunther, MS, CGC