Topics in Medical Genetics I
Mondays 1-4pm; MSB B.610
*Fridays 12pm – 2pm; MSB B.603
*Bioinformatics Room; MSB 3rd Floor
*Alternate dates/times/rooms are listed in the syllabus below

Course Coordinators:
Jennifer Lemons, MS, CGC
Jennifer.M.Lemons@uth.tmc.edu
Leslie Dunnington, MS, CGC
Leslie.A.Newman@uth.tmc.edu

Learning Objectives
This class is designed to provide the essential medical genetics information needed as a genetic counselor. It is critical to understand the etiologies and presentations of the diseases discussed in order to be a successful genetics clinician. The objectives of this class are:
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 scenario in order to accurately assess the medical and family histories
3. To analyze and interpret information for use in determining differential diagnoses

Class 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. 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.

Attendance
Attendance is mandatory for all assigned classes. Missing more than one class for any reason (excused or unexcused) 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.

Assignments:
1. Readings as assigned by lecturer.
2. Practice Guidelines Assignment – Due November 5th – Each student has been assigned a Practice Guideline for a specific condition. Guidelines can be found on the Google Drive. Please
read and become familiar with your assigned guideline. Answer the questions listed below and be prepared to turn in your answers and present your assignment to your classmates on November 5th. Each student will have 10 minutes to present and 5 minutes for questions.

** Be mindful that while most information in practice guidelines do not often change, some molecular genetics information and genetic testing strategies for conditions have changed and are not updated in the guidelines. Please refer to more frequently updated resources, such as Gene Reviews, to become familiar with the most current molecular genetics information and genetic testing strategies for your assigned condition.

Achondroplasia – Kaitlyn Amos
Down syndrome – Caroline Bertsch
Fragile X – Wendi Betting
Marfan syndrome – Sarah Burke
Neurofibromatosis type I – Aranza Gonzalez Cendejas
Prader-Willi syndrome – Addison Johnson
Turner syndrome – Luke Kruidenier
Williams syndrome – Brad Power
Hemihyperplasia – Emily Stiglich
Noonan syndrome – Autumn Vara

I. Provide a brief clinical description of your condition. Please include major findings, clinical criteria for diagnosis (if applicable), genetic etiology and prevalence.

II. According to your guideline, what are the recommended methods for diagnosing the condition? If genetic testing is recommended, list two genetic testing laboratories that offer the test you need.

III. Aside from the information in question 1, what additional information from your guideline would you provide to the parents of a patient who is newly diagnosed (less than 1 year for DS and achondroplasia; 5-13 years old for all others) with the condition.

IV. Pick 10 medical terms specific to your condition found in the guideline, and define them in patient friendly language. For example, in CF, meconium is a word that you could explain to the parents as baby’s first stool after birth.

3. Vocabulary Quizzes- 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 12 of these words. The quiz will be in a free answer format.

<table>
<thead>
<tr>
<th>Vocabulary Quiz Dates</th>
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<tbody>
<tr>
<td>Quiz 1  9/21/2018</td>
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<td>Quiz 2  10/1/2018</td>
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<td>Quiz 3  10/8/2018</td>
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<td>Quiz 4  10/29/2018</td>
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<td>Quiz 5  11/19/2018</td>
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4. **Exams** – There will be a midterm and a final exam. They will be cumulative for their respective block.  
   **Dates:** October 15th and December 3rd

**Grading**

Students will be graded on level of participation, attendance, and completion/quality of course assignments.

<table>
<thead>
<tr>
<th>Component</th>
<th>Weight</th>
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<tbody>
<tr>
<td>Block I Exam</td>
<td>30%</td>
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<tr>
<td>Block II Exam</td>
<td>30%</td>
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<tr>
<td>Practice Guidelines</td>
<td>20%</td>
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<tr>
<td>Vocabulary Quizzes</td>
<td>15%</td>
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<tr>
<td>Participation</td>
<td>5%</td>
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<tr>
<td><strong>Total</strong></td>
<td>100%</td>
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100-90% = A  
89-80% = B  
79-70% = C  
69-60% = D  
<60 = F

**Textbooks:**

Gardner, Sutherland, and Schaffer (2012).

Chromosome Abnormalities and Genetic Counseling, ISBN# 978-0195375336


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

<table>
<thead>
<tr>
<th><strong>Pediatric/ Specialty/ General texts</strong></th>
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<tbody>
<tr>
<td>Management of Genetic Syndromes</td>
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<tr>
<td>The Metabolic and Molecular Basis Inherited Disease</td>
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<tr>
<td>Atlas of Metabolic Diseases</td>
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<tr>
<td>Human Malformation and Related Anomalies</td>
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<tr>
<td>Smith's Recognizable Patterns Of Human Malformation 7-ed</td>
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<tr>
<td>Genetics in Primary Care &amp; Clinical Medicine</td>
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<td>Chromosome Abnormalities and Genetic Counseling</td>
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<td>Emery and Rimoin's Principles and Practice of Medical Genetics</td>
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<td>Unbalanced Chromosome Aberrations in Man</td>
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<td>Radiology of Syndromes, Metabolic Disorders &amp; Skeletal Dysplasias</td>
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<tr>
<td>Syndromes of the Head and Neck</td>
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<tr>
<td>Introduction to Risk Calculation in Genetic Counseling</td>
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<tr>
<td>Practical Genetic Counseling</td>
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### Hematology of Infancy and Childhood

**Nathan & Orkin**

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<tr>
<th>Growth curves</th>
<th>GGC</th>
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**Resources:**

- www.geneclinics.org
- [http://www.acmg.net/resources/policy-list.asp](http://www.acmg.net/resources/policy-list.asp)
- www.kumc.edu
- [http://www.nsgc.org/about/position.cfm](http://www.nsgc.org/about/position.cfm)
- www.nsgc.org
- www.abgc.net
- [http://www.childrenshearthistute.org/educate/heartwrk/hearthse.htm](http://www.childrenshearthistute.org/educate/heartwrk/hearthse.htm)
- [www.reprotox.org](http://www.reprotox.org) username: 20050001 password GENE11
- [http://www.possumcore.com](http://www.possumcore.com) username: uthealth password: possum321

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**LECTURE SCHEDULE - BLOCK II**

1pm-4pm Mondays

<table>
<thead>
<tr>
<th>Date/Room</th>
<th>Topic/Readings</th>
<th>Facilitator(s)</th>
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| 8/27/2018 B.610 | **Molecular Genetics I and II**  
Thompson & Thompson  
Chapters 2, 3 & 4 | James Hixson, PHD  
James.E.Hixson@uth.tmc.edu |
| 9/03/2018 | **LABOR DAY - CANCELLED** | |
| 9/10/2018 B.610 | **Cytogenetics I**  
Thompson & Thompson  
Chapter 5 & 6  
- Chapter 2  
- Chapter 3 – pp 52-56 on mosaicism  
- Chapter 3 – pp 46-52 on Structural Imbalance  
- Chapters 5 & 6 on autosomal and sex chromosome translocations | Dr. Jan Smith  
JS5@bcm.edu |
<table>
<thead>
<tr>
<th>Date</th>
<th>Time</th>
<th>Topic/Reading Material</th>
<th>Instructor/Contact Information</th>
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<tbody>
<tr>
<td>9/17/2018</td>
<td>B.610</td>
<td>Cytogenetics II&lt;br&gt;See previous lecture for readings.</td>
<td>Dr. Jan Smith&lt;br&gt;<a href="mailto:JS5@bcm.edu">JS5@bcm.edu</a></td>
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<tr>
<td>9/21/2018</td>
<td>Friday 12pm – 2pm B.603</td>
<td>Variant Interpretation&lt;br&gt;<a href="http://varnomen.hgvs.org/">http://varnomen.hgvs.org/</a></td>
<td>Myla Ashfaq, MS, CGC&lt;br&gt;<a href="mailto:Myla.ashfaq@uth.tmc.edu">Myla.ashfaq@uth.tmc.edu</a></td>
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<tr>
<td>9/24/2018</td>
<td>B.610</td>
<td>1. Genetic Testing Laboratory Techniques&lt;br&gt;2. Patterns of Inheritance&lt;br&gt;- Thompson and Thompson Chapters 7 &amp; 8</td>
<td>Molly Daniels, MS, CGC&lt;br&gt;<a href="mailto:MSDaniel@mdanderson.org">MSDaniel@mdanderson.org</a>&lt;br&gt;Kate Mowrey, MS, CGC&lt;br&gt;<a href="mailto:Kate.Mowrey@uth.tmc.edu">Kate.Mowrey@uth.tmc.edu</a></td>
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<tr>
<td>9/28/2018</td>
<td>Friday 12pm – 2pm B.603</td>
<td>Next Generation Sequencing</td>
<td>Michelle Jackson&lt;br&gt;<a href="mailto:MJackson@ambrygen.com">MJackson@ambrygen.com</a></td>
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<tr>
<td>10/1/2018</td>
<td>B.610</td>
<td>Typical Child Development: Newborn through Early Childhood&lt;br&gt;AAP (2010) Your Baby’s First Year – Copies in UTPB Suite</td>
<td>Jennifer Lemons, MS, CGC&lt;br&gt;<a href="mailto:Jennifer.m.lemons@uth.tmc.edu">Jennifer.m.lemons@uth.tmc.edu</a></td>
</tr>
<tr>
<td>10/8/2018</td>
<td>B.610</td>
<td>1. Newborn Screening&lt;br&gt;1 – 2:30pm&lt;br&gt;2. Biochemical Testing&lt;br&gt;2:30pm – 4pm&lt;br&gt;A. Genetic Counseling in a Busy Metabolic Practice; J of Genetic Counseling (2011) 20:20-22; Hartley, J., Greenberg, C., Mhanni, A.</td>
<td>Leslie Dunnington, MS, CGC&lt;br&gt;<a href="mailto:Leslie.A.Newman@uth.tmc.edu">Leslie.A.Newman@uth.tmc.edu</a>&lt;br&gt;Malorie Jones, MS, CGC&lt;br&gt;<a href="mailto:Malorie.A.Hensley@uth.tmc.edu">Malorie.A.Hensley@uth.tmc.edu</a>&lt;br&gt;Jennifer Lemons, MS, CGC&lt;br&gt;<a href="mailto:Jennifer.m.lemons@uth.tmc.edu">Jennifer.m.lemons@uth.tmc.edu</a></td>
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<tr>
<td>Date</td>
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<tr>
<td>10/15/2018</td>
<td>MIDTERM EXAMINATION</td>
<td>B.610</td>
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| 10/22/2018 | Medical Genetics Evaluation and Dysmorphology | 1. Myla Ashfaq, MS, CGC  
                                |                                                                         | 2. Medical Genetics Genetic Counselors |
| 10/26/2018 | Database Lecture                         | Myla Ashfaq, MS, CGC  
                                |                                                                         |  
| 10/29/2018 | Common Genetic Syndromes: Multiple Congenital Anomalies (MCA) | 1. Katie Gunther, MS, CGC  
                                |                                                                         | 2. Katie Gunther, MS, CGC |
| 11/2/2018  | Common Genetic Syndromes: Dermatologic and Radial Ray | Victoria Wagner, MS, CGC  
                                |                                                                         |  
| 11/5/2018  | In-Class Discussion: Practice Guidelines Presentations | Genetic Counseling Students |
| 11/12/2018 | Chromosome Abnormalities Required reading:  | Dr. Hecht  
                                |                                                                         | 1. Linden MG et al (2002) Genetic Counseling for Sex  
                                |                                                                         |  

**Vocabulary Quiz #3**

**Vocabulary Quiz #4**


8. Torfs CP and Christianson RE


Optional reading:


| 11/19/2018  
B.610 | 1. **Readers, Writers, Erasers, and Remodelers**  
2. **Common Genetic Syndromes:** Neuromuscular and Brain | 1. Jennifer Lemons, MS, CGC  
[Jennifer.m.lemons@uth.tmc.edu](mailto:Jennifer.m.lemons@uth.tmc.edu)  
2. Kate Mowrey, MS, CGC  
[Kate.Mowrey@uth.tmc.edu](mailto:Kate.Mowrey@uth.tmc.edu) |
| 11/26/2018  
B.610 | 1. **Imprinting Disorders**  
Elhamamsy. Role of DNA Methylation in imprinting | 1. David Rodriguez-Buritica MD  
[David.F.RodriguezBuritica@uth.tmc.edu](mailto:David.F.RodriguezBuritica@uth.tmc.edu)  
2. Katie Gunther, MS, CGC |

2. **Common Genetic Syndromes**: Overgrowth

Kathryn.A.Gunther@uth.tmc.edu

12/3/2018 B.610 FINAL EXAMINATION