Fall 2021
GS11 1172: Prenatal Genetic Counseling
Credit Hours: _2_____
Meeting Location (Building/Room # or WebEx/Zoom):
_McGovern Medical School B.610

Program Required Course: _X_ Yes ____ No
Approval Code _X__ Yes ____ No
(If yes, the Course Director or the Course Designee will provide the approval code.)

Audit Permitted: _____ Yes __X___ No
Classes Begin: August 17, 2021
Classes End: December 07, 2021
Final Exam Week: December 07, 2021

Class Meeting Schedule:

Day	Time
Tuesdays, Class Meeting	2:00-4:00 p.m.
Tuesdays, MultiD Conference	4:00- 5:00 p.m.

Course Director:
Claire N. Singletary, MS, CGC
Program Director, UT Genetic Counseling Program
Professor, Pediatrics and Obstetrics/Gynecology
McGovern Medical School, UTHealth
Email Address: Claire.N.Singletary@uth.tmc.edu
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Course Co-Director/s:
Jennifer Czerwinski, MS, CGC
Associate Director, UT Genetic Counseling Program
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NOTE: Office hours are available on request. Please email me to arrange a time to meet.

Instructor/s:

1. Meagan Choates, MS, CGC
   McGovern Medical School
   Meagan.Giles@uth.tmc.edu
2. Blair Stevens, MS, CGC
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   McGovern Medical School
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4. Lara Friel, MD, PhD
   McGovern Medical School
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5. Tony Jonhson, DO
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   Baylor Genetics Laboratories
   mburgess@baylorgenetics.com
7. Sandra Darilek, MS, CGC
   Baylor College of Medicine
   sdarilek@bcm.edu

Course description: This course provides an in-depth review of current topics in prenatal genetic counseling, including screening and diagnostic testing, ultrasound findings, and carrier screening. Students are expected to gain an appreciation for more complex prenatal issues that impact prenatal practice and to work on critical thinking skills.

Textbook/Supplemental Reading Materials (if any)

- Hogge WA (2017) Sanders' Structural Fetal Abnormalities, ISBN 978-1259641374
- Gardner and Amor (2018) Gardner and Sutherland's Chromosome Abnormalities and Genetic Counseling ISBN# 978-0199329007

Course Objective:

Upon successful completion of this course, students will have the depth and breath of knowledge of prenatal genetic counseling in order to identify and assess appropriate genetic testing options and to assess the probability of a condition with a genetic component in the prenatal setting.

***Specific Learning Objectives*:**

1. To demonstrate a depth and breadth of understanding and knowledge of prenatal genetics to include AMA, serum screening, NIPT, ultrasound abnormalities
2. To identify and assess genetic testing options in prenatal setting, including the analytic validity, clinical validity, and clinical utility.
3. To critically assess the genomic scientific literature in the prenatal field
4. To assess individuals' and their relatives' probability of conditions with a genetic component or carrier status in a prenatal setting
5. To work on critical thinking skills which allow effective evaluation of counseling sessions.

Student responsibilities and expectations:

Students enrolled in this course will be expected to perform the following activities each week.

1. Read the assigned readings prior to each class
2. Prepare for and take course exams based on course lectures/ readings.
3. Attend and participate in class field trips
4. Develop unique work on assignments and submit in a timely fashion (see grading)cs

Students are expected to complete all assigned reading material prior to class. While you may work and discuss all course materials and assignments in groups, all final products must be your own. Plagiarism and failure to properly cite scientific literature and other sources will not be tolerated and are grounds for dismissal from the course and further GSBS disciplinary action. Cheating or engaging in unethical behavior during examinations (quizzes and final) will be grounds for dismissal from the course without credit and further GSBS disciplinary action. 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. Exceptions will only be made for an illness with a doctor's note. Being tardy to two classes by more than 5 minutes is considered an unexcused absence.

Grading System: Letter Grade (A-F); 90-100 A, 80-89 B, 70-79 C, 60-69 D,<60 F	
Student Assessment and Grading Criteria : (May include the following:)	
Prenatal Literature Assignment (5%)	Review your assigned article as directed below and bring typed/written answers to be collected at the beginning of class. Bring a second copy to refer back to during the in-class discussion.
Ultrasound Scenario (5%)	Answer the following questions for your assigned anomaly. The goal of this assignment is to create a resource that may be used in clinic when ultrasound abnormalities arise.
MCA Assignment (5%)	The goal of this assignment is to think about how to approach a case in which more than one ultrasound abnormality is present. Cases will be distributed the week before the class discussion takes place.
FTS/NIPT/Amnio graphic/chart (5%)	Create a visual aid/chart that you could use with patients that gives pertinent information comparing and contrasting First Trimester Screen vs Non Invasive Prenatal Testing vs Amnio for a singleton pregnancy. This should be a companion to your prenatal outline
AMA Outline (10%)	Develop an outline from scratch for use in AMA session in the first trimester (38 y/o at 12 weeks for discussion of various screening and diagnostic testing options).
Carrier screening assignment (15%)	Create a reference document that compares core/ACOG testing (CF, SMA, Hemoglobin beta chain for sickle cell/beta thal, alpha thal as needed, AJ diseases as needed) and expanded options with X-linked conditions and complete the scenarios with risk assessment.
Baby center reflection (5%)	Write a summary (no more than 3 pages) that explains what you think the benefits of sites/apps like this are for pregnant women, what concerns/risks you foresee. In addition, specifically discuss the accuracy of the genetic screening and testing information presented.
MidTerm Exam (20%) Final Exam (20%)	The exams will be based on the information presented/discussed in class as well as information contained in assigned readings. Each facilitator will be asked to submit questions for the exam. There will be multiple choice, fill in the blank and short answer questions. A significant portion of the questions will be modeled after board-style questions in order to make students more familiar with the format of questions they may see on their boards. Please visit the ABGC website (www.abgc.net) to view some sample board questions. As information on complex anomalies will be included on the final exam,

	cumulative knowledge from the simple ultrasound anomalies presented in the first part of the course will be needed on the final exam.
Workshop (5%)	Attend the prenatal workshop and participate in the role plays as listed below. This is a completion grade.
Prenatal Folder (5%)	Create a folder on the drive with resources you want at your fingertips as you go on prenatal rotation. This is designed to help you organize and prepare for Intro rotations and you should continue to add to it as you progress through the program.
Attendance	Required: 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. Exceptions will only be made with a doctor's note.

CLASS SCHEDULE

Day/ Date	Duration (Hr)	Lecture Topic	Lecturer/s
8/17	**2.5**	**Intro to Pregnancy, Prenatal Terminology, & Pregnancy Wheel (1 hr) In class exercise** **Advanced Maternal Age & Diagnostic Prenatal Testing (1.5 hr)** 1. Hook EB (1981) Rates of chromosome abnormalities at different maternal ages. Obstet Gynecol, 58:282-285. 2. ACOG (2016) Practice Bulletin No. 162 Summary: Prenatal Diagnostic Testing for Genetic Disorders. *Obstet & Gynecol*, 2016 May;127(5):976-8. doi: 10.1097/AOG.0000000000001438. 3. Wapner et al (2012) Chromosomal Microarray versus karyotyping for prenatal diagnosis. *NEJM*, 367, 2175-2184. 4. CDC Morbidity and Mortality Weekly Report (1995) Chorionic Villus Sampling and Amniocentesis: Recommendations for Prenatal Counseling. 44, No RR-9.	**Claire Singletary** **Jen Czerwinski**
8/24	**3**	**Screening for Aneuploidy: Serum Screens and NIPT** 1. Wilson K et al (2013) NSGC Practice Guideline: Prenatal Screening and Diagnostic Testing Options for Chromosome Aneuploidy *J Genetic Counsel*, 22: 4-15. 2. ACOG (2020) Practice Bulletin No. 226 Summary: Screening for Fetal Aneuploidy. *Obstetrics and Gynecology*, 2020 Oct 136 (4)epub ahead of print Aug2020	**Claire Singletary**

		3. Gil et al (2017) Analysis of cell-free DNA in maternal blood in screening for aneuploidies: updated meta-analysis. *Ultrasound Obstet Gynecol*, 50, 302-314. 4. Gil et al (2019) Screening for trisomies by cfDNA testing of maternal blood in twin pregnancy: update of the Fetal Medicine Foundation results and meta-analysis. Ultrasound Obstet Gynecol, 53, 734-742. 5. Artieri et al (2017) Noninvasive prenatal screening at low fetal fraction: comparing whole-genome sequencing and single-nucleotide polymorphism methods. *Prenat Diag*, 37, 482-490.	
8/31	**3**	**Discuss Primary Literature Assignment (1 hr)** Student Presentations - see assignment **NIPT: Monogenic Disorders** 1. Chiu EKL et al (2018) cfDNA screening and diagnosis of monogenic disorders - where are we heading? *Prenat Diagn*, 38, 52-58. 2. Zhang J et al (2019) Non-invasive prenatal sequencing for multiple Mendelian monogenic disorders using circulating cell-free fetal DNA. *Nature Med*. 25, 439-447. 3. Itzep N et al (2018) [Poster] A Novel Next-Generation Sequencing Based Assay for Non-Invasive Prenatal Testing of Sickle Cell Disease without Paternal DNA, ASHG. *(see also Tsao et al if interested)*	**Claire Singletary**
9/7	**2.5**	**Prenatal Ultrasound: Normal Anatomy** 1. ACOG Practice Bulletin: Ultrasonography in Pregnancy (2016) *Obstet & Gynecol*, 128 (6), 241-256. 2. Breathnach et al (2007), The second trimester genetic sonogram. *Am J Med Gen,* 145C: 62-72. **Ultrasound: Soft Signs** 1. Review - ACOG (2016) Practice Bulletin No. 163 Summary: Screening for Fetal Aneuploidy. *Obstetrics and Gynecology*, 2016 May;127(5):979-81. doi: 10.1097/AOG.0000000000001439. 2. Agthaokleous M (2013) Meta-analysis of second-trimester markers for trisomy 21. *Ultr Obstet Gynecol*, 41, 247-261. 3. Benacerraf (2005), The role of the second trimester genetic sonogram in screening for fetal Down syndrome. *Semin Perinatol,* 29 (6) 386-94. 4. Reference: *Clinical Significance and GC for Common Ultrasound Findings* from NSGC Prenatal SIG	**Meagan Choates**

9/14 **multiD**	**2**	**Ultrasound: Head & Heart, Neural Tube** 1. McKechnie, et al. (2012), Neonatal outcome of congenital ventriculomegaly. *Semin Fetal Neonatal Med*, 17 5), 301-7. 2. First Trimester Scan, Nicolaides (see drive) Chapter 3 3. D'Antonio, F., Pagani, G., Familiari, A., Khalil, A., Sagies, T.L., 2016. Outcomes associated with isolated agenesis of the corpus callosum: a meta-analysis. Pediatrics 138, e20160445 4. Maarse W, et al.(2012), A systematic review of associated structural and chromosomal defects in oral clefts: when is prenatal genetic analysis indicated? J Med Genet, 49, 490–498. 5. Patel A, Costello JM, Backer CL, Pasquali SK, Hill KD, Wallace AS, et al. Prevalence of noncardiac and genetic abnormalities in neonates undergoing cardiac operations: analysis of The Society of Thoracic Surgeons Congenital Heart Surgery Database. Ann Thorac Surg. 2016;102(5):1607–1614. 6. Loffredo C. A., Chokkalingam A., Sill A. M., Boughman J. A., Clark E. B., Scheel J., et al. (2004). Prevalence of congenital cardiovascular malformations among relatives of infants with hypoplastic left heart, coarctation of the aorta, and d-transposition of the great arteries. *Am. J. Med. Genet. Part A* 124A, 225–230. 7. Mardy AH, Chetty SP, Norton ME, Sparks TN. A system-based approach to the genetic etiologies of non-immune hydrops fetalis. Prenat Diagn. 2019 May 13. doi: 10.1002/pd.5479 8. Moldenhaeur JS & Adzick NS. Fetal surgery for myelomeningocele: After the management of the MOMS Study. Seminars Fetal & Neonatal. 2017. 360-366.	**Blair Stevens**
9/21 **multiD**	**2**	**Ultrasound: Abdomen, Renal, Urogenital** 1. Harman, C. R. (2008). Amniotic fluid abnormalities. In Seminars in perinatology (Vol. 32, No. 4, pp. 288-294). WB Saunders. 2. Leeuwen, L., & Fitzgerald, D. A. (2014). Congenital diaphragmatic hernia. Journal of paediatrics and child health, 50(9), 667-673. 3. Prefumo, F., & Izzi, C. (2014). Fetal abdominal wall defects. Best practice & research Clinical obstetrics & gynaecology, 28(3), 391-402.	**Shannon Mulligan**

		a. Helpful supplement: Carli et al 2020 Prenatal features in Beckwith-Wiedemann syndrome and indications for prenatal testing 4. Talati, A. N., Webster, C. M., & Vora, N. L. (2019). Prenatal genetic considerations of congenital anomalies of the kidney and urinary tract (CAKUT). Prenatal diagnosis, 39(9), 679-692. 5. Smet, M. E., Scott, F. P., & McLennan, A. C. (2020). Discordant fetal sex on NIPT and ultrasound. Prenatal diagnosis, 40(11), 1353-1365.	
9/28 **3-5**	**2**	**Carrier Screening:** **Core Diseases of CF, SMA, Hemoglobinopathies; Unity** 1. ACOG Committee Opinion 691: Carrier Screening for Genetic Conditions, Obstet Gynecol 2017, reaffirmed 2019	**Meagan Choates**
10/5 **multiD**	**2**	**ACOG/ACMG vs Expanded Panels** 1. ACOG Committee Opinion 690: Carrier screening in the age of Genomic Medicine. Obstet Gyencol 2017, 2. Lazarin GA et al. An empirical estimate of carrier frequencies for 400+ causal Mendelian variants: results from an ethnically diverse clinical sample of 23,453 individuals. Genet Med, 15, 178-186. 3. Stevens B, Krstic N, Jones M, Murphy L, Hoskovec J. Finding middle ground in constructing a clinically useful expanded carrier screening panel. Obstet Gynecol 2017, 130, 279–84.	**Malorie Jones**
10/12 **2-5 pm**		**Ultrasound: Skeletal** 1. Dighe et al (2008), Fetal Skeletal Dysplasia: An Approach to Diagnosis with Illustrative Cases. *RadioGraphics,* 28 (4) 1061-77. 2. Krakow et al (2009), Guidelines for the prenatal diagnosis of fetal skeletal dysplasia. *Genet Med,* 11 (2) 127-33. **Introduction to Working up a Multiple Congenital Anomalies case**	**Theresa Wittman** **Jen Czerwinski**
10/19 **multiD**		**Ultrasound: Multiple Congenital Anomalies** **MCA Presentations** 1. Viora et al (2007), Trisomy 18: Fetal Ultrasound Findings at Different Gestational Ages. *Am J Med Genet A,* 143A: 553-557. 2. Wapner et al (2012), Chromosomal Microarray versus karyotyping for prenatal diagnosis. *NEJM*, 367, 2175-2184. *[review again from AMA lectures]*	**Jen Czerwinski**

		3. Donnelly JC et al (2014), Association of Copy Number Variants with Specific Ultrasonographically Detected Fetal Anomalies. *Obstet & Gynecol,* 124 (1), 83-90. 4. ACOG Committee Opinion: The Use of Chromosomal Microarray Analysis in Prenatal Diagnosis (2013) *Obstet & Gynecol*, 122(6): 1374-7. 5. Carss et al (2014), Exome sequencing improves genetic diagnosis of structural fetal abnormalities revealed by ultrasound. *Hum Mol Genet*, 23(12): 3269-3277.	
10/22 1-3pm B.625		**MIDTERM EXAM**	**Jen/Claire**
11/2		**No class this date of 11/2 - see 11/12**	
11/9 multiD		**Unusual Results: Mosaicism, markers, microarray, etc..** 1. Muzzey et al (2017) Understanding the Basics of NGS: From Mechanism to Variant Calling. *J Genet Counsel* 2. Eggermann et al (2015), Mosaicism and UPD in Prenatal Diagnosis. *Trends in Molecular Medicine*, 21 (2) 77-86 3. Skim Chapters 16, 17 and 27 in Gardner, Sutherland and Shaffer's Chromosome Abnormalities and Genetic Counseling. particularly pgs 442-454 and 472-476 (4th edition). **Prenatal WES** 1. ISPD and SMFM (2018) Joint Position Statement from the International Society for Prenatal Diagnosis (ISPD), the Society for Maternal Fetal Medicine Foundation (SMFM) and the Perinatal Quality Foundation (PDGF) on the use of genome-wide sequencing for fetal diagnosis. *Prenat Diagn*, 38, 6-9. 2. Best S et al (2018) Promises pitfalls and practicalities of prenatal whole exome sequencing. *Prenat Diagn*, 38, 10-19. 3. Normand et al (2018) Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. *Genome Medicine*, 10:74.	**Theresa Wittman**
11/12 12-2pm		**Assisted Reproductive Technology** 1. Practice Committee of American Society for Reproductive Medicine; Practice Committee of Society for Assisted Reproductive Technology. Preimplantation	**Sandra Darilek**

(in lieu of class 11/2)		genetic testing: a Practice Committee opinion. *Fertil Steril.* 2008 Nov;90(5 Suppl):S136-43 PMID: 19007612 2. ACOG Committee Opinion No. 430: preimplantation genetic screening for aneuploidy. *Obstet Gynecol.* 2009 Mar;113(3):766-7 PMID: 19300349 3. Dahdouh EM at al (2015). Technical Update: Preimplantation Genetic Diagnosis and Screening. *J Obstet Gynaecol Can.* 2015 May;37(5):451-63. PMID: 26168107 4. Kohn TP et al (2016). Genetic counseling for men with recurrent pregnancy loss or recurrent implantation failure due to abnormal sperm chromosomal aneuploidy J Assist Reprod Genet. 2016 May;33(5):571-6 PMID: 27020275 5. Greco E, Minasi MG, Fiorentino F. Healthy babies after intrauterine transfer of mosaic aneuploid blastocysts. N Engl J Med. 2015 Nov 19;373(21):2089-90. PMID: 26581010 6. Besser AG, Mounts EL. Counselling considerations for chromosomal mosaicism detected by preimplantation genetic screening. Reprod Biomed Online. 2017 Apr;34(4):369-374. PMID: 28129970	
11/16		**Role Plays**	**GCs**
11/23 **Class 8-10am**		**Fetal Intervention** 1. Adzick NS et al (2011) A Randomized Trial of Prenatal versus Postnatal Repair of Myelomeningocele. *NEJM*, 364,11, 993-1004. 2. Morris RK (2013) Percutaneous vesicoamniotic shunting versus conservative management for fetal lower urinary tract obstruction (PLUTO): a randomised trial. *Lancet*, 382 (9903) 1496-506. 3. Deprest J (2014) Prenatal management of the fetus with isolated congenital diaphragmatic hernia in the era of the TOTAL trial. *Sem Fetal Neonat Med*, 19, 338-348. 4. Senat (2004) Endoscopic Laser Surgery versus Serial Amnioreduction for Severe Twin to Twin Transfusion syndrome. NEJM, 351. 136-144	**Dr. Tony Johnson**
11/30 **multiD**		**Maternal Conditions Affecting the Fetus: Diabetes, Epilepsy, Lupus, Antibodies, Thrombophilias, Recurrent Loss** 1. Cassina et al (2013). Pregnancy outcome in women exposed to antiepileptic drugs: teratogenic role of	**Dr. Lara Friel**

		maternal epilepsy and its pharmacologic treatment. *Reproduct Toxicol,* 39, 50-57. 2. HAPO Research Study Group (2008). Hyperglycemia and Adverse Pregnancy Outcomes. *NEJM*, 358, 1991-2002. 3. Moise KJ et al (2012). Management and prevention of red cell alloimmunization in pregnancy. *Obstet Gynecol*, 120, 1132-1139. 4. Laurino MY et al (2005). Genetic Evaluation and counseling of couples with recurrent miscarriage: recommendation of the National Society of Genetic Counselors. *J Genet Counsel*, 14, 165-181. 5. Browse the Pregnant with Cancer Network website: http://www.hopefortwo.org/	
12/7		**FINAL EXAM**	**Jen/Claire**

Assignments:

Due date	**Points**	**Assignment**
8/31	**5**	**Prenatal Literature Assignment** Read your assigned article and consider the following questions: Over what year(s) was the study completed? What type of study is it? (prospective/retrospective, randomized/non-randomized) How many subjects are there? Is there a control group(s)? What is the inclusion/exclusion criteria? Are the findings significant? Would you feel comfortable quoting this data to a patient? Why or why not? What are the strengths and limitations to this study? Bring typed/written answers to be collected at the beginning of class. Bring a second copy to refer back to during the in-class discussion.

Student	Article	Topic
Jordan	Malone, F. D., Canick, J. A., Ball, R. H., Nyberg, D. A., Comstock, C. H., Bukowski, R., … First- and Second-Trimester Evaluation of Risk (FASTER) Research Consortium. (2005). First-trimester or second-trimester screening, or both, for Down's syndrome. The New England Journal of Medicine, 353(19), 2001–2011. http://doi.org/10.1056/NEJMoa043693	serum screen
Jasmine	Dugoff, L., & Society for Maternal-Fetal Medicine. (2010). First- and second-trimester maternal serum markers for aneuploidy and adverse obstetric outcomes. Obstetrics and Gynecology, 115(5), 1052–1061. http://doi.org/10.1097/AOG.0b013e3181da93da	serum screen

		Cindy	Palomaki et al (2012), DNA sequencing of maternal plasma reliably identifies trisomy 18 and trisomy 13 as well as Down syndrome: an international collaborative study. *Genetics in Medicine*	NIPT validation
		Tessa	Pergament et al (2014) Single Nucleotide Polymorphism Based Noninvasive Prenatal Screening in a High Risk and a Low Risk Cohort. Obstet Gynecol, 2, 210-219.	NIPT validation
		Emily	Mazloom et al (2013) Noninvasive prenatal detection of sex chromosomal aneuploidy by sequencing circulating cell free DNA from maternal plasma *Prenat Diagn* 33, 591-597	NIPT validation
		Jack	Zhao C, Tynan J, Ehrich M, et al. Detection of fetal subchromosomal abnormalities by sequencing circulating cell-free DNA from maternal plasma. Clin Chem. 2015;61:608.	microdel NIPT
		Yusra	Wapner et al (2015) Expanding the scope of noninvasive prenatal testing: detection of fetal microdeletion syndromes. *Amer J Obstet Gynecol*, 212, 332e1-9.	microdel NIPT
		Erin	Helgeson, J., Wardrop, J., Boomer, T., Almasri, E., Paxton, W. B., Saldivar, J. S., Dharajiya, N.,Monroe, T. J., Farkas, D. H., Grosu, D. S., and McCullough, R. M. (2015) Clinical outcome of subchromosomal events detected by whole-genome noninvasive prenatal testing. Prenat Diagn, doi: 10.1002/pd.4640.	microdel NIPT
		Maddi	Lefkowitz RB et al (2016) Clinical validation of noninvasive prenatal test for genomewide detection of fetal copy number variants. *Amer J Obstet Gynecol*, 215, 227.e1-16.	genome NIPT
		Latonya	Bianchi, D. W., Chudova, D., Sehnert, A. J., Bhatt, S., Murray, K., Prosen, T. L., . . . Halks-Miller, M. (2015). Noninvasive prenatal testing and incidental detection of occult maternal malignancies. JAMA, 314(2), 162. [see also Carlson LM et al (2018) Maternal Malignancy Evaluation After Discordant Cell-Free DNA Results. *Obstet Gynecol,* 131, 3, 464-468]	NIPT & cancer
9/28	**5**	**Ultrasound Scenario** Answer the following questions for your assigned anomaly. The goal of this assignment is to create a resource that may be used in clinic when ultrasound abnormalities arise. Once approved by the coordinators, these scenarios will be made available to each student. Your patient is a 30 y/o woman of Mexican ancestry at 18 weeks gestation. ● Describe the anomaly (what is it, how does it happen): ● What trisomy/syndrome is at the top of your differential? ● What is the likelihood that your patient has an aneuploidy (trisomy 21, 13 and/or 18) given the finding? Give a specific number (or range of numbers) that you would quote to the patient. ● What other syndromes are high enough in your differential that you would mention them to a patient and/or offer testing to rule them out? How likely are these syndromes? (e.g. 22q11, a single gene disorder, etc). What screening or testing is available?			

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What is the prognosis for this baby if the finding is isolated (no syndrome)? How likely is this outcome?
Provide a visual aid or drawing that you would use with a patient for this condition.
List at least 3 primary literature references that you used.

<table>
<tr><td>AV canal defect – Latonya
Coarctation of the aorta – Maddi
Echogenic bowel – Erin
Increased Nuchal Fold – Yusra
Absent Nasal Bone – Jack</td><td>Ventriculomegaly at 12 mm – Emily
Spina bifida (open L3-5) – Tessa
Cystic hygroma – Cindy
Choroid plexus cyst - Jasmine
Unilateral cleft lip - Jordan</td></tr>
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<tr><td>9/14</td><td>10</td><td>Prenatal outline/flowchart/table
Each student is expected to develop an outline/flowchart/table from scratch for use in AMA session in the first trimester (38 y/o at 12 weeks for discussion of NIPT and diagnostic testing options). Resources for developing a resource:
see appendix 3 for example template
pg 189 in Uhlmann – table 6-1 on prenatal genetics patients
pg 153 in Weil – section on prenatal diagnosis counseling</td></tr>
<tr><td>9/14</td><td>5</td><td>Comparison Chart: FTS, NIPT, and amnio
Create a visual aid/chart/graphic that you could use with patients that gives pertinent information comparing and contrasting First Trimester Screening, Non Invasive Prenatal Testing, and Amnio for a singleton pregnancy. This should be a companion to your prenatal outline</td></tr>
<tr><td>10/19</td><td>5</td><td>Multiple Congenital Anomaly Case Preparation Assignment
Each individual will be given a case to work-up. The goal of this assignment is to think about how to approach a case in which more than one ultrasound abnormality is present. Read the case you are assigned (will be handed out October 12) and think about what information you would present to your patient. Things you should consider are the condition(s) you are most concerned about, appropriate testing to offer your patient (be specific – not just amnio but what specific chromosome study, CMA, DNA test, etc) and resources and/or referrals you would make. You will turn in the written part of your assignment at the beginning of class. You should be prepared to discuss your thoughts about your case during class.</td></tr>
<tr><td>11/2</td><td>15</td><td>Carrier screening assignment
A. Create a reference document that compares the following options:
Core/ACOG testing (CF, SMA, Hemoglobin beta chain for sickle cell/beta thal, alpha thal as needed, AJ diseases as needed)
UT custom panel (Invitae and/or Myriad)
Expanded option with X linked conditions (Invitae and/orMyriad)
(see chart template on the drive in assignments as a guide)</td></tr>
</table>

		B. Create a general visual aid for use in clinic that would help you explain recessive inheritance/ the different carrier screening options to a patient. **C. Case scenarios to write a brief/bulleted summary:** 1. You have an G1P0 26 y/o African American patient referred to you who has silent alpha thalassemia on an expanded panel done at her doctor's office that includes HBB, CF and SMA. Her partner is of African American ancestry and has no testing on record. What risk numbers would you present? a. likelihood he is carrier of a clinically relevant hemoglobinopathy b. likelihood fetus is affected c. If he tests negative, what is his residual risk to be a carrier and risk to pregnancy? d. What specific testing would you offer to him? 2. You have a G3P2 33 y/o Latina patient referred to you due to finding the SMA SNP on her carrier screening panel at her OB's office. The remainder of her expanded screen is normal, including CF and HBB. Her form indicates her partner is also Latino. What risk numbers would you present to the patient? a. Likelihood he is a carrier of SMA b. Likelihood fetus is affected c. If he tests negative, what is his residual risk to be a carrier and risk to pregnancy? 3. You are seeing a G1P0 29 y/o White patient referred to you for a likely pathogenic variant in DMD found on her expanded carrier screen at her doctor's office. The remainder of her expanded Natera panel is negative, including CF and SMA. Her NIPT that was drawn at the same time was negative and states that the fetus is expected to be male. What is the current (potential) risk to the fetus? What testing options would you present to the patient and how would you discuss the fact that the variant is likely pathogenic rather than pathogenic?
11/16	**5**	**Prenatal Explanation Role Plays** – completion/participation grade <u>Patient Scenario 1</u>: Advanced Maternal Age for Possible Amniocentesis Name: Janet Smith [husband Chris Smith not present] DOB: 2/12/1982 LMP: 07/19/21 EDC: 4/25/2022 G1P0 Occupation: Elementary school teacher Religion: Protestant Scenario: You are meeting with Mrs. Smith to review her age related risk to have a baby with a chromosome problem and discuss her option of NIPT versus amniocentesis for diagnostic testing. You need to explain:

		1. contracting to determine how she feels about testing 2. age related risk 3. chromosome problems related to age 4. amniocentesis a. how the procedure is done b. the risks c. the benefits 5. explore alternative of NIPT and ultrasound as screens if appropriate based on contracting You do not need to talk about quadruple marker screening or any other type of serum screening. The patient did not do a first trimester screen or NIPT because she thought she wanted diagnostic testing by amniocentesis after talking to her obstetrician. As the date for the amnio has approached, she has become a little more nervous about the procedure. You will need to help the patient make a decision about testing. You do not have to take her family history in the interest of time. <u>Patient Scenario 2:</u> carrier screening: core/ACOG vs expanded panel Name: Michelle Davis [husband Will Davis not present] DOB: 8/24/1988 G0P0: Preconception patient Occupation: NICU nurse Religion: Baptist Scenario: You are meeting with Mrs. Davis to discuss her screening options for preconception carrier screening (compare/contrast core panel vs expanded) 1. genes/ recessive inheritance 2. core conditions – CF, SMA, Hemglobinopathies methodology 3. expanded panel – X linked conditions, conditions with risks to carrier 4. likelihood of finding a carrier 4. options for FOB testing (tandem, sequential) 5. next steps if positive Focus on how to explain/compare/contrast the two major options. You will need to explain the options and help the patient make a decision. You do not need to take a family history in the interest of time.
12/7	**5**	**Baby Center Reflection** email prenatalfall2021@gmail.com password - utgcpprenatal2021 Original information: Learn about what women and their partners hear about pregnancy, screening, testing, and development by following the pregnancy updates at https://www.babycenter.com/. We created an email unrelated to your general email so that you don't get spam forever about pregnancy and babies. Please start viewing the updates as of July 1st. Plan to log in once per week to see the first trimester updates before class starts. There will be an assignment at the end of the semester to

		reflect on the information (helpfulness, accuracy, concerns). You can view the weekly emails from babycenter in the gmail account and also look at all the types of spam that start going to our group gmail (prenatalfall2021@gmail.com (UTGCP Prenatal Class) password utgcpprenatal2021). This is the same email and password for the babycenter site if you prefer to log in directly or download the app. Your LMP was 4/26/21 and due date is 1/31/22. Assignment: When viewing the emails/site weekly, keep notes about what you found interesting, helpful, and scary about the information. If they gave specific information on genetic screening or testing, make note of details. Upon reflection, write a summary (no more than 3 pages) that explains what you think the benefits of sites/apps like this are for pregnant women, what concerns/risks you foresee. What was helpful for you to understand about pregnancy from this assignment? In addition, specifically discuss the accuracy of the genetic screening and testing information presented. What misinformation did you find? How will knowing this impact your practice as a prenatal genetic counselor?
12/7	**5**	**Prenatal Rotation Folder** Throughout this course you will be introduced to resources that might be helpful to have in clinic, such as risk charts, listings of medications, and likelihood ratios for some of the ultrasound soft signs. You will also be creating items that will be useful to refer to in clinic, such as your NIPT charts and ultrasound factsheets. We recommend thinking outside of the box--ask counselors you observe or other students what they have found helpful. Feel free to incorporate items from other classes (ex: pedigree information from Introduction to Genetic Counseling) and think of how it may be best to organize all the resources. Think to yourself "if I was in clinic alone or had a last minute add-on, what information would I want to have at my fingertips?" Remember, this assignment is to HELP you organize and prepare for your introductory rotations. The assignment will largely be graded on completion and the class coordinators will give you feedback on what other resources may be helpful. Please create a folder on the Google drive with all of your resources and share it with Jen and Claire. You will later add your head rotation supervisors as they will look at your rotation folder again at the conclusion of your introductory prenatal rotation.