Etiology and Characteristics of Infants and Young Children with Disabilities

Chromosome Deletions and Abnormalities

Sample Syllabus

Course Description

This course is designed to provide students with information about identifying what chromosome deletion and abnormality is, traits chromosome deletions have in common, and lifelong impacts of individuals and families who have chromosome deletions and abnormalities. This course will focus on contemporary issues surrounding chromosome deletions and abnormalities will be addressed, including, but not limited to family lifestyle, early childhood development and learning, and an overview of chromosome deletions. Students will learn and practice to support families of children with chromosome deletions and abnormalities.

Required Texts

McKinlay Gardner, R. J., Sutherland, G. R., & Gardner, L. G. (2012). *Chromosome abnormalities and genetic counseling (*4th ed.). Oxford University Press.

Course Learning Objectives

As a result of active participation and successful completion of course requirements, students will be able to:

1. Identify chromosome deletion and abnormality.
2. List traits deletions and abnormalities have in common.
3. Define accompanying conditions children can exhibit.
4. List lifelong impacts for children with chromosome deletions and abnormalities.

Readings

Carruth, L, L., Reisert, I., & Arnold, A. P. (2002). Sex chromosome genes directly affect brain sexual differentiation. *Nature Neuroscience, 5*(10), 933-934. https://doi.org/10.1038/nn922

Gilmore, L. (2017). Supporting families of children with rare and unique chromosome disorders. *Research and Practice in Intellectual and Developmental Disabilities, 5*(1), 8-16. https://doi.org/10.1080/23297018.2017.1360152

He, W., Sun, X., Liu, L., Li, M., Jin, H., & Wang, W. H. (2014). The prevalence of chromosomal deletions relating to developmental delay and/or intellectual disability in human euploid blastocysts. *PloS ONE*, *9*(1), e85207. https://doi.org/10.1371/journal.pone.0085207

Winsor, E. (2004). Book review of Handbook of chromosomal syndromes. *American Journal of Medical Genetics, 12*(4a), 105-106. https://doi.org/10.1002/ajmg.a.20335

Course Schedule

Week	Topic	Readings	Activities
1	History, theory, and applied directions including the field of child development, historical foundations, and basic issues related to chromosome deletions	McKinlay et al., 2012, Ch. 1 Winsor, 2003	Discussion on prior knowledge/experience regarding chromosome deletion Review class expectations
2	Biological foundations, genetic foundations	McKinlay et al., 2012, Ch. 2-3 He et al., 2014	**Discussion Prompt**: What insights do chromosomal deletions and rare syndromes offer about the biological mechanisms underlying developmental delays and intellectual disabilities (He et al., 2014; Winsor, 2004)?
3	Understanding risk figures and autosomal reciprocal	McKinlay et al., 2012, Ch. 4-5	Bring one article on a chromosome deletion to class to share with your peers.

Week	Topic	Readings	Activities
	translocations, impact on childbirth once a family understands they will have a child with a chromosome abnormality, birth complications; a look ahead to heredity, environment, and behavior; translocations.		Share 3-5 facts about the selected chromosome deletion
4	Translocations, sex chromosomes, fissions, isochromosomes. Understanding complications and how it impacts the lifestyle of the families after birth	McKinlay et al., 2012, Ch. 6-8	Using the selected article from week 3, create a short, 5 question quiz (multiple choice, fill in the blank, true/false)
5	Inversions, insertions, and autosomal ring chromosomes	McKinlay et al., 2012, Ch. 9-11	**Discussion Prompts**: • How do structural chromosomal changes—such as inversions and insertions—contribute to developmental delay and intellectual disability? • What are autosomal ring chromosomes, and how do they disrupt gene expression and neurological development?
6	Arrangement and rearrangement of complex, prenatal sex and parental autosomal aneuploidy	McKinlay et al., 2012, Ch. 12-14	Students will reach out to a local or school club organization that works with children with disabilities, specifically

Week	Topic	Readings	Activities
	chromosomes. knowledge of other chromosome abnormalities		individuals with chromosome deletions to volunteer at least once during the semester. Picture proof for documentation and a 2-page minimum double-spaced reflection regarding the experience to address these questions will be assessed: a) Briefly describe the organization you volunteered with and how did it change your view of working with children with chromosome deletions or disabilities? b) What is something you noticed while interacting with the children or adults? c) When did you feel most useful? d) What skills did you learn, and where can the skills you learned while volunteering be used in the future with this population? e) What was the most rewarding and challenging aspect about volunteering and working with

Week	Topic	Readings	Activities
			children and families who have chromosome deletions? Alternatives, such as in the case of a global pandemic, will be determined by discretion of the instructor of record.
7	Fragile X syndrome, childhood development with individuals diagnosed with variant chromosomal syndromes. Understandings of development with chromosome abnormalities and how it impacts the lifestyle	McKinlay et al., 2012, Ch. 15-17 https://documentaryheaven.com/cant-stop-eating/ https://topdocumentaryfilms.com/the-ghost-in-our-genes/	Students will locate and watch a documentary, episode, or podcast regarding chromosome deletion. A one-page minimum, double-spaced reflection will be required (provide link to documentary, episode or podcast).
8	Early intervention and intellectual development, advocacy in the community and how to support parents and - families who have children with chromosome abnormalities, emphasis on Down Syndrome	McKinlay et al., 2012, Ch. 18 Gilmore, 2017	**Myths vs. facts interview**
9	Impact on chromosomal disorders and sex development, instability syndrome	McKinlay et al., 2012, Ch. 19-20 Carruth et al., 2002	In-Class Activity: Role Play **Role-Play Activity: Genetic Counseling Session Simulation** **Objective:** Students will simulate a genetic

Week	Topic	Readings	Activities
			counseling session to explore how structural chromosomal abnormalities (inversions, insertions, ring chromosomes) are communicated to families and how they relate to neurodevelopmental outcomes. **Roles:** 1. **Genetic Counselor** – A graduate student trained in genetics, responsible for explaining the chromosomal findings and their implications. 2. **Parent(s)** – A student or pair of students portraying caregivers of a child recently diagnosed with a rare chromosomal disorder. 3. **Developmental Specialist** – A student who provides insight into potential developmental delays, intellectual disabilities, and intervention strategies. 4. *(Optional)* **Medical Geneticist** – A student who can elaborate on the

Week	Topic	Readings	Activities
			biological mechanisms behind the chromosomal abnormality.
			Scenario Setup: • The child has been diagnosed with a **ring chromosome 15** and shows signs of developmental delay. • Genetic testing also revealed a **chromosomal insertion** on chromosome 7. • The family is seeking clarity on what this means for their child's development and future.
			Instructions: 1. Each group receives a **brief case summary** and relevant background from the readings (e.g., Gilmore, He et al., Winsor). 2. Students prepare for their roles using the readings to guide their explanations and responses.

Week	Topic	Readings	Activities
			3. Role-play the session for 10–15 minutes. 4. After the role-play, groups reflect on: o What communication strategies were effective? o What challenges arose in explaining complex genetic concepts? o How did the team balance scientific accuracy with empathy?
10	Imprinting disorders, disorders associated with genomic imprinting, genomic imprinting	McKinlay et al., 2012, Ch. 21-22	Presentation planning
11	Learning how to support families who have infertility, pregnancy loss; understanding the impact for reproductive failure on families	McKinlay et al., 2012, Ch. 23	**Presentations**
12	Prenatal diagnoses: understanding counseling and screening measurements, diagnostic assessments and	McKinlay et al., 2012, Ch. 24-25	**Presentations** Write a reflection (2-3 pages) about key events and information learned during

Week	Topic	Readings	Activities
	procedures, understanding the family's role with professionals		one presentation and how the audience was impacted.
13	Development of genetic diagnoses, gender stereotypes, influences on genetic diagnoses. Role of social media in play for advocacy for families and children with chromosome abnormalities	McKinlay et al., 2012, Ch. 26	**Awareness social media post or email**
14	Origins and functions of the family, family as a social system, socialization within the family, family lifestyles and transitions, vulnerable families, child maltreatment. Understanding access families have, supports and therapy services available	McKinlay et al., 2012, Ch. 27	**Case study**
15	Wrap-up of the course readings and family structure. Understanding damage from exposure to extrinsic agents	McKinlay et al., 2012, Ch. 28	

Instructions for Assignments

1. **Myths vs. Facts Interview (week 8)**

   Students will be given a myths vs fact "cheat sheet" where the student will be instructed to find one person to interview about common myths regarding chromosome deletions and abnormalities. Discuss the Audio/Video release required for interview. Students will video or audio record the interview and submit the video or audio file. This is targeted to be a fun and interactive piece in order to help test the students' skills, and to advocate and promote awareness about chromosome deletions. Students will be encouraged to interview someone who is not in the course (e.g., significant other, family member, friend) to prevent bias.

2. **Presentations (weeks 10-12)**

   Students will create and present a 10-12 minute presentation on the assigned chromosome deletion. Students will have the option of doing the presentation individually, or as a group. There will be one week dedicated to planning.

3. **Awareness Social Media Post or Email (week 13)**

   Students will be instructed to create a social media post (Twitter, Facebook, Instagram, TikTok) -if they have one—about chromosome deletion awareness. Turn in a screen shot of your social media post.

   Alternative: Create an email with visuals to send to 10 people (that are not family) to promote what chromosome deletion is and the impacts on individuals and families.

4. **Case Study (week 14)**

   Students will be given a case study and instructed to reflect on it. Describe what you would do if you were in the family's shoes and trying to provide accommodations and accessibility for their family. What would you want the SLP/PT/OT/Psychologist and/or medical doctor or primary care provider want to know? Discuss collaboration, including barriers and successes.

Course Grading

Assignments are due by 11:59pm on the indicated date. Late assignments without previous written approval of the instructor will incur a 50% penalty for the first time and lose all points beyond. This instructor and learners are required to adhere to the University's Academic Integrity policy. Any plagiarism will not be tolerated and referred to the Academic Integrity Office. The learner will be given an "F" in the course and be recommended to the Student Conduct Office.

Learners will be evaluated based upon the assignments described below. The plus/minus grading system and scale is as follows:

Letter Grade	Grade Range
A	93 and above
A-	90-92
B+	88-89
B	83-87
B-	80-82
C+	78-79
C	73-77
C-	70-72
D	60-69
F	59 and below

Component	Weight for final grade
Reflections (readings, experiences)	20%
Discussions	20%
Presentations	30%
Myths vs. facts interview	5%
Awareness social media post or email	5%
Case study	20%

Resources

Resources to supplement the Syllabus:

- CEC Initial Practice-Based Professional Preparation Standards for EI/ECSE (2020)
- Center for Parent Information and Resources
- Early Childhood Recommended Practices Modules (Module 1)
- Learn the Signs. Act Early
- Understanding Rare Chromosome and Gene Disorders
- Ip36 Deletion
- National Center for Advancing Translational Sciences
- Prader-Willi Syndrome
- Angelman Syndrome
- 22q11.2 deletion syndrome
- 5q deletions: Current knowledge and future directions
- Professional Standards and Competencies for Early Childhood Educators
- Cognitive and Behavior profiles

Journals in EI/ECSE

Below are examples of journals that publish topics about EI/ECSE. Faculty may want to explain how to use articles to support practice and to examine current research. Clarify the difference between practitioner-based journals and research-based journals.

- *Exceptional Parent Magazine*
- *Infants and Young Children*
- *International Journal of Early Childhood Special Education*
- *Intervention in School and Clinic*
- *Journal of Early Hearing Detection and Intervention*
- *Journal of Early Intervention*
- *Journal of Special Education Technology*
- *Rural Special Education Quarterly*
- *Teaching Exceptional Children*
- *Topics in Early Childhood Special Education*
- *Young Children*
- *Young Exceptional Children*

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