ECiDC Early Childhood Intervention Doctoral Consortium

Creating Leaders in Intensive Interventions for Infants, Young Children and Their Families

Etiology and Characteristics of Infants and Young Children with Disabilities

Fragile X Syndrome

Sample Syllabus

Course Description

This course is designed to provide information on supporting developmentally and individualized care to infants and young children with Fragile X syndrome needs. Students will learn about the early identification process and subsequent support needs for children diagnosed with Fragile X. As well as interventions to support developmental progress for young children and their families.

Required Texts

Hagerman. R. J., & Hagerman. H. P. (2002). *Fragile X syndrome—Diagnosis, treatment, and research* (3rd ed.). John Hopkins University Press. https://doi.org/10.56021/97380801868436

Course Learning Objectives

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

1. Explain the importance of the newborn's behavioral language and its implications for care delivery.
2. Identify the evidence for brain protection based on behavioral cue-based, individualized care.
3. Explain the importance of system-wide change required for consistent brain protective, individualized, behavior-based, family-integrated care.
4. Describe newborn screening (NBS) and identify challenges currently facing NBS.
5. Describe Fragile X syndrome as a prototype of some of these challenges.
6. Describe Early Check, a newborn screening research project.
7. Raise questions about the role of early intervention in a new world of medicalization of treatment.
8. Justify the importance of newborn screening for developmental disabilities and delays for early intervention services.

Readings

Bagni, C., Tassone, F., Neri, G., & Hagerman, R. (2012). Fragile X syndrome: causes, diagnosis, mechanisms, and therapeutics. *The Journal of clinical investigation*, *122*(12), 4314-4322**.**

Bailey, D. B., Hebbeler, K., Olmsted, M. G., Raspa, M., & Bruder, M. B. (2008). Measuring family outcomes: Considerations for large-scale data collection in early intervention. *Infants & Young Children*, *21*(3), 194-206.

Hatton, D. D., Bailey Jr, D. B., Roberts, J. P., Skinner, M., Mayhew, L., Clark, R. D., ... & Roberts, J. E. (2000). Early intervention services for young boys with fragile X syndrome. Journal of Early Intervention, 23(4), 235-251.

Hardiman, R. L., & McGill, P. (2018). How common are challenging behaviours amongst individuals with fragile X syndrome? A systematic review. *Research in developmental disabilities*, *76*, 99-109.

Mirrett, P. L., Roberts, J. E., & Price, J. (2003). Early intervention practices and communication intervention strategies for young males with fragile X syndrome. *Language, Speech, and Hearing Services in Schools*, *34*(4), 320-331.

Tassone, F. (2014). Newborn screening for fragile X syndrome. *JAMA neurology*, *71*(3), 355-359.

Course Schedule

Week	Topics	Readings	Activities
1	Introduction of class	Review syllabus, assignments	• Syllabus discussion on Attendance, Grades, Textbook requirement, Communication, etc. • Discussion on prior knowledge/experience regarding the topic
2	The Physical and Behavioral Phenotype	Hagerman & Hagerman, (2002). Chapter 1 Bagni et al., 2012 Tassone, 2014	**Discussion Prompt**: Based on Hagerman & Hagerman's Ch 1, critically examine the interplay between the physical and behavioral phenotypes associated with the condition. How do the molecular and neurobiological underpinnings of Fragile X contribute to the observable traits in affected individuals? In your response, consider the following: • The role of FMR1 gene mutation and its impact on FMRP production. • Common physical characteristics (e.g., facial

			features, connective tissue involvement) and how they may vary across age and gender. • Behavioral manifestations such as anxiety, hyperactivity, and social avoidance, and their potential neurodevelopmental origins. • How understanding the phenotype informs diagnosis, intervention strategies, and future research directions.
3	The Molecular Biology of the Fragile X Mutation	Hagerman & Hagerman, (2002). Chapter 2	• **Discussion Prompt**: Chapter 2 outlines the molecular basis of Fragile X syndrome, focusing on the FMR1 gene mutation and its downstream effects. How does this molecular mechanism contribute to the neurodevelopmental and behavioral phenotype observed in individuals with Fragile X syndrome? • In your response, consider: • How the absence of FMRP affects synaptic development and plasticity. • Implications for diagnosis, carrier screening, and therapeutic strategies.

4	Review the presentation and come up with a glossary list.	RTI International: Lessons learned from newborn screening for Fragile X Syndrome	• Glossary list: Create a glossary including term, definition and reference/resource.
4	Epidemiology - Stephanie Sherman	Hagerman & Hagerman, (2002). Chapter 3	
5	FMR1 Protein Studies and Animal Model for Fragile X Syndrome	Hagerman & Hagerman, (2002). Chapter 4	**Discussion Prompt**: Based on Bagni et al. (2012), discuss how dysregulation of synaptic plasticity and protein synthesis contributes to the cognitive and behavioral phenotype of Fragile X syndrome. What are the implications of these mechanisms for developing targeted therapeutics, and how might future treatments address both molecular and behavioral symptoms?

6	Brain Structure and Functions of FMR1 Protein - Scott Irwin, Roberto Galvez, Ivan Jeanne Weiler, Andrea Beckel-Mitchener, and William Greenough	Hagerman & Hagerman, (2002). Chapter 5	• **In-class Activity: Concept Mapping**
			  • Instructions: Students work in small groups to create a concept map that links:
			  ○ FMR1 gene mutation
			  ○ FMRP function
			  ○ Synaptic plasticity
			  ○ Dendritic spine morphology
			  ○ Affected brain regions (e.g., hippocampus,

			cerebellum, prefrontal cortex) ○ Behavioral outcomes (e.g., learning deficits, anxiety, sensory hypersensitivity) • Materials: Large paper or digital whiteboard tools (e.g., Miro, Jamboard) • Objective: Each group presents their map and explains one key pathway from gene to behavior.
7	Review one of the learning modules	Learning Module: Understanding the basics of Fragile X Learning Module: Fragile X Syndrome Washington University	• Reflect and discuss three ideas/information from the multimedia document resource and post it. • Review & respond to at least two peers' work.
8	Infant Massage Therapy	Goldson (1999), Chapter 4 Bailey et al., 2008	• Discussion Prompt: Drawing from Bailey et al. (2008), evaluate the challenges and ethical considerations in measuring family outcomes in large-scale early intervention programs for children with Fragile X syndrome. How can data collection methods be

			designed to capture meaningful, culturally sensitive, and actionable insights?
9	The Treatment of Emotional and Behavioral Problems	Hagerman & Hagerman, (2002). Chapter 9 Hardiman & McGill, 2018	**State FXS report** **Discussion Prompt:** Based on Hardiman & McGill (2018), critically assess the prevalence and types of challenging behaviors in individuals with Fragile X syndrome. What are the implications for caregivers, educators, and clinicians, and how can behavior support plans be individualized to reduce risk and improve quality of life?
10	Vestibular Stimulation as a Neurodevelopmental Intervention with Preterm Infants	Goldson (1999). Chapter 5	• Discussion Prompt: Reflecting on Tassone (2014), debate the ethical, clinical, and logistical considerations of implementing newborn screening for Fragile X syndrome. What are the potential benefits and risks, and how might early identification influence intervention strategies and family planning?
11	An Integrated Approach to Intervention	Hagerman & Hagerman, (2002). Chapter 10 Hatton et al., 2000	**Role-Play: Interdisciplinary Team Meeting** **Activity:** Students take on roles (e.g., geneticist, speech-language pathologist, early intervention

			specialist, parent advocate) and simulate a team meeting to design a care plan for a child recently diagnosed with Fragile X.
12	Academic Interventions	Hagerman & Hagerman, (2002). Chapter 11 Mirrett et al., 2003	• **Online discussion**: Discuss how you can implement what you have learned from chapters 10&11. Look for resources to provide one example of a script on how and what you would explain to a parent of a child with FXS. Respond to two peers' posts.
13	FMRI Gene Expression and Prospects for Gene Therapy	Hagerman & Hagerman, (2002). Chapter 12	• Group Activity: **Debate: Newborn Screening Ethics** ○ Divide students into two groups to debate the implementation of newborn screening for Fragile X syndrome (Tassone, 2014). One side argues for universal screening; the other against it.
14	Review RTI International website	RTI International | Research Institute	• **Discussion Prompt**: Discuss three things you found that were interesting and will benefit your professional growth from the RTI International website.
15	Finals Week		**Policy Brief; Reflection**

Instructions for Assignments

1. **Policy Brief**

Title: *Policy Brief: Enhancing Early Intervention for Fragile X Syndrome*
Instructions: Write a 1–2 page policy brief that includes:

1. **Issue Overview**
    - Briefly describe Fragile X syndrome and its impact.
2. **Evidence Summary**
    - Use findings from Hatton et al. (2000) and Bailey et al. (2008).
3. **Recommendations**
    - Propose 2–3 actionable policy changes at the state-level.
    - Justify with data and anticipated outcomes based on the readings and course materials.
4. **Conclusion**
    - Summarize the importance of early intervention and family-centered care.

2. **Discussion Group Participation**

To prepare for the discussion sessions, students will be required to complete the readings leading up to the discussion session. Students will be asked to respond to the discussion prompt by Friday evening and give substantive feedback to 2 (or more) classmates by Sunday evening. Submit the discussion questions by 5 pm the day before the discussion. Students receive points for timely and in-depth discussion responses. Evidence of integrating reading and applying theory to practice is necessary to receive full points. Students will receive points based on the quality of feedback given to peers.

3. **Reflection**

Review and reflect on one of the learning modules on FXS.
Write a one-page reflection and post in Discussion. Follow the table of contents and reflect on each session. Discuss how you can apply and implement what you have learned from the materials. Provide one example of what you will use to communicate with a parent of an infant with FXS. Respond to two peers' posts.

4. **State FXS Report**

Identify credible sources (e.g. .gov, or journal articles) to collect information on your State's key FXS statistics (find the most recent year). For example, descriptive statistics on FXS and newborn screening. Share your statistics with 2 other people.

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
Discussions (including responses to peers)	25%
Presentations (in class/video, quizzes)	20%
Reflection	15%
State FXS report	20%

Resources

Resources to supplement the Syllabus:

- National Fragile X Foundation
- The Fragile X Society
- American Academy of Pediatrics
- Learning Module: Understanding the basics of Fragile X
- Learning Module: Fragile X Syndrome Washington University
- Fragile X Syndrome Medical Home Portal

Journals in Fragile X and 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.

- *American Journal of Medical Genetics*
- *American Journal of Intellectual and Developmental Disabilities*
- *Child Development*
- *Exceptional Parent Magazine*
- *Infants and Young Children*
- *International Journal of Early Childhood Special Education*
- *Journal of Early Intervention*
- *Journal of Developmental and Physical Disabilities*
- *Journal of Developmental & Behavioral Pediatrics*
- *Journal of Intellectual & Developmental Disability*
- *Journal of Neurodevelopmental Disorders*
- *Journal of Pediatrics*
- *Journal of Pediatric Nursing*
- *Journal of Special Education Technology*
- *Pediatric Neurology*
- *Pediatric Research*
- *Rural Special Education Quarterly*
- *Teaching Exceptional Children*
- *Topics in Early Childhood Special Education*
- *Young Children*
- *Young Exceptional Children*

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