

Etiology and Characteristics of Infants and Young Children with Disabilities

Rett Syndrome

Sample Syllabus

Course Description

This course is designed to increase students' comprehensive understanding of Rett syndrome. This course will explore the biological, psychological, and social implications of Rett syndrome, centering the individual and family systems. Students are expected to gain an understanding of the diagnostic process and genetic etiology in efforts to understand the underlying biologic factors at play. Students will learn bodily systems that are affected in this syndrome and the functional implications, with attention given to the way the bodily systems interact with each other.

Course Learning Objectives

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

1. Recognize diagnostic characteristics of Rett syndrome.
2. Discuss impacts on body systems and functions and comorbidities/co-occurring conditions which are observed with Rett syndrome.
3. Identify evidence based therapeutic interventions for activities such as mobility, activities of daily living and communication.
4. Identify parent perceptions and support resources.

Readings

- Barnes, K. V., Coughlin, F. R., O'Leary, H. M., Bruck, N., Bazin, G. A., Beinecke, E. B., Walco, A. C., Cantwell, N. G., & Kaufmann, W. E. (2015). Anxiety-like behavior in Rett syndrome: Characteristics and assessment by anxiety scales. *Journal of Neurodevelopmental Disorders*, 7, (30). Epub. <https://doi.org/10.1186/s11689-015-9127-4>
- Bartolotta, T. E., Zipp, G. P., Simpkins, S. D., & Glazewski, B. (2011). Communication skills in girls with Rett syndrome. *Focus on Autism and Other Developmental Disabilities*, 26(1), 15–24. <https://doi.org/10.1177/1088357610380042>
- Borloz, E., Villard, L., & Roux, J. C. (2021). Rett syndrome: Think outside the (skull) box. *Faculty Reviews* 10(59). <https://doi.org/10.12703/r/10-59>
- Downs, J., Parkinson, S., Ranelli, S., Leonard, H., Diener, P., & Lotan, M. (2014). Perspectives on hand function in girls and women with Rett syndrome. *Developmental Neurorehabilitation*, 17(3), 210-217. <https://doi.org/10.3109/17518423.2012.758183>

- Fu, C., Armstrong, D., Marsh, E., Lieberman, D., Motil, K., Witt, R., Standridge, S., Lane, J., Dinkel, T., Jones, M., Hale, K., Suter, B., Glaze, D., Neul, J., Percy, A., & Benke, T. (2020). Multisystem comorbidities in classic Rett syndrome: A scoping review. *BMJ Paediatrics Open*, 4, e000731. <https://doi.org/10.1136/bmjpo-2020-000731>
- Isaacs, J. S., Murdock, M., Lane, J., & Percy, A. K. (2003). Eating difficulties in girls with Rett syndrome compared with other developmental disabilities. *Journal of the American Dietetic Association*, 103(2), 224–230. <https://doi.org/10.1053/jada.2003.50026>
- Leonard, H., Ravikumara, M., Baikie, G., Naseem, N., Ellaway, C., Percy, A., Abraham, S., Geerts, S., Lane, J., Jones, M., Bathgate, K., Downs, J., & Telethon Institute for Child Health Research (2013). Assessment and management of nutrition and growth in Rett syndrome. *Journal of Pediatric Gastroenterology and Nutrition*, 57(4), 451–460. <https://doi.org/10.1097/MPG.0b013e31829e0b65>
- Neul, J. L., Kaufmann, W. E., Glaze, D. G., Christodoulou, J., Clarke, A. J., Bahi-Buisson, N., Leonard, H., Bailey, M. E. S., Schanen, N. C., Zappella, M., Renieri, A., Huppke, P., Percy, A. K., & RettSearch Consortium. (2010). Rett syndrome: Revised diagnostic criteria and nomenclature. *Annals of Neurology*, 68(6), 944-950. <https://doi.org/10.1002/ana.22124>
- Oddy, W. H., Webb, K. G., Gordon, B., Thompson, S. M., Reilly, S., Fyfe, S. D., Young, D., Anderson, A. M., & Leonard, H. (2007). Feeding experiences and growth status in a Rett syndrome population. *Journal of Pediatric Gastroenterology and Nutrition*, 45(5), 582-590. <https://doi.org/10.1097/MPG.0b013e318073cbf7>
- Smeets, E. E. J., Pelc, K., & Dan, B. (2011). Rett syndrome. *Molecular Syndromology*, 2, 113-127. <https://doi.org/10.1159/000337637>
- Townend, G. S., Bartolotta, T. E., Urbanowicz, A., Wandin, H., & Curfs, L. M. G. (2020). Development of consensus-based guidelines for managing communication of individuals with Rett syndrome. *Augmentative and Alternative Communication*, 36(2), 71-81. <https://doi.org/10.1080/07434618.2020.1785009>

Accommodations

Section 504 of the Rehabilitation Act of 1973 and the American with Disabilities Act of 1990 require the institution to provide academic adjustments or accommodations for students with documented disabilities. Students seeking academic adjustments or accommodations must self-identify with the Coordinator of Services for Students with Disabilities on the appropriate campus.

Attendance

Since many of the classes involve activities, videotapes, discussion, etc., regular attendance is vital to gain maximum benefit. Because this is a 15-week course, each class member needs to

make every effort to attend each class. Anyone who misses more than one class will lose 5 points for each class missed after the first absence. Significant tardiness or early departure beyond 15 minutes will count as an absence. If you know ahead of time you will not be in class, please contact the instructor the day before the class.

Communicating with the Instructor

All correspondence from the instructor to the students by way of announcements, updates, assignments, and so forth, will be communicated via school email or online learning management system if not verbally in class. It is the student's responsibility to ensure their university e-mail is correct. Failure to do so may result in missing important information that could negatively impact your grade. Please be mindful of professionalism.

Course Outline

The following schedule is subject to change. For any course syllabi posted prior to the beginning of the semester, the course instructor reserves the right to make changes prior to or during the semester. The course instructor will notify students, via e-mail or verbal announcements when changes are made in the requirements and/or grading of this course. ***The course instructor reserves the right to revise the schedule/assignments if needed.***

Course Schedule

Week	Topic	Reading	Activities
1	Background and overview of Rett syndrome		<ul style="list-style-type: none"> Find one research article regarding the genetic make-up of Rett syndrome
2	Genetic diagnostic process	Neul et al., 2010 International Rett Syndrome Foundation: About Rett Syndrome	Discussion Prompt: Neul et al. (2010) revised the diagnostic criteria for Rett Syndrome to reflect a broader and more nuanced understanding of its clinical presentation. As future educators, how might these changes influence your approach to identifying, supporting, and advocating for students with Rett Syndrome in inclusive educational settings?
3	Early neuromotor signs	Smeets et al., 2012 Barnes et al., 2015	<ul style="list-style-type: none"> Guest presentation: Neurodevelopmental Pediatrician
4	Early intervention	When in Doubt, Reach Out: Teaming Strategies for Inclusive Early Childhood Settings NAEYC	In-Class Activity: Role Play Name: Lily Age: 4 years Diagnosis: Rett Syndrome Strengths: Enjoys music, responds to visual cues, smiles and vocalizes when engaged Challenges: Limited verbal communication, hand stereotypies, occasional seizures, difficulty with motor planning Assign students to play the following team members: <ul style="list-style-type: none"> Early Intervention Provider (EI) Preschool Teacher Speech-Language Pathologist (SLP) Occupational Therapist (OT) Family Member (Parent or Guardian) Program Administrator or Inclusion Specialist

Week	Topic	Reading	Activities
			<p>The team is meeting for Lily's Individualized Family Service Plan (IFSP) transition meeting as she prepares to enter a preschool classroom. The goal is to:</p> <ul style="list-style-type: none"> • Review Lily's current needs and strengths • Discuss seizure management and communication strategies • Plan inclusive classroom supports • Ensure family voice is central in decision-making <ul style="list-style-type: none"> • Identify the members of an EI team • Brief presentation of roles and responsibilities of team members
5	School evaluations and programming	School - International Rett Syndrome Foundation	<ul style="list-style-type: none"> • Identify benefits and limitations of classroom environments and interventions
6	Body systems	<p>Borloz et al., 2021</p> <p>Downs et al., 2014</p>	<ul style="list-style-type: none"> • Evidence based practice studies • Review body systems that can be impacted by Rett syndrome
7	Adaptive equipment	<p>Top 5 Equipment Categories and Recommendations for Rett Syndrome</p> <p>Rett syndrome aids and adaptations</p>	<p>Discussion Prompt: The websites outlines key equipment categories—such as seating, mobility, and communication devices—that can significantly improve quality of life for children with Rett Syndrome. As future early childhood educators or interventionists, how can you collaborate with families and interdisciplinary teams to ensure that assistive equipment is integrated meaningfully into inclusive classroom environments?</p> <ul style="list-style-type: none"> • Positioning, mobility, splinting

Week	Topic	Reading	Activities
8	Feeding and nutrition	Isaacs et al., 2003; Leonard et al., 2013; Oddy et al., 2007 Rett Syndrome News: Rett Syndrome and Diet	Discussion Prompt: Feeding challenges and growth concerns are common in children with Rett Syndrome, as highlighted by readings this week. How can early childhood educators and interventionists collaborate with families and healthcare providers to support safe, responsive, and inclusive mealtime experiences in early learning settings?
9	Communication	Bartolotta et al., 2011 National Joint Committee for the Communication Needs of Persons with Severe Disabilities (2016): Communication Bill of Rights	Discussion Prompt: Bartolotta et al. (2011) highlight the unique and often subtle communication behaviors of girls with Rett Syndrome, including nonverbal cues, vocalizations, and eye gaze. As future educators and interventionists, how can you recognize and respond to these communication attempts in inclusive early childhood settings to promote engagement, learning, and social connection?
10	Literacy instruction	Center for Literacy and Disability Studies (unc.edu)	<ul style="list-style-type: none"> Discuss models of literacy instruction, motor/visual adaptations
11	AAC evaluations and processes	Townsend et al., 2020	<ul style="list-style-type: none"> Discuss AAC process, funding opportunities, and special considerations for RTT <p>Discussion Prompt: Townend et al. (2020) emphasize the importance of individualized, multimodal communication strategies for individuals with Rett Syndrome. As future educators and interventionists, how can you apply these consensus-based guidelines to foster meaningful communication in inclusive early childhood settings? Also, what does “presumed competence” mean, and how does it shape your approach to communication?</p>
12	New diagnosis		<ul style="list-style-type: none"> Group Activity: New Diagnosis

Week	Topic	Reading	Activities
			<ul style="list-style-type: none"> Discuss family perspectives, navigating emotions, finances, medical concerns, school decisions that follow a new diagnosis of RTT
13	Case study		<ul style="list-style-type: none"> Case study <ul style="list-style-type: none"> Review case study in groups and respond to discussion questions 1-2 page reflection of Group Activity due
14	Parent perspectives		<ul style="list-style-type: none"> Guest presentation(s): Family members
15	Discipline-specific supports; lifespan development perspective		<ul style="list-style-type: none"> Presentation: Professional Perspectives

Instructions for Assignments

1. Group Activity: New Diagnosis:

Understand the family perspectives of navigating emotions, finances, medical concerns, school decisions, etc. following a new diagnosis of Rett syndrome.

Directions:

1. Work with your group to find resources describing how to support families through a new medical diagnosis.
2. Practice active listening and motivational interviewing, role playing a provider and a family member in the context of giving/receiving a new diagnosis.
3. Identify 3 resources that exist in the local area to provide a family member regarding services or supports for their child.
4. Turn in a 1-2 page individual reflection based on your group discussion (due at the beginning of the next class period).

2. Presentation: Professional Perspectives:

Synthesize the information learned throughout the course to develop and share a 10-minute presentation from the perspective of your individual discipline.

Directions:

1. Identify an opportunity that your discipline could work with an individual with Rett syndrome and/or family.
2. Create a slide show presentation about intervention or support from your discipline.
3. Include practice settings where you would work with the families.
4. Create a handout of resources that are specific to your discipline to share with your classmates.

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
Professional conduct (preparation for class, in-class discussion)	20%
In-class group work/practice activities (EBP practice, reviewing documents, reflections)	30%
New Diagnosis Presentation	25%
Professional Perspectives Presentation	25%

Resources

Resources to supplement the Syllabus:

- [International Rett Syndrome Foundation](#)
- [Rett Syndrome Research Trust](#)

Journals in EI/ECSE

Below are examples of journals that publish topics about motor disabilities and 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.

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| • <i>Exceptional Parent Magazine</i> | • <i>Journal of Special Education Technology</i> |
| • <i>Infants and Young Children</i> | • <i>Rural Special Education Quarterly</i> |
| • <i>International Journal of Early Childhood Special Education</i> | • <i>Teaching Exceptional Children</i> |
| • <i>Intervention in School and Clinic</i> | • <i>Topics in Early Childhood Special Education</i> |
| • <i>Journal of Early Hearing Detection and Intervention</i> | • <i>Young Children</i> |
| • <i>Journal of Early Intervention</i> | • <i>Young Exceptional Children</i> |
| • <i>Journal of Pediatrics</i> | |

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