

Etiology and Characteristics of Infants and Young Children with Disabilities

Joubert Syndrome and Related Disorders

Sample Syllabus

Course Description

This course is designed to provide students with knowledge and skills to work with children who are diagnosed with Joubert syndrome or a related disorder. This course will focus on content knowledge surrounding the syndrome to build a foundation of understanding. Students will learn about information pertaining to the diagnosis process and strategies in working with young children diagnosed with Joubert syndrome will be discussed. Future implications for research will also be addressed.

Required Text

Tompkins, K. (2014). *Purple stew: A journey to Joubert syndrome*. Lulu Press. (ebook)
<http://www.ktompkins.com/purple-stew>

Course Learning Objectives

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

1. Explain the main characteristics of Joubert syndrome.
2. Identify strategies that can be used when working with children diagnosed with Joubert syndrome.
3. Develop Individualized Family Service Plan goals appropriate for an identified child with Joubert syndrome.

Readings

Alam, S., Khatoon, F., & Khan, N. (2021). Joubert syndrome: A case report. *Bulletin of Faculty of Physical Therapy*, 26(18). <https://doi.org/10.1186/s43161-021-00039-7>

Bachmann-Gagescu, R., Dempsey, J. C., Bulgheroni, S., Chen, M., D'Arrigo, S., Glass, I. A., Heller, T., Heon, E., Hildebrandt, F., Joshi, N., Knutzen, D., Kroes, H. Y., Mack, S. H., Nuovo, S., Parisi, M. A., Snow, J., Summers, A. C., Symons, J. M., Zein, W. M., Boltshauser, E., ... Doherty, D. (2019). Healthcare recommendations for Joubert syndrome. *American Journal of Medical Genetics Part A*, 182(1), 229-249.
<https://doi.org/10.1002/ajmg.a.61399>

- Brancati, F., Dallapiccola, B., & Valente, E. M. (2010). Joubert syndrome and related disorders. *Orphanet Journal of Rare Diseases*, 5(20). <https://doi.org/10.1186/1750-1172-5-20>
- Dempsey, J. C., Phelps, I. G, Bachmann-Gagescu, R., Glass, I. A., Tully, H. M., & Doherty, D. (2017). Mortality in Joubert syndrome. *American Journal of Medical Genetics Part A*, 173(5), 1237-1424. <https://doi.org/10.1002/ajmg.a.38158>
- Doherty D. (2009). Joubert syndrome: Insights into brain development, cilium biology, and complex disease. *Seminars in Pediatric Neurology*, 16(3), 143–154. <https://doi.org/10.1016/j.spen.2009.06.002>
- Doherty, D., Millen, K. J., & Barkovich, A. J. (2013). Midbrain-hindbrain malformations: Advances in clinical diagnoses, imaging, and genetics. *The Lancet Neurology*, 12(4), 381-393. [https://doi.org/10.1016/S1474-4422\(13\)70024-3](https://doi.org/10.1016/S1474-4422(13)70024-3)
- Joubert, M., Eisenring, J. J., & Andermann, F. (1969). Familial agenesis of the cerebellar vermis: A syndrome of episodic hyperpnea, abnormal eye movements, ataxia, and retardation. *Neurology*, 19(9), 813-825. <https://doi.org/10.1212/WNL.19.9.813>
- Parisi, M. A., Doherty, D., Chance, P. F., & Glass, I. A. (2007). Joubert syndrome (and related disorders) (OMIM 213300). *European Journal of Human Genetics*, 15, 511-521. <https://doi.org/10.1038/sj.ejhg.5201648>
- Radha Rama Devi, A., Mohammad Naushad, S., & Lingappa, L. (2020). Clinical and molecular diagnosis of Joubert syndrome and related disorders. *Pediatric Neurology*, 106, 43-49. <https://doi.org/10.1016/j.pediatrneurol.2020.01.012>
- Steinlin, M., Schmid, M., Landau, K., & Boltshauser, E. (1997). Follow-up in children with Joubert syndrome. *Neuropediatrics*, 28(4), 204-211. <https://doi.org/10.1055/s-2007-973701>

Course Schedule

Week	Topic	Readings and Online Activities	In Class Activities and Assignments
1	Introductions & Overview	<i>Purple Stew</i> pages 1-8 What is JS?	Class Introductions; Class Discussion on Content
2	Characteristics of Joubert Syndrome	Doherty, 2009; Doherty et al., 2013 Parisi et al., 2007	Discussion Prompt: Drawing from Doherty (2009) and Doherty et al. (2013), discuss how disruptions in primary cilia function contribute to the

Week	Topic	Readings and Online Activities	In Class Activities and Assignments
			structural brain abnormalities observed in Joubert syndrome. How do advances in neuroimaging and genetic diagnostics enhance our understanding of these malformations, and what are the implications for clinical management and early intervention?
3	Characteristics (continued) & Diagnostic Process	<p><i>Purple Stew</i> pages 9-46</p> <p>Radha Rama Devi et al., 2020</p> <p>Joubert Syndrome Foundation: Words from our experts</p>	<p>Book Discussion; Class Discussion on Content</p> <p>Guided Reading Reflection:</p> <p>Each student writes a brief reflection (1–2 paragraphs) on the following prompt:</p> <p><i>How does the author’s journey through diagnosis reflect broader challenges faced by families navigating rare diseases like Joubert syndrome? What emotions, uncertainties, or turning points stood out to you?</i></p>
4	<p>Living with Joubert Syndrome</p> <ul style="list-style-type: none"> • Kidney disease • Liver disease • Respiratory abnormalities • Vision issues 	Bachman-Gagescu et al., 2019	Class Discussion on Content
5	Related Disorders	<p>UW: What is rhombencephalosynapsis?</p> <p>UW: What is pontine tegmental cap dysplasia?</p>	In-Class Group Activity: Related Disorders

Week	Topic	Readings and Online Activities	In Class Activities and Assignments
		UW: What is Chudley-McCullough syndrome?	
6	Developmental Indicators	<i>Purple Stew</i> 47-54	<p>Book Discussion; Class Discussion on Content</p> <p>In-Class Activity: Systems Mapping Activity Each group creates a visual map of the systems the author interacted with (e.g., medical, educational, therapeutic). Include:</p> <ul style="list-style-type: none"> • Points of access • Barriers encountered • Supportive individuals or institutions • Gaps in coordination or communication
7	Supports in Early Intervention	Resources by State	In-Class Lecture: Supports in EI; Class Discussion on Content
8	Collaboration and Teaming	Birth to Three Evidence Supported Teaming Alam et al., 2021	<p>In-Class Activity: In small groups, <i>considering the case report by Alam et al. (2021) on Joubert syndrome, discuss how interdisciplinary teaming can be applied to support infants and toddlers with rare neurodevelopmental disorders. What unique challenges arise when coordinating care for children with complex medical and developmental needs, and</i></p>

Week	Topic	Readings and Online Activities	In Class Activities and Assignments
			<p><i>how can early intervention teams ensure culturally responsive, evidence-based support for families?</i></p> <p>In your response, consider:</p> <ul style="list-style-type: none"> • The role of physical therapy and other disciplines in managing motor and developmental delays in Joubert syndrome. • How teaming practices can address diagnostic uncertainty and evolving needs. • Strategies for integrating family voice and priorities into service planning. • The importance of communication, role clarity, and shared goals in transdisciplinary care.
9	Case Study	Case study video Joubert case study	<p>In-Class Activity: Based on the video: Break into small groups and discuss:</p> <ul style="list-style-type: none"> • What were the most striking features of the child's presentation?

Week	Topic	Readings and Online Activities	In Class Activities and Assignments
			<ul style="list-style-type: none"> How did the family describe their journey through diagnosis and care? What interdisciplinary supports were evident or missing?
10	Supports in Education	“I am Isabella” Review of Education Supports	Discussion Prompt: What are the key components of effective educational planning for students with rare neurodevelopmental disorders, and how can individualized supports be balanced with inclusive practices?
11	Case Study Presentations		Case Study DUE Group Case Study Presentations
12	Case Study Presentations		Group Case Study Presentations
13	In-Class Group Work		Develop IFSP goals based on group case study. Class discussion on IFSP goals.
14	Research	Doherty et al., 2005 Dempsey et al., 2017	Class Discussion on Content
15	Group Summary Presentations		Copy of Presentation DUE

**The course syllabus is a general plan for the course; deviations announced to the class by the instructor(s) may be necessary. This syllabus is subject to change. **

Instructions for Assignments

1. Class Participation

Readings should be completed prior to each class session.

Classes will primarily consist of discussions and activities. Participation in the in-class activities and discussions will contribute to your participation grade.

Case Study

This is a group assignment. Develop a case study for a child with Joubert syndrome. Use information from class readings, discussions or based on professional experience. Create a 10-12 minute PowerPoint presentation to share with the class about your student.

Areas covered should include Medical and Developmental History, Family Priorities, Individual and Family Goals & Objectives, Embedded Routines.

Group Summary Presentations

Students will break up into groups and choose a topic from the course schedule to summarize at the end of the semester. Students will use knowledge from the readings, material presented, discussions, the case study and related materials to summarize and present to the class. Students should be prepared to present for 10-12 minutes on their topic. Copies of the presentations will be shared with the class.

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

Letter Grade	Grade Range
C	73-77
C-	70-72
D	60-69
F	59 and below

Component	Weight for final grade
Class participation/discussion	40%
Case study	30%
Group summary presentation	30%

Resources

Resources to supplement the Syllabus:

- [University of Washington's Hindbrain Malformation Research Program](#)
- [Joubert Syndrome & Related Disorders Foundation](#)
- [National Organization for Rare Disorders: Joubert Syndrome](#)

Journals in Joubert Syndrome

Below are examples of journals that publish topics about Joubert syndrome. 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: Seminars in Medical Genetics*
- *Cureus Journal of Medical Science*
- *GeneReviews*
- *Journal of Medical Cases*
- *Journal of Pediatric Neurosciences*
- *Medicine*
- *Neuropediatrics*
- *Orphanet Journal of Rare Diseases*
- *Pediatric Neurology*
- *Seminars in Pediatric Neurology*
- *The Lancet Neurology*

Journals in EI/ECSE

While the above journals are useful in healthcare and case studies, the following journals are appropriate for early intervention/early childhood special education practitioners. The below journals may have useful information in working with children who have physical and developmental characteristics associated with Joubert syndrome. 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*

This is a product of the Early Childhood Intervention Doctoral Consortium (ECiDC), a project of the [A.J. Pappanikou Center for Excellence in Developmental Disabilities](#) at [UConn Health](#). The Center is funded through cooperative agreement number H325H190004 from the [Office of Special Education Programs](#), U.S. Department of Education. Materials and opinions expressed herein do not necessarily represent the Department of Education's position or policy.