AIR-P LEND Seminar Series:
Genetics

Purpose: This document corresponds with the Genetics video in the AIR-P LEND Seminar Series. It outlines the video's content and provides suggestions on how to use this resource.

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Research Node: Genetics

Summary: In this video, the AIR-P Genetics Research Node leaders will discuss topics related to genetic testing and autism, including: (1) barriers to genetic testing access; (2) types of genetic testing and their clinical utility in medical practice; (3) awareness about genetic risk and clinical knowledge for medical providers; and (4) broad implementation and ethical implications of genetic testing.

Learning objectives:

- Understand why genetic testing for autistic individuals and their families is important
- Understand the importance of taking a lifespan approach when considering genetic testing for autistic individuals
- Understand important ethical considerations related to genetic testing for autistic individuals

Outline:

Section of Video	Content Outline & Talking Points	Presenter(s)	Time allotted*

General Overview	1. Welcome
	   a. Node leaders introduce AND describe themselves (for those with low vision or who are blind).
	2. Context
	   a. Give context on AIR-P.
	      i. Autism Intervention Research Network on Physical Health | AIR-P Network (ucla.edu)
	      ii. AUCD - Autism Intervention Research Network on Physical Health (AIR-P)
	      iii. Network of researchers
	   b. Describe the video's topic.
	3. Overview of Node
	   a. Topic 1: Diagnostic journey barriers to genetic testing access
	      i. (Costs, knowledge about options, etc.).
	   b. Topic 2: Types of genetic testing and their clinical utility in medical practice.
		Facilitator:
		Gabriel Dichter
		Professor
		UNC-Chapel Hill Psychiatry and Carolina Institute for Developmental Disabilities; NC LEND Faculty
		
		Node Leader:
		Julian A Martinez, MD, PhD
		Associate Professor, Human Genetics, Pediatrics and Psychiatry
		UCLA
			2-5 min

	c. Topic 3: General population literacy awareness about genetic risk and clinical knowledge and genetic counseling gap for medical providers
	     i. (Informed consent, interpretation, risks and limitations of genetic testing).
	d. Topic 4: Broad implementation and ethical implications of genetic testing
	     i. (Vulnerable populations, stigmatization, privacy and confidentiality, family issues, obligation to disclose, premature marketing, reliable interpretation).
	4. Learning objectives (see above)		

How is AIR-P addressing research gaps in this node?	1. Topic 1: What proportion of autistic people find answers from genetic tests? Which specific demographic or clinical factors (e.g., sex, age of diagnosis, "simplex" vs. "multiplex" autism, ID status, dysmorphology, co-occurring conditions, etc.) best predict a high likelihood of a positive genetic result? 2. Topic 2: What is the clinical utility, measurable benefit, and impact of positive genetic tests on clinical care (psychological, plan and cost of care, early diagnosis, prognosis)? Based on these benefits, how "cost-effective" are these tests when applied at a population level? 3. Topic 3: What interventions can improve family history self-knowledge, natural history and phenotypic spectrum of rare conditions that present with neurodevelopmental differences, and access to underserved populations? 4. Topic 4: Can practice guidelines be developed that ensure ethical research and clinical use and implementation of genetic data analysis?	<u>Node leader:</u> **Julian A Martinez, MD, PhD** Associate Professor, Human Genetics, Pediatrics and Psychiatry UCLA	3-5 min

Panel Discussion	1. Introductions
	   a. CRE leads/co-leads and panel discussants introduce AND describe themselves (see example above).
	2. Structure/facilitator notes:
	   a. Topic 1: Parent vs provider vs personal perspectives on genetic testing.
	   b. Topic 2: Should every family with an autistic child (or adult) be offered genetic testing?
	   c. Topic 3: What are the ethical concerns associated with genetic data and what can be done to address them?
	   d. Topic 4: How can we improve access to genetic testing?
	3. Closing summary
		<u>Facilitator:</u>
		Gabriel Dichter
		Professor
		UNC-Chapel Hill Psychiatry and Carolina Institute for Developmental Disabilities; NC LEND Faculty
		
		<u>Panel:</u>
		
		AUTISTIC SELF-ADVOCATE:
		Zachary J Williams, MD/PhD Candidate, Neuroscience and Hearing & Speech Sciences
			25-35 min

| | | Vanderbilt University School of Medicine

CAREGIVER:
Felicia Williams
NC LEND Graduate, Certified Community Health Worker, and Educator
Connection and Resource Specialist for the Autism Society of NC

PROFESSIONAL:
Julian A Martinez, MD, PhD
Associate Professor, Human Genetics, Pediatrics and Psychiatry
UCLA | |
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Tips to Use this Resource:

- Recruit a local autistic self-advocate to be present when you expect to review this video. This self-advocate can provide the perspective of an autistic individual for this topic.
- Recruit a local caregiver of an autistic individual when you expect to review this video to provide a family perspective.
- Recruit a local content expert to be present when you expect to review this video. This local content expert may help facilitate your seminar and address how the topic can be applied locally at your LEND. Schedule them now to be sure they are available to attend your seminar!

Sample discussion questions:

1. What are some reasons that an autistic individual and/or their family may or may not want genetic testing?
2. What are some questions that an autistic individual and/or their family may want to consider asking their provider prior to receiving genetic testing?
3. Should genetic testing be considered the standard of care for any autistic individual?

Contextual Articles:

- Savatt, J. M., & Myers, S. M. (2021). [Genetic testing in neurodevelopmental disorders](#). An accessible overview of different genetic tests and their role in diagnostic workup of NDDs, along with discussion of existing guidelines.
- Thapar, A., & Rutter, M. (2021). [Genetic advances in autism](#). Thorough discussion of autism genetics.
- **Optional**: Havdahl, A., Niarchou, M., Starnawska, A., Uddin, M., van der Merwe, C., & Warrier, V. (2021). [Genetic contributions to autism spectrum disorder](#). A technically more advanced overview of autism genetics.