3q29 Deletion Syndrome
Family Web-Meeting Series

This series is sponsored by:
The Emory 3q29 Project
Department of Human Genetics,
Emory University

It is funded through a Patient-Centered Outcomes Research Institute (PCORI) Eugene Washington PCORI Engagement Award (EAIN #00097).
Housekeeping

• Be sure to mute your microphone when not speaking to minimize background noise.

• We will watch the chat throughout the meeting. Use the chat to:
  • post comments or questions
  • private message questions for us to raise during the session

• Clinicians or healthcare workers attending who are not registered in the 3q29 Registry:
  • Please private message me with your name and email to include you on future communications.

• Tell us what you think. Following the session you may receive a 3-question (2 minute) survey via email for feedback on this session.
We are interested in the topics, issues, and research priorities that are important to you.

This meeting will be recorded. We will also make notes about important ideas and issues that come up that could be addressed in research.

If slides, recordings, or content from this session are made available, they will not include images, voices, or names that could allow identification of individual participants.

You may choose to turn off audio and video feeds or leave the meeting at anytime without penalty.
A quick poll ...

... help us know who is here.
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Unless otherwise noted, all meetings are planned for 12-1:30 pm Eastern Time.
Additional session description and speaker bios will be available on the website: https://genome.emory.edu/3q29/for-families/3q29-families-meeting/
3q29 Deletion Syndrome

Cognitive Profile & Social Disability

Dr. Cheryl Klaiman
Dr. Celine Saulnier
Dr. Stormi White

Emory 3q29 Project
Overview

• Cognitive Profile

• Social Disabilities & Autism Spectrum Disorder

• Question & Answer
Two Reminders

• Findings described here are based upon average (group) performance not individual performance
  • As researchers, we are interested in group performance to understand what behaviors may be attributed to 3q29 deletion.

• Within each group, individual performance may vary, so...
  • As a parent, it is important to consider your individual loved one’s strengths and vulnerabilities.
  • We hope one day to better understand why individual performance varies
Cognitive & Related Abilities Profile

- Intellectual Ability or Cognition (IQ)
  - The skills we use to perform tasks that involve perceiving, remembering, thinking, reasoning, and learning about the world.
  - Often organized into “verbal” and “nonverbal” abilities

- Adaptive Behavior/Adaptive Functioning
  - The skills we use to navigate everyday life.
  - Skills are *conceptual* (e.g., reading, time, money), *social*, and *practical* (e.g., personal care, safety)

- Intellectual Disability (ID)
  - When BOTH cognition and adaptive functioning are significantly delayed

- Graphomotor & Motor Coordination Skills
  - Small-motor skills that are required for tasks like handwriting

- Visual Perception Skills
  - Visual scanning and matching skills
Intellectual Ability (IQ)

Average in the general population = 100

2 SD below the mean = 70

34% of individuals in our study qualify for diagnosis of Intellectual Disability (ID)
Verbal-Nonverbal Difference

On average, verbal and nonverbal subtest scores are 14 points apart (in either direction)

Positive values: verbal subtest score is higher than nonverbal
N = 19, 59%

Negative values: verbal subtest score is lower than nonverbal
N = 10, 31%
Adaptive Behavior

Overall adaptive functioning is consistent with intellectual ability

Average in the general population = 100

2 SD below the mean = 70

Avg= 73
Range 40-99

Avg= 74
Range 48-110
Graphomotor skills are lower than expected given overall intellectual ability.
Graphomotor weakness reflects poor motor coordination skills more so than visual perception skills.
Visual-motor integration

Draw this:
Visual-motor integration

Draw this:

- 15 years
- 10 years
- 7 years
- 18 years
- 6 years
- 18 years
- 16 years
- 18 years

00:15:40
Summary

• Overall IQ score must be interpreted carefully
  • A discrepancy between verbal and nonverbal ability may lead to over- or under-estimation of ability in some areas.

• Adaptive functioning
  • Identify meaningful ways to strengthen adaptive skills and provide reasonable accommodations (e.g., Velcro vs shoelaces)

• Graphomotor vulnerabilities may be underestimated given IQ
  • Early and ongoing occupational therapy supports may be beneficial
  • In educational settings, consider ways to emphasize learning component vs graphomotor demands of task/assignment
Social Disabilities and ASD

- **Autism Spectrum Disorder (ASD):** A neurodevelopmental disorder characterized by:
  1. Deficits in social communication, interaction, and play skills
  2. Restricted, repetitive, and stereotyped behaviors
- Symptoms are present from early childhood
Social disability in 3q29 deletion syndrome

36% of our study sample qualify for an ASD diagnosis using gold-standard instruments.

39% of males
2.7% in general population
14x enriched

30% of females
0.7% in general population
42x enriched
Social Responsiveness Scale

Pollak RM et al, Mol Autism, 2019
Social Responsiveness Scale: 3q29 Deletion Syndrome

Pollak RM et al, Mol Autism, 2019
Social Responsiveness Scale

Severity

3q29 Deletion:
ASD diagnosis reported

3q29 Deletion:
No ASD diagnosis reported

Control

Moderate/Severe

Normal/Mild

Pollak RM et al, Mol Autism, 2019
A Unique Autism Profile

These data will be validated by direct clinical assessment

Contact information

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