3q29 Deletion Syndrome
Family Web-Meeting Series

It was 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.

Timestamp audio file
00:00:00
Web-Meeting Permission Form*

*emailed with meeting link

• 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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<td>Overview of 3q29 Deletion</td>
<td>Dr. Jennifer Mulle</td>
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<td>Th, 12/3/2020</td>
<td>How your health needs are addressed in research</td>
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<td>W, 1/13/2021</td>
<td>Cognitive Profile &amp; Social Disability</td>
<td>Drs. Celine Saulnier, Cheryl Klaiman, &amp; Stormi White</td>
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<td>Tu, 2/2/2021</td>
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<td>TBD-Aug 2021</td>
<td>Dating &amp; Interpersonal Relationships-Parent Session</td>
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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
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).

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

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

Graphomotor weakness reflects poor motor coordination skills more so than visual perception skills.

Average in general population

2 SD below the mean = 70

Overall score
Mean: 69
Range 45-103

Visual Perception
Mean: 74
Range 45-101

Motor Coordination
Mean: 61
Range 45-90
Visual-motor integration

Draw this:
Visual-motor integration

Draw this:
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

- **Severity**
  - Moderate/Severe
  - Normal/Mild

**Graph:**
- Y-axis: SRS Score
- X-axis: Age

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

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

Pollak RM, et al, “Neuropsychiatric phenotypes and novel features of ASD in 3q29 Deletion Syndrome: Results from the 3q29 Registry,” Mol Autism, In Press, 2019
Contact information

Jennifer Mulle, co-PI
Department of Human Genetics
404-727-3042
jmulle@emory.edu

Melissa Murphy, co-PI
Department of Human Genetics
404-727-3446
Melissa.murphy@emory.edu