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 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.
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.

Timestamp in audio file:
00:00:09
A quick poll ...

... help us know who is here.
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<tr>
<th>Date</th>
<th>Topic</th>
<th>Presenters</th>
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<tr>
<td>Th, 10/29/2020</td>
<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>
<td>Dr. Sharron Close</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>
<td>Anxiety, ADHD, and other neuropsychiatric symptoms</td>
<td>Drs. Lindsey Burrell, Elaine Walker, &amp; Joe Cubells</td>
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<td>M, 3/29/2021</td>
<td>3q29 Awareness Day Celebration</td>
<td>Parent Advocates</td>
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<td>M, 5/3/2021</td>
<td>Mental Health &amp; Psychosis</td>
<td>Drs. Elaine Walker &amp; Joe Cubells</td>
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<td>Th, 6/25/2021</td>
<td>Medical and Physical Symptoms</td>
<td>Drs. Rossana Sanchez &amp; Michael Gambello</td>
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<td>TBD-Aug 2021</td>
<td>Dating &amp; Interpersonal Relationships-Parent Session</td>
<td>Dr. Opal Ousley</td>
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<td>TBD-Sept 2021</td>
<td>Dating &amp; Interpersonal Relationships-Young Adult Session</td>
<td>Dr. Amanda Palmer</td>
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<td>F, 9/10/2021</td>
<td>Sharing Research Findings: How to spread the word</td>
<td>Drs. Jennifer Mulle &amp; Melissa Murphy</td>
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<td>TBD-Oct 2021</td>
<td>3q29 Family Camp Weekend</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

Anxiety, ADHD, and Other Neuropsychiatric Symptoms

February 2, 2021

Dr. Lindsey Burrell
Dr. Joseph Cubells
Dr. Elaine Walker
Any GI symptom, 81%

- Failure to thrive in infancy, 9%
- Feeding problems beyond infancy, 16%
- Constipation, 41%
- Failure to thrive beyond infancy, 41%
- Reflux, 50%
- Feeding problems in infancy, 59%

60% require a feeding tube.
At least 1 anxiety disorder, 40%
18% have more than 1 anxiety disorder
(7% for individuals ages 3-17 years in general population)

Social Anxiety Disorder: 6%
Separation Anxiety: 12.5%
Specific Phobia: 19%
Generalized Anxiety Disorder: 22%

We do not see an increase in depression. In 3q29 deletion syndrome, anxiety seems to be uncoupled from depression.
3q29 Deletion: ADHD

- ADHD in 3q29 Deletion: 63%
- ADHD in general population: 9%
- ADHD inattentive type: 31%
- ADHD combined type: 25%
- ADHD hyperactive/impulsive: 3%
3q29 Deletion: Enuresis

Average age at toilet training: 4 years
Range: 2 years to “not yet”

Enuresis: a repeated inability to control urination.

22% of individuals with 3q29 deletion qualify for a diagnosis of enuresis, aged 6 – 17 years.
Questions Raised At Prior Meetings

• How does anxiety typically present in 3q29 deletion?

• What are some of the biological contributors to anxiety?

• What are the psychological effects of anxiety and mood disorders? How do these conditions play out over the lifetime?

• What treatments and medications are best for addressing anxiety?

• Are there interventions/treatments for improving GI and feeding-related issues?

• How can we communicate effectively with providers regarding the seriousness of the challenges our child faces?