

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.

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.

3q29 Web Meeting Series 2020-2021

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/>



Date	Topic	Presenters
Th, 10/29/2020	Overview of 3q29 Deletion	Dr. Jennifer Mulle
Th, 12/3/2020	How your health needs are addressed in research	Dr. Sharron Close
W, 1/13/2021	Cognitive Profile & Social Disability	Drs. Celine Saulnier, Cheryl Klaiman, & Stormi White
Tu, 2/2/2021	Anxiety, ADHD, and other neuropsychiatric symptoms	Drs. Lindsey Burrell, Elaine Walker, & Joe Cubells
M, 3/29/2021	3q29 Awareness Day Celebration	Parent Advocates
M, 5/3/2021	Mental Health & Psychosis	Drs. Elaine Walker & Joe Cubells
Th, 6/25/2021	Medical and Physical Symptoms	Drs. Rossana Sanchez & Michael Gambello
TBD-Aug 2021	Dating & Interpersonal Relationships-Parent Session	Dr. Opal Ousley
TBD-Sept 2021	Dating & Interpersonal Relationships-Young Adult Session	Dr. Amanda Palmer
F, 9/10/2021	Sharing Research Findings: How to spread the word	Drs. Jennifer Mulle & Melissa Murphy
TBD-Oct 2021	3q29 Family Camp Weekend	

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

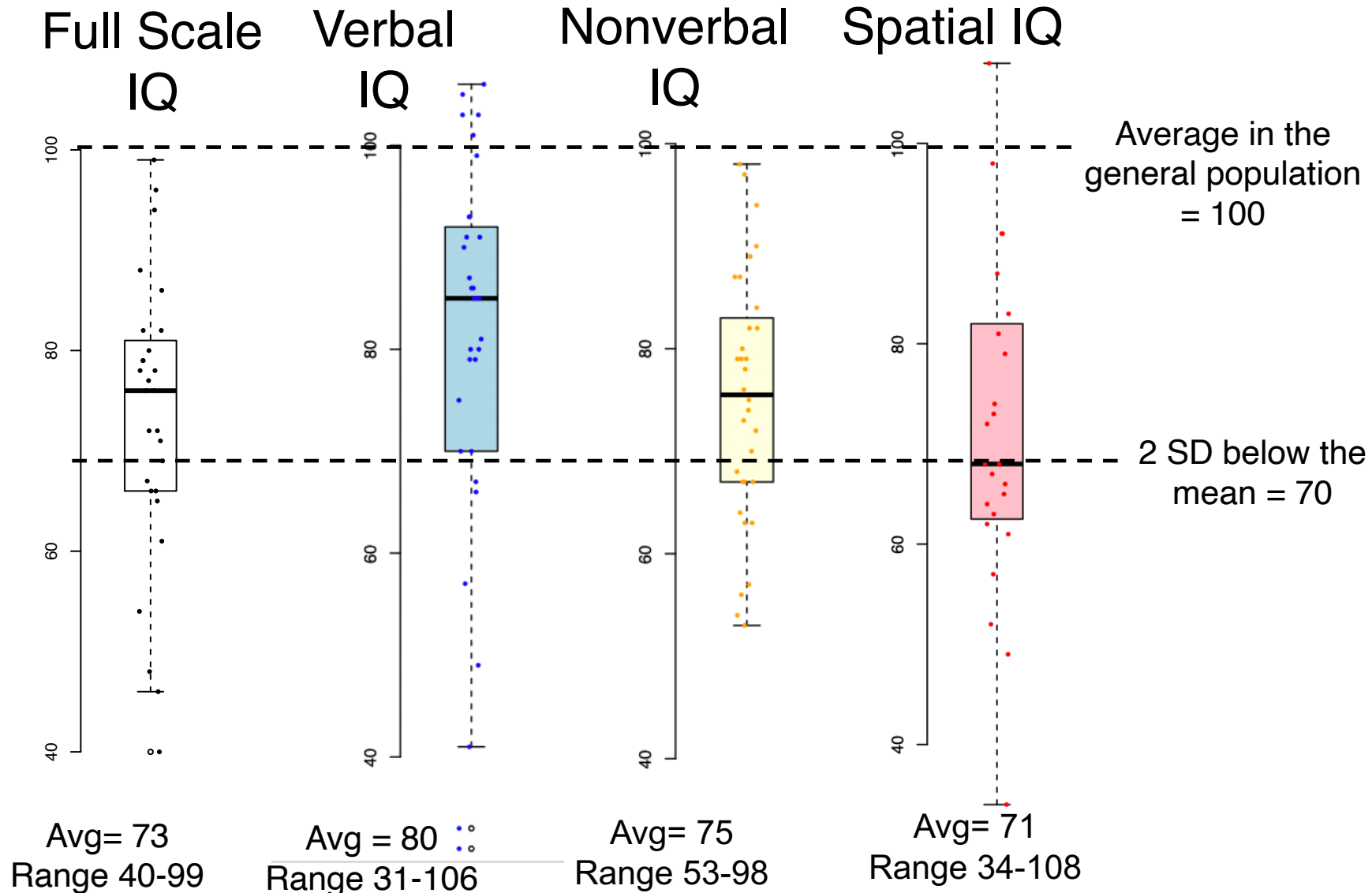
- Findings described here are based upon average (group) performance not individual performance
 - As researchers, we are interested in group performance to understand what behaviors may be attributed to 3q29 deletion.
- Within each group, individual performance may vary, so...
 - As a parent, it is 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



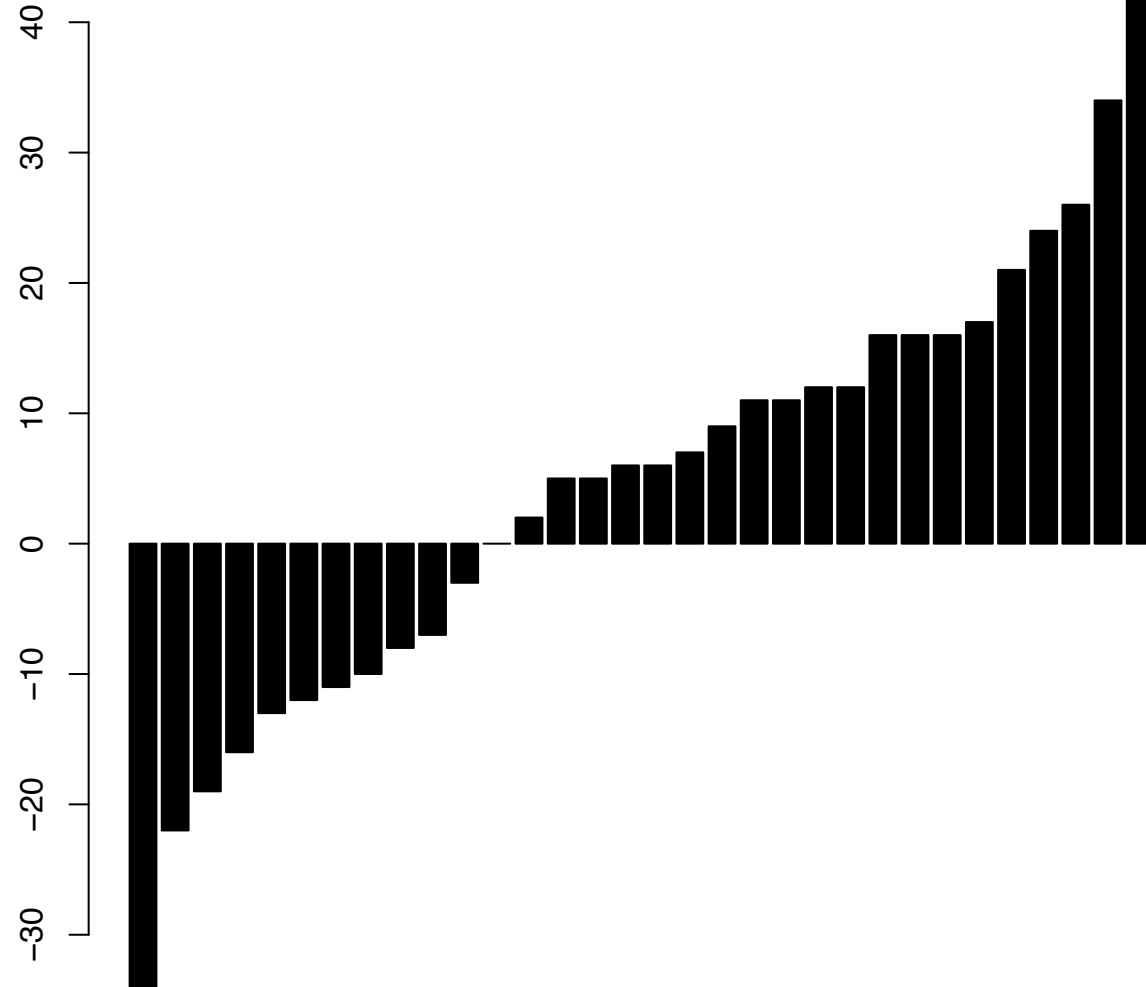
- Intellectual Ability or Cognition (IQ)
 - The skills we use to perform tasks that involve perceiving, remembering, thinking, reasoning, and learning about the world.
 - Often organized into “verbal” and “nonverbal” abilities
- Adaptive Behavior/Adaptive Functioning
 - The skills we use to navigate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



**34% of individuals
in our study qualify
for diagnosis of
Intellectual
Disability (ID)**

Verbal-Nonverbal Difference



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

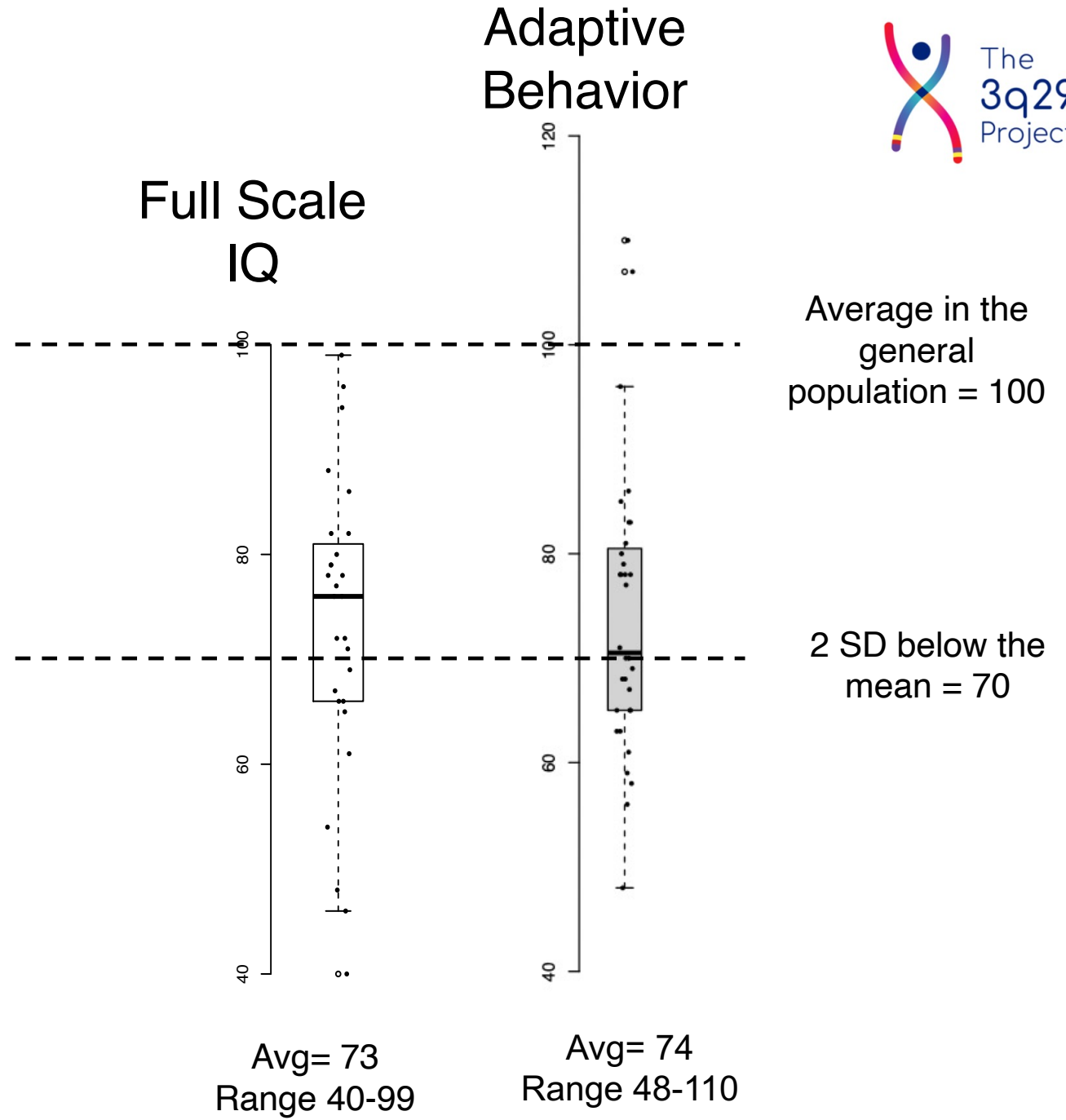
**Positive values: verbal
subtest score is
higher than nonverbal
N = 19, 59%**

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

Adaptive Behavior



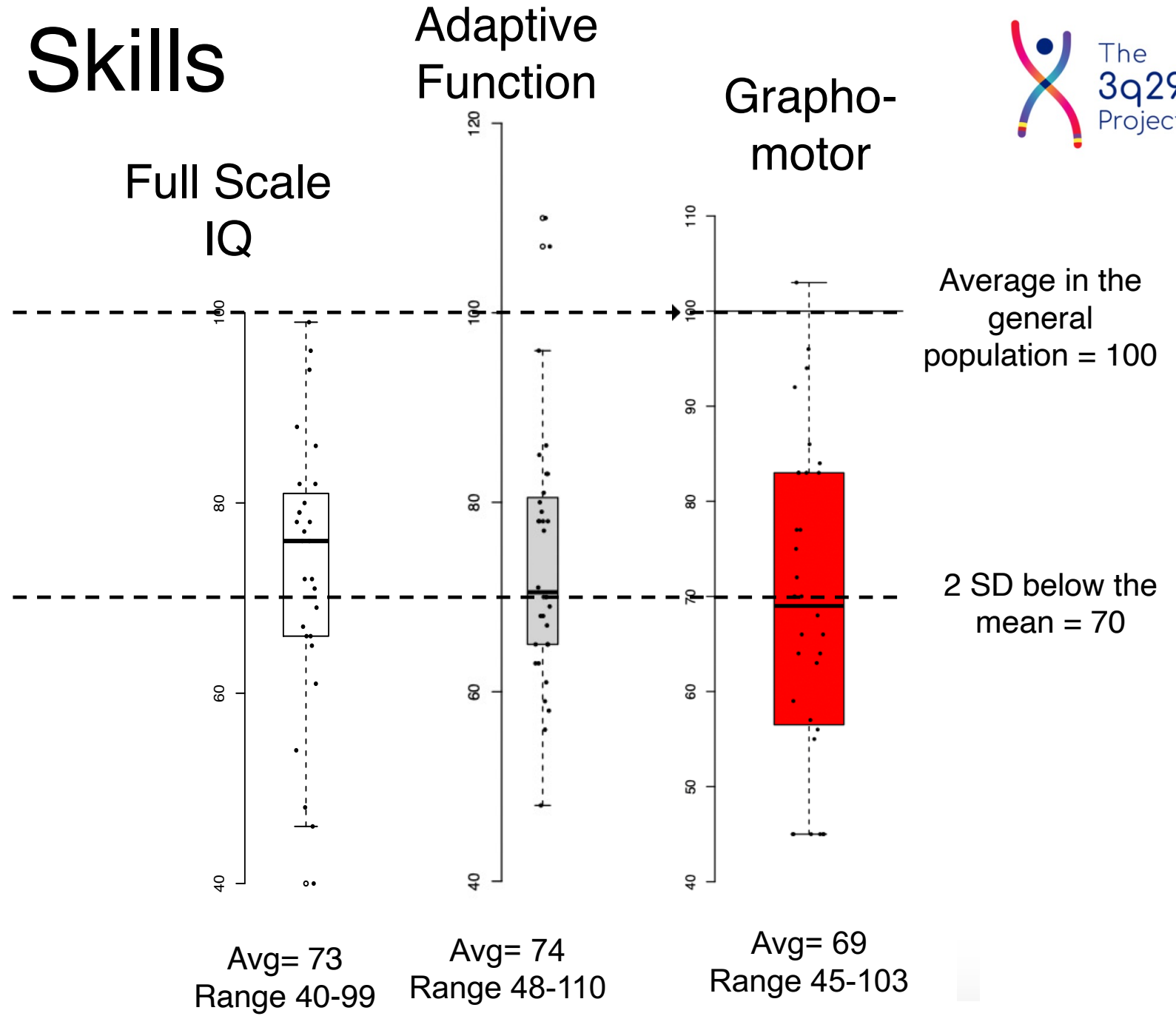
Overall adaptive functioning is consistent with intellectual ability



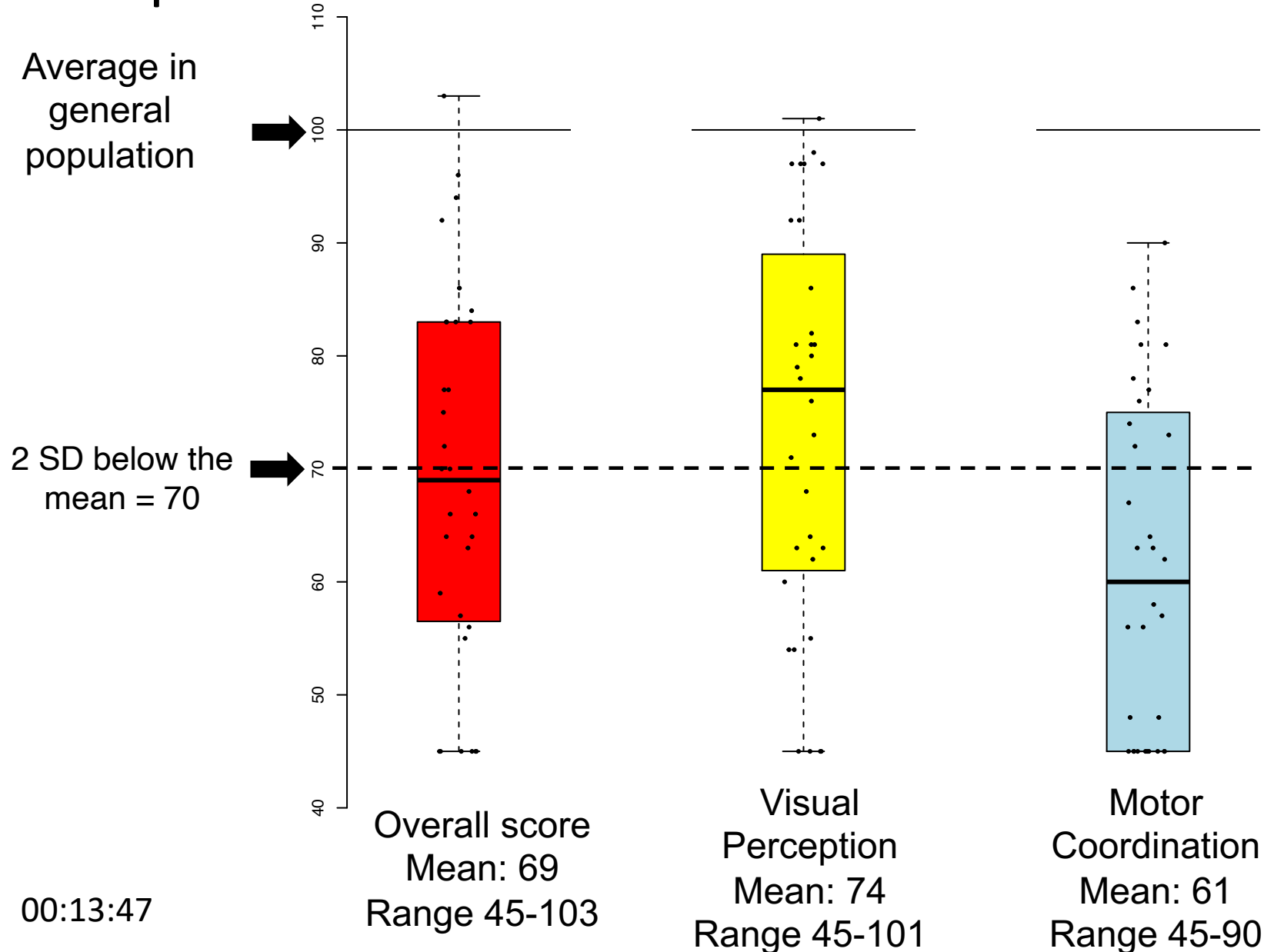
Graphomotor Skills



Graphomotor skills are lower than expected given overall intellectual ability.



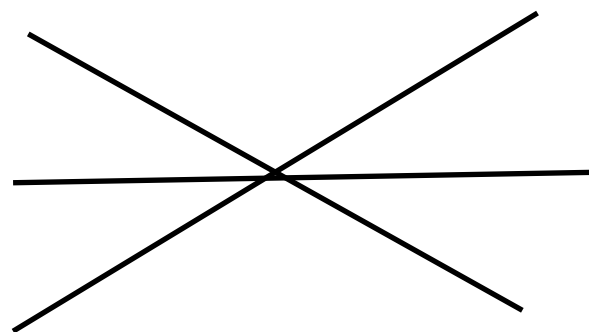
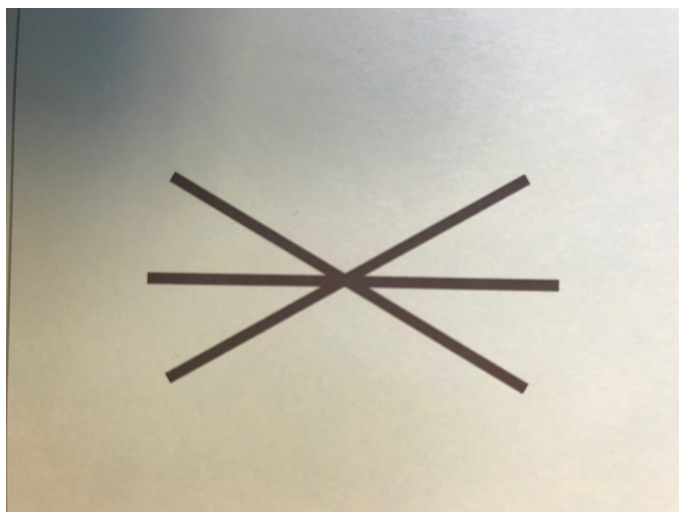
Graphomotor weakness



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

Visual-motor integration

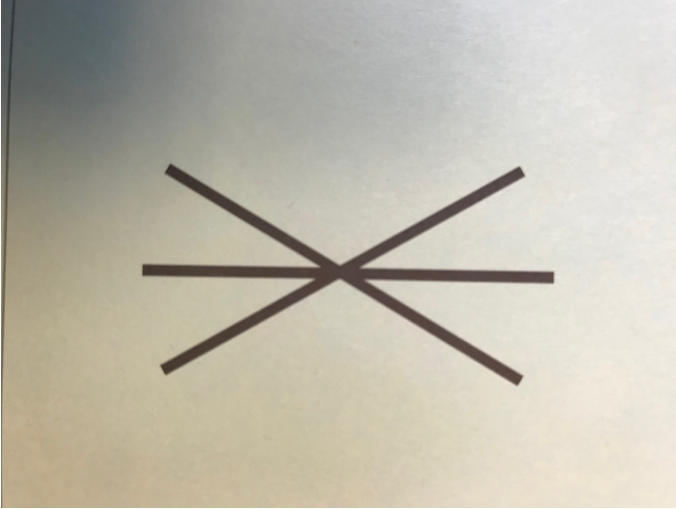
Draw this:



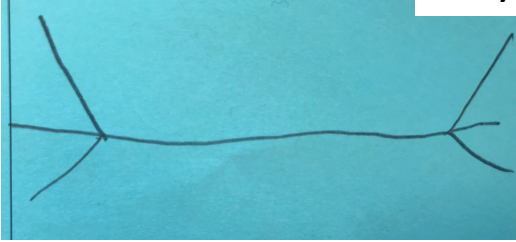
Visual-motor integration



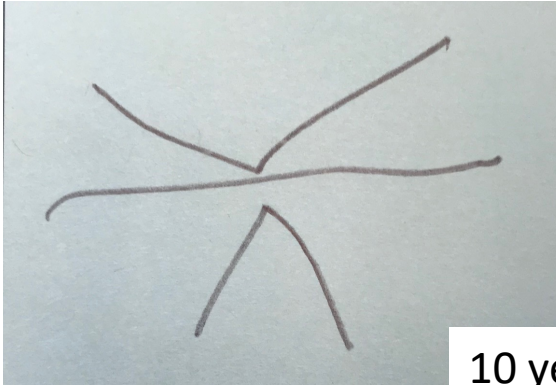
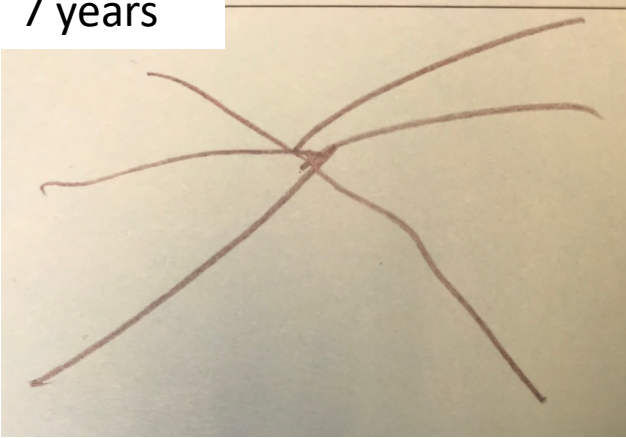
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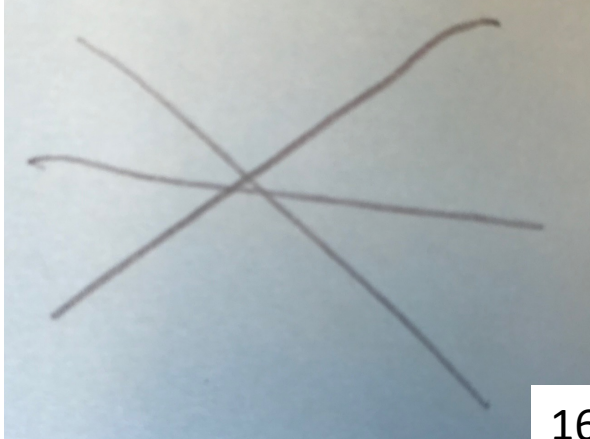
15 years



7 years

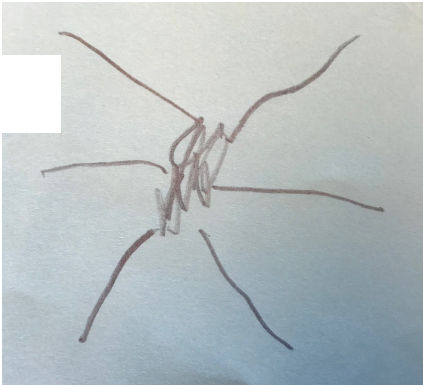


10 years



16 years

6 years



18 years



18 years



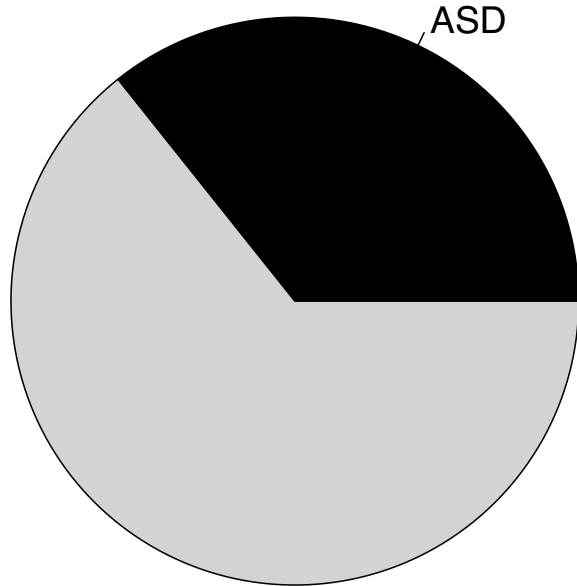
Summary

- 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

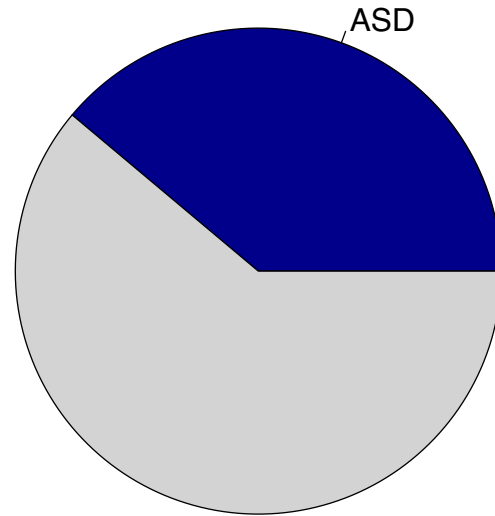
Social Disabilities and ASD

- 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

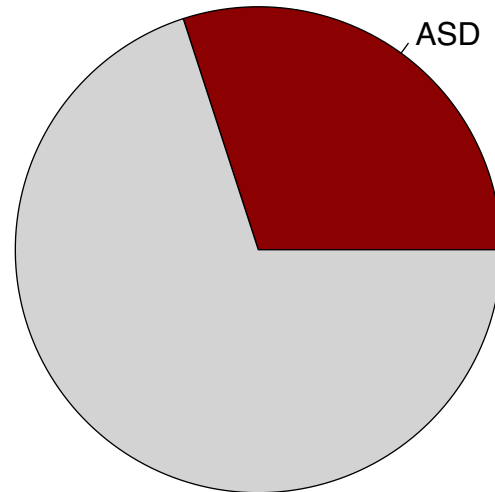
Social disability in 3q29 deletion syndrome



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

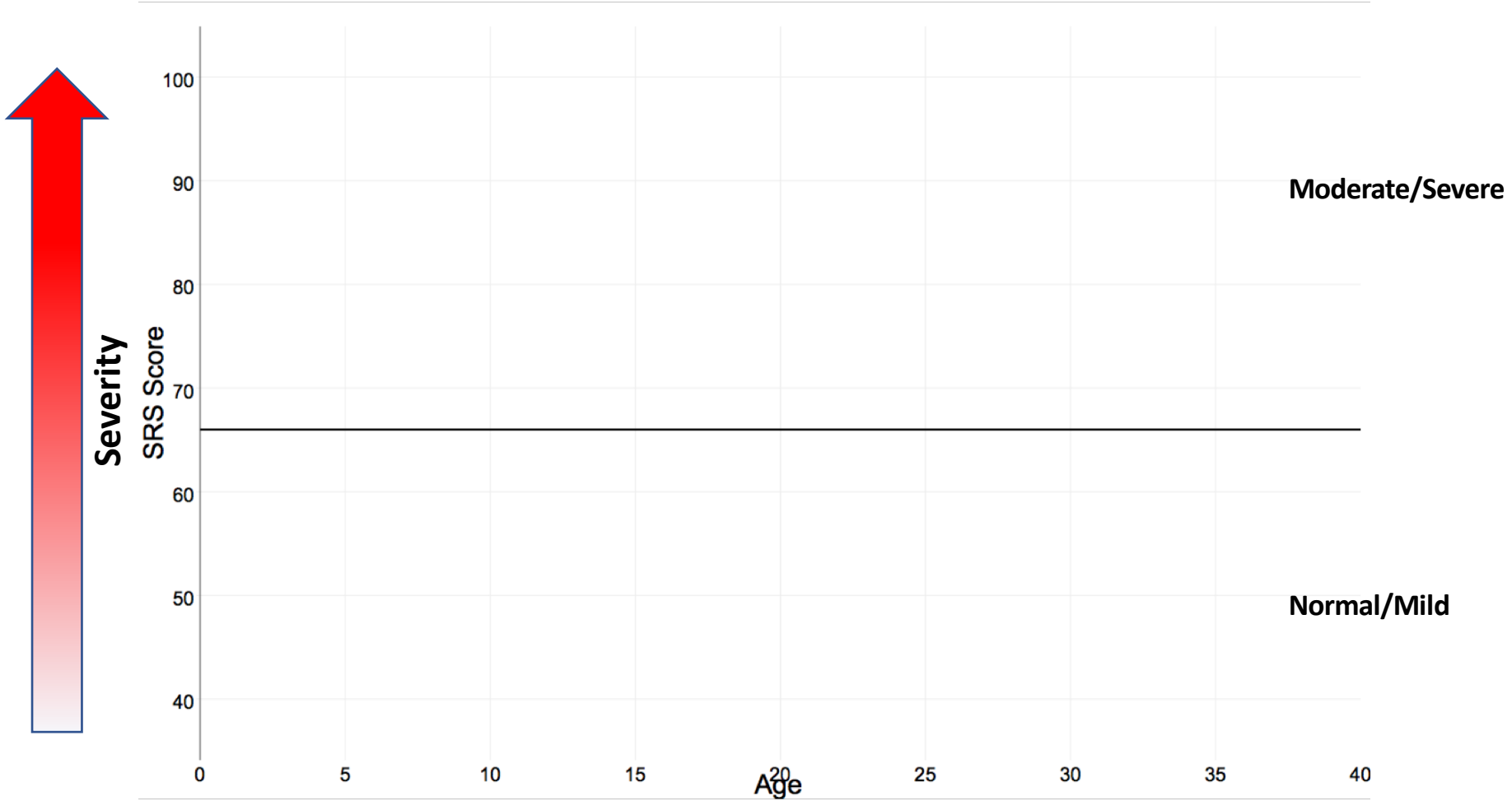


39% of males
2.7% in general population
14x enriched



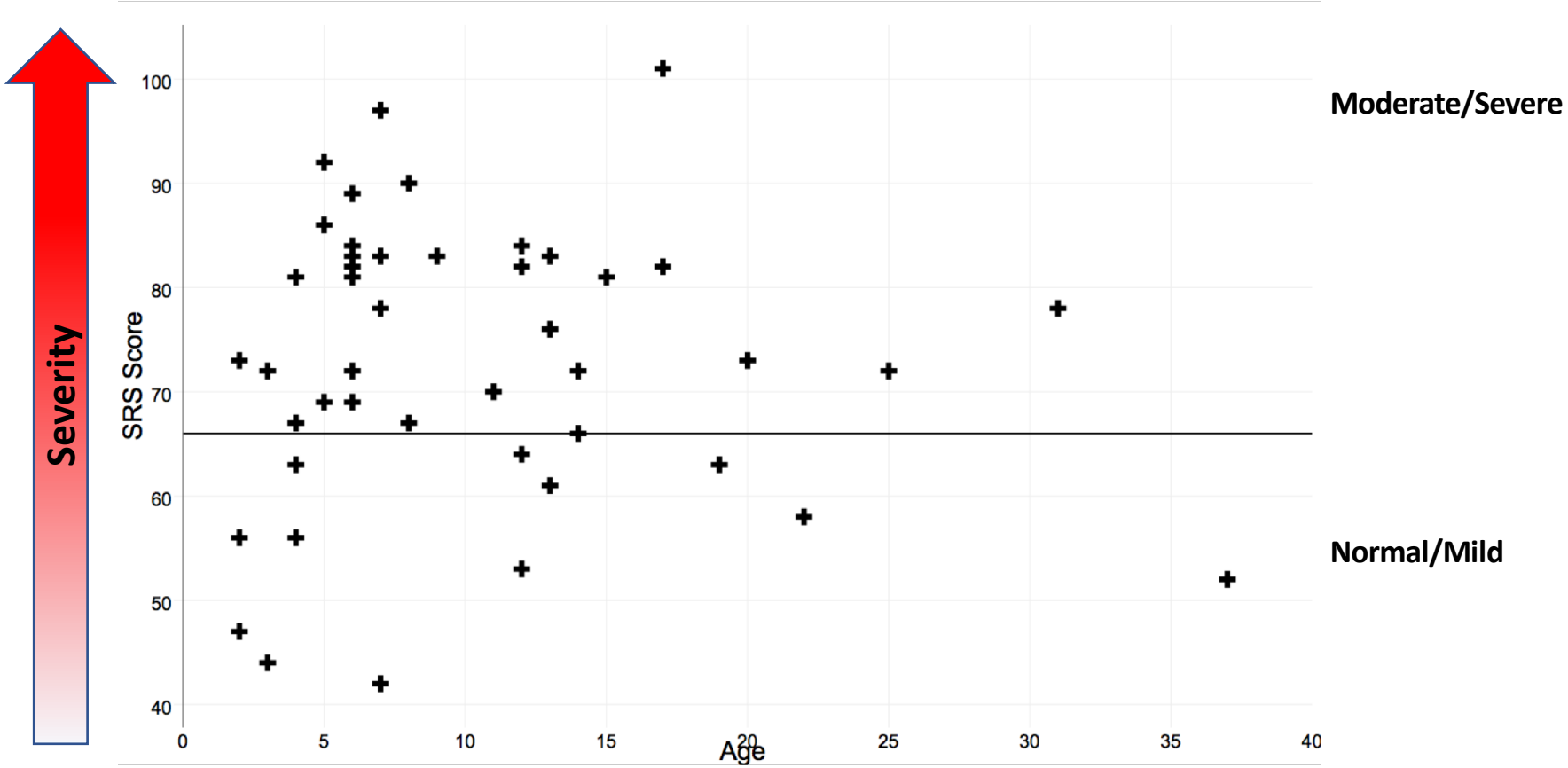
30% of females
0.7% in general population
42x enriched

Social Responsiveness Scale



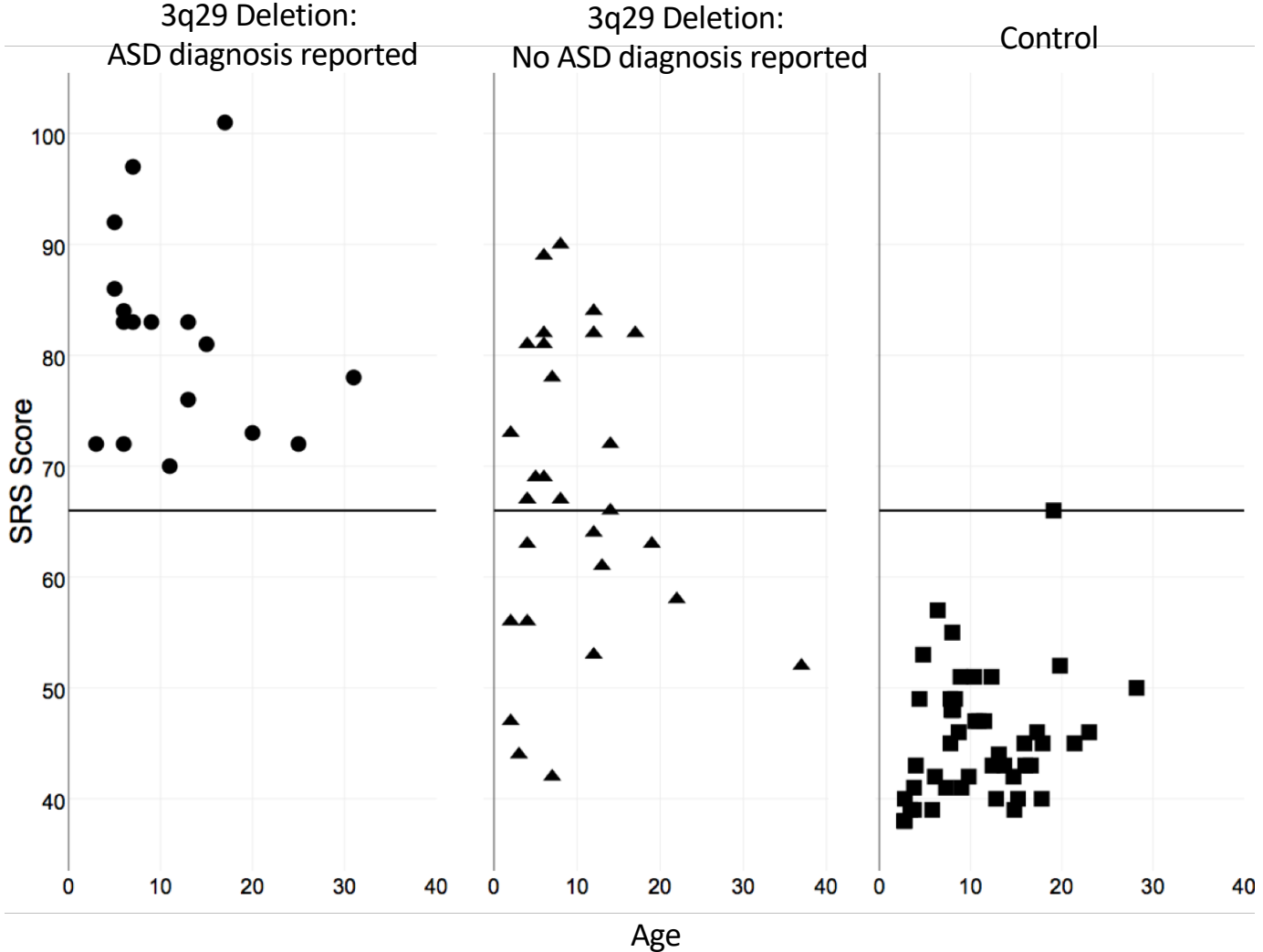
**Pollak RM et al,
Mol Autism, 2019**

Social Responsiveness Scale: 3q29 Deletion Syndrome



Pollak RM et al,
Mol Autism, 2019

Social Responsiveness Scale



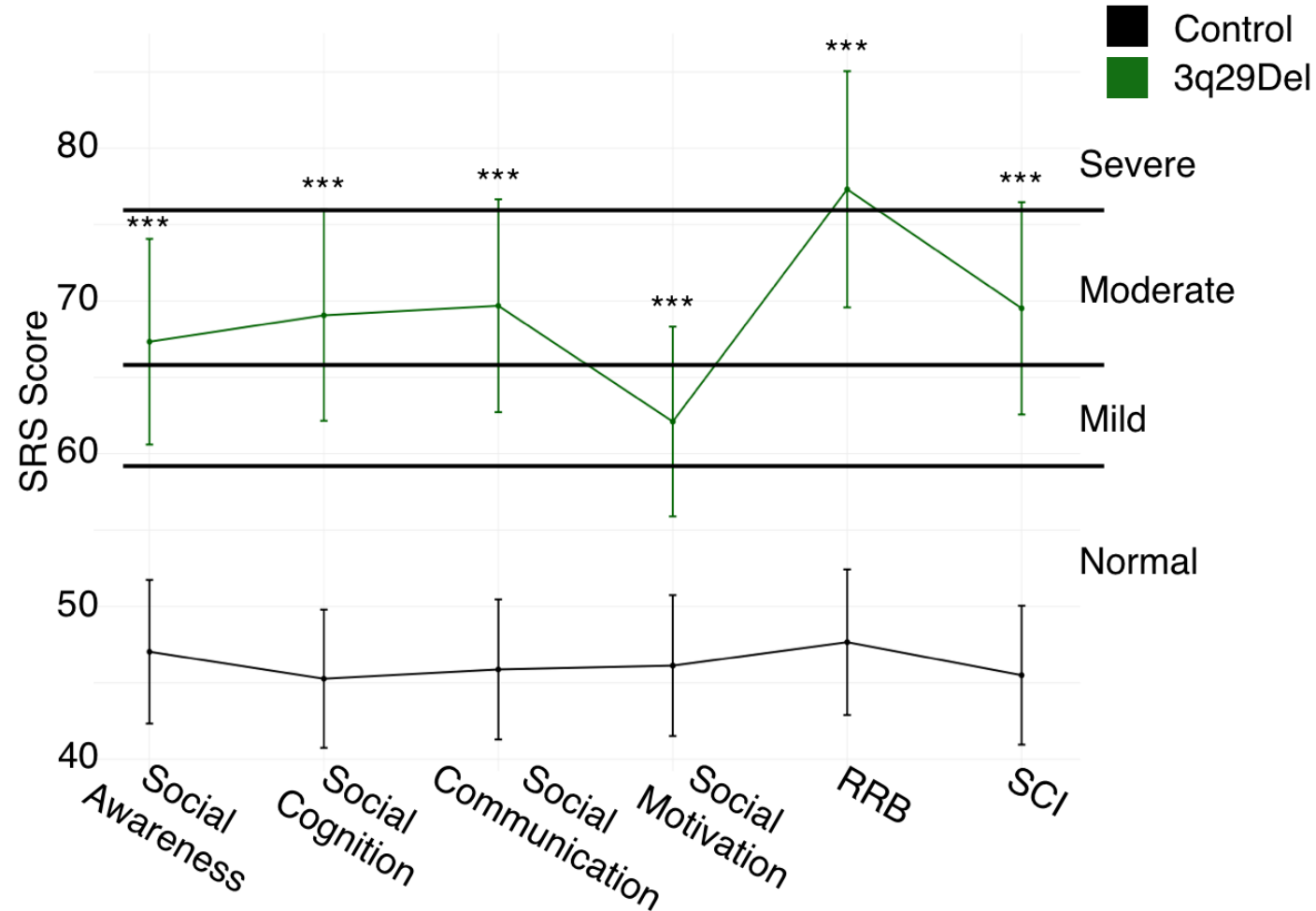
Moderate/Severe

Normal/Mild



Pollak RM et al, *Mol Autism*, 2019

A Unique Autism Profile



These data will be validated by direct clinical assessment

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