Cerebellum is more than a motor control center

- Cerebellum may have fundamental cognitive and emotional functions (Klein et al 2016)

  - Includes disturbances of executive function and impaired spatial cognition

Is cerebellar dysfunction in the causal pathway for 3q29 deletion syndrome phenotypes? Area for future study
Your participation has given us an important clue!
Conclusions

• ADHD, graphomotor weakness, and executive function deficits are present in 3q29 deletion syndrome
• There is a unique profile of cognitive vulnerability
• High burden of ASD in females
• Hypothesis: Cerebellum is a site of dysregulation

• Direct evaluation of study subjects reveals nuances of behavior, inspires data-driven hypotheses for mechanistic investigation
3q29 deletion syndrome: 2010

- Developmental Delay
- Intellectual Disability
- Features of ASD

- Increased risk for schizophrenia

Birth -> Childhood -> Adulthood
3q29 deletion syndrome: 2020

Developmental Delay
Intellectual Disability
Features of ASD

Birth

Increased risk for schizophrenia

Childhood

Increased risk for neuropsychiatric phenotypes, especially anxiety disorders

Adhd

Graphomotor Weakness

Executive Function Deficits

Adaptive Behavior Deficits

Dental issues

Cognitive Vulnerabilities

Specific timeline of delayed milestones

Feeding disorders, failure to gain weight

Reduced birth weight

Sex-dependent effects of neurodevelopmental phenotypes

Adulthood

We’re going to need a bigger graphic....