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
Preliminary Neuroimaging Results

A consistent cerebellar phenotype: volumetric reduction

T1-weighted sMRI; N = 24
Arachnoid cysts

-most often described as “benign”

-in 1113 healthy young adults who were scanned as part of the Human Connectome Project, 11 were found to have arachnoid cysts in the “posterior fossa”

-in 23 subjects with the 3q29 deletion, we identified 7 posterior fossa arachnoid cysts

-while these are considered benign, the high rate of these findings in our sample is surprising, and may be an important clue about what the 3q29 deletion is doing.
The 3q29 Deletion Mouse

3q29 mouse has deficits in spatial learning and memory, social interaction, acoustic startle, and amphetamine sensitivity

Cerebellar deficits are present in the 3q29 deletion mouse model

Genotype differences in absolute cerebellar volume:
- WT: 65.08 ± 2.1 mm³
- Mut: 56.02 ± 1.1 mm³

T-test, p = 0.00016

Esra Sefik
Neuroscience
PhD Candidate
Cerebellum is emerging as a site of intense interest for SZ, ASD

Pubmed search for "cerebellum autism" = 991 citations
Pubmed search for "cerebellum schizophrenia" = 1291 citations

The Cerebellum, Sensitive Periods, and Autism

Altered cerebellar and cerebellar autism-related

Dysfunctional cerebellar Purkinje cells contribute to autism-like behaviour in Shank2-deficient

Cerebellar volume and cerebellocerebral structural covariance in schizophrenia: a multisite mega-analysis of 983 patients and 1349 healthy controls