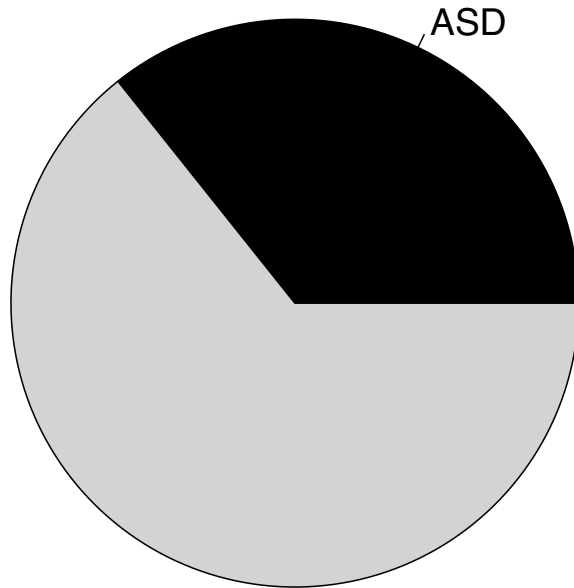
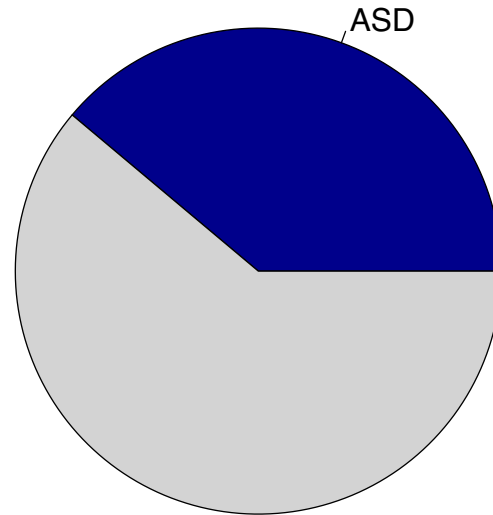


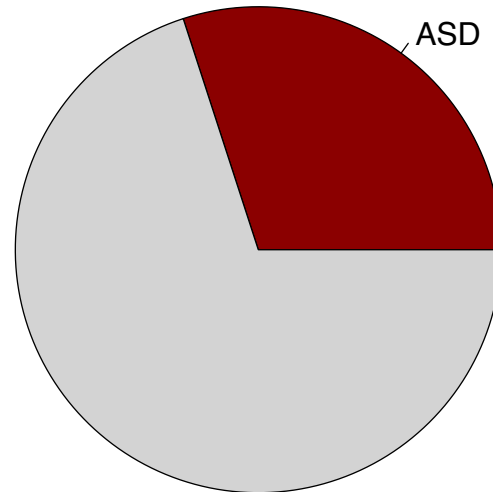
Social disability in 3q29 deletion syndrome



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

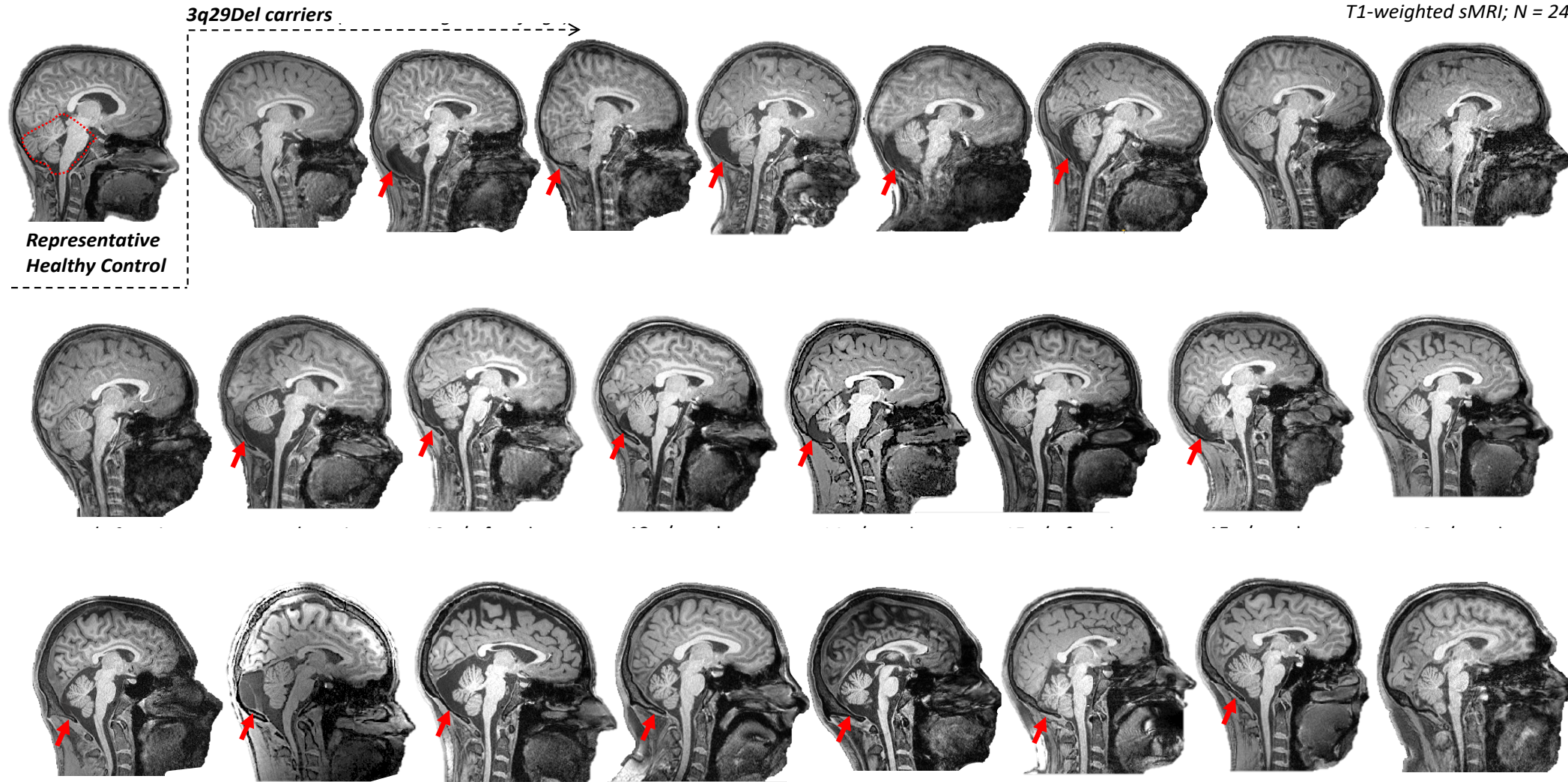


39% of males
2.7% in general population
14x enriched



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

Preliminary Neuroimaging Results



A consistent cerebellar phenotype: volumetric reduction

Arachnoid cysts

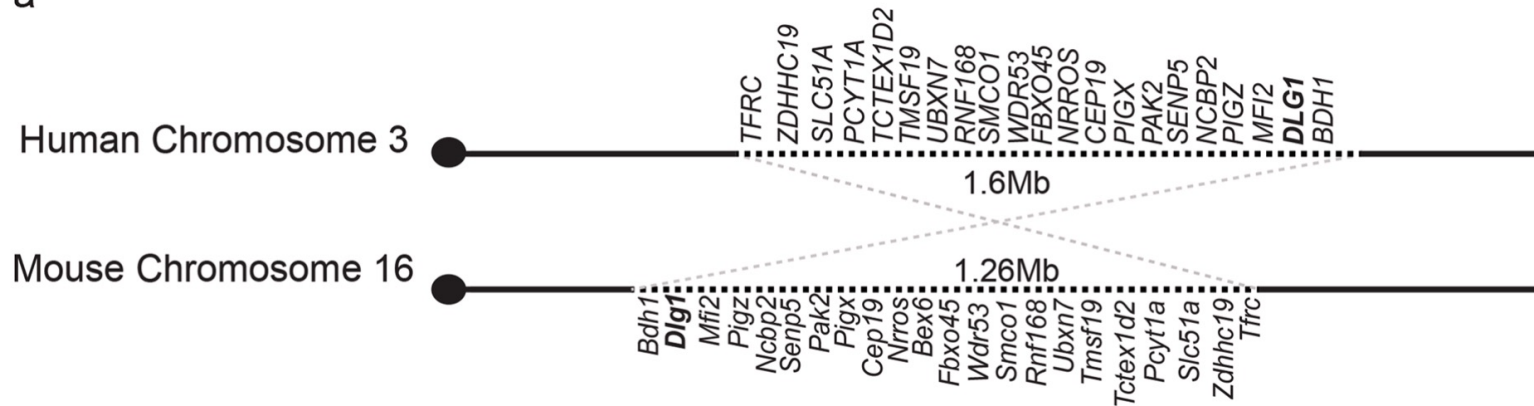
- most often described as “benign”
- in **1113 healthy young adults** who were scanned as part of the Human Connectome Project, **11** were found to have arachnoid cysts in the “posterior fossa”
- in **23 subjects with the 3q29 deletion**, we identified **7** posterior fossa arachnoid cysts
- while these 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



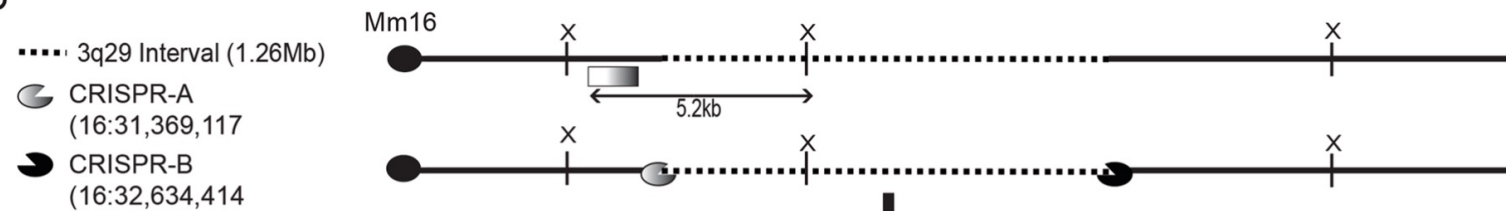
The
3q29
Project

a



Tamara Caspary

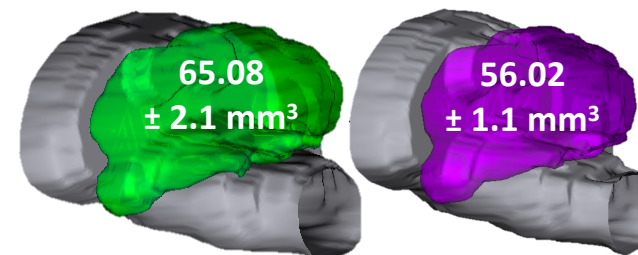
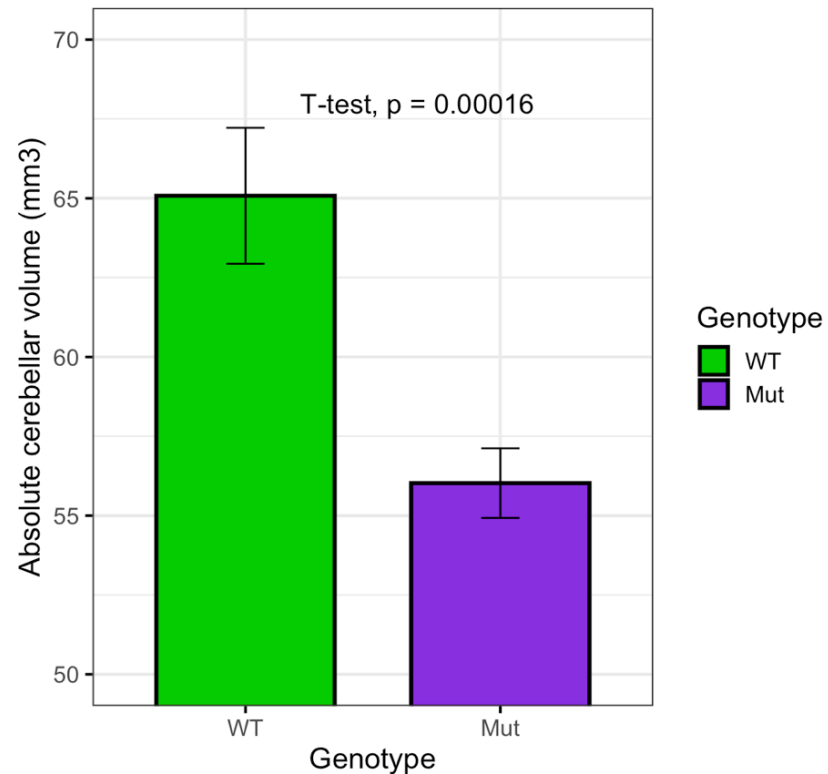
b



David Weinschenker

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



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

Samuel S.-H.
1Princeton Ne
*Corresponde
<http://dx.doi.c>

nature
neuroscience

Article | Published: 30 Oc

**Altered cereb
and cerebella
autism-relate**

Catherine J. Stoodley ,
Liu, Mary Beth Nebel, Jeni
Cano, Juan M. Pascual, St



ARTICLE

Received 9 Mar 2016 | Accepted 19 Jul 2016 | Published 1 Sep 2016

DOI: 10.1038/ncomms12627

OPEN

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

Saša Peter^{1,*}, Michiel M. ten Brinke^{2,*}, Jeffrey Stedehouder³, Claudia M. Reinelt⁴, Bin Wu², Hait
Kuikui Zhou², Henk-Jan Boele², Steven A. Kushner³, Min Goo Lee⁵, Michael J. Schmeisser^{4,6},
Tobias M. Boeckers⁴, Martijn Schonewille², Freek E. Hoebeek² & Chris I. De Zeeuw^{1,2}

Molecular
Psychiatry

Original Article | Published: 16 May 2017

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

T Moberget , N T Doan, D Alnæs, T Kaufmann, A Córdova-Palomera, T V
Lagerberg, J Diedrichsen, E Schwarz, M Zink, S Eisenacher, P Kirsch, E G Jönsson,
H Fatouros-Bergman, L Flyckt, KaSP, G Pergola, T Quarto, A Bertolino, D Barch, A
Meyer-Lindenberg, I Agartz, O A Andreassen & L T Westlye