Foxp1 Syndrome

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Overview

- Individuals with Foxp1-related disorder
- A mouse model for Foxp1-related disorder syndrome
- Stem cells in Foxp1-related disorder

Phenotyping Battery

Domain	Measure
Global Cognitive Ability	Stanford Binet Intelligence Scales, Fifth Edition or Mullen Scales for Early Learning
Adaptive Behavior	Vineland Adaptive Behavior Scales, Second Edition
Language	Peabody Picture Vocabulary Test, Fourth Edition Expressive Vocabulary Test, Second Edition Vineland and Mullen Subscales
Motor Functioning	Beery Visual-Motor Integration Test, Sixth Edition Mullen and Vineland Subscales Developmental Coordination Disorder Questionnaire
Autism Symptoms	Autism Diagnostic Observation Schedule, Second Edition (ADOS-2) Autism Diagnostic Interview-Revised (ADI-R) Social Responsiveness Scale, Second Edition (SRS-2) Repetitive Behavior Scales-Revised
Other Symptoms	Behavior Assessment Scale for Children, Second Edition (BASC-2) Behavior Rating Inventory of Executive Functioning (BRIEF) Child Behavior Checklist (CBCL) Aberrant Behavior Checklist (ABC) Child & Adolescent Symptom Inventory, Fifth Edition (CASI-5) Sensory Profile Questionnaire- Short Form

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Summary of Findings

- Foxp1 phenotype based on prospective data collection (N=4):
  - Delay in achievement of motor and language milestones
    - Motor deficits later characterized by visual-motor integration difficulties
    - Expressive Language stronger than receptive language on clinician-administered assessments
  - Mild-to-moderate intellectual disability
  - Autism Spectrum Disorder traits
  - Comorbid symptoms: anxiety, obsessive-compulsive behavior, attention problems, and externalizing symptoms
- An additional 4 families scheduled for the next 2 months

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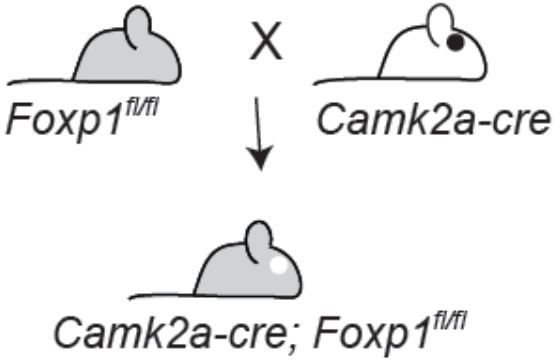
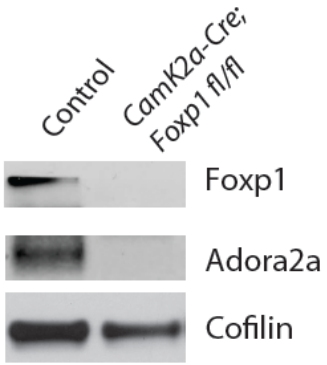
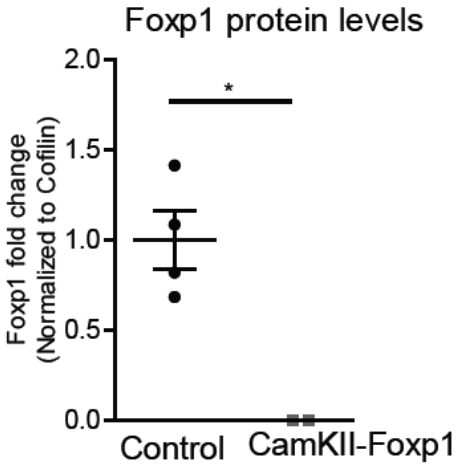
Introducing a Foxp1 mutation in mice







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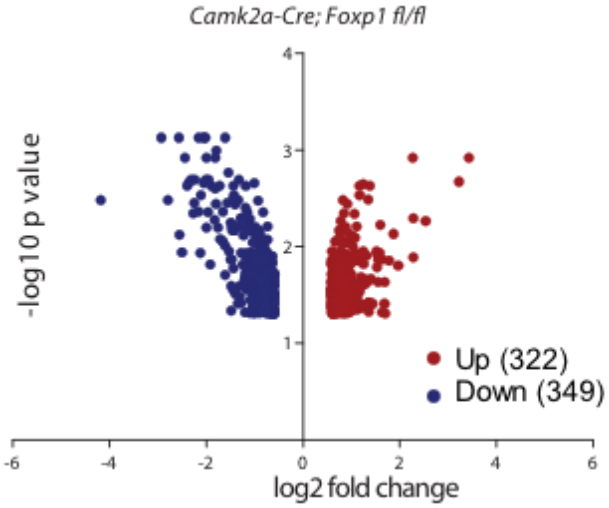
Changes in brain molecules in Foxp1 mutant mice

- Schaefer and colleagues have created a mouse with a Foxp1 mutation in order to understand brain changes associated with Foxp1 mutation
- The mouse is currently being characterized for changes in brain molecules, brain cells, and behavior
- For example, Schaefer and colleagues are examining the molecular changes in brain cells, as a result of Foxp1 loss, in order to understand the downstream effects



Josefa Sullivan and Anne Schaefer

Stem cells

- We are collecting blood from all families that consent
- For two families so far, we have blood from mother, father, affected child and a sibling
- For these families we have reprogrammed the blood cells into stem cells
- These cells can now be made into nerve cells in petri dishes to understand how loss of Foxp1 leads to changes in nerve cell function

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Acknowledgements

- Thank you to the Whitney family for providing support for these studies
- Thank you to the Seaver Foundation for supporting our genetics-first approaches in autism and related neurodevelopmental disorders
- And a special thanks to the families that participate in our studies

Please join us!

- For questions or comments, please email me at admin.buxbaum@mssm.edu
- If you are interested in participating in research, please contact us at theseavercenter@mssm.edu or 212-241-0961
- The Associates Committee is a group of committed stakeholders that drive the Center's scientific mission forward through strategic and philanthropic support.
  - If you are interested in learning more about the Associates Committee, please contact Savannah Lennertz at savannah.lennertz@mssm.edu or 212-241-0349
- To make a contribution to the Seaver Center, please visit https://philanthropy.mountsinai.org/seaver