Macrocephaly in Autism

Summary

- **Study director:** Elliott Sherr, MD PhD
- **Sponsor:** NIH and private foundations and grants
- **Recruiting?** Yes
- **Official study title:** Brain Imaging and Cell Signaling: Insights into the Biology of Autism
- **Conditions studied:** Autism spectrum disorders with macrocephaly
- **Purpose:** This research study is looking at biochemical changes in the blood as well as brain anatomy in order to learn more about autism spectrum disorder (ASD). This would yield better understanding of a disorder that affects an estimated one in 88 children in the United States, but largely remains understood. By investigating the structural and functional brain imaging in a cohort of idiopathic ASD patients who have macrocephaly, we are optimistic that we can establish a link between common biochemical and abd brain developmental pathways that are disrupted in ASD, and thus, plot a path to targeted treatment.

Eligibility

- **Inclusion criteria:**
  - Children aged 6–13, both affected individuals and healthy controls
    - **Affected participants:**
      - Diagnosis of ASD and/or developmental delay, or a sibling of someone affected by ASD
      - Macrocephalic (Head circumference in the 90th percentile or above)
      - Able to undergo blood draw and brain imaging
      - We prefer parents of an affected participants to participate in the blood draw as well
    - **Control participants:**
      - No significant diagnoses
      - Macrocephalic (Head circumference in the 90th percentile or above)
      - Able to undergo blood draw and brain imaging
      - Must be available to travel to UCSF Mission Bay for the study procedures
- **Exclusion Criteria:** None

What is Involved?

- Blood draw
- Sharing of medical records
- Neuropsychological questionnaires
- In-person testing including: cognitive testing and autism spectrum disorder assessment
- Comprehensive brain imaging including: fMRI, DTI, EEG and MEG
○ A total of 2–8 hours of participating

Contact Information

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