

Neurophysiologic Biomarkers in Rett Syndrome

NCT05932589

Status	RECRUITING
Sponsor	Children's Hospital of Philadelphia
Enrollment	202 participants

Key Eligibility Criteria

Inclusion (3)

- Rett Group: Females ages 3-18 (inclusive) with a clinical diagnosis of RTT with a likely pathogenic or known pathogenic variant in MECP2.
- Likely Rett Group: Females from 1 year to ≤ 5 years of age with MECP2 variant if regression has not yet occurred or child is within 6 months of last skill loss.
- Typically developing (TD) Group: Females age matched to RTT population (1-18) with no developmental or cognitive concerns as assessed using the Child/Adult Behavioral Checklist, Survey of Well-Being of Young Children (≤ 5 yo), or the Wide Range Achievement Test-4 (≤ 5 yo).

Exclusion (7)

- Rett and Likely Rett Groups:
- Presence of a duplication in MECP2 or any other identified pathogenic mutation in another gene.
- Active medical conditions not typically found in RTT.
- Typically Developing Group:
- Score below norms on the performance tests
- ... and 2 more (see full listing online)

Locations (6 total)

Children's Hospital of Los Angeles, Los Angeles, California, United States
Children's Hospital Colorado, Aurora, Colorado, United States
Boston Children's Hospital, Brookline, Massachusetts, United States
... and 3 more locations