

Clinical and Molecular Biomarker Studies in RAI1 (Retinoic Acid-Induced 1) -Related Disorders

NCT06274164

Status RECRUITING
Sponsor Baylor College of Medicine
Enrollment 90 participants

Key Eligibility Criteria

Inclusion (9)

- Patient group:
- Patients who have RAI1-related disorder confirmed by genetic testing including karyotyping, fluorescence in situ hybridization (FISH), array Comparative Genomic Hybridization (aCGH), single nucleotide polymorphism (SNP) array and next generation sequencing performed by a Clinical Laboratory Improvement Amendments (CLIA)-certified laboratory.
- Grossly intact hearing and vision as per parent report
- Age between 1 month to 60 years old
- Able to complete the study (i.e., travel to site and spend 1 day in Houston)
- ... and 4 more (see full listing online)

Exclusion (6)

- Patient group:
- Contraindication for blood draw or skin biopsy as determined by the enrolling provider (e.g., bleeding diathesis)
- Patients who are at high risk including ventilator/tracheostomy dependent, poorly controlled endocrine disorders, and unstable seizures (will be assessed by neurologist), end-stage renal disease.
- Participation in any investigational treatment study
- Control group:
- ... and 1 more (see full listing online)

Locations (1 total)

Texas Children's Hospital, Houston, Texas, United States