Rett Syndrome Registry

Status: RECRUITING

Eligibility Criteria

Sex: ALL
Age: 0 years to 99 years old
Healthy Volunteers: This study is NOT accepting healthy volunteers
Inclusion Criteria:

- Male or female with a pathologic loss of function alteration of MECP2

Exclusion Criteria:

- Male or female with a gain of function alteration of MECP2, including those with MEPC2 duplication or triplication

Conditions & Interventions

Conditions:
Rett Syndrome, Rett Syndrome, Atypical, Genetic Disease, Genetic Diseases, X-Linked, Intellectual Disability, Neurobehavioral Manifestations, Neurologic Manifestations, Neurologic Disorder, Neurodevelopmental Disorders, Nervous System Diseases
Keywords:
Rett syndrome, MECP2, Neurodevelopmental disorder, Registry, Natural History Study

More Information

Description:
The Rett Syndrome Registry is a longitudinal observational study of individuals with MECP2 mutations and a diagnosis of Rett syndrome. Designed together with the IRSF Rett Syndrome Center of Excellence Network medical directors, this study collects data on the signs and symptoms of Rett syndrome as reported by the Rett syndrome experts and by the caregivers of individuals with Rett syndrome. This study will be used to develop consensus based guidelines for the care of your loved ones with Rett syndrome and to facilitate the development of better clinical trials and other aspects of the drug development path for Rett syndrome.

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