We are committed to…

… understanding the relationship between a person's genes and severe rare neurodevelopmental disorders – namely Rett syndrome-like phenotypes and early-onset severe epilepsies – in order to help patients and families.

Understanding the causes of Rett-like phenotypes and severe epilepsy may in the future improve the ability to diagnose and treat children with these rare neurodevelopmental disorders.

Interested in learning more?

For more information on our study (HSM#12-00798) or on how to participate go to:

http://pintolab.mssm.edu

We are happy to answer any questions you may have

ndd.project@mssm.edu

(or contact us at 212-824-9065; Monday to Friday 8:00am to 4:00pm)

Participating Institutions

seaver autism center
for Research & Treatment at Mount Sinai
Committed to Research. Dedicated to Care.

Mount Sinai *The Mindich Child Health and Development Institute*

NeuroDevelopment

Genetics of Rett-like phenotypes and severe epilepsies

Icahn School of Medicine at Mount Sinai

What are we studying?

Rett syndrome is a rare genetic disorder of the central nervous system that causes developmental delay. Children initially have a normal development, but between 6-24 months of age their development stagnates or regresses. Mutations in three genes, *MECP2*, *CDKL5* and *FOXG1* can cause the disorder, but in a significant proportion of children with Rett-like phenotypes mutations in these genes cannot be identified. Epilepsy often occurs in Rett syndrome or Rett-like phenotypes.

We are interested in learning how the genetic changes can affect brain development and subsequently, lead to disease. This study will examine similarities and differences in the genes of people with Rett-like phenotypes and/or severe epilepsy and their family members.

What is involved?

Subjects and their family members will donate a blood sample, in the same way blood is drawn at a doctor's office.

All samples will be given an anonymous code number, and used only for research purposes. No names or personal information will be linked to the samples.

Why is this important?

In a significant number of individuals with a Rett-like phenotype and/or severe epilepsy, genetic changes cannot be identified.

This study will use your samples for research purposes only, meaning that what we learn from the analysis of an individual's sample has no direct benefit for the individual.

However, by gathering information from many individuals, we hope to learn more about how changes affecting genes can lead to these rare disorders.

We are recruiting people with Rett-like phenotypes and/or severe epilepsies for a systematic genetic screening using high-throughput sequencing approaches and evolutionary concepts to identify novel genes and factors involved in these diseases.

The participation of many individuals with these disorders and their family members is the necessary foundation for making these discoveries.

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GCO# 12-1490

Who can participate?

- Children and young adults with a diagnosis of severe epilepsy or a Rett-like phenotype without mutations in *MECP2* and *CDKL5* are eligible to participate.
- Parents and family members of those with these disorders are eligible to participate whether or not they have the disorder. Research is usually more informative when both parents are enrolled in their child's study.

If I participate, what will I do?

If you agree to participate, a questionnaire will be completed, you will participate in an interview, and donate a blood sample.

Who should I contact?

If you are interested in participating, please call or email us.

Who is the research team?

The Pinto laboratory is a research team led by Dr. Dalila Pinto.

We are located at the:
Icahn School of Medicine at Mount Sinai
Department of Psychiatry
Hess Center for Science and Medicine
1470 Madison Avenue, 8th floor
10029New York City
212-824-9065
(Mon to Fri- 8:00AM-4:00PM)