OUR RESEARCH PROGRAM

The Neurodevelopmental Disorders Research Program at the Icahn School of Medicine at Mount Sinai leads cutting edge research in the field of genetics and genomics. Working together with families, our aim is to identify genes that affect neurodevelopment and advance clinical treatments.

Led by Dr. Dalila Pinto, the Neurodevelopmental Disorders Research Program is home to a multidisciplinary team of researchers and physicians who use genetics, molecular biology, and neuroimaging to advance our understanding of neurodevelopment.

Our research program focuses on the study of neurodevelopmental conditions, including:

- Autism Spectrum Disorders
- Pediatric Epilepsy
- Developmental Delay
- Intellectual Disability
- Specific Conditions including:
  - Landau-Kleffner Syndrome
  - Lennox-Gastaut Syndrome
  - Rett-like Syndrome (*MECP2*-negative)

Visit our website
HTTPS://PINTOLAB.MSSM.EDU/
to learn more about current studies
and how to participate.

Like us on Facebook.
Facebook.com/NDDProgram

Follow us on Twitter.
Twitter.com/NDDProgram

Icahn School of Medicine at Mount Sinai

PINTO LABORATORY
One Gustave L. Levy Place
Box 1230
New York, NY 10029-6574
NDD.Project@mssm.edu
212-824-9065

The Neurodevelopmental Disorders Research Program

Icahn School of Medicine at Mount Sinai

The Neurodevelopmental Disorders Research Program

Genetics Study of
Neurodevelopmental Conditions

Your DNA could help us better understand the genetics of neurodevelopment!

If you, or one of your family members, has been diagnosed with a neurodevelopmental condition, you may be eligible to participate in our research study!

Participation in our study takes roughly
30-60 minutes and involves a:

- Blood Draw
- Medical History Questionnaire
- Brief Physical Exam

Additionally, we will ask for copies of your:

- Previous Neuropsychological Evaluations
- Individualized Educational Program (IEP) Report
- Previous Genetic Testing (when applicable)

WHY SHOULD MY FAMILY PARTICIPATE?

The genetic alterations involved in neurodevelopmental disorders have yet to be fully identified.

This study will use your samples for research purposes. Relevant results may be shared with your physician . Study results are not diagnostic.

By gathering information from many
individuals, we hope to learn more about how changes in genes can lead to neurodevelopmental disorders. Mapping both shared and distinct changes in families will inform about the biological basis of neurodevelopmental disorders.

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

HOW IS MY IDENTITY PROTECTED?

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 . You may also remain anonymous should you choose.

WHAT IS WHOLE-EXOME OR WHOLE-GENOME SEQUENCING?

Whole genome sequencing is used to "read" your entire genetic code or "genome". Your genome is made up of a chemical called deoxyribonucleic acid (DNA), which is stored in almost every cell in your body.

Whole-exome sequencing focuses only on the parts of your genome that contain genes (or “exome”), which encode the
information to make all proteins in your body.

Sequencing "reads" each letter of your DNA (genome or exome) and finds
changes (also called “variants” or “mutations”) in your genes that may cause disease or affect your risk for a disease.

WHAT IS A *DE NOVO* VARIANT?

***De novo* variants** are genetic alterations that have not been inherited from your biological parents. Also called new mutations or new variants, *de novo* variants are believed to play a prominent role in neurodevelopmental disease.

For more information on how to
participate, please contact our research team at
212-824-9065
or e-mail us at
NDD.PROJECT@MSSM.EDU