

**We are  
committed to...**

... understanding the relationship between a person's genes and neurodevelopmental disorders in order to help patients and families.

The neurodevelopmental disorders we study are autism spectrum disorders (ASD) with epilepsy.

Understanding the causes of these disorders may in the future improve the ability to diagnose and treat children with these disorders.

## Interested in learning more?

For more information on our study (GCO# 15-1766) 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](mailto:ndd.project@mssm.edu)

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

## Participating Institutions



**Mount Sinai** *The Mindich Child Health and Development Institute*



## Genetics of Autism Spectrum Disorders (ASD) and Epilepsy (or ASD-Epilepsy)



**Icahn School of Medicine at Mount Sinai**

## What are we studying?

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Autism spectrum disorders (ASD) with epilepsy is frequent in children and can have a genetic basis.

We are interested in learning how genetic changes can affect brain development, and, subsequently, lead to disease. This study will examine similarities and differences in the genes of people with ASD and epilepsy (ASD-epilepsy) and their family members.

ASD and epilepsy can co-occur in the same family or the same person, suggesting that they may be caused by similar or related genetic changes. By mapping both shared and distinct changes we will better understand the biological basis of each disorder.

## What is involved?

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Subjects with ASD-epilepsy 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?

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In a large percentage of ASD-epilepsy patients, the genetic changes have not yet been 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 ASD-epilepsy.

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

## IRB Approved

**11/21/2016 -11/20/2017**



## Who can participate?

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- Children and young adults with a diagnosis of autism and epilepsy (ASD-epilepsy) with *unknown* cause.
- Parents and family members of those with autism and epilepsy (ASD-epilepsy) are eligible to participate whether or not they have ASD-epilepsy. Research is usually more informative when both parents are enrolled in their child's study.

## If I participate, what will I do?

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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?

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

## Who is the research team?

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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  
New York, NY 10029

☎ 212-824-9065  
Monday to Friday 8:00am-4:00pm