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Sinai**

*Seaver Autism  
Center for Research  
and Treatment*

**Seaver Autism Center**

26<sup>TH</sup> ANNUAL

# **Advances in Autism Conference**

TOPIC

**Convergence in Autism  
From Many Genes to Few Pathways**

COURSE DIRECTOR

**Joseph D. Buxbaum, PhD**

**TUESDAY, MAY 24, 2022**

etc.venues | 601 Lexington, New York, NY 10022 | Online



# Advances in Autism Conference

Tuesday, May 24, 2022

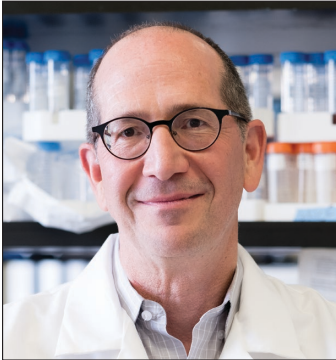
## SCHEDULE

<b>10:30 – 10:40 AM</b>	<b>Opening Remarks</b> Joseph D. Buxbaum, PhD
<b>10:40 – 11:05 AM</b>	<b>100s of Genes Identified in Autism</b> Joseph D. Buxbaum, PhD
<b>11:05 – 11:30 AM</b>	<b>Connecting Rare Autism Mutations to Common Pathways</b> Matthew Lalli, PhD
<b>11:30 – 11:40 AM</b>	<b>10-MINUTE BREAK</b>
<b>11:40 – 12:05 PM</b>	<b>Using Frogs to Translate Autism Genetics to Actionable Biology</b> Helen Willsey, PhD
<b>12:05 – 12:30 PM</b>	<b>Scalable Investigation of Autism Risk Genes in the Developing Brain</b> Xin Jin, PhD
<b>12:30 – 1:00 PM</b>	<b>LUNCH BREAK</b>
<b>1:00 – 1:25 PM</b>	<b>Building the Human Cortex: Molecular Logic of Neural Circuit Formation and Evolution</b> Nenad Sestan, MD, PhD
<b>1:25 – 1:50 PM</b>	<b>Studying Convergence in Autism Using 3D Models of Brain Development</b> Aaron Gordon, PhD
<b>1:50 – 2:00 PM</b>	<b>10-MINUTE BREAK</b>
<b>2:00 – 2:25 PM</b>	<b>Redefining Autism – Using the Neuroanatomy to Create Autism Subgroups</b> Jacob Ellegood, PhD
<b>2:25 – 2:30 PM</b>	<b>5-MINUTE BREAK</b>
<b>2:30 – 3:00 PM</b>	<b>All Speaker Panel</b>
<b>3:00 – 3:10 PM</b>	<b>Closing Remarks</b> Joseph D. Buxbaum, PhD

# Advances in Autism Conference

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## CONFERENCE SPEAKERS



### **Joseph D. Buxbaum, PhD**

Director, Seaver Autism Center for Research and Treatment at Mount Sinai  
Professor and Vice Chair for Research and Vice Chair for Mentoring, Department of Psychiatry

Joseph D. Buxbaum is a Professor of Psychiatry, Genetics and Genomic Sciences, and Neuroscience, and serves as the Director of the Seaver Autism Center for Research and Treatment and Vice Chair for Research and Mentoring of the Department of Psychiatry. Dr. Buxbaum is a renowned molecular geneticist whose research aims to understand the molecular and genetic basis of autism spectrum disorder (ASD) and other neurodevelopmental disorders, with the goal of developing novel therapeutics. Dr. Buxbaum is a founder and communicating Principal Investigator of the Autism Sequencing Consortium, currently analyzing whole exome sequencing from 65,000 individuals to identify ASD genes. In addition, his lab has numerous human stem cell lines ongoing and has characterized more

than a dozen rodent models for ASD and associated disorders. Dr. Buxbaum received his BSc in Math and Biology from Touro College, and his MSc and PhD in Neurobiology from the Weizmann Institute of Science in Israel. Dr. Buxbaum completed a Postdoctoral Fellowship in Molecular and Cellular Neuroscience at the Rockefeller University. Dr. Buxbaum was elected to the National Academy of Medicine in 2015 and was elected a Fellow of the American College of Neuropsychopharmacology in 2011 and a Fellow of the International Society for Autism Research in 2019.



### **Jacob Ellegood, PhD**

Research Associate, Department of Neurosciences and Mental Health, Hospital for Sick Children

Jacob Ellegood is a Research Associate at the Mouse Imaging Centre. Jacob joined MICE in January 2008 as a Postdoctoral Fellow. He received his PhD in Medical Sciences / Biomedical Engineering from the University of Alberta in Edmonton. He received his BSc in Physics from the University of Lethbridge, Alberta in 2002. His research aims to redefine the autism phenotype by examining the neuroanatomy in genetically altered mouse models relevant to Autism. He does this using advanced magnetic resonance imaging techniques to measure and compare volume and micro-structural changes throughout the brain. Jacob has currently examined over 150 different genetic models related to autism.



### **Aaron Gordon, PhD**

Project Scientist, UCLA

Dr. Gordon trained in the lab of Prof. Ori Peles at the Weizmann Institute before moving to the lab of Prof. Dan Geschwind at UCLA. During his time in the Geschwind lab Dr. Gordon has published a number of paper revolving around validating and using cortical organoid models to study brain development. He is especially interested in understanding how these models can be used to investigate early brain development changes in autism.

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## CONFERENCE SPEAKERS



### **Xin Jin, PhD**

Assistant Professor, Scripps Research

Xin Jin is an Assistant Professor in the Dorris Neuroscience Center and Department of Neuroscience at Scripps Research. She was previously a Junior Fellow at Harvard Society of Fellows working with the groups of Paola Arlotta, Feng Zhang, and Aviv Regev at the Broad Institute and Harvard University. She applies a scalable genetic screening tool, in vivo Perturb-seq, to study the function of a panel of Autism-associated risk genes in the developing neocortex with single-cell resolution. Xin obtained her PhD in neuroscience from The Rockefeller University, and her BSc in chemistry from MIT.



### **Matthew Lalli, PhD**

Instructor, Seaver Autism Center for Research and Treatment at Mount Sinai

Matthew Lalli, PhD, is an Instructor at the Seaver Autism Center for Research and Treatment at the Icahn School of Medicine at Mount Sinai where he is working with Dr. Joseph Buxbaum to study the functions of risk genes implicated in ASD using human brain cells. Matthew earned his PhD at the University of California Santa Barbara where he used induced pluripotent stem cells to study the rare neurodevelopmental disorder Williams syndrome. He trained as a Postdoctoral Scholar in the Department of Genetics at Washington University in St. Louis where he developed a new functional genomics technology using CRISPR/Cas9 and single-cell RNA-sequencing to elucidate the functions of many ASD risk genes in human neurons. Matthew is a Seaver Fellow and a NARSAD 2021 Young Investigator.



### **Nenad Sestan, MD, PhD**

Harvey and Kate Cushing Professor of Neuroscience,  
Professor, Comparative Medicine, Genetics, and Psychiatry,  
Kavli Institute for Neuroscience, Yale School of Medicine

Dr. Sestan obtained his MD from the University of Zagreb and his PhD in neurobiology from Yale University. Nenad Sestan's laboratory investigates how neural circuits form within the developing cerebral cortex, the outside part of the mammalian brain that processes our senses, commands motor activity, and helps us perform higher-order cognitive functions like language. He also studies how neural circuits were modified during human evolution and may become compromised in neuropsychiatric disorders. Recently, his laboratory developed a technology, they named BrainEx, that can restore circulation and cellular functions in the porcine brain after prolonged circulatory arrest and anoxia. This

finding also indicates that cell death in the mammalian brain takes place over a longer period of time than previously thought, and that brain cells are considerably more robust to ischemic injury than previously assumed.

Nenad Sestan is the recipient of several international honors and awards, including memberships in the National Academy of Medicine, Croatian Academy of Sciences and Arts, and Connecticut Academy of Science and Engineering, Nature's 10 who mattered in science, Constance Lieber Prize, Krieg Cortical Discoverer Prize, NARSAD Distinguished Investigator, McDonnell Scholar, and Krieg Cortical Scholar. He has been a member of the BrainSpan, BRAIN Initiative Cell Census Network, and PsychENCODE consortia.



# Advances in Autism Conference

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## CONFERENCE SPEAKERS



### **Helen Willsey, PhD**

Assistant Professor at UCSF & Investigator at Chan Zuckerberg Biohub

Helen Willsey, PhD, is an Assistant Professor in the Department of Psychiatry and Behavioral Sciences at UCSF and a Chan Zuckerberg Biohub Investigator. She has developed a high-throughput *Xenopus tropicalis* frog model system to 1) study many large-effect autism risk genes in parallel during brain development, and 2) screen small molecules to identify resiliency pathways to counter these effects. Her work has pinpointed forebrain neurogenesis as a nexus of risk in autism and estrogen signaling as a potential mitigating pathway. Dr. Willsey completed her postdoctoral training with Dr. Matthew State at UCSF in 2021, receiving the Psychiatry Department Trainee Research Award. She received a PhD in Genetics and the Carolyn Slayman Prize from Yale University in 2015. She received a BS degree in Biology and the Edward C. Horn Memorial Prize from Duke University in 2009.



# Advances in Autism Conference

Tuesday, May 24, 2022

## The Seaver Autism Center would like to acknowledge the members of its Associates Board for their generous contributions:

Alison Singer, Chair

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The Seaver Associates Board is a group of committed stakeholders—parents, grandparents, siblings, and others—who want to learn more and do more to support their loved ones with autism and to support the work at the Seaver Center.

For more information on how to join this group or support the Seaver Center, please contact Sarah Lynch, Communications and Marketing Associate, at [sarah.lynch@mssm.edu](mailto:sarah.lynch@mssm.edu) or 212-241-0349.



# Advances in Autism Conference

Tuesday, May 24, 2022

The Seaver Autism Center would like to thank the sponsors below for their generous support of the 26th annual Advances in Autism Conference

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Understanding  
ADNP SYNDROME  
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The Seaver Autism Center for Research and Treatment at Mount Sinai would like to thank the Beatrice and Samuel A. Seaver Foundation for their ongoing support and generosity since the founding of the Center in 1993. With their support, we have been able to make great strides in helping individuals with autism. We are honored by the Foundation's ongoing support, and we appreciate the opportunity provided by the 26th annual Conference to recognize their generosity.



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# ADNP Kids Research Foundation

Understanding **ADNP SYNDROME** / *Helsmoortel - VanDerAa Syndrome*



## What is ADNP Syndrome?

**ADNP Syndrome** (also known as Helsmoortel-VanDerAa Syndrome, HDVAS) is an extremely rare complex neurological genetic disorder caused by a mutation to the ADNP (Activity Dependent Neuroprotective Protein) gene. (estimated prevalence - 1 in 27,000 children in US & Europe)

The ADNP gene on chromosome 20q13 is crucial in the formation and maturation of the brain. When mutated, it can disrupt brain development, brain function and many other areas of the body. Most mutations are a spontaneous (de novo) change and it is equally seen in males and females.

ADNP Syndrome can cause the following conditions and affect the following systems:

- Neurological System
- Cardiovascular System
- Endocrine System
- Gastrointestinal System
- Immune System
- Gross Motor
- Fine Motor
- Oral Motor Planning
- Intellectual Delay
- Speech Delay
- Muscle Tone
- Vision / Hearing
- Growth Delay
- Sleep Disturbances
- Autism

ADNP is thought to be mutated in at least 0.17% of genetic autism cases, making it one of the most frequent ASD-associated genes known to date. Life expectancy is unknown and unique to the child's underlying conditions. *Children have similar features to Angelman Syndrome, Prader Willi Syndrome, Kleeftstra Syndrome, Smith-Magenis Syndrome, Williams, SYNGAP and Phelan-McDermid Syndrome.*

**UNIQUE BIOMARKER:** A recent study found that 81% of children with ADNP Syndrome have "early teeth eruption". Baby teeth come in extremely quickly, the teeth are usually very small, jagged, and with color differences. Most ADNP children have a full mouth of teeth by their 1<sup>st</sup> birthday, including molars. (Early tooth eruption isn't seen in any other syndrome making it a unique & early biomarker for ADNP)

## Treatment:

There is currently NO CURE or FDA approved treatment for ADNP Syndrome, however, the Seaver Autism Center has begun the worlds first drug trial for treatment, Phase 2 study of ketamine for ADNP syndrome

The treatment of individuals with ADNP Syndrome should be symptomatically directed towards the needs of each individual. Physical therapy, occupational therapy, behavioral therapy, sensory processing therapy, feeding therapy during infancy, music therapy and water therapy may all be useful in helping children with ADNP Syndrome reach their full potential. Specialized treatment for speech is extremely important because children with ADNP show symptoms of oral apraxia and dysarthria. Many individuals have quite severe difficulty in planning and coordinating the movement necessary for speech. These conditions are usually seen in traumatic brain injury patients who require aggressive rehabilitation therapy.

In ADNP, there are associated life threatening conditions including heart abnormalities, respiratory problems, sleep apnea, seizures, compromised immune systems, and complications from surgeries that require treatment from relevant specialists, such as neurologists, cardiologists, and surgeons.



TO LEARN MORE ABOUT ADNP SYNDROME VISIT

[www.adnpkids.com](http://www.adnpkids.com) | [www.adnpfoundation.org](http://www.adnpfoundation.org)

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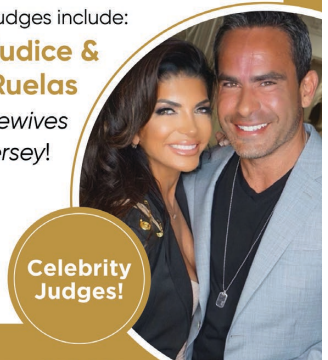
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Contact Julie Berger for more information:  
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was created by parents of children diagnosed with DDX3X Syndrome to support children like theirs, as well as adults, who have been diagnosed with this condition and the families that love them. As part of our mission, the Foundation supports existing research, education, and treatment programs focused on DDX3X Syndrome.

## DDX3X Syndrome

is a rare disease caused by a spontaneous mutation within the DDX3X gene at conception or can be inherited. DDX3X Syndrome is often misdiagnosed as autism spectrum disorder, cerebral palsy, Rett Syndrome, Dandy-Walker Syndrome, or a generic developmentally delayed label.



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Email [asgenetics@autismspeaks.org](mailto:asgenetics@autismspeaks.org) to learn more.



The Autism Care Network, a learning health system, offers a data registry to conduct clinical research and trials through a growing network of premier autism clinical and research sites across North America.

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Phelan-McDermid syndrome



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**What is Phelan-McDermid syndrome ?**

Phelan-McDermid Syndrome (PMS) is a rare genetic condition caused by a deletion or other structural change of the terminal end of chromosome 22 in the 22q13 region or a disease-causing mutation of the SHANK3 gene. PMS is sometimes called 22q13 Deletion Syndrome.

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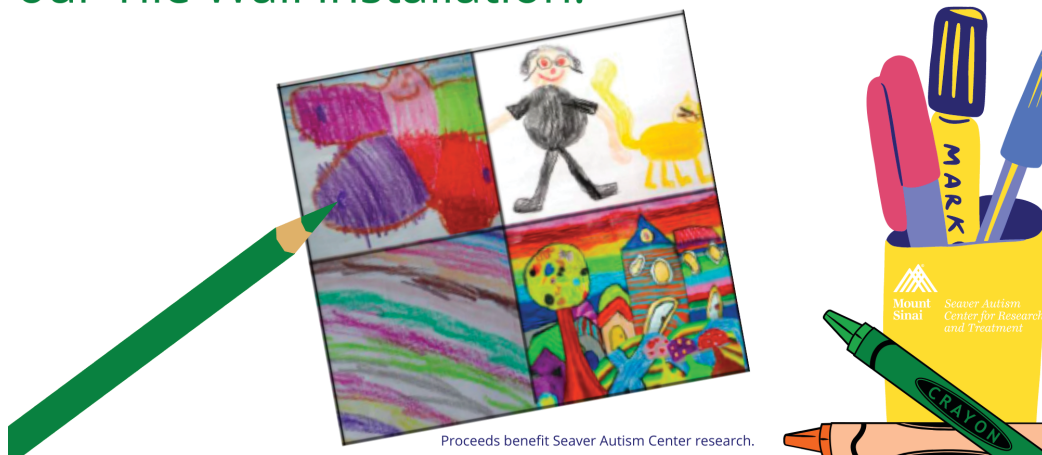
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# **Glossary of Autism-related Terms**

# Glossary of Autism-related Terms

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## **22q13 deletion syndrome**

Also known as Phelan-McDermid syndrome, a genetic disorder caused by a deletion of Shank3 on chromosome 22, characterized by general hypotonia, absent to delayed speech, and global developmental delays. Errors on the same gene are associated with autism spectrum disorder (ASD), so Phelan-McDermid Syndrome is considered a cause of ASD, accounting for about 1% of cases.

## **Aberrant Behavior Checklist – Community Version (ABC-CV)**

A parent report instrument with 5 subscales (irritability, social withdrawal, hyperactivity, stereotypic behavior, and inappropriate speech). It was developed for use with individuals with intellectual disability and is also frequently used in ASD.

## **ADNP (Activity Dependent Neuroprotective Protein) gene**

A gene linked to autism that provides instructions for making a protein that helps control the activity (expression) of other genes through a process called chromatin remodeling. By regulating gene expression, the ADNP protein is involved in many aspects of development. It is particularly important for regulation of genes involved in normal brain development, and it likely controls the activity of genes that direct the development and function of other body systems.

## **ADNP Syndrome**

A rare neurodevelopmental disorder caused by a mutation in the ADNP (Activity Dependent Neuroprotective Protein) gene, which affects brain formation and development, as well as brain function.

## **Allele**

One of two or more forms of a given gene; each gene can have different alleles and different alleles can result in different traits.

## **AMPA receptor**

A type of transmembrane receptor for glutamate that mediates excitatory synaptic transmission in the central nervous system.

## **Amygdala**

A part of the brain located in the front part of the temporal lobe that is part of the limbic system and involved in the processing and expression of emotions, especially anger and fear.

## **Apraxia**

Loss or impairment of the ability to execute complex coordinated movements without muscular or sensory impairment.

## **Asperger's Disorder**

An autism spectrum disorder characterized by significant difficulties in social interaction, along with restricted and repetitive patterns of behavior and interests. In earlier versions of the DSM, it was distinguished from Autistic Disorder by the absence of language delay and intellectual disability.

## **Astroglia**

Characteristic star-shaped glial cells in the brain and spinal cord that perform many functions, including: biochemical support of endothelial cells which form the blood-brain barrier; provision of nutrients to the nervous tissue; maintenance of extracellular ion balance; repair of the brain and spinal cord following traumatic injuries.

## **Attention Deficit Hyperactivity Disorder (ADHD)**

A neurobehavioral developmental disorder primarily characterized by attentional problems, hyperactivity, and impulsiveness.

## **Autism Centers of Excellence (ACE)**

The Autism Centers of Excellence (ACE) Program is a trans-NIH program that supports large-scale multidisciplinary studies on ASD. ACE research centers foster collaboration between teams of specialists who share the same facility to address a particular research problem in depth. ACE research networks consist of researchers at many facilities throughout the country who work together on a single research question.

### **Autism Spectrum Disorder (ASD)**

Autism Spectrum Disorder (ASD) is a group of developmental disorders characterized by widespread deficits in social interactions, communication, and restricted interests and repetitive behavior. The latest edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-5) no longer contains separate criteria for autism, Asperger's Syndrome, and Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS). They are now subsumed within the broader category of ASD.

### **Baclofen**

A muscle relaxer and an anti-spastic agent, used to treat muscle symptoms caused by multiple sclerosis, including spasm, pain, and stiffness.

### **Biomarker**

Refers to a broad subcategory of medical signs – that is, objective indications of medical state observed from outside the patient – which can be measured accurately and reproducibly.

### **Brain & Behavior Research Foundation (BBRF)**

A private not-for-profit organization. It is the largest donor-supported organization that supports research on brain and behavior disorders. Its raised funds for scientific research into the causes, cures, treatments and prevention of severe psychiatric brain and behavior disorders. Prior to 2011, the organization was known as Formerly known as National Alliance of Research on Schizophrenia and Depression (NARSAD).

### **CHARGE syndrome**

A syndrome caused by a genetic disorder – “CHARGE” is an acronym for congenital features seen in a number of newborn children, including Coloboma of the eye, Heart defects, Atresia of the nasal choanae, Retardation of growth and/or development, Genital and/or urinary abnormalities, and Ear abnormalities and deafness. These features are no longer used in making a diagnosis of CHARGE syndrome, but the name remains.

### **Childhood Disintegrative Disorder (CDD)**

A rare pervasive developmental disorder characterized by late onset (>3 years of age) of development delays in language, social function, and motor skills. Also known as Heller's Syndrome and disintegrative psychosis.

### **Children's Yale-Brown Obsessive Compulsive Scale modified for pervasive developmental disorders (CYBOCS-PDD)**

A questionnaire-based measure of obsessive and compulsive symptoms.

### **Chromosome microarray**

A laboratory technique that is used for the identification of structural alterations of the chromosomes, including deletions or duplications of chromosomes segments. It is often used as a diagnostic tool in individuals with unexplained intellectual disability and autism spectrum disorder.

### **Clinical Global Impressions (CGI) scale**

The CGI Scale (Guy 1976) is a standardized assessment tool that allows the clinician to rate the severity of illness, change over time, and efficacy of medication, taking into account the patient's clinical condition and the severity of side effects. The CGI Scale is widely used in clinical psychopharmacology trials as an outcome measure.

### **Comorbid**

Coexisting or concomitant illness or symptoms in addition to the primary disease.

### **Control group**

In a clinical study, this is the group that does not receive the active treatment, in order to determine the effectiveness of the treatment being tested.

### **Copy number variation (CNV)**

A type of genetic variation due to an abnormal number of copies of a chromosomal region, including deletions (removal of the region) and duplications (gain of extra copies).

### **Cornelia de Lange syndrome**

A rare genetic syndrome associated with autism and characterized by distinctive facial appearance, growth deficiency, feeding difficulties, psychomotor delay, behavioral problems, and malformations that mainly involve the upper extremities.

### **Corpus callosum**

The arched bridge of nervous tissue that connects the two brain hemispheres, allowing communication between the right and left sides of the brain.

### **CSF**

Cerebral Spinal Fluid (CSF) clear bodily fluid that occupies the subarachnoid space and ventricles in the brain and spinal cord. The CSF acts to cushion the brain inside the skull.

### **CYFIP1 heterozygotes**

Cytoplasmic Functional Mental Retardation-1 Interacting Protein 1 is the protein encoded by the CYFIP1 gene. Mutations in CYFIP1 are associated with autism and a mouse model with one copy of CYFIP1 missing is called a heterozygote.

### **Cysteine**

A non-essential amino acid synthesized in humans.

### **DDX3X gene**

A gene linked to intellectual disability and autism that encodes a conserved DEAD-box RNA helicase which is important in a variety of cellular processes, including transcription, splicing, RNA transport, and translation.

### **DDX3X Syndrome**

DDX3X syndrome is a recently discovered disorder in females with developmental delay and/or intellectual disability. The first girls and women with this disorder were reported in 2015. DDX3X syndrome occurs when one of the two copies of the DDX3X gene has lost its normal function.

### **De novo mutation**

An alteration in a gene that is present for the first time in one family member as a result of a mutation in a germ cell (egg or sperm) of one of the parents or in the fertilized egg itself

### **Diffusion Tensor Imaging (DTI)**

A magnetic resonance imaging (MRI) technique that enables the measurement of the diffusion of water in tissue in order to produce images of neural tracts.

### **Dizygotic (DZ) twins**

Commonly known as fraternal twins, this happens when two eggs are independently fertilized by two different sperm cells. Dizygotic twins share the same amount of genetic material as non-twin siblings (50%).

### **Double blind treatment**

A clinical trial where neither the investigator nor the subjects know which condition they are assigned to (i.e., control or experimental group).

### **Down Syndrome**

A genetic syndrome characterized by intellectual disability, low muscle tone, heart defects, increased risk of thyroid disease, increased risk of some types of cancers, and differences in facial features.

### **Dual diagnosis**

Co-occurring disorders

### **Duplications**

Any duplication of a region of DNA that contains a gene; it may occur as an error in recombination, a transposition event, or the duplication of an entire chromosome.

**Electroencephalography (EEG)**

A measure of electrical activity of the brain waves that is typically used to evaluate seizure disorders.

**Epidemiological studies**

A study on human populations which attempts to link human health effects to a specified cause.

**Epidemiology**

The study of factors affecting the health and illness of populations, and serves as the foundation and logic of interventions made in the interest of public health and preventative medicine.

**Epilepsy**

A neurological disorder characterized by recurrent episodes of seizures manifesting with symptoms that can vary from person to person.

**Etiology**

The study of the causes of diseases.

**FOXP1 gene**

A gene linked to autism that belongs to subfamily P of the forkhead box (FOX) transcription factor family. Forkhead box transcription factors play important roles in the regulation of tissue- and cell type-specific gene transcription during both development and adulthood.

**FOXP1 Syndrome**

A genetic disorder caused by a mutation in the FOXP1 (forkhead box protein P1) gene, which causes intellectual disability (ID) and language impairment.

**Fragile X syndrome**

A genetic disorder caused by mutation of the FMR1 gene on the X chromosome. Aside from intellectual disability, prominent characteristics of the syndrome include an elongated face, large or protruding ears, flat feet, larger testes (macroorchidism), low muscle tone, and autism.

**Frontal lobes**

One of the four major lobes of the brain, located at the front of each cerebral hemisphere and positioned anterior to (in front of) the parietal lobes and above and anterior to the temporal lobes (i.e. directly behind the forehead or “temple”).

**Functional Magnetic Resonance Imaging (fMRI)**

A type of specialized magnetic resonance imaging (MRI) scan. It measures brain activity by detecting changes in blood oxygenation and flow that occur in response to neural activity

**Fusiform gyrus**

A part of the brain located on the ventral surface of the temporal lobe. The fusiform gyrus plays an important role in face recognition.

**Genotype**

The genetic makeup, as distinguished from the physical appearance, of an organism or a group of organisms.

**Glutathione**

A tripeptide antioxidant.

**Heritability**

The proportion of phenotypic variation in a population that is attributable to genetic variation among individuals.

**Het mice**

Refers to mice that are heterozygous for a particular gene – see heterozygote.



**Heterogeneous disorder**

A disorder that has multiple origins.

**Heterozygote**

An organism is heterozygous for a particular gene when two different alleles occupy the gene's position (locus) on the homologous chromosomes.

**Hippocampus**

A convoluted, seahorse-shaped structure in the temporal lobe of the brain. It forms part of the limbic system and is involved in the processing of emotions and memory.

**Idiopathic**

Of unknown cause.

**Indel**

A type of genetic variation that is due to the duplication (insertion) or removal (deletion) of a small region of DNA, typically inside a gene. It can result in a genetic lesion and often causes the loss of functionality of the protein encoded by the gene.

**Institutional Review Board (IRB)**

A committee that has been formally designated to approve, monitor, and review biomedical and behavioral research involving humans with the aim to protect the rights and welfare of the research subjects.

**Insulin-like Growth Factor (IGF-1)**

A hormone that is similar in structure to insulin and plays an important role in growth. It is produced in the liver and its release is stimulated by growth hormone.

**Intellectual disability**

A neurodevelopmental disorder characterized by deficits in intellectual and cognitive abilities and a lack of skills required for daily living; these symptoms can range from moderate to severe.

**Intrauterine growth**

The size of a baby as a function of time since conception.

**Inverse agonist**

A pharmacological agent that binds to the same receptor as an agonist but reverses the activity of the receptors.

**Limbic system**

A group of interconnected structures of the brain including the hypothalamus, amygdala, and hippocampus that are located beneath the cortex, are common to all mammals, and are associated with emotions such as fear and pleasure, memory, motivation, and various autonomic functions.

**Long Term Depression (LTD)**

The process of a lasting decrease in synaptic signal strength between neurons. LTD is a form of learning and memory.

**Long Term Potentiation (LTP)**

The process of long-lasting enhancement of signal transmission between neurons. This process underlies forms of synaptic plasticity and learning and memory.

**Macrocephaly**

Abnormally enlarged head.

**Magnetic Resonance Imaging (MRI)**

A technique that uses a magnetic field and radio waves to create detailed images of the brain and body.

### **Magnetic Resonance Spectroscopy (MRS)**

A noninvasive technique that is similar to magnetic resonance imaging (MRI) but uses the concentrations of certain brain metabolites to study tissues of the human body and brain as opposed to using the signal from hydrogen protons to form anatomic images as in MRI.

### **Messenger RNA (mRNA)**

The form of RNA that mediates the transfer of genetic information from the cell nucleus to ribosomes in the cytoplasm, where it serves as a template for protein synthesis. It is synthesized from a DNA template during the process of transcription.

### **Metabolic disorders**

When abnormal chemical reactions in the body disrupt metabolism (the process the body uses to get or make energy from food). Examples include phenylketonuria (PKU) and thyroid conditions.

### **Methyl CpG binding protein 2 (MeCP2)**

A gene that causes Rett Syndrome when mutated and is essential for the normal function of nerve cells.

### **Microdeletion**

The loss of a tiny piece of a chromosome, a piece so small its absence is not apparent on ordinary examination (using a regular light microscope to look at chromosomes prepared in the usual fashion).

### **Microglia**

A type of cell in the brain and spinal fluid that acts to prevent infection and decrease inflammation in order to prevent damage to neural tissue.

### **Minocycline**

A broad spectrum tetracycline antibiotic.

### **Mitochondrial disorders**

A group of disorders relating to the mitochondria, which are organelles that act to convert the energy of food molecules into a type of energy that powers most cell functions.

### **Model system**

An experimental system used by researchers to investigate a biological process and often model a human disease. The systems can range from cells (e.g., the stem cells derived from the skin biopsies of a patient) to organisms, including invertebrate (e.g., fruit fly) and vertebrates (e.g., mouse and rats).

### **Monozygotic (MZ) twins**

This happens when one fertilized egg splits into two. Monozygotic twins are “identical” and share 100% of their genes.

### **mTOR**

A protein which regulates cell growth, cell proliferation, cell motility, cell survival, protein synthesis, and transcription.

### **NAA**

N-Acetyl-Aspartate – synthesized from the amino acid aspartic acid and plays a critical role in the formation of myelin in the brain. NAA also gives off the largest chemical signal in MRS (see above).

### **National Alliance for Research on Schizophrenia and Depression (NARSAD)**

A private, not-for-profit organization. It is the largest donor-supported organization that supports research on brain and behavior disorders. It raises funds for scientific research into the causes, cures, treatments and prevention of severe psychiatric brain and behavior disorders. In 2011, the organization rebranded itself and became the Brain & Behavior Research Foundation.

**National Institute of Child Health and Human Development (NICHD)**

One of 27 research institutes and centers that comprise the National Institutes of Health (NIH) which conducts and supports laboratory research, clinical trials, and epidemiological studies that explore health processes. It also examines the impact of disabilities, diseases, and variations on the lives of individuals.

**National Institute of Environmental Health Sciences (NIEHS)**

One of the 27 component organizations of the NIH whose mission is to reduce the burden of human illness and disability by understanding how the environment influences the development and progression of human disease.

**National Institute of Mental Health (NIMH)**

One of the 27 component organizations of the NIH and the largest research organization in the world specializing in mental illness.

**National Institute of Neurological Disorders and Stroke (NINDS)**

One of the 27 component organizations of the NIH which conducts and supports research to better understand traumatic brain injury and the biological mechanisms underlying damage to the brain.

**Neurodevelopmental disorders (NDD)**

A group of brain disorders with onset in the developmental period, often manifesting before the child enters the grade school. Symptoms can range from specific deficits to more broad impairments, and different NDD can co-exist in the same child. Intellectual disability and ASD belong to this group of disease.

**Neurofibromatosis**

A genetically-inherited disorder in which the nerve tissue grows tumors (i.e., neurofibromas) that may be harmless or may cause serious damage by compressing nerves and other tissues.

**Neuronal plasticity**

Refers to the ability of the brain to change as a function of experience. The brain's neuronal connections are able to change by adding, removing, or forming new cells.

**Neuropsychiatric syndromes**

A term referring to a group of brain-based disorders which manifest a combination of both neurological and psychiatric symptoms.

**Obsessive-Compulsive Disorder (OCD)**

A mental disorder characterized by intrusive thoughts (obsessions) that produce anxiety, and by repetitive behaviors (compulsions) aimed at reducing anxiety.

**Office of Mental Retardation and Developmental Disabilities (OMRDD)**

An independent agency in the state of New York whose mission is to provide services and conduct research for those with mental retardation and developmental disabilities. It is now called the Office of People With Developmental Disabilities (OPWDD)

**Oxytocin**

A mammalian hormone that acts primarily as a neurotransmitter in the brain. It is best known for its role in female reproduction (e.g., uterine contraction and milk let-down), but studies have also demonstrated its role in various behaviors, including social recognition, anxiety, trust, love, and maternal-infant attachment.

**Pathophysiology**

The group of biological processes and events occurring in an organism (physiology) in a disease state (pathology). For example, the pathophysiology of autism comprises the functional changes occurring in the body of a person with autism.

**Perseverating**

To repeat something insistently or redundantly

**Pervasive Developmental Disorder – Not Otherwise Specified (PDD-NOS)**

An autism spectrum disorder (ASD) characterized by social, language, and behavioral impairment. Patients with PDD-NOS have characteristics of autism, but do not fit full criteria according to the Diagnostic and Statistical Manual of Mental Disorders (DSM).

**Phelan-McDermid syndrome**

See 22q13 deletion syndrome.

**Phenotype**

The observable physical or biochemical characteristics of an organism, as determined by both genetic makeup and environmental influence.

**Phenylketonuria (PKU)**

A genetic disorder in which the body lacks the enzyme necessary to metabolize phenylalanine to tyrosine. Left untreated, the disorder can cause brain damage and progressive mental retardation as a result of the accumulation of phenylalanine and its breakdown products.

**Placebo**

An inactive substance or preparation used as a control in an experiment or test to determine the effectiveness of a given intervention.

**Polysomnography (PSG)**

A sleep study used as a diagnostic tool in sleep medicine.

**PP-LFS-induced LTD**

Paired-pulse low-frequency stimulation induced long term depression – see LTD.

**Precision medicine**

An emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person.

**Protein synthesis inhibitor**

A substance which stops or slows the growth or proliferation of cells by disrupting the processes that lead directly to the generation of new proteins.

**Psychoactive drug**

A drug that can produce mood changes or distorted perception.

**Psychotropic drug**

A drug that affects mental activity, behavior, or perception.

**Rare disorder**

A disease or disorder is defined as rare in the USA when it affects fewer than 200,000 Americans at any given time.

**Repetitive Behavior Scale – Revised (RBS-R)**

A rating tool that captures repetitive behaviors in autism.

**Rett Syndrome**

Also known as Rett's Disorder, a neurodevelopmental disorder characterized by autistic features, small hands and feet, and a deceleration of the rate of head growth (including microcephaly in some). Repetitive hand movements such as mouthing or wringing and breathing changes are also noted.

**Rodent model**

A mouse or rat used during the research and investigation of human disease, for the purpose of better understanding the disease without risk of causing harm to a human being during the process.

**Schizophrenia**

A chronic psychiatric disorder characterized by difficulties in recognizing and interpreting what is real, with symptoms including hallucinations, delusions, abnormal social and emotional behavior, and disordered thinking.

**Serotonin**

A neurotransmitter, derived from tryptophan, that is involved in sleep, depression, memory, and other neurological processes.

**Serotonin reuptake inhibitor (SSRI)**

A class of drugs that prolong the action of serotonin in the brain by inhibiting its reabsorption by neurons.

**SHANK3 gene**

A gene located on chromosome 22 (q13) that is mutated or deleted in Phelan-McDermid syndrome/22q13 deletion syndrome as described above.

**Short-chain acyl-coenzyme A dehydrogenase deficiency (SCADD)**

A fatty acid oxidation disorder which affects enzymes required to break down a certain group of fats called short chain fatty acids.

**Single nucleotide variation (SNV)**

A type of genetic variation that is due to the substitution of a single unit (nucleotide) within a gene. The substitution can be benign or can result in a genetic lesion because it alters or destroys the functions of the protein encoded by the gene.

**Stimming**

Repetitive body movement that is hypothesized to stimulate one or more senses. The term is shorthand for self-stimulation. Repetitive movement, or stereotypy, is often referred to as stimming under the hypothesis that it has a function related to sensory input.

**Stoppage**

In autism, “stoppage” usually refers to the observation that many families stop having additional children after a child with autism is diagnosed.

**Studies to Advance Autism Research and Treatment (STAART)**

In 2000, Congress passed the Children’s Health Act, legislation that mandated, among many things, the establishment of a new autism research network – at least five centers of excellence in autism research. In response, the five Institutes of the NIH Autism Coordinating Committee (NIMH, NICHD, NINDS, & NIEHS) implemented the STAART network program. Each center contributes to the autism research base in the areas of causes, diagnosis, early detection, prevention, and treatment of ASD.

**Synaptic plasticity**

The ability of the connection, or synapse, between two neurons to change in strength.

**Tardive dyskinesia**

A disorder characterized by restlessness and involuntary rolling of the tongue or twitching of the face, trunk, or limbs, usually occurring as a complication of long-term therapy with antipsychotic medication.

**Telescoping**

The tendency of most people, when looking back to events in the past, to move the dates in the past closer to the present.

**Temporal lobe**

The lower lateral lobe of either cerebral hemisphere, located in front of the occipital lobe and containing the sensory center of hearing in the brain.

**Teratogen**

A drug or other substance capable of interfering with the development of a fetus, causing birth defects.

**Theory of Mind**

The ability to understand the mental states – beliefs, feelings, intentions, etc. – of the self and others.

**Titration (in reference to medications)**

The gradual increasing of medication dose to carefully adjust from low dosage to therapeutic levels. A slow titration helps the body adapt to the medication and to reduce common side effects.

**Translational Research**

The process of applying knowledge from basic biology and clinical trials to techniques and tools that address critical medical needs.

**Treatment Emergent adverse effects**

In a clinical trial, any event not present prior to the initiation of the treatments or any event already present that worsens in either intensity or frequency following exposure to the treatments.

**Tuberous sclerosis complex**

A genetic disorder that causes non-malignant tumors to form in many different organs, primarily in the brain, eyes, heart, kidney, skin and lungs. Tuberous sclerosis is caused by a mutation in one of two genes, TSC1 and TSC2, which encode proteins that act as tumor growth suppressors and regulate cell proliferation and differentiation, and can present with autism.

**Turner syndrome**

A congenital condition of females associated with a defect or an absence of an X-chromosome, characterized by short stature, webbed neck, low set ears, broad chest, sexual underdevelopment, amenorrhea, heart disease, and endocrine disorders like hypothyroidism and diabetes.

**Uncinate Fasciculus (UF)**

A hook-shaped bundle of long association fibers connecting the frontal lobe with the anterior portion of the temporal lobe of the brain.

**Whole exome sequencing (WES)**

A technology that decodes the most meaningful fraction of the DNA of an individual, the exome. The human genome includes about 22,000 protein-coding genes. Each gene contains exons, functional units that translate the genetic information encrypted in each gene into a protein with specific functions in the cell. The entire gene repertoire of an individual is called the genome, and the collection of all exons is the exome.

**Williams syndrome**

A genetic neurodevelopmental disorder caused by a deletion of genetic material on chromosome 7 and characterized by a distinctive, “elfin” facial appearance, along with a low nasal bridge; an unusually cheerful demeanor and ease with strangers; and developmental delay coupled with unusual language skills. Patients are also at higher risk of cardiovascular problems, gastrointestinal problems, hypercalcemia, diabetes, and autism.

*The Seaver Autism Center would like to thank Terri Rosenblum for her contributions to this glossary.*





# Advances in Autism Conference

Tuesday, May 24, 2022

Notes

A series of horizontal lines for taking notes.













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