24TH ANNUAL

Seaver Autism Center

Advances in Autism Conference

THURSDAY, SEPTEMBER 17, 2020

TOPIC

Profound Autism
Understanding Needs and Tailoring Support

VIRTUAL CONFERENCE

COURSE DIRECTOR
Joseph D. Buxbaum, PhD
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CONFERENCE SPEAKERS

Evdokia Anagnostou, MD is a Child Neurologist and Professor of Pediatrics at the University of Toronto and Assistant Director of Holland Bloorview’s Research Institute. As a Senior Clinician Scientist, she co-leads of the Autism Research Centre (ARC) at Holland Bloorview and University of Toronto. She holds a Canada Research Chair in translational therapeutics in Autism Spectrum Disorder (ASD) and the Dr. Stuart D. Sims Chair in Autism at Holland Bloorview. With a passion for improving outcomes and quality of life for children with autism spectrum disorders and their families, Dr. Anagnostou has received extensive international funding (~40 million dollars over the last 10 years) to understand how our emerging knowledge of genomic and systems biology findings in ASD and related developmental conditions translate into novel treatments for children. She is well published (~130 publications, 2 books, several book chapters) and speaks around the world on autism research and health systems innovation. She currently serves as the Global Senior Leader for the International Society for Autism Research (INSAR), representing North America.

Joseph D. Buxbaum, PhD 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 is the Deputy Chair 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 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 38,000 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.

Eron Friedlaender, MD, MPH is a Professor of Clinical Pediatrics at the University of Pennsylvania Perelman School of Medicine and an attending physician in the Division of Emergency Medicine at the Children’s Hospital of Philadelphia. Her research has centered on how conditions in the built environment relate to injury risk as well as documenting ways in which individuals with autism are vulnerable within health care systems. She leads program development supported by ongoing research initiatives to shape a comprehensive approach to the care of children with autism and related developmental disabilities within the hospital environment. Much of this work centers on translating successful interventions for children with autism within educational systems to health care settings. Eron has experience in qualitative research methodology in injury-related investigation as well as in directing needs assessments of individuals with social disabilities and among health care providers. As an advocate for those with autism, she has developed and taught models for community inclusion nationally. Eron also has advanced interdisciplinary programming within the hospital system for educational, quality and safety initiatives. She now serves as a core member of the cross-disciplinary team at MIXdesign, an inclusive-design consultancy of experts in architecture, design, diversity and inclusion, and policy. Dr. Friedlaender is also leading interdisciplinary investigation into the educational, quality, systems and care practices embedded in clinical event debriefings and serves as a trained facilitator in master communication skills.
CONFERENCE SPEAKERS

Ana Kostic, PhD is a clinical scientist with experience in drug development, biomarkers, and patient selection. She recently joined Icahn Medical School at Mount Sinai as Director of Drug Discovery and Development at the Seaver Autism Center for Research and Treatment. The main focus of her research will be to identify potential drug candidates for treatment of autism, design experimental strategies for testing in neuronal cell systems and animal models as well as discovery and validation of molecular biomarkers in autism.

Prior to assuming her current position, Dr. Kostic spent eleven years in the biotech/pharmaceutical industry working in various roles across preclinical, clinical and precision medicine at Regeneron Pharmaceuticals and as Senior Director of Translational Medicine at Kiniksa Pharmaceuticals. Dr. Kostic received her PhD and postdoctoral training in molecular and cell biology at Columbia University.

Amy S.F. Lutz’s writing about severe autism has been featured on many platforms, including Psychology Today, The Atlantic, Slate, and Spectrum. Her first book, Each Day I Like It Better: Autism, ECT, and the Treatment of Our Most Impaired Children, was published in 2014 and her second book, a collection of essays called We Walk: Life with Severe Autism, will be published in October. She is a founding board member of the National Council on Severe Autism (NCSA) and is currently pursuing her doctorate in the history of medicine at the University of Pennsylvania. She lives outside Philadelphia with her husband and five children.

Paige Siper, PhD, is a licensed clinical psychologist, Chief Psychologist of the Seaver Autism Center for Research and Treatment, and an Assistant Professor in the Department of Psychiatry. She has expertise in the diagnosis, neuropsychological assessment, and treatment of children and adults with a variety of neurodevelopmental disorders (NDDs). Dr. Siper’s research focuses on sensory processing and biomarker discovery using electrophysiological and behavioral approaches. Dr. Siper is the co-developer of the Sensory Assessment for Neurodevelopmental Disorders (SAND), which is the first clinician-administered observation and corresponding caregiver interview to quantify sensory reactivity symptoms specific to NDDs.
Helen Tager-Flusberg, PhD received her Bachelors in Science in Psychology from University College London, and her doctorate in Experimental Psychology from Harvard University. Since 2001 she has been at Boston University, where she is now Professor of Psychological and Brain Sciences, and Director of the Center for Autism Research Excellence. She has devoted her lengthy career to conducting research on autism and other neurodevelopmental disorders including children with developmental language disorders and genetic syndromes, exploring variability in phenotypic expression and investigating developmental and intervention-based changes in language and social cognition using behavioral and brain imaging methodologies. Her research has been funded by the federal government and private foundations and she has led several multi-site multidisciplinary autism research programs including NIH-sponsored CPEA, STAART and ACE Centers. She has edited seven books and written over 200 journal articles and book chapters. She is the Past President of INSAR (2011-2013) and received the INSAR Lifetime Achievement Award in 2020 for her lasting contributions to research on autism. She serves on the editorial board of several professional journals and is Section Editor (Cognition and Behavior) for the Journal of Neurodevelopmental Disorders. She regularly presents her work at scientific and professional conferences and to parent advocacy groups and other stakeholders in the US and in countries around the world.

Audrey Thurm, PhD is Director of the Neurodevelopmental and Behavioral Phenotyping Service in the Office of the Clinical Director, part of the National Institute of Mental Health (NIMH)'s Intramural Research Program (IRP). After receiving a BS in human development from Cornell University, she received training in child clinical psychology at DePaul University, trained as an intern at Boston Children’s Hospital/Harvard Medical School, and conducted a post-doctoral fellowship at Johns Hopkins School of Medicine. She has been at NIMH since 2002, serving in the extramural program until 2006, at which time she moved to the IRP to engage in research on autism spectrum disorder (ASD) and other related neurodevelopmental disorders. Through the Neurodevelopmental and Behavioral Phenotyping Service, Dr. Thurm’s research interests focus on evaluating and improving upon diagnostic and cognitive assessment instrumentation through longitudinal studies of risk and characterization of neurodevelopmental disorders. In addition to studying the prodromal and post-diagnostic characterization of idiopathic ASD, studies also focus on phenotypic explorations of genetic disorders associated with Intellectual Disability and ASD. A goal of this research is to improve instrumentation to allow for more finely-tuned developmental assessments that distinguish various phenotype-genotype relationships and serve as useful treatment outcome measures.
The Seaver Autism Center would like to acknowledge the members of its Associates Board for their generous contributions:

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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 or 212-241-0349.
The Seaver Autism Center would like to thank the sponsors below for their generous support of the 24th annual Advances in Autism Conference.
The Seaver Autism Center thanks the Seaver Foundation!

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 24th annual Conference to recognize their generosity.
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 underlining conditions. Children have similar features to Angelman Syndrome, Prader Willi Syndrome, Kleefstra 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 1st 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.
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CureSHANK has a singular purpose:
To accelerate the development of treatments for Phelan-McDermid Syndrome and SHANK-related disorders. Our approach is to identify and fund projects that overcome critical barriers to successful drug development and to coordinate scientific efforts to improve efficiency and speed in the field.

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Pioneering personalized epigenetic medicine in autism
Rumi Scientific is pleased to be involved with Dr. Buxbaum and supporting the Seaver Autism Center in their important and innovative work.

Learn more at autismspeaks.org/research
At NeuCyte, we are working to make a difference in the lives of patients suffering from neurological disorders. Our unique technology allows us to develop human neural in vitro disease models using patient-derived cells. By focusing on specific phenotypes identified in disease relevant cells, we aim to better understand the underlying disrupted biology leading to these disorders. This knowledge gives us unique and novel perspectives on treatment development. We are actively pursuing drug discovery programs for several genetically defined neurological diseases.

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Ovid Therapeutics is honored to be part of this community. More than ever, together we are strong, and our team is inspired by every family and the work of Seaver Autism Center for Research and Treatment. Thank you for including Ovid in this important event.
Thank You Seaver Autism Center
for your partnership and contributions to the understanding and therapeutic treatment of Phelan-McDermid Syndrome.

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.

For more information visit PMSF.ORG

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It is Seaver Autism Center’s goal to be the international leaders for precision medicine in autism and related disorders.

To learn more about the Center and our ongoing studies, please call 212-241-0961 or email theseavercenter@mssm.edu.
Glossary of Autism-related Terms
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 regulating 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 balance; repair of the brain and spinal cord following traumatic injuries.

Attention Deficit Hyperactivity Disorder (ADHD)
A neurobehavioral developmental disorder primarily characterized by attentional, 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 faculty to address a particular research problem in depth. ACE research networks consist of researchers at many faculties 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 interaction, communication, and restricted interests and repetitive behaviors. 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 indicators 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 chromosome 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 Funtional 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 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.

**FOX1 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 or removal of a small region of DNA, typically inside a gene. It can result in a genetic lesion and often causes the loss of function of the protein encoded by the gene.

**Institutional Review Board (IRB)**
A committee that has been formally designated to approve, monitor, and review all medical 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 invertebrates (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 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 reproductive functions (e.g., uterine contractility 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 perceptions.

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 variant (SNV)
A type of genetic variant 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 function 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.
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