



**Mount
Sinai**

*Seaver Autism
Center for Research
and Treatment*

Seaver Autism Center
27TH ANNUAL
**Advances in
Autism Conference**

TOPIC

**Precision Medicine
in Profound Autism**

COURSE DIRECTOR

Joseph D. Buxbaum, PhD

TUESDAY, MAY 23, 2023
VIRTUAL EVENT



Advances in Autism Conference

Tuesday, May 23, 2023

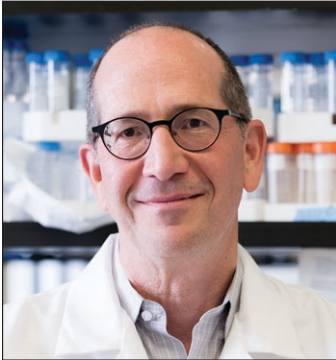
SCHEDULE

10:30 – 10:40 AM	Opening Remarks Alex Kolevzon, MD
10:40 – 11:05 AM	Genetics—First Approaches to Therapeutics in Autism Joseph D. Buxbaum, PhD
11:05 – 11:30 AM	Modelling Neurodevelopmental Disorders in a Dish Nael Nadif Kasri, PhD
11:30 – 11:40 AM	10-MINUTE BREAK
11:40 – 12:05 PM	Making a New Medicine Bill Polvino, MD
12:05 – 12:30 PM	Building a Pharmaceutical Industry, Academia, and Patient Advocacy Ecosystem for the Efficient Development of Therapeutics Rachel Groth, PhD
12:30 – 1:00 PM	LUNCH BREAK
1:00 – 1:25 PM	Predicting Treatment Response to Growth Hormone in Phelan-McDermid Syndrome and Idiopathic Autism Alex Kolevzon, MD
1:25 – 1:50 PM	Milestones on the Path Towards Genetic Interventions for Autism Tim W. Yu, MD, PhD
1:50 – 2:00 PM	10-MINUTE BREAK
2:00 – 2:25 PM	Rett Syndrome, from Gene Discovery to First Approved Treatment: Opportunities and Challenges for Future Clinical Trials Jeffrey Neul, MD, PhD
2:25 – 2:30 PM	5-MINUTE BREAK
2:30 – 3:00 PM	All Speaker Panel
3:00 – 3:10 PM	Closing Remarks Alex Kolevzon, MD

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CONFERENCE SPEAKERS



Joseph D. Buxbaum, PhD

Professor of Psychiatry, Genetics and Genomic Sciences, and Neuroscience
Director, Seaver Autism Center for Research and Treatment

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 Vice Chair for Research and Vice Chair for Mentoring in the Department of Psychiatry at the Icahn School of Medicine at Mount Sinai. Dr. Buxbaum is a renowned molecular geneticist whose research aims to understand the molecular and genetic basis of autism 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 autism genes. In addition, his lab has numerous human stem cell lines

ongoing and has characterized more than a dozen rodent models for autism and associated disorders. In 2015, Dr. Buxbaum was elected to the National Academy of Medicine.



Rachel Groth, PhD

Vice President of Research, BridgeBio

Rachel Groth, PhD, is currently Vice President of Research at BridgeBio in Palo Alto, CA. She has more than a decade of experience leading drug discovery and early development efforts in industry across diverse therapeutic areas (e.g., neuroscience, immunology, renal, and oncology) and drug modalities (e.g., small molecules, monoclonal antibodies, and antisense oligonucleotides). Before joining BridgeBio, Rachel held roles of increasing responsibility at Pfizer and Biogen. Rachel earned her BA in Biology and Neuroscience from Macalester College, her PhD in Neuroscience from the University of Minnesota, and completed her postdoctoral training in Molecular and Cellular Physiology at Stanford University.



Nael Nadif Kasri, PhD

Professor of Medical Neuroscience for Neurodevelopmental Disorders, Department of Human Genetics Radboudumc Donders Institute for Brain, Cognition and Behaviour

Nael Nadif Kasri, PhD, is a professor of Medical Neuroscience for Neurodevelopmental Disorders in the Department of Human Genetics at Radboudumc Donders Institute for Brain, Cognition and Behaviour in the Netherlands. The focus of his current research is to understand the synaptic basis of neurodevelopmental disorders using in vitro human stem cell-based models. He studied biochemistry followed by a PhD (2004) at the KU Leuven. After his PhD he worked as a postdoctoral researcher (2010) at Cold Spring Harbor Laboratory, where he studied the role of disease genes in excitatory synapses. In 2011 he received the Hypatia Fellowship from the Radboud Medical Centre and started his independent research group.

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Alex Kolevzon, MD

Clinical Director, Seaver Autism Center for Research and Treatment at Mount Sinai
Director of Child and Adolescent Psychiatry, Mount Sinai Health System

Alex Kolevzon, MD is a child and adolescent psychiatrist and Professor of Psychiatry and Pediatrics at the Icahn School of Medicine at Mount Sinai. He serves as the Clinical Director of the Seaver Autism Center for Research and Treatment at Mount Sinai and as the Director of Child and Adolescent Psychiatry for the Mount Sinai Health System. His research interests pertain to understanding the neurobiology of autism and related neurodevelopmental disorders, biomarker discovery, and developing novel therapeutics. Most recently, his group has focused on studying specific rare genetic forms of autism, including Phelan-McDermid syndrome, ADNP syndrome, and FOXP1 syndrome in order to explore targets for pharmacological intervention. Dr. Kolevzon leads the Experimental Therapeutics Program at the Seaver Autism Center which conducts studies that range from small pilot trials to multi-centered pivotal FDA studies. He also leads a national rare disease clinical research network dedicated to clinical trial readiness and studying the natural history of Phelan-McDermid syndrome. He is a frequently invited speaker regionally, nationally, and internationally and is also extremely committed to medical student and residency education as an active teacher, mentor, and clinical supervisor.



Jeffrey Neul, MD PhD

Director, Vanderbilt Kennedy Center
Annette Shaffer Eskind Professor of Pediatrics, Vanderbilt University Medical Center

Jeffrey Neul, MD PhD attended medical and graduate school at the University of Chicago and then completed child neurology residency at Baylor College of Medicine and Texas Children's Hospital. He subsequently was a post-doctoral fellow in the laboratory of Huda Zoghbi at Baylor where his interest in Rett syndrome began. He is currently the Director of the Vanderbilt Kennedy Center and the Annette Shaffer Eskind Professor of Pediatrics at Vanderbilt University Medical Center. His clinical practice and research focus is on Rett syndrome and related genetically based neurodevelopmental disorders. His laboratory-based research explores disease mechanisms, tests new therapeutics, and discovers and evaluates biomarkers of disease improvement in Rett syndrome. His clinical research focuses on longitudinal natural history studies, development of outcome measures and biomarkers, and conducting clinical trials in novel therapeutic approaches to Rett syndrome.



Dr. Bill Polvino

CEO, Bridge Medicines

Dr. Bill Polvino, is a pharmaceutical start-up entrepreneur having financed and run biopharma companies as CEO and having raised approximately \$300 million over the course of his career. Dr. Polvino is presently CEO and on the board of directors at Bridge Medicines, he is executive chairman of the board at Traverse Biotech and he is a member of the board of directors at SAB Biotherapeutics. Prior to joining Bridge Medicines, Bill was CEO of a publicly traded company, Veloxis Pharmaceuticals, where he oversaw the successful development and commercial launch of the company's first drug product.

Previously Bill held senior positions with Helsinn Therapeutics, Merck, Wyeth and Theravance. Bill is a board-certified physician. He completed his residency in Internal Medicine at Massachusetts General Hospital and his fellowship in Clinical Pharmacology at the National Institute of Health, USA where he worked on the first human gene therapy protocols. He received his undergraduate degree in biology

from Boston College, his medical degree from Rutgers Medical School and is an inventor on six US patents.



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CONFERENCE SPEAKERS



Tim Yu, MD, PhD

Associate Professor, Division of Genetics & Genomics, Boston Children's Hospital & Harvard Medical School

Tim Yu, MD, PhD is an Associate Professor for the Division of Genetics & Genomics at Boston Children's Hospital & Harvard Medical School. His research group works at the intersection of genomics, informatics, and neurobiology to better understand and treat rare neurologic disease. A graduate of Harvard College, he completed MD-PhD training at UC San Francisco and neurology residency at Massachusetts General Hospital and Brigham and Women's Hospital. Research efforts span the range from gene discovery for autism and other neurodevelopmental disorders to clinical genome interpretation to the advancement of novel models for advancing individualized genomic medicine. He is staff physician at Boston Children's Hospital, an Associate Member of the Broad Institute, and founder of the N-of-1 Collaborative.



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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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Inna and Josh Needelman

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 Manager, at sarah.lynch@mssm.edu or 212-241-0349.



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**The Seaver Autism Center would like to thank
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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 27th annual Conference to recognize their generosity.



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The Gut-Brain Connection in Autism

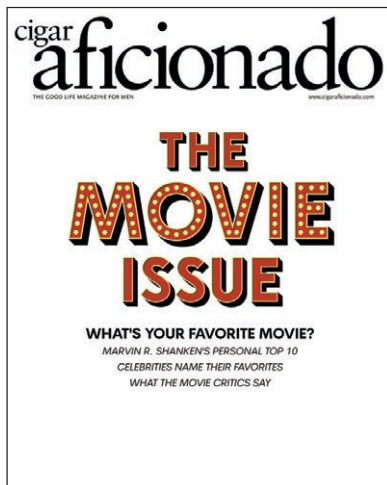
Did you know that our gut and our brain are connected? In fact, scientists have discovered that a new potential treatment leveraging this microbiome gut-brain connection may improve behaviors such as aggression, emotional meltdowns, and self-injury, in children with Autism Spectrum Disorder. A clinical trial is underway based on this research.

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Thank you to everyone at the Seaver Center for your excellent work toward making lives better for the families in our community.

-- Phelan-McDermid Syndrome Foundation



Glossary of Autism-related Terms

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 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.

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