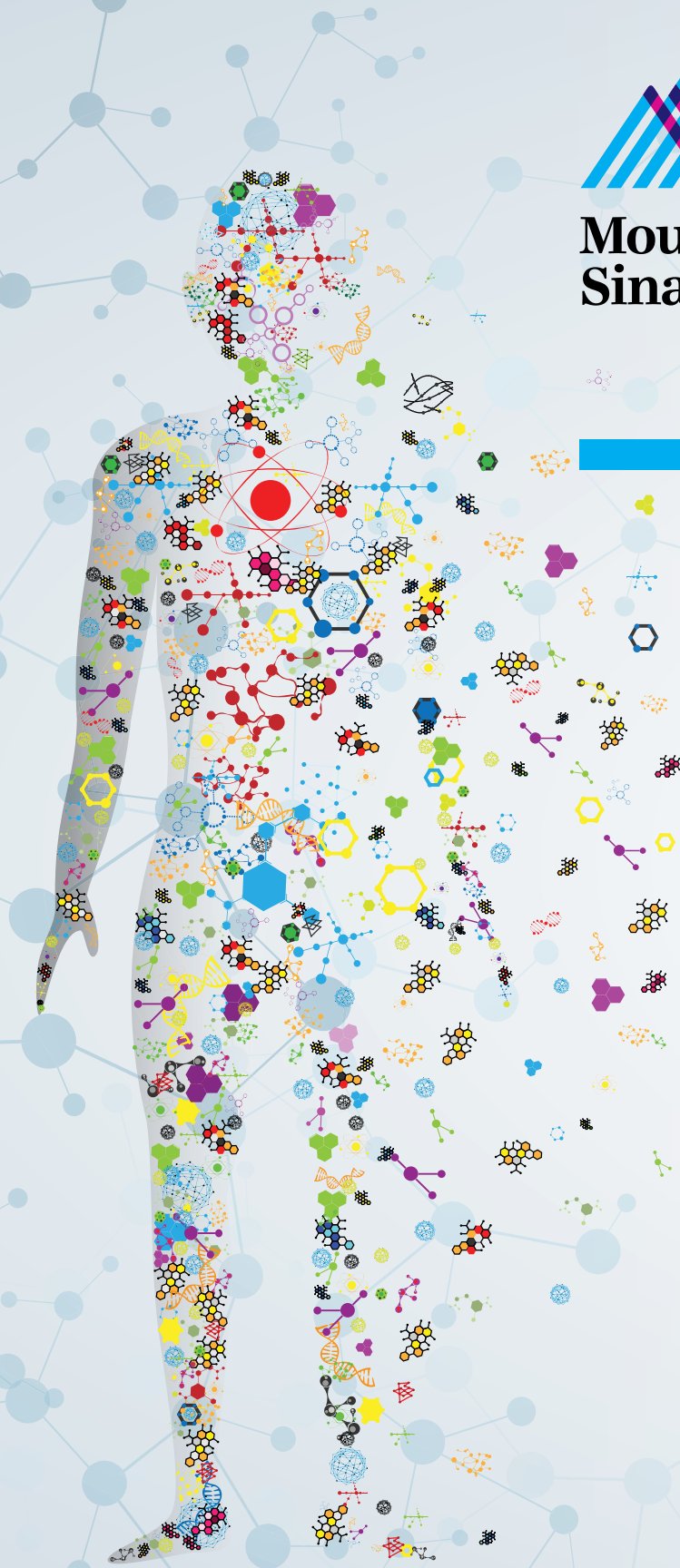

Mount Sinai

Seaver Autism Center for Research and Treatment

Seaver Autism Center

28TH ANNUAL

Advances in Autism Conference

TOPIC

Precision Therapeutics in Profound Autism

COURSE DIRECTOR

Joseph D. Buxbaum, PhD

THURSDAY, APRIL 18, 2024

New York Academy of Medicine
New York, NY

Advances in Autism Conference

Thursday, April 18, 2024

SCHEDULE

9:00 – 9:20 AM	**REGISTRATION**
9:30 – 9:35 AM	**Opening Remarks** Joseph D. Buxbaum, PhD
9:35 – 10:05 AM	**Gene Discovery as a Basis for Precision Medicine in Autism** Joseph D. Buxbaum, PhD *Director, Seaver Autism Center*
10:05 – 10:35 AM	**Biomarkers in Profound Autism: What, Why, and How?** Paige Siper, PhD *Chief Psychologist, Seaver Autism Center*
10:35 – 11:05 AM	**Multipronged Approach to Drug Discovery at the Seaver Autism Center** Ana Kostic, PhD *Director, Drug Discovery and Development, Seaver Autism Center*
11:10 – 11:30 AM	**Selections from "My World," an original musical by Jake Gluckman** Jake Gluckman *Clinical Research Coordinator, Seaver Autism Center*
11:30 – 12:30 PM	**LUNCH BREAK**
12:30 – 1:00 PM	**Today's Exome; Tomorrow's Insights and Innovations for Patients with Autism** Margo Gallegos, MS, CGC *Rare Disease Genomic Science Liaison, Ambry Genetics*
1:00 – 1:30 PM	**JAG201 for the Treatment of SHANK3 Haploinsufficiency** Daniel Gallo, PhD *Vice President, Medical Affairs, Jaguar Gene Therapy*
1:30 – 2:00 PM	**Bridging Gaps in CNS Disorders: The Power of Human Stem Cell Models** Samuele Marro, PhD *Assistant Professor, Neuroscience; Co-Director, Stem Cell Engineering Core, Icahn School of Medicine at Mount Sinai*
2:15 – 2:45 PM	**Speaker Panel**
2:45 – 3:00 PM	**Closing Remarks**

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, Psychiatry, Genetics and Genomic Sciences, and Neuroscience &
Vice Chair, Research and Mentoring, Psychiatry, Icahn School of Medicine at Mount Sinai

Dr. Buxbaum is a renowned molecular neuroscientist whose research aims to understand the molecular and genetic basis of autism spectrum disorder (ASD) and associated 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 60,000 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 International Society for Autism Research in 2019. Dr. Buxbaum is the author of more than 300 publications, and he is co-editor-in-chief of the journal Molecular Autism.

Margo Gallegos, MS, CGC

Rare Disease Genomic Science Liaison, Ambry Genetics

Margo Gallegos joined Ambry Genetics in 2020 as the Rare Disease and Cardiology Genomic Science Liaison for the Northeast United States territory. She previously worked as a pediatric genetic counselor at Children's National Medical Center in Washington, D.C, and an oncology genetic counselor at Anne Arundel Medical Center in Annapolis, MD. Her volunteer interests include public policy and licensure of genetic counselors both at the state and federal level. Margo received her Bachelors of Science degree in Cell Biology and Genetics from University of Maryland, College Park. She earned her Masters of Science degree in Genetic Counseling from University of South Carolina School of Medicine and is certified by the American Board of Genetic Counseling.

Dan Gallo, PhD

Head of Medical Affairs and Clinical Strategy, Jaguar Gene Therapy

Dan Gallo joined Jaguar Gene Therapy in January of 2022, where he has led medical affairs and clinical development strategy. During his tenure with Jaguar, Dan has worked closely with academic investigators and collaborators, including the Developmental Synaptopathies Consortium to develop and advance Jaguar's clinical program for JAG201 in SHANK3haploinsufficiency. Dan has a PhD in Cell and Molecular Biology from Northwestern University and has over 12 years of industry experience leading and supporting drug development in neurology and rare diseases.

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CONFERENCE SPEAKERS

Ana Kostic, PhD

Director, Drug Discovery and Development, Seaver Autism Center for Research and Treatment at Mount Sinai
Associate Professor, Psychiatry, Icahn School of Medicine at Mount Sinai
Director, Business Development & Licensing, Mount Sinai Innovation Partners

Ana Kostic is an Associate Professor of Psychiatry at the Icahn School of Medicine at Mount Sinai, and the Director of Drug Discovery and Development at the Seaver Autism Center for Research and Treatment. Dr. Kostic is a clinical scientist with expertise in drug development, biomarkers,patient selection and stratification. The main focus of her research is to identify potential drug candidates for treatment of autism, design experimental strategies for testing in neuronal cell systems and animal models, as well as to discover and validate molecular biomarkers in autism. Specifically, her group is interested in ADNP, DDX3X, FOXP1, and Phelan-McDermid syndromes, the most common single-gene causes of autism.

Prior to joining Mount Sinai, 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.

Samuele Marro, PhD

Assistant Professor, Neuroscience, Co-Director, Stem Cell Engineering Core,
Icahn School of Medicine at Mount Sinai

Dr. Marro is a distinguished cellular neuroscientist and an expert in pluripotent stem cell biology and genome editing. With over a decade of groundbreaking research, Dr. Marro has made significant strides in understanding cell identity and neurodevelopmental disorders. His pioneering work in direct lineage reprogramming has opened new avenues for regenerative medicine, notably in converting somatic cells into induced neuronal cells (iN cells) through innovative cross-lineage transdifferentiation techniques.

Under the mentorship of renowned scientists Marius Wernig and Nobel laureate Thomas C. Südhof, Dr. Marro has contributed extensively to the development of transcription factor-based protocols for generating specific neuron types from human induced pluripotent stem cells (iPSCs). His expertise in creating and optimizing protocols spans various cell types, including neurons, monocytes, macrophages, and microglia, underscoring their versatility in the field.

Currently leading two dynamic teams, Dr. Marro explores synaptic plasticity and its disruptions in Fragile X syndrome (FXS) within the Neuroscience Department of the Icahn School of Medicine at Mount Sinai. Their research is at the forefront of identifying the molecular mechanisms underlying homeostatic synaptic plasticity using advanced models like iPSC-derived neurons and cortical 3D organoids. Additionally, as co-director of the Stem Cell Engineering Core at Mount Sinai, Dr. Marro champions the application of gene editing to model and treat genetic disorders, having overseen the training of scientists and the generation of over 200 iPSC clones and 100 gene-edited loci since 2020.

Dr. Marro's contributions to neuroscience and stem cell research are not only pivotal in understanding complex neurological conditions but also instrumental in advancing therapeutic approaches for their treatment.

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CONFERENCE SPEAKERS

Paige Siper, PhD

Chief Psychologist, Seaver Autism Center for Research and Treatment at Mount Sinai
Associate Professor, Psychiatry, Icahn School of Medicine at Mount Sinai

Dr. Siper is a licensed clinical psychologist, Chief Psychologist of the Seaver Autism Center for Research and Treatment, and an Associate 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, including in profoundly affected participants. She 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.

SPECIAL PERFORMANCE

Selections from "My World," an original musical by Jake Gluckman

Jake Gluckman

Clinical Research Coordinator, Seaver Autism Center for Research and Treatment at Mount Sinai

Jake Gluckman is a Clinical Research Coordinator at Mount Sinai's Seaver Autism Center working directly with Dr. Dorothy Grice and the Tics, OCD, and Related Disorders Program. Before pivoting careers towards pursuing medicine, Jake studied musical theater performance and composition at Yale College and graduated in 2021 with a BA in American Studies. For his work on My World, an original musical, Jake received the Norman Holmes Pearson Price for best senior essay in American Studies, the Beekman Cannon Friends of Music Prize, and the Joseph Lentilhon Selden Memorial Award. In March 2023, he presented selections from the show to the Yale Child Study Center Grand Rounds. Upon graduating from Yale, Jake received the Yale Glee Club Service Through Music Fellowship to work as Programs Fellow for Hear Your Song, a nonprofit that empowers children and teens with serious illnesses through collaborative songwriting. After his fellowship, Jake completed his premedical coursework at Bryn Mawr College's Premedical Postbaccalaureate Program. He is thrilled to be starting medical school this summer and hopes to pursue a specialty in Pediatrics or Child and Adolescent Psychiatry.

About "My World"

Jake started writing the music, lyrics, and book for "My World" in 2017 after he heard his brother, Sam, who is autistic, deliver a speech at his graduation from Vista Life Innovations, a program that helps adults with special needs create a path towards independence. In the speech, Sam articulated a desire to become a marine biologist, and Jake realized for the first time that Sam had dreams that were larger than the life he was currently living. Written in collaboration with an artist on the spectrum, Sair Kaufman, "My World" follows Benjy, an ambitious and curious young autistic adult who, like Sam, dreams of becoming a marine biologist. Along the way, Benjy finds his first autistic friend, navigates his first crush, and he learns to express himself more authentically to his family. Today, Jake will perform a selection of songs from the show and will give brief narrations of the major scenes of the show in between. He will also share about his brother's reaction to the show, and how his brother has grown since the show's premiere in 2019. Jake dedicates this performance of "My World" to Sam, who has generously allowed Jake to share this version of his story over the past five years, and whose idealism and kindness inspires everyone around him.

Advances in Autism Conference

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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

Mary Lou and Tony Cancellieri

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Brian Fiumara

Sara Karp Golden

Rebecca Goodman-Stephens

Martin Lomazow

Shawna Makover

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.

Advances in Autism Conference

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The Seaver Autism Center would like to thank the sponsors below for their generous support of the 28th annual Advances in Autism Conference:

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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 28th annual Conference to recognize their generosity.

Mount Sinai

Hazel and Marvin R. Shanken
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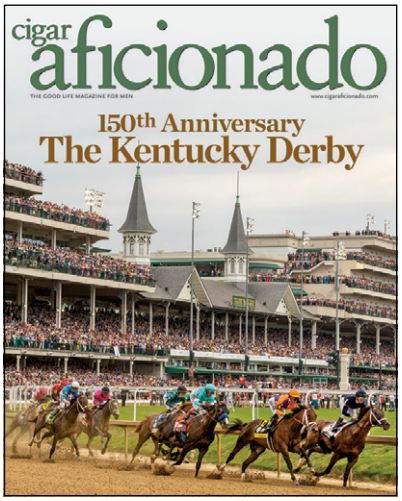

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CureSHANK is proud to support the
Seaver Autism Center for Research and Treatment!
We are so grateful for all that the Seaver Team does to help all of us living with and suffering from neurodevelopmental disorders.

JOIN US

OVERVIEW

Join us for the Phelan-McDermid Syndrome Drug Development Symposium, a global gathering to focus on the development of treatments for PMS. Open to academia and industry alike, the Symposium will be held at MassBioHub in Cambridge, MA.

Beginning with an evening poster reception on Thursday, April 25th, it will be followed on Friday, April 26th by presentations, panel discussions, and Q&A. A limited number of travel scholarships will be available to young investigators whose posters are accepted for presentation.

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Phelan-McDermid Syndrome
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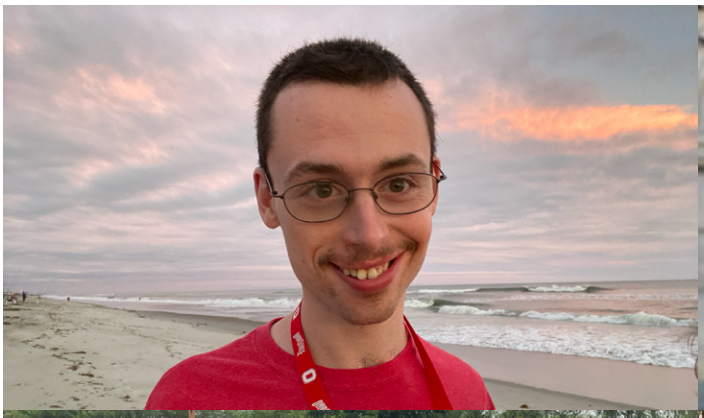

Thank you,

Seaver Autism Center

for your partnership and contributions to the understanding and therapeutic treatment of Phelan-McDermid syndrome.

Phelan-McDermid syndrome is a rare genetic disorder that can affect many critical functions in a person's body, from learning and communicating to eating and sleeping.

The Phelan-McDermid Syndrome Foundation is doing everything it takes to make today better and the future brighter for everyone living with this complex condition – from the moment of diagnosis to the delivery of treatments and cures.

Learn more at **pmsf.org**

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Visit: **AutismBrainNet.org**

Autism BrainNet is a program of the Simons Foundation Autism Research Initiative (SFARI).

SIMONS FOUNDATION

Rumi Scientific is pleased to be working with the Seaver Autism Center to develop a pipeline for discovering therapeutics for autism.

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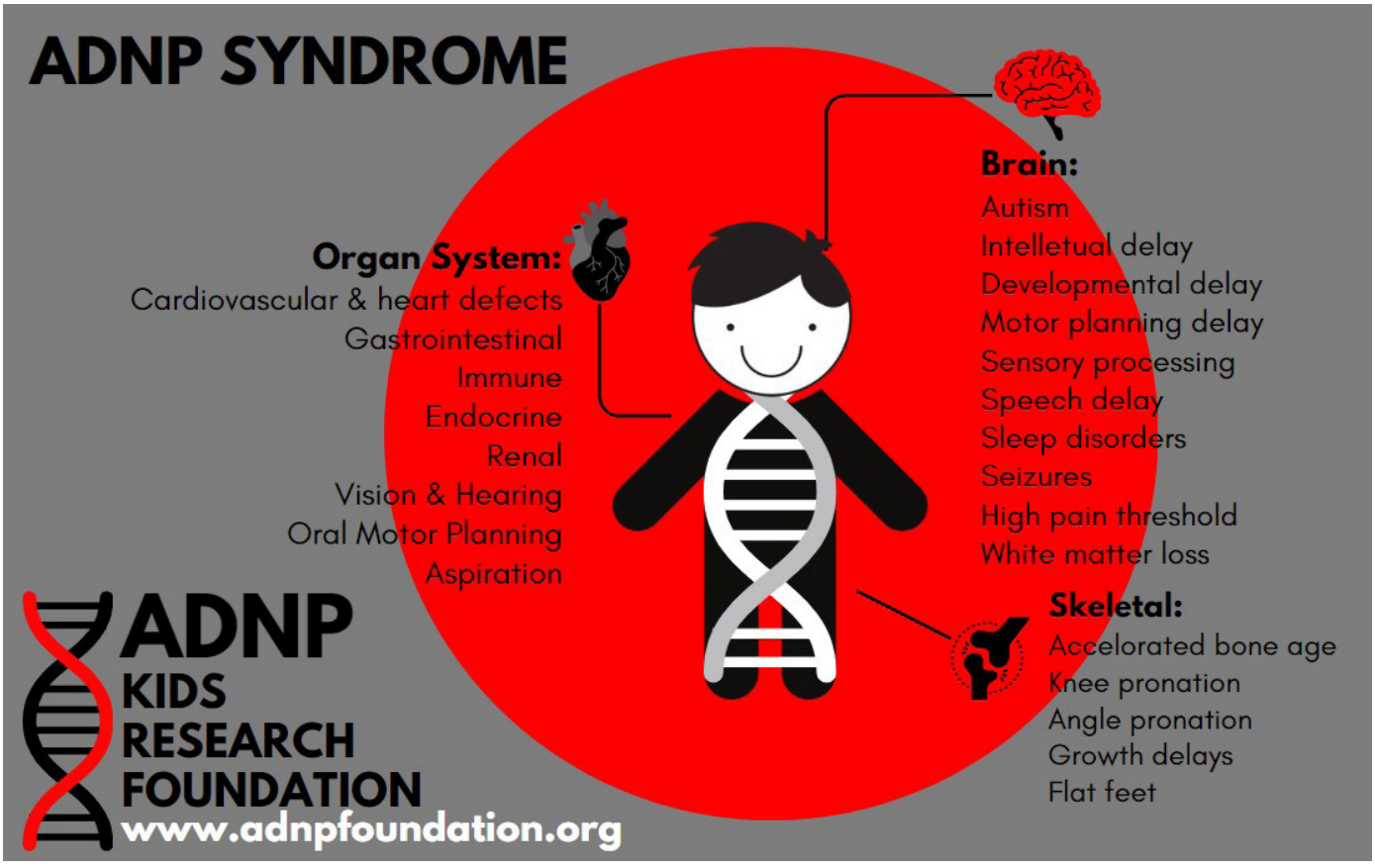

neuren

pharmaceuticals

Improving the Lives of People with Neurodevelopmental Disabilities

Neuren is developing new therapies for debilitating neurodevelopmental disorders emerging in early childhood, characterised by impaired connections and signalling between brain cells. The first, DAYBUE™ (trofinetide), was approved by US FDA as the 1st and only treatment for Rett syndrome[1], launched by partner Acadia in April 2023.

2 novel drugs, targeting **6** disorders, all with **Orphan Drug** designation

NNZ-2591

Phelan-McDermid Syndrome

Neuren has recently announced positive results in a Phase 2 trial of NNZ-2591 in Phelan-McDermid syndrome[2] and is planning a Phase 3 trial pending consultation with the FDA

Phase 2 trials ongoing:

- Pitt Hopkins
- Angelman
- Prader-Willi

Neuren is pleased to support the work of the Seaver Autism Center for Research and Treatment

1 Acadia Pharmaceuticals holds exclusive worldwide license; currently approved in US only

2 Phelan-McDermid syndrome Phase 2 trial top-line results presentation (https://neurenpharma.com/pdf/a651ed08-6f05-4a9d-a16f-2607b70066ea/Investor-presentation-18-December-2023.pdf)

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 subarachniod 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 intellecutal 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 actvity 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 causes 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.

Obessive-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 profileration 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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Advances in Autism Conference

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Notes

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