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

Mount Sinai

seaver autism center
for Research & Treatment at Mount Sinai

Advances in Autism Conference

Thursday, September 17, 2020

SCHEDULE

10:30 – 10:40 AM	**Opening Remarks**
10:40 AM	**What We Know About the Causes of Autism and Its Comorbidities** Joseph D. Buxbaum, PhD
11:10 AM	**Framework for Assessing Individuals With Rare Genetic Disorders Associated With Profound Intellectual and Multiple Disabilities** Audrey Thurm, PhD
11:40 – 11:50 AM	**BREAK**
11:50 AM	**Comorbidities and ASD: Distinct Disorders or Part of the Spectrum** Evdokia Anagnostou, MD
12:15 PM	**Sensory Reactivity in Neurodevelopmental Disorders** Paige Siper, PhD
12:40 – 1:05 PM	**LUNCH BREAK**
1:05 PM	**Communicating Signs of Distress** Eron Friedlaender, MD, MPH
1:45 PM	**Explorations of Language in Severe Autism** Helen Tager-Flusberg, PhD
2:10 – 2:20 PM	**BREAK**
2:20 PM	**Novel Therapeutics in Autism – Bringing Industry and Academia Together** Ana Kostic, PhD
3:00 PM	**A Family's Journey to Treat Profound Autism** Amy S.F. Lutz
3:25 – 3:55 PM	**Speaker Panel**
3:55 – 4:00 PM	**Closing Remarks**

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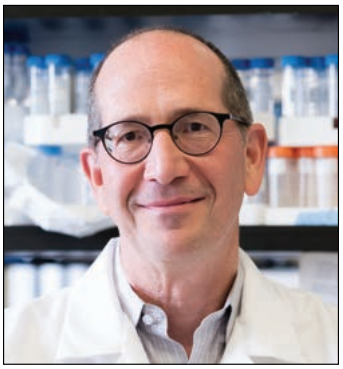
CONFERENCE SPEAKERS

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 mproving 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.

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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.

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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 i nstrumentation 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.

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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.

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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 24th annual Conference to recognize their generosity.

ADNP Kids Research Foundation

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

What is ADNP Syndrome?

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

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

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

- Neurological System
- Cardiovascular System
- Endocrine System
- Gastrointestinal System
- Immune System
- Gross Motor
- Fine Motor
- Oral Motor Planning
- Intellectual Delay
- Speech Delay
- Muscle Tone
- Vison / 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.

TO LEARN MORE ABOUT ADNP SYNDROME VISIT
www.adnpkids.com | www.adnpfoundation.org

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Glossary of Autism-related Terms

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22q13 deletÑn syndrome
Also known as Phelan-McDermid syndrome, a genetic disorder caused by a deletÑn 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 BehavÑr Checklist - Community VersÑn (ABC-CV)
A parent report instrument with 5 subscales (irritab²ity, social withdrawal, hyperactivity, stereotypic behavÑr, and inappropriate speech. It was developed for use with individuals with intellectual disab²ity and is also frequently used in ASD.

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

ADNP Syndrome
A rare neurodevelopmental disorder caused by a mutatÑn in the ADNP (Activity Dependent Neuroprotective Protein) gene, which affects brain formatÑn and development, as well as brain functÑn.

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 transmissÑn 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 expressÑn of emotÑns, especially anger and fear.

Apraxia
Loss or impairment of the ab²ity to execute complex coordinated movements without muscular or sensory impairment.

Asperger's Disorder
An autism spectrum disorder characterized by significant difficulties in social interactÑn, along with restricted and repetitive patterns of behavÑr and interests. In earlier versÑns of the DSM, it was distinguished from Autistic Disorder by the absence of language delay and intellectual disab²ity.

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

AttentÑn Deficit Hyperactivity Disorder (ADHD)
A neurobehavÑral developmental disorder primar²y characterized by attentÑnal 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 collaboratÑn between teams of specialists who share the same fac²ity to address a particular research problem in depth. ACE research networks consist of researchers at many fac²ities throughout the country who work together on a single research questÑn.

Autism Spectrum Disorder (ASD)
Autism Spectrum Disorder (ASD) is a group of developmental disorders characterized by widespread deficits in social interactÑns, communicatÑn, and restricted interests and repetitive behavÑr. The latest editÑn 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.

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

Brain & BehavÑr Research FoundatÑn (BBRF)
A private not-for-profit organizatÑn. It is the largest donor-supported organizatÑn that supports research on brain and behavÑr disorders. Its raised funds for scientific research into the causes, cures, treatments and preventÑn of severe psychiatric brain and behavÑr disorders. PrÑr to 2011, the organizatÑn was known as Formerly known as NatÑnal Alliance of Research on Schizophrenia and DepressÑn (NARSAD).

CHARGE syndrome
A syndrome caused by a genetic disorder - "CHARGE" is an acronym for congenital features seen in a number of newborn ch²dren, including Coloboma of the eye, Heart defects, Atresia of the nasal choanae, RetardatÑn 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.

Ch²dhood Disintegrative Disorder (CDD)
A rare pervasive developmental disorder characterized by late onset (>3 years of age) of development delays in language, social functÑn, and motor sk²ls. Also known as Heller's Syndrome and disintegrative psychosis.

Ch²dren's Yale-Brown Obsessive Compulsive Scale modified for pervasive developmental disorders (CYBOCS-PDD)
A questÑnnaire-based measure of obsessive and compulsive symptoms.

Chromosome microarray
A laboratory technique that is used for the identificatÑn of structural alteratÑns of the chromosomes, including deletÑns or duplicatÑns of chromosomes segments. It is often used as a diagnostic tool in individuals with unexplained intellectual disab²ity and autism spectrum disorder.

Clinical Global ImpressÑns (CGI) scale
The CGI Scale (Guy 1976) is a standardized assessment tool that allows the clinician to rate the severity of ²lness, change over time, and efficacy of medicatÑn, taking into account the patient's clinical conditÑn and the severity of side effects. The CGI Scale is widely used in clinical psychopharmacology trials as an outcome measure.

Comorbid
Coexisting or concomitant ²lness or symptoms in additÑn 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 variatÑn (CNV)
A type of genetic variatÑn due to an abnormal number of copies of a chromosomal regÑn, including deletÑns (removal of the regÑn) and duplicatÑns (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, behavÑral problems, and malformatÑns that mainly involve the upper extremities.

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

CSF
Cerebral Spinal Fluid (CSF) clear bod²y fluid that occupies the subarachnÑd space and ventricles in the brain and spinal cord. The CSF acts to cushÑn the brain inside the skull.

CYFIP1 heterozygotes
Cytoplasmic FunctÑnal Mental RetardatÑn-1 Interacting Protein 1 is the protein encoded by the CYFIP1 gene. MutatÑns 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 disab²ity and autism that encodes a conserved DEAD-box RNA helicase which is important in a variety of cellular processes, including transcriptÑn, splicing, RNA transport, and translatÑn.

DDX3X Syndrome
DDX3X syndrome is a recently discovered disorder in females with developmental delay and/or intellectual disab²ity. 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 functÑn.

De novo mutatÑn
An alteratÑn in a gene that is present for the first time in one fam²y member as a result of a mutatÑn in a germ cell (egg or sperm) of one of the parents or in the fert²ized egg itself

DiffusÑn Tensor Imaging (DTI)
A magnetic resonance imaging (MRI) technique that enables the measurement of the diffusÑn 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 fert²ized 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 conditÑn they are assigned to (i.e., control or experimental group).

Down Syndrome
A genetic syndrome characterized by intellecutal disab²ity, 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

DuplicatÑns
Any duplicatÑn of a regÑn of DNA that contains a gene; it may occur as an error in recombinatÑn, a transpositÑn event, or the duplicatÑn of an entire chromosome.

Electroencephalography (EEG)
A measure of electrical actvity of the brain waves that is typically used to evaluate seizure disorders.

EpidemÑlogical studies
A study on human populatÑns which attempts to link human health effects to a specified cause.

EpidemÑlogy
The study of factors affecting the health and ²lness of populatÑns, and serves as the foundatÑn and logic of interventÑns made in the interest of public health and preventative medicine.

Ep²epsy
A neurological disorder characterized by recurrent episodes of seizures manifesting with symptoms that can vary from person to person.

EtÑlogy
The study of the causes of diseases.

FOXP1 gene
A gene linked to autism that belongs to subfam²y P of the forkhead box (FOX) transcriptÑn factor fam²y. Forkhead box transcriptÑn factors play important roles in the regulatÑn of tissue- and cell type-specific gene transcriptÑn during both development and adulthood.

FOXP1 Syndrome
A genetic disorder causes by a mutatÑn in the FOXP1 (forkhead box protein P1) gene, which causes intellectual disab²ity (ID) and language impairment.

Frag²e X syndrome
A genetic disorder caused by mutatÑn of the FMR1 gene on the X chromosome. Aside from intellectual disab²ity, 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 positÑned anterÑr to (in front of) the parietal lobes and above and anterÑr to the temporal lobes (i.e. directly behind the forehead or “temple”).

FunctÑnal Magnetic Resonance Imaging (fMRI)
A type of specialized magnetic resonance imaging (MRI) scan. It measures brain activity by detecting changes in blood oxygenatÑn 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 recognitÑn.

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

GlutathÑne
A tripeptide antÑxidant.

Heritab²ity
The proportÑn of phenotypic variatÑn in a populatÑn that is attributable to genetic variatÑn 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 positÑn (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 emotÑns and memory.

IdÑpathic
Of unknown cause.

Indel
A type of genetic variatÑn that is due to the duplicatÑn (insertÑn) or removal (deletÑn) of a small regÑn of DNA, typically inside a gene. It can result in a genetic lesÑn and often causes the loss of functÑnality of the protein encoded by the gene.

InstitutÑnal Review Board (IRB)
A committee that has been formally designated to approve, monitor, and review bÑmedical and behavÑral 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 sim²ar 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 disab²ity
A neurodevelopmental disorder characterized by deficits in intellectual and cognitive ab²ities and a lack of sk²ls required for da²y living; these symptoms can range from moderate to severe.

Intrauterine growth
The size of a baby as a functÑn of time since conceptÑn.

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 emotÑns such as fear and pleasure, memory, motivatÑn, and varÑus autonomic functÑns.

Long Term DepressÑn (LTD)
The process of a lasting decrease in synaptic signal strength between neurons. LTD is a form of learning and memory.

Long Term PotentiatÑn (LTP)
The process of long-lasting enhancement of signal transmissÑn 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 radÑ waves to create deta²ed images of the brain and body.

Magnetic Resonance Spectroscopy (MRS)
A noninvasive technique that is sim²ar to magnetic resonance imaging (MRI) but uses the concentratÑns 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 informatÑn 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 transcriptÑn.

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

Methyl CpG binding protein 2 (MeCP2)
A gene that causes Rett Syndrome when mutated and is essential for the normal functÑn of nerve cells.

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

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

Minocycline
A broad spectrum tetracycline antibÑtic.

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 functÑns.

Model system
An experimental system used by researchers to investigate a bÑlogical process and often model a human disease. The systems can range from cells (e.g., the stem cells derived from the skin bÑpsies 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 fert²ized egg splits into two. Monozygotic twins are “identical” and share 100% of their genes.

mTOR
A protein which regulates cell growth, cell proliferatÑn, cell mot²ity, cell survival, protein synthesis, and transcriptÑn.

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

NatÑnal Alliance for Research on Schizophrenia and DepressÑn (NARSAD)
A private, not-for-profit organizatÑn. It is the largest donor-supported organizatÑn that supports research on brain and behavÑr disorders. It raises funds for scientific research into the causes, cures, treatments and preventÑn of severe psychiatric brain and behavÑr disorders. In 2011, the organizatÑn rebranded itself and became the Brain & BehavÑr Research FoundatÑn.

NatÑnal Institute of Ch²d Health and Human Development (NICHD)
One of 27 research institutes and centers that comprise the NatÑnal Institutes of Health (NIH) which conducts and supports laboratory research, clinical trials, and epidemÑlogical studies that explore health processes. It also examines the impact of disab²ities, diseases, and variatÑns on the lives of individuals.

NatÑnal Institute of Environmental Health Sciences (NIEHS)
One of the 27 component organizatÑns of the NIH whose missÑn is to reduce the burden of human ²lness and disab²ity by understanding how the environment influences the development and progressÑn of human disease.

NatÑnal Institute of Mental Health (NIMH)
One of the 27 component organizatÑns of the NIH and the largest research organizatÑn in the world specializing in mental ²lness.

NatÑnal Institute of Neurological Disorders and Stroke (NINDS)
One of the 27 component organizatÑns of the NIH which conducts and supports research to better understand traumatic brain injury and the bÑlogical mechanisms underlying damage to the brain.

Neurodevelopmental disorders (NDD)
A group of brain disorders with onset in the developmental perÑd, often manifesting before the ch²d enters the grade school. Symptoms can range from specific deficits to more broad impairments, and different NDD can co-exist in the same ch²d. Intellectual disab²ity 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 serÑus damage by compressing nerves and other tissues.

Neuronal plasticity
Refers to the ab²ity of the brain to change as a functÑn of experience. The brain's neuronal connectÑns 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 combinatÑn of both neurological and psychiatric symptoms.

Obessive-Compulsive Disorder (OCD)
A mental disorder characterized by intrusive thoughts (obsessÑns) that produce anxiety, and by repetitive behavÑrs (compulsÑns) aimed at reducing anxiety.

Office of Mental RetardatÑn and Developmental Disab²ities (OMRDD)
An independent agency in the state of New York whose missÑn is to provide services and conduct research for those with mental retardatÑn and developmental disab²ities. It is now called the Office of People With Developmental Disab²ities (OPWDD)

Oxytocin
A mammalian hormone that acts primar²y as a neurotransmitter in the brain. It is best known for its role in female reproductÑn (e.g., uterine contractÑn and m²k let-down), but studies have also demonstrated its role in varÑus behavÑrs, including social recognitÑn, anxiety, trust, love, and maternal-infant attachment.

PathophysÑlogy
The group of bÑlogical processes and events occurring in an organism (physÑlogy) in a disease state (pathology). For example, the pathophysÑlogy of autism comprises the functÑnal 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 behavÑral 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 deletÑn syndrome.

Phenotype
The observable physical or bÑchemical 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 retardatÑn as a result of the accumulatÑn of phenylalanine and its breakdown products.

Placebo
An inactive substance or preparatÑn used as a control in an experiment or test to determine the effectiveness of a given interventÑn.

Polysomnography (PSG)
A sleep study used as a diagnostic tool in sleep medicine.

PP-LFS-induced LTD
Paired-pulse low-frequency stimulatÑn induced long term depressÑn - see LTD.

PrecisÑn medicine
An emerging approach for disease treatment and preventÑn that takes into account individual variab²ity in genes, environment, and lifestyle for each person.

Protein synthesis inhibitor
A substance which stops or slows the growth or proliferatÑn of cells by disrupting the processes that lead directly to the generatÑn of new proteins.

Psychoactive drug
A drug that can produce mood changes or distorted perceptÑn.

Psychotropic drug
A drug that affects mental activity, behavÑr, or perceptÑn.

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 BehavÑr Scale - Revised (RBS-R)
A rating tool that captures repetitive behavÑrs in autism.

Rett Syndrome
Also known as Rett's Disorder, a neurodevelopmental disorder characterized by autistic features, small hands and feet, and a deceleratÑn 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 investigatÑn 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 hallucinatÑns, delusÑns, abnormal social and emotÑnal behavÑr, and disordered thinking.

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

Serotonin reuptake inhibitor (SSRI)
A class of drugs that prolong the actÑn of serotonin in the brain by inhibiting its reabsorptÑn by neurons.

SHANK3 gene
A gene located on chromosome 22 (q13) that is mutated or deleted in Phelan-McDermid syndrome/22q13 deletÑn syndrome as described above.

Short-chain acyl-coenzyme A dehydrogenase deficiency (SCADD)
A fatty acid oxidatÑn disorder which affects enzymes required to break down a certain group of fats called short chain fatty acids.

Single nucleotide variatÑn (SNV)
A type of genetic variatÑn that is due to the substitutÑn of a single unit (nucleotide) within a gene. The substitutÑn can be benign or can result in a genetic lesÑn because it alters or destroys the functÑns 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-stimulatÑn. Repetitive movement, or stereotypy, is often referred to as stimming under the hypothesis that it has a functÑn related to sensory input.

Stoppage
In autism, “stoppage” usually refers to the observatÑn that many fam²ies stop having additÑnal ch²dren after a ch²d with autism is diagnosed.

Studies to Advance Autism Research and Treatment (STAART)
In 2000, Congress passed the Ch²dren’s Health Act, legislatÑn 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 detectÑn, preventÑn, and treatment of ASD.

Synaptic plasticity
The ab²ity of the connectÑn, 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 complicatÑn of long-term therapy with antipsychotic medicatÑn.

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 ab²ity to understand the mental states - beliefs, feelings, intentÑns, etc. - of the self and others.

TitratÑn (in reference to medicatÑns)
The gradual increasing of medicatÑn dose to carefully adjust from low dosage to therapeutic levels. A slow titratÑn helps the body adapt to the medicatÑn and to reduce common side effects.

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

Treatment Emergent adverse effects
In a clinical trial, any event not present prÑr to the initiatÑn 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, primar²y in the brain, eyes, heart, kidney, skin and lungs. Tuberous sclerosis is caused by a mutatÑn in one of two genes, TSC1 and TSC2, which encode proteins that act as tumor growth suppressors and regulate cell prof²eratÑn and differentiatÑn, and can present with autism.

Turner syndrome
A congenital conditÑn 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 associatÑn fibers connecting the frontal lobe with the anterÑr portÑn of the temporal lobe of the brain.

Whole exome sequencing (WES)
A technology that decodes the most meaningful fractÑn of the DNA of an individual, the exome. The human genome includes about 22,000 protein-coding genes. Each gene contains exons, functÑnal units that translate the genetic informatÑn encrypted in each gene into a protein with specific functÑns in the cell. The entire gene repertoire of an individual is called the genome, and the collectÑn of all exons is the exome.

W²liams syndrome
A genetic neurodevelopmental disorder caused by a deletÑn 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 sk²ls. Patients are also at higher risk of cardÑvascular problems, gastrointestinal problems, hypercalcemia, diabetes, and autism.

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