Editorial

On behalf of the entire CNL team, we are delighted to present the first edition of THE CNL- the official newsletter of the Cognitive Neurophysiology Laboratory and the Human Clinical Phenotyping Core at Albert Einstein College of Medicine. As a lab, we continuously strive to connect with the wonderful communities that make our research possible. From our participation in local events to our engagement on social media, we work to bridge the worlds of science and community, which can sometimes feel so far apart. With this semi-annual newsletter, we intend to strengthen that bridge by sharing with you our interests, projects, and findings in a way that is both inviting and accessible. Each edition will highlight a subset of our work, beginning with this first edition, which focuses on our rare disease and autism spectrum disorder projects. To our participants near and far, we thank you, and hope this newsletter excites you about the studies you have contributed to and those that are yet to come. To our colleagues and members of the scientific community, we hope this encourages you to learn more about the neurodevelopmental and neuropsychiatric disorders we are investigating.

Stay safe and see you soon,

Alaina Berviti, Ana Francisco, Filip de Sanctis, & Sophie Molholm
The CNL stands with #BlackLivesMatter

As a lab, we stand in solidarity with the Black community, alongside Einstein and Montefiore, in fighting against social and racial injustice. In reflecting on how we can use our resources to uplift the Black community, we have developed the CODE Committee. This committee will lead the CNL in expanding our scope of Community Outreach and increasing our Diversity Efforts through various initiatives, which will be announced via social media and future newsletters. We are invested in the successful implementation of CODE initiatives, and welcome suggestions and collaborations from all participants, colleagues, and community members.

The CNL #ShutDownSTEM on Wednesday, June 10th, to reflect on how we, as a lab, can foster an environment of anti-racism.

The CNL, alongside the Einstein and Montefiore community, observed a moment of silence on Friday, June 5th, in memory of George Floyd and countless others.
Our mission

The mission driving our research is to understand the neural processes underlying perception and cognition in the human brain, and to define how these differ in (and contribute to) neurodevelopmental and neuropsychiatric conditions. To this end, we use state-of-the-art brain imaging tools, such as electroencephalography (EEG) and magnetic resonance imaging (MRI), in concert with body/eye tracking, task guided behavior, standardized neuropsychological and clinical tests, and genetic information. We apply these tools to study attention, executive function, multisensory and sensorimotor integration, and basic sensory processing. We work with populations that span from childhood to older adults, and include genetic conditions such as 22q11.2 deletion syndrome and Rett syndrome, idiopathic neurodevelopmental and neuroconditions including autism and schizophrenia, and degenerative conditions such as multiple sclerosis. In our clinical work we collaborate with essential clinical partners at Montefiore, Jacobi, and University of Rochester Medical Center. Our ultimate goal is to increase basic understanding of brain function and, in turn, to use this knowledge for the development of markers of risk and disease progression and of optimized and targeted intervention programs in clinical conditions.

2019

2019 was a great year! Check the numbers below!

Our work would not be possible without you and, therefore, we would like to extend our most sincere gratitude to the participants and their families for their interest, their involvement, and their time.
Meet the group

In each newsletter, we will be introducing our select members of our lab! In this issue, you'll meet our coordinators, technicians, and administrative staff. Say hello to Alaina, Cat, Douwe, and Rachel!

Humans of the CNL

Due to COVID-19 our lab members are working from home and communicating on ZOOM. Fortunately, we have been able to continue our research by conducting interviews and questionnaires with participants over the phone and online!

Not pictured: John Foxe (Founder & Primary Investigator), Emily Bates (Student Trainee)
The Cognitive Neuroscience Society (CNS) hosts an annual conference where its members gather to present their latest work. This year, the CNL had plans to travel to the Boston conference in March to present a total of seven posters, each showcasing different projects we’ve been working on. Due to the COVID-19 outbreak, the conference has since been postponed to May and moved online. Nevertheless, we are eager to present our work, safely at home.

Check our posters below!
Click to enlarge and to see video presentations of some of the posters.
In honor of Rare Disease Day, we are showcasing our projects on cystinosis, rett syndrome, and 22q11.2 deletion syndrome.

Rare disease families across the US visited the CNL in 2019. We look forward to meeting more in 2020!
Cystinosis is a rare genetic disease affecting 1 in 100,000-200,000 live births in the United States. It is a lysosome storage disorder caused by a mutation in the CTNS gene on chromosome 17. This gene encodes a protein called cystinosin. Cystinosin transports an amino acid called cystine out of an intracellular compartment, the lysosome. If the cystinosin protein is absent or not fully functional, cystine accumulates in the lysosome and forms crystals, damaging the cells. Cystinosis affects different organs of the body!

Click here for a description of our research on cystinosis.

Earlier this year, Ana Francisco participated in the #GreatGivers campaign for the Cystinosis Research Network. Check it here!

A special thanks to Dr. Kaskel and to the Cystinosis Research Network for all their support!

Rett syndrome is a rare genetic neurological disorder affecting 1 in 10,000 female births. It is even rarer in boys. Rett is caused by mutations on the X chromosome on a gene called MECP2. It leads to mild/severe impairments, affecting nearly every aspect of the child’s life: their ability to talk, eat, and even breathe. Rett is often diagnosed in children between 6 to 18 months as they begin to miss developmental milestones or lose abilities they had acquired.

Click here for a description of our research on Rett. This work is done in collaboration with the Rett Syndrome Center at the Children’s Hospital at Montefiore.
22q11.2 deletion syndrome

22q11.2 deletion syndrome (also referred to as velocardiofacial syndrome (VCFS) or DiGeorge syndrome) is a disorder caused by a small missing piece of chromosome 22. Affecting 1 in 4,000 people worldwide, this syndrome is almost as common as Down syndrome. 22q11.2 deletion can result in about 200 mild to serious health and developmental issues in children, which include: growth delays, feeding problems, congenital heart disease, gastrointestinal difficulties, serious breathing concerns, cleft and craniofacial issues, calcium deficiencies, immune deficiencies, kidney problems, and skeletal anomalies. This list also includes the possibility of speech, developmental and cognitive delays, as well as ADHD, Autism, and anxiety and other psychiatric disorders.

Click here for a description of our research on 22q11.2 deletion syndrome. This work is done in collaboration with the Montefiore-Einstein Regional Center for 22q11.2 deletion syndrome at the Children’s Hospital at Montefiore.

22Q★TEXAS

We had an amazing time at the 22q Camp Texas! Thank you for having us!

ANKS1B

The ANKS1B syndrome is caused by mutations in a gene with the same name. There is still a lot to be understood about this syndrome, but it appears to be very rare. In the families we were able to identify and test, ANKS1B is characterized by neurodevelopmental differences including Autism, ADHD, and speech and motor deficits. Our aim is to continue to characterize ANKS1B and understand how the syndrome affects individuals and families. We are using different methods to accomplish this goal, both in humans and in mouse and cell models.

In collaboration with Bryen Jordan’s group, we published the first paper on this syndrome last year.
In this issue, we are spotlighting our ongoing project with the Autism Center of Excellence. This project has two main goals, the first of which aims to understand the role of genetics in the development of Autism in those with African (African, African-American, Afro-Caribbean, Afro-Latino/a) heritage. The role of genetics in Autism has been a topic of interest for some time, but minority populations have been widely underrepresented in this research. Since we know various racial backgrounds have distinct genetic makeups, our project intends to fill the gaps in our knowledge of the genetic impact on Autism in these populations.

Secondly, our project aims to understand the barriers to diagnosis and treatment of Autism in these groups. Previous research has shown that minority children are, on average, diagnosed with Autism over a year later than children of Caucasian decent. Therefore, we are interested in the cultural, societal, and economic factors which contribute to this discrepancy.

As we move towards developing targeted treatments and interventions for Autism, we need to ensure they will work for everyone. If you’re interested in learning more about our project, please contact tracie.ebalu@einsteinmed.org.

Meet Tracie, the ACE study coordinator!
Tracie’s responsibilities include recruitment and scheduling, administering clinical interviews, and data management. Tracie is interested in cognitive neurodevelopment in children and youth. In her spare time, Tracie enjoys solo-travelling, cooking, swimming, salsa dancing, and playing volleyball.

Einstein is one of six universities across the US collaborating on this exciting project.
We measured brain activation using electroencephalography (EEG) in individuals with Multiple Sclerosis (MS) and healthy controls (HCs) performing a cognitive task while standing (single task) or walking (dual-task). We say that the brain processes multi-task information efficiently when the number of mistakes in the cognitive task while standing is equal or even less than the number of mistakes in the cognitive task while walking. In HCs, we found a relationship between changes in brain activation and better cognitive performance during dual-tasking, but there was no evidence of such relationship in MS. Our data could suggest that individuals with MS are less able to leverage brain resources in multi-task situations, but the comparison was somewhat unfair as our sample was composed of individuals with a diagnosis between 2 and 15 years' prior to study participation (we are currently enrolling individuals during early-stage MS!). Our studies have the potential to lead to improved diagnosis of cognitive and motor difficulties in MS and more effective interventions. With a better understanding of how the brain responds to and processes multi-task information, clinicians can begin to develop interventions that are based on known neurophysiological profiles.

We studied auditory processing and sensory memory in cystinosis. While basic auditory processing is fully functional, children and adolescents with cystinosis seem to have some difficulties with sensory memory. Sensory memory is the shortest type of memory and it helps keeping track of information in the millisecond rate. Because basic and complex aspects of brain processing are related and co-dependent (for example, a problem in a more basic process can hinder the processing at a more complex level), difficulties at this very basic level of memory could impact, for example, working memory—crucial to fulfill day-to-day tasks and to academic success. Remarkably, though, when we did this same task in adults with cystinosis (publication coming soon!!), their sensory memory appeared to be intact. This means that though some difficulties are present in younger individuals with cystinosis (which may have a negative impact in both school and home contexts and should be considered when developing education plans, for instance), they appear to be solved in adulthood.
Recent publications

We used EEG, a non-invasive method which allows you to record the electrical activity of the brain, to understand basic auditory processing and sensory memory in people with 22q11.2DS. While basic auditory processing refers to the initial steps your brain takes to comprehend sound in your environment, sensory memory is the stage of memory that allows you to briefly remember these sounds. Our findings suggest that these two processes are overall functional in adolescents and adults with 22q11.2DS, at least in those individuals with less severe conditions. However, if there is any sign of psychosis, which is more prevalent in 22q11.2DS than in the general population, the auditory responses become reduced. We need larger groups of individuals with and without psychotic symptoms to confirm that these brain responses are indeed able to distinguish between those two groups, but these are important findings in trying to better understand the development of schizophrenia in 22q11.2DS.

Sensory Processing Disorder (SPD) is characterized by over or under responsiveness to sensory stimulation (sound, light, texture, etc). It is thought that this responsiveness is due to the nervous system’s inability to regulate sensory input from the environment, but the exact mechanism of how this happens is still unknown. We sought to better understand how exactly sensory information is processed in individuals with SPD in comparison to typically developing individuals and individuals diagnosed with Autism, which is itself partly characterized by dysregulation of sensory processing. Here, we found that people with Autism and SPD process auditory (alone) and visual (alone) stimuli similarly to those that are typically developing. However, processing a joint audio-visual stimulus is a little different than processing a stimulus that is only auditory or only visual. In typically developing people, we see that audio-visual stimuli are processed much faster than stimuli that are only auditory or visual. That is, there seems to be an advantage to processing multi-sensory information over single-sensory information. Our study found evidence that people with Autism and SPD also benefit from processing multi-sensory information. However, this benefit is significantly reduced in comparison to that of their typically developing peers.
Online resources for children & caregivers navigating stress and changes from COVID-19.

WE ARE IN THIS TOGETHER

GoNoodle: free movement, yoga, and mindfulness videos for kids at home.

Remote education resource center: educational resources for all grade levels.

"Caring for Each Other" by Sesame Street: ebooks, coloring pages, games, and videos for children featuring their favorite Sesame Street characters.

Coronavirus social story: a book explaining the pandemic in simple language.


Daily schedule maker: schedule templates for children at home.

Free & discounted services: a listing of entertainment, fitness, & mindfulness services.

Rare Diseases Clinical Research Network: Consider participating in research on how COVID-19 has impacted rare disease patients and their families.

#IStayHomeForRare: A campaign to support individuals living with rare diseases during this challenging time.

*Disclaimer: the CNL does not endorse nor sponsor the programs listed on this page. The listing is only to inform individuals and caregivers of resources from which they or their families may benefit.

This year, 22q at the Zoo was 22q and Zoom!