Center Updates

**Our new IDDRC website is live!** Bookmark the website [link](#), start browsing through the [core services](#) pages, stay in the know with the numerous [events and seminars](#), and many more announcements on our [news](#) page.

**We've moved!** The IDDRC team has moved to a newly renovated office location at 1 Autumn Street.

Research News

**#1 in US News Survey**

Congratulations to the Departments of Neurology and Neurosurgery for maintaining the #1 ranking in the US News survey for 12 consecutive years. US News rankings are no longer based solely on reputation, but are a composite assessment of the hard work everyone does every day. Patient volume, quality of care, clinical outcomes, clinical and basic research, teaching, and reputation as a center for referral of complex patients are among the many factors taken into account. Remaining the top program is a testament to the incredible work of each and every member of our Department!

*Scott Pomeroy, MD, PhD, Neurologist-in-Chief*

**Dr. Clifford Woolf elected to the American Academy of Arts and Sciences**

Congratulations to Clifford Woolf, MB, BCh, PhD, on his election to the American Academy of Arts and Sciences. This honor signifies the high regard in which he is held by leaders in the field and members throughout the nation. We are lucky to have him here at BCH!
A team of researchers at Boston Children’s Hospital and the Broad Institute have successfully found a technique, known as base editing, to restore hearing in mice. IDDRC PI, Jeffrey Holt, PhD, confirms that, “This is a big step beyond the field of hearing restoration and for the broader field focused on treatment of genetic disorders.” Read the BCH Press Release.

The tiny choroid plexus protects the prenatal brain — but may also pass on inflammation from the mother.
As a result of this groundbreaking study, Dr. Maria Lehtinen, PhD, Neurobiologist at Boston Children’s Hospital, informs us that “there is a correlation between maternal illness during pregnancy and autism." Read the BCH Press Release.

Kleefstra Syndrome Awareness Day – 17th September
To mark Kleefstra Syndrome Awareness Day (17th September), the Translational Neuroscience Center at Boston Children’s Hospital announced a Kleefstra Syndrome Family and Scientific Virtual Meeting to be held on February 4, 2021. This meeting will be hosted by the TNC in partnership with the patient advocacy group, IDefine. Please sign up for IDefine email updates to register. Scientific and clinical experts, including Professor Tjitske Kleefstra of Radboud University, Nijmegen, will update participants on the latest research into this rare genetic disorder. This initiative will be supported by an award from the ICCTR.

Defining the Clinical, Molecular and Imaging Spectrum of Adaptor Protein Complex 4-Associated Hereditary Spastic Paraplegia
Over 80 types of Hereditary Spastic Paraplegias (HSP) are the leading cause of neurodegeneration in children. There is a poor understanding of how clinical HSP forms correlate with genetic variants associated with loss-of-function in the AP-4 protein complex. A recent publication in BRAIN by the Sahin group offers systematic documentation of the clinical presentation and natural history of AP-4-related HSP developed by an international cohort. Read more here.

2020 STED Grants
A total of $20,000 was awarded to Drs. Jonathan Lipton, Pankaj Agarwal, Xi He, Hisashi Umemori, Mustafa Sahin, Maria Lehtinen, and Michela Fagiolini for projects involving super resolution imaging. These funds were made possible by the generous funds from Boston Children’s Equipment and Core Resources Allocation Committee (ECRAC) Institutional Core Fund as well as the F.M. Kirby Neurobiology Center at Boston Children’s.

Dr. Hensch and colleagues PNAS Paper
Takao Hensch, PhD, is a co-author of an important review article on Critical Period Regulation Across Multiple Timescales on PNAS: Find the article here.

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On Thursday, October 1st, a group of clinical researchers from Neurology gave presentations to the FM Kirby Center for Neurobiology faculty, both to introduce their work as well as to forge collaborations. The following presentations were made:
Caitlin Rollins, MD: Fetal Brain Development in Congenital Heart Disease
April Levin, MD: Biomarkers for Neurodevelopmental Disorders
Alexander Cohen, MD, PhD: Bedside to Bedside Translational Neuorimaging: asking causal questions about ASD symptoms using ‘other’ clinical cohorts
Heather Olson, MD: Genotype-phenotype analysis in Epilepsy: CDKL5 deficiency disorder

New Trials in our Center

NeuroNEXT 107
Dr. Lisa Prock, MD, MPH, (Developmental Medicine, BCH) is the PI of a phase II double blind
placebo-controlled flexible-dose study, with an open-label extension studying the effects of the drug AFQ056 (a mGluR5 negative modulator) in conjunction with intensive language intervention on language learning in children with Fragile X Syndrome (FXS). Participants have full mutation Fragile X Syndrome and are between 3 and 6 years old at time of enrollment. The 22-month study tests response to the study medication as well as a parent-implemented language intervention, being taught to parents by a speech language pathologist.

**JASPER in Down Syndrome**

Dr. Nicole Baumer, MD, MEd, (Neurology & Developmental Medicine, BCH) is the PI of this study, and the goal is to determine whether JASPER, an intensive, targeted early behavioral intervention can improve joint engagement, joint attention, play skills, behavioral and emotional regulation, social communication skills, and developmental trajectories in young children with Down syndrome. Also explored is the potential use of EEG and ERP as outcome measures, and whether this approach may help elucidate mechanisms of change in behavior and development, and may help explain differences in development of social communication skills in individuals with DS. Participants are between 24 and 48 months old at time of enrollment, and are randomized to begin the intervention immediately, or to begin after a 6-month delay.

**TSC CALM Study**

Dr. Jurriaan Peters, MD, PhD, (Neurology, BCH) is the PI of a phase II open label clinical drug trial with a long term extension period studying the effects of the drug Ganaxolone (an allosteric GABA type A modulator) as an adjunct therapy in Tuberous Sclerosis Complex (TSC)-related epilepsy. Participants have a clinical or genetic diagnosis of TSC and are between 2 and 65 years old at time of enrollment. The objective of this 3-month trial followed by 6 month extension is to assess the safety and efficacy of Ganaxolone for the treatment of primary seizure types.

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**Events and Seminars**

*Find below our upcoming Neurobiology and Translational Neuroscience seminars.*

*Don’t forget to Zoom in!*

**Neurobiology seminars**

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<tr>
<th>Date</th>
<th>Speaker</th>
<th>Institution</th>
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<tbody>
<tr>
<td>10/26</td>
<td>Diana Bautista, PhD - UC Berkeley</td>
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<tr>
<td>11/2</td>
<td>Chinfei Chen, MD, PhD – Boston Children’s Hospital</td>
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<td>11/9</td>
<td>Botond Roska, MD, PhD - Friedrich Miescher Institute for Biomedical Research</td>
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<td>11/16</td>
<td>Lisa Goodrich, PhD – Harvard University</td>
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<td>11/30</td>
<td>Kenichi Ohki, MD, PhD – Utokyo</td>
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<td>12/7</td>
<td>Fred Rieke, PhD – University of Washington</td>
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<td>12/14</td>
<td>Jeff Lichtman, MD, PhD – Harvard University</td>
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Where: Zoom
When: 12.15pm - 1.15pm

*Find more Boston Children’s Hospital Neurobiology & Translational Neuroscience Center events [here](#).*

**Translational Neuroscience seminars**

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<tr>
<th>Date</th>
<th>Speaker</th>
<th>Institution</th>
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<tr>
<td>11/10</td>
<td>Michael Fox, MD, PhD – Brigham and Women’s Hospital</td>
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<tr>
<td>12/8</td>
<td>Jeremy Schmahmann, MD – Massachusetts General Hospital</td>
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Where: Zoom
When: 12pm - 1pm

*Find more Boston Children’s Hospital FM Kirby Neurobiology Center events [here](#).*
Introduction to Drug Discovery for Neuroscientists- 2020 Fall Series. This fall mini-series will be a weekly lecture on the basics of preclinical drug discovery for the neuroscience community. It is intended to give translational neuroscientists insight into the drug discovery process and an overview of the industrial landscape in neuroscience. Lectures from industry experts will be offered to cover the basics of the drug discovery pipeline from target selection to clinical trials. The lectures will be sequential, so we encourage you to attend all 6 sessions. Registration is required. Find more information here.

"World Family Celebration" Saturday, October 24, 10:30am, EST
Our 10th Blue Sky Day will be special in spite of changes made necessary by the Pandemic.
On Saturday, October 24 we will come together as a community to honor and to celebrate our very special children. Joining in our virtual observance will be families from around the world who are also facing the challenges of Rett syndrome.
We are thrilled that Dr. Djukic, the originator of Blue Sky, will also be participating in our program. Find more info here.

Featured Publications (2020)

Congratulations on your excellent work, Investigators!


Boxer LD, Renthal W, Greben AW, Whitwam T, Silberfeld A, Stroud H, Li E, Yang MG, Kinde B, Griffith EC, Bonev B, Greenberg ME. MeCP2 Represses the Rate of Transcriptional Initiation of Highly Methylated


Morton SU, Maleyeff L, Wypij D, Yun HJ, Newburger JW, Bellinger DC, Roberts AE, Rivkin MJ, Seidman JG, Seidman CE, Grant PE, Im K. Abnormal Left-Hemispheric Sulcal Patterns Correlate with Neurodevelopmental Outcomes in Subjects with Single Ventricular Congenital Heart Disease. Cereb


