One Mission: End AHC!

YOUR ALTERNATING HEMIPLEGIA OF CHILDHOOD FOUNDATION NEWSLETTER May/June 2018
2000 Town Center ■ Suite 1900 ■ Southfield, Michigan 48075

A MESSAGE FROM THE PRESIDENT

AHCF Goes to New Jersey Next Week

The time has finally arrived for the Family Meeting to get started. Next week, the largest number of AHC families ever to gather in one place will occur in Iselin, New Jersey.

The AHCF Board of Directors and Family Meeting Committee have been working very hard to make sure this meeting is another outstanding opportunity for the AHC community.

We are very excited to meet new families and visit with old friends.

For those families not able to travel to New Jersey, there is an opportunity to view most of the session over the internet.

Simply follow the instructions listed in the opposite graphic and you will be able to hear from AHC specialists and guest speakers.

After the meeting, we also look forward to recapping some of the "greatest hits" from the meeting in future issues of the newsletter. Key an eye out for more information in the days and weeks to come.

AHC FUNdraising - SUPPORT THE AHCF WITH THE NFL

An Amazing Opportunity for All of our East Coast Supporters

Come join us for a fun day at MetLife stadium to cheer on the New York Jets.

That's right! Support the AHC Foundation and enjoy an amazing day in New Jersey watching NFL football.

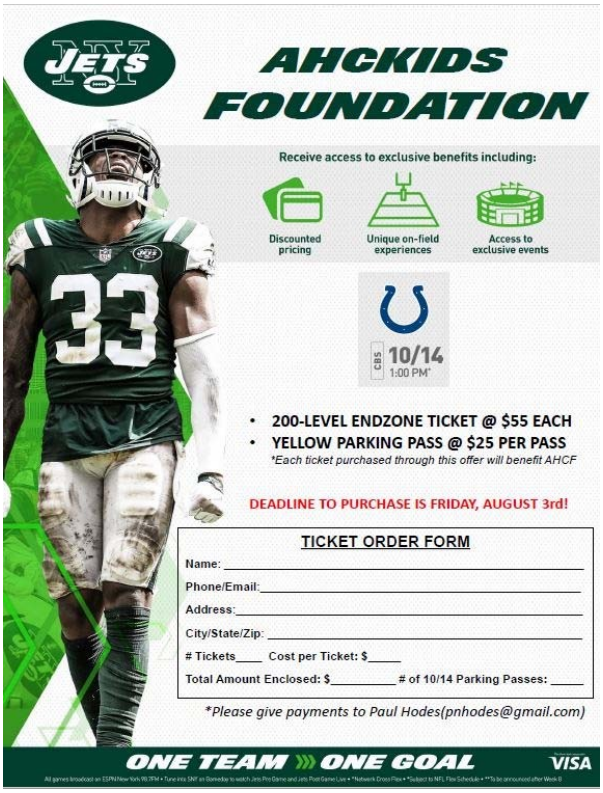
Call your friends. Gather your family. Invite your coworkers. Bring together a group of people to have fun while supporting the AHC community.

It's so simple.

All tickets **must be ordered by Friday, August 3rd.**

If you are interested in purchasing tickets, contact Paul Hodes at pnhodes@gmail.com

Thank you and hope to see you at the game.



AHC FUNdraising – NEW ENGLAND WALK A HUGE SUCCESS

The Gerber Family Held another Event in Honor of Son, Sean

On Sunday, May 20th, Olga and Bill Gerber hosted the **AHCF New England Walk** at Rotary Pavilion in Nevers Park, in South Windsor Connecticut.

The walk was held to honor their son Sean who is a huge Red Sox fan. There was a walk from 10:00 to 12:00 pm, but there was also a lot of fun for family and friends with music and inflatables.

The AHCF is grateful for the Gerber's efforts in helping to raise money for AHC research. Thanks for everything you do to help End AHC!

Thanks to Bill & Olga Gerber and Paul & Renee Hodes for continuing to find new and exciting ways to raise money for our ONE MISSION: End AHC!

AHCF IN ACTION – AT THE FAMILY MEETING

Board of Directors Give Notice of Meeting

The Annual Business Meeting of the AHCF Board of Directors will take place Friday, June 22, 2018 at 8:00 p.m. local time at the Renaissance Woodbridge Hotel in Iselin, New Jersey, in association with the AHCF Family Meeting.

The Annual Business Meeting will include the treasurer's report, nomination of board members, a strategic plan initiative, old and new business, and announcement of election results.

If you have any questions about the Annual Business Meeting, please contact Vicky Platt, Secretary, at vicky@ahckids.org or 847-337-9044. Please look to future issues of the newsletter for more information

AHC COMMUNITY CONNECTIONS

Dr. Kenneth Silver Speaks About AHC to Med Students

This spring, **Dr. Kenneth Silver spoke at Ann & Robert H. Lurie Children's Hospital of Chicago** to a group of interested doctors and medical students about the rare and interesting disorder, Alternating Hemiplegia of Childhood. Here is some of what he shared with this fabulous audience.

Alternating Hemiplegia of Childhood **(AHC) is a complex and difficult disease to diagnose.** Families go through many years of challenges in diagnosis in addition to caring for and treating their children.

The diagnosis of AHC is established by applying specific clinical criteria.

These include symptoms, which starts before 18 months of age, of repeated attacks of hemiplegia involving either side of the body, other repetitive movements, episodes of bilateral hemiplegia, disappearance of symptoms upon sleeping and resume after awakening, evidence of developmental delay and other neurological abnormalities. The collection of these symptoms and signs when clustered together suggest the specific syndrome such as AHC. However most of our patients only have some of these described signs and symptoms especially during the time when the infant presents for evaluation.

Because of this variability in the signs and symptoms of AHC it can be difficult to identify this disorder at its earliest presentation. When an infant with AHC presents to a clinician the initial manifestations are paroxysmal (intermittent) with stiffening episodes and/or abnormal eye movements. These features would make the clinician instantly diagnose seizures. This is because clinicians are concerned about the potential harmful effects of seizures on the developing brain; the seizures can recur and can be controlled with medications.

When repetitive movements occur in infants most of time these are on the basis of epileptic seizures so that is what the clinician expects. **Suspicions of AHC should arise when in spite of adequate anticonvulsant medication the seizures continue.** Additionally if an episode is captured while performing an EEG and there are no epileptic discharges to confirm that these movements are on the basis of epilepsy then other diagnoses should be considered.

Come Hear More from Dr. Silver in Person at the AHCF 2018 Family Meeting.
Through research, education and family support, we have ONE MISSION: END AHC!

AHC INFO EXCHANGE

ATP1A3 Mouse Model Research Published this Month

On June 11th, Epilepsia released an **important new article using a mouse model of the D801N mutation,** which is the most common mutation causing AHC. The title of the article is, "Mechanisms of increased hippocampal excitability in the Mashl+/- mouse model of Na+ /K+ -ATPase dysfunction."

The objective of the paper was to evaluate the mechanisms underlying such increased excitability they studied mice carrying the D801N mutation, the most common mutation causing human disease, specifically alternating hemiplegia of childhood (AHC) including epilepsy. Because the gene is expressed in all neurons, particularly γ-aminobutyric acid (GABA)ergic interneurons, they hypothesized that the pathophysiology would involve both pyramidal cells and interneurons and that fast-spiking interneurons, which have increased firing rates, would be most vulnerable.

During the project they observed that juvenile knock-in mice carrying the above mutation reproduce the human phenotype of AHC. They then demonstrated in the CA1 region of these mice the following findings as compared to wild type: (1) Increased number of spikes evoked by electrical stimulation of Schaffer collaterals; (2) equalization by bicuculline of the number of spikes induced by Schaffer collateral stimulation; (3) reduced miniature, spontaneous, and evoked inhibitory postsynaptic currents, but no change in excitatory postsynaptic currents; (4) robust action potential frequency adaptation in response to depolarizing current injection in CA1 fast-spiking interneurons; and (5) no change in the number of pyramidal cells, but reduced number of parvalbumin positive interneurons.

This work was incredibly significant because their data indicate that, in the genetic model of Atp1α3 mutation, there is increased excitability and marked dysfunction in GABAergic inhibition. This supports the performance of further investigations to determine if selective expression of the mutation in GABAergic and or glutamatergic neurons is necessary and sufficient to result in the behavioral phenotype.

Researchers working on this project include:

Arsen S. Hunanyan Ashley R. Helseth Elie Abdelnour Bassil Kherallah Monisha Sachdev Leeyup Chung Melanie Masoud Jordan Richardson Qiang Li J. Victor Nadler Scott D. Moore Mohamad A. Mikati

The AHCF is grateful to all of the researchers working towards our One Mission: End AHC!

We are looking for suggestions on which topics to present during future webinars.

If you have ideas about what would be most helpful to you and other AHC families please let us know.