



STEP UP 4 AHC

FUNDING THE AAV PROJECT AND AHC RELATED RESEARCH

AHC Foundation Newsletter

July 2019

EXCITING CHANGES FOR AHCF

Board of Directors' Election Seat New President – Josh Marszalek



It has been seven years since we had a change in leadership in the president's office and we're excited to see where we go under the leadership of Josh Marszalek.

Here's a little bit about Josh in his own words.

I am Joshua Marszalek, and I am honored to be the new president of AHCF. I was **first introduced to the foundation in 2012 when our daughter Kathryn was diagnosed with AHC.** Like all AHC families, we had many questions, and AHCF was our greatest resource (and still is!)

I joined the AHCF board of directors in 2013 and have **previously served as Secretary and Vice President.** I am a licensed architect, and own my own architecture practice, Rural Valley Architecture. Prior to that, I have worked in Connecticut and at CSO Architects in Indianapolis, IN.

My wife, Laura and I, live in Connersville, IN with our son Levi and our daughter Kathryn. I am excited about the future for our children. There are many families and donors who are passionate about ending AHC, and by working collaboratively, it is something we will accomplish! **Our work will continue to align with our mission through research, education, and family support to END AHC.**

A FUN FUNDRAISER THIS AUGUST

Drink a Pint and Grab a Bite for AHC

A fun and community building fundraising event will take place on **Saturday, August 10th at the Embers Tap House in Lockport, Illinois.**

For a \$40 donation, drinks and food will be provided from 6:00 to 10:00 pm. It is an exciting and fun way to support the AHC Foundation and those suffering from AHC.

But, if you can't join us, copy us. That's right. Plan your own evening celebrating AHC patients and their families in your own hope town. It's a great time and a great way to raise money for AHC research with the AHC Foundation.



AHCF
Serving the
International
AHC community
since 1993



AHCF
Newsletter



Sharing
information for
advocates,
caregivers,
professionals
and families.



Thank you to
everyone who
helped raise
funds during
the month of
July.

You are doing
the most
important
work for AHC
research and
your efforts
are greatly
appreciated.

THANKS!

FUNDRAISING OPPORTUNITY FOR AAV PROJECT CONTINUES
Second Matching Grant Challenge Made to AHC Community

The Blockbuster Summer Sequel is here!

We have another opportunity to double our donations this summer!

Thanks to the Wedum Family Foundation, we will have all donations made from June 1st thru August 31 matched **up to \$25,000!**

This means, that the price of a single movie ticket (\$12) becomes \$24. A family of four trip to the movies becomes \$96. Or, more simply, a \$100 donation gets matched and we now have \$200.

These funds raised this summer will go towards the AAV Gene Therapy Project and other related research.

Encourage your friends & family to donate this summer. It is double the fun. Simply donate, online (www.ahckids.org or Facebook) by mail, or through personal (peer-to-peer) pages linked to the AHCF.

With your help we can have a blockbuster of a summer raising money for research and get that much closer to a treatment for AHC.

AHC COMMUNITY CONNECTIONS
Golf Outing to Benefit AHCF in August

The 8th Annual Illinois Golf Outing will take place on Friday, August 2nd at Deer Creek Golf Course in University Park, IL. A dinner is served at 3:00 pm and many prizes are awarded too. If you would like to join us, please reach out to Gene Andrasco at gene@ahckids.org.



The May 2019

\$50,000 Challenge was met by the AHC community.

In total, we raised \$109,196 because of your efforts.

AWESOME JOB!

We now need your help on a new challenge finishing up in the month of August.

AHCF 2019

Directors:

- Gene Andrasco
- Sharon Ciccodicola
- Cate Cohen
- Lynn Egan
- Heather Gates
- Bill Gerber
- Rik Greenwood
- April Hawk
- Renee Hodes
- Shannon Leigh
- Vicky Platt
- Josh Marszalek
- Meredith Schalick

PUBLICATION FROM JAPANESE RESEARCHERS FOCUSES ON THE HEART **Additional Sleep Studies Recommended**

In May 2019, an article titled, “**Heart Rate Variability in a Patient with Alternating Hemiplegia,**” was published in the journal *Intractable & Rare Diseases Research*. The paper looked at the issue of autonomic nervous system dysfunction being associated with AHC even though there are no reports of heart rate variability (HRV) in patients with AHC.

They found, an elevated heart rate lasting one to two minutes and sometimes longer, was primarily observed at night while the patient was sleeping. Large fluctuations in HRV, including low- and high-frequency components, were primarily observed while the patient was sleeping but suppressed during paralytic attacks.

These results confirm the presence of an autonomic nervous system disorder in AHC. Because large variation of the autonomic nervous function was observed at night, the pathophysiological function should be investigated for 24 hours.



We extend our thanks to the Japanese researchers for sharing this important work with our community.

GENE THERAPY 101 FOR THE AAV PROJECT **Some Basic Principles of Gene Therapy**

The AHC Community has come together in an amazing fashion to raise money for the AAV Gene Therapy Project. We are happy to share with you some basic principals of gene therapy.

The first principal we'll cover is Adeno-Associated Viral Vectors

Adeno-associated viruses, from the parvovirus family, are small viruses with a genome of single stranded DNA. **These viruses can insert genetic material at a specific site on chromosome 19 with near 100% certainty.** There are a few disadvantages to using AAV, including the small amount of DNA it can carry (low capacity) and the difficulty in producing it. **This type of virus is being used, however, because it is non-pathogenic (most people carry this harmless virus).** In contrast to adenoviruses, most people treated with AAV will not build an immune response to remove the virus and the cells that have been successfully treated with it.

Several trials with AAV are on-going or in preparation, mainly trying to treat muscle and eye diseases; the two tissues where the virus seems particularly useful. However, clinical trials have also been initiated where AAV vectors are used to deliver genes to the brain. This is possible because **AAV viruses can infect non-dividing (quiescent) cells, such as neurons in which their genomes are expressed for a long time.**

The above information is from Gene Therapy Net.com



Help AHC research efforts grow this season and donate to the AHCF.

AHC BIBLIOGRAPHY UPDATED FOR LAST 6 MONTHS**Check out the Latest Research Publications**

The AHC Foundation is pleased to provide the AHC Community with a list of research articles published over the years on AHC-related topics. While this bibliography is in no way complete, it is a comprehensive record of academic and scientific publications relevant to our community.



Categories Include: AAV, ATP1A2, ATP1A3, Behavior, Dystonia, Epilepsy, Genetics, Medication, Migraine, Treatment, and General Research.

Here is a peak at the five most recent articles.

Lardinois KL, Guge N. **“Physical Therapy for a Patient With Alternating Hemiplegia of Childhood: A Case Report.”** *Pediatric Physical Therapy*. Jul 2019; 31(3): E8-E14. Abstract: <https://insights.ovid.com/pubmed?pmid=31220019>

Domenger C, Grimm D. **“Next-generation AAV Vectors – Don’t Judge a Virus (only) by its Cover.”** *Hum Mol Genet*. 2019 Jul 1. Ahead of print. Abstract: <https://www.ncbi.nlm.nih.gov/pubmed/31261383>

Yang X, Yang X, Chen J, Li S, Zeng Q, Huang AY, Ye AY, Yu Z, Wang S, Jiang Y, Wu X, Wu Q, Wei L, Zhang Y. **“ATP1A3 Mosaicism in Families with Alternating Hemiplegia of Childhood.”** *Clin Genet*. 2019 Jul; 96(1): 43-52. Abstract: <https://www.ncbi.nlm.nih.gov/pubmed/30891744>

Pattali R, Mou Y, Li XJ. **“AAV9 Vector: A Novel Modality in Gene Therapy for Spinal Muscular Atrophy.”** *Gene Ther*. 2019 Jun 26. Ahead of print. Abstract: <https://www.ncbi.nlm.nih.gov/pubmed/31238336>

Crudele JM, Chamberlain JS. **“AAV-based Gene Therapies for the Muscular Dystrophies.”** *Hum Mol Genet*. 2019 Jun 25. Ahead of print. Abstract: <https://www.ncbi.nlm.nih.gov/pubmed/31238336>

OUR CONGRATULATIONS TO DR. ERIN HEINZEN

Irving Institute Announces 2019 Irving Scholars

On May 24, 2019, the Irving Institute for Clinical and Translational Research, together with Dean Lee Goldman, announced that Dr. Erin Heinzen was a 2019-2022 Irving Scholars awardee.

Recognizing the critical importance of training young clinical investigators, Herbert and Florence Irving created the Irving Scholars. The program is at Columbia University Irving Medical Center. **Scholars are selected based on research proposals that reflect independent, well-developed scientific initiative in clinical investigation.**



**Congrats
Dr. Heinzen**



**It is time to
join the
movement.**

**Be part of the
team
making the
AAV Project
a reality.**

**Contact
Josh
Marszalek for
more info
joshua@
ahckids.org**



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