



AHC Foundation Newsletter January 2019

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As 2019 Gets Underway the New Year is an Amazing Time of Great Advances for the Entire AHC Community and the AHC Foundation

As we pass the six month milestone for the AAV Project, we wanted to provide the AHC community with a brief update and some clarity about next steps. Since June 2018, the AHC Foundation, Cure AHC, and Hope for Annabel have collaborated on a gene therapy effort using Adeno Associated Virus (AAV) as a delivery system for functioning ATP1A3 to compensate for the mutated ATP1A3 associated with AHC. **Our collaboration has involved pooling our financial resources and leveraging connections with scientists around the world to work together.**

To eventually and hopefully get to a clinical trial with AHC patients, the AAV Project will have several phases with multiple steps in each phase. We are in just the first phase of **the AAV Project where we aim to develop a viral vector to deliver extra ATP1A3 in mice to assess whether AHC symptoms improve after treatment.**



In Phase One, the three foundations, along with contributions from AHC organizations in the United Kingdom and Ireland, have already funded over \$225,000 to complete the following steps:

1. Partnered with Dr. Steve Gray at UT-Southwestern who will help us lead the AAV Project – completed October 2018.
2. Designed and produced four viral vectors to deliver functioning ATP1A3 – completed November 2018.
3. Injected non-diseased mice using the four viral vectors to deliver functioning ATP1A3 – completed November 2018.
4. Assess which vector did the best job in delivering extra ATP1A3 to targeted areas of the mouse brain – started December 2018 and will be done March 2019.

More coverage of the AAV Project on the next page.

AHCF
Serving the
International
AHC community
since 1993



To comment on the newsletter contact:
Vicky Platt at
vicky@ahckids.org



To verify the financial strength of our foundation, check out our status with GuideStar or Great NonProfits.

AAV PROJECT UPDATE CONTINUED
Continuing the Great News on a Great Opportunity in 2019



The next step will cost approximately \$500,000 and is scheduled to start April 1, 2019 at Jackson Labs.

We will be selecting the best performing vector to deliver extra ATP1A3 to mice with AHC, and then **perform a variety of behavior experiments to see whether the AHC symptoms show improvement.** If the AAV treatment shows significant improvement in the mice with AHC, we would be on our way to the next phase of testing and closer to a clinical trial.

As a result of our fundraising efforts, AHC Foundation, Cure AHC, and Hope for Annabel have approximately \$250,000-300,000 currently available to fund this next step of the AAV Project.

While we are hopeful that continued fundraising will result in filling the gap in the coming weeks, we need every family to keep contributing to this effort.

Families and organizations wanting to support the AAV Project should feel comfortable directing fundraising efforts to one, two or all three of the foundations: AHC Foundation, Cure AHC, or Hope for Annabel.

For questions about the AAV Project or specific fundraising efforts, contact

Lynn Egan (lynn@ahckids.org), Josh Marszalek (joshua@ahckids.org) or Meredith Schalick (meredith@ahckids.org)

To donate and help us reach our goal, click www.ahckids.org

Thank you in advance for your support.



It is time to join the movement.

- **Be part of the team making the AAV Project a reality.**
- **Contact Lynn Egan for more info**

SHARE AHC INFO

If you know of friends, family teachers, or therapists who would benefit from our newsletter, share this issue with them & ask them to join our mailing list.

The AAV Project has been designed in a very **SMART** way.

The goals of the project give our community a great chance at determining if gene therapy is a viable treatment option for AHC.

Be part of the AAV Project's success and join us today.

S	Specific
M	Measurable
A	Attainable
R	Relevant
T	Time-Sensitive

Through YOUR Support and Fundraising Efforts
The AHCF Board of Directors Authorized the Funding of the Following Research
Projects in January 2019.

- Northwestern Univ. grant with Dr. George – Second Half \$79,714
- Vanderbilt Univ. grant with Dr. Ess – Second Half \$42,107
- Northwestern Univ. grant with Dr. Thomas Holmes \$37,723
- Univ. of Texas SW grant with Dr. Gray for AAV Project \$17,036
 - IHC AAV Project \$13,650
- Harvard Univ. grant with Dr. Swoboda for database \$30,000



CELEBRATE this amazing achievement in AHC research.
It belongs to all of us.



AHCF
Donated over \$220,000 to fund AHC research in the month of January because of your hard work!

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AHCF CELEBRATES INTERNATIONAL AHC DAY THIS MONTH
A Webinar to Help Parents was Held on January 17th – Is it AHC?



The AHC Foundation kicked 2019 off to a strong start with an incredibly beneficial event in advance of AHC Day.

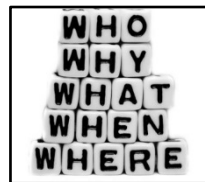
We were so fortunate to have **Dr. Matt Sweney**, our medical advisory board chair, lead us for one hour of valuable education in our first webinar of the new year!

As we all know, our AHC children and young adults are unique in many ways, and even within the realm of AHC, there are so many different presentations of this disease.

We frequently have questions about symptoms and behaviors our children are exhibiting, all with the underlying bigger question- “Is this AHC or something else entirely?”

This webinar looked to help answer our questions, shed some light, and lessen our confusion, so we can move forward with more direction in the management of AHC and our child.

We must thank Dr. Sweney for leading this webinar and helping our families in such a generous way. Keep an eye out for when the webinar is available on our website.



Do you have questions about AHC?

Are you looking for people who understand life with AHC?

We're here to help.
www.ahckids.org

AHC RESOURCES FROM THE AHCF**Bibliography on AHC Research Updated Through December 2018**

We are pleased to provide an update to the AHC Bibliography of research articles published on AHC-related topics. **Below are November & December papers** and the complete bibliography can be viewed on our website at www.ahckids.org.



Topics covered in the full bibliography are: AAV, ATP1A2, ATP1A3, Behavior, Dystonia, Epilepsy, Genetics, Medication, Migraine, Treatment, and General.

Panagiotakaki E. “Alternating Hemiplegia of Childhood: The Gap Between Paroxysmal Manifestations and Non-Paroxysmal Characteristics.” *Developmental Medicine & Child Neurology*. 2018 Dec 13. Ahead of print.

Shrivastava A, Triller A, Melki R. “Cell Biology and Dynamics of Neuronal Na⁺/K⁺-ATPase in Health and Diseases.” *Neuropharmacology*. 2018 Dec 11. Abstract:

<https://www.sciencedirect.com/science/article/pii/S0028390818309079?via%3Dihub>

Hakim CH, Wasala NB, Nelson CE, Wasala LP, Yue Y, Louderman JA, Lessa TB, Dai A, Zhang K, Duan D. “AAV CRISPR Editing Resuces Cardiac and Muscle Function for 18 Months in Dystrophic Mice.” *JCL Insight*. 2018 Dec 6; 3(23). Fulltext: <https://insight.jci.org/articles/view/124297>

Polanowska K, Dziezyc K, Rosewich H, Ohlenbusch A, Seniow J. “Alternating Hemiplegia of Childhood in Two Adult Patients with a Mild Syndrome.” *Cognitive and Behavioral Neurology*. Dec 2018; 31(4): 214-219. Abstract: <https://insights.ovid.com/pubmed?pmid=30562231>

Sampedro Castaneda M, Zanuteli E, Scalco RS, Scaramuzzi V, Marquest Caldas V, Conti Reed U, da Silva AMS, O’Callaghan B, Phadke R, Bugiardini E, Sud R, Matthews E. “A Novel ATP1A2 Mutation in a Patient with Hypokalaemic Periodic Paralysis and CNS Symptoms.” *Brain*. 2018 Dec; 141 (12): 3308-3318. Full Text: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6262219/pdf/awv283.pdf>

Kros L, Lykke-Hartmann K, Khodakhah K. “Increased Susceptibility to Cortical Spreading Depression and Epileptiform Activity in a Mouse Model for FHM2.” *Scientific Reports*. 2018; 8:16959. Fulltext: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6240030/pdf/41598_2018_Article_35285.pdf

Li SP, Zhang YH, Yang XL, Chen JY, Zeng Q, Zhang J, Wu XR. “Genotype-phenotype Correlation in Patients with Alternating Hemiplegia of Childhood.” *Zhonghua Er Ke Za Zhi* (Chinese). 2018 Nov 2; 56(11): 811-817. Abstract: <https://www.ncbi.nlm.nih.gov/pubmed/30392204>

Ishihara N, Inagaki H, Miyake M, Kawamura Y, Yoshikawa t, Kurahashi H. “A Case of Early Onset Life-threatening Epilepsy Associated with a Novel ATP1A3 Gene Variant.” *Brain & Development*. 2018 Nov 1; S0387-7604. Abstract: [https://www.brainanddevelopment.com/article/S0387-7604\(18\)30256-0/fulltext](https://www.brainanddevelopment.com/article/S0387-7604(18)30256-0/fulltext)

The Board of Directors
of the AHC Foundation wish
everyone in the AHC Community a
very Happy & Healthy
New Year.



May 2019 be filled with new adventures
which bring you joy & happiness.

Gene, Shannon, Sharon, Cate, Lynn,
Heather, Bill, Rik, April, Renee, Josh,
Mario, Vicky, Carol, and Meredith

AHC COMMUNITY CONNECTIONS

Have Fun Exchanging Valentine's Day Wishes with AHC Friends

It's that time of year to think about **our AHC Valentine's Day card exchange!**

For those new to the group, our children and adults send cards (homemade or bought), photos, photo cards, drawing, stickers, whatever they like to each person participating.

This started over 10 years ago by Heather Gates to keep our children connected between family meetings. It is a favorite of Heather's daughter and she saves all her cards (photo of a few below). This is open to all ages and we have many from Canada and few other countries who participate.

If you are interested, please send your mailing address, child's name and age to heather@ahckids.org. The deadline to let Heather know is **February 2nd**. Heather will then send a list of all participating to your e-mail.

Thanks, Heather, for coordinating this activity once again.



AHCF PUTS THE FUN IN FUNDRAISING

Win Amazing Quilt Raffle with Warriors for Kathryn Going on Now

This year we will be raffling off **"Dreamcatcher"**, a 100" by 100" quilt made using Hoffman Bali Watercolors, 100% cotton hand-dyed batik fabrics produced in Bali. **The Raffle will be drawn February 2nd, 2019** at 7 pm during the Warriors for Kathryn event in Connersville, Indiana, **but you need not be present to win.**



To learn more and enter for your chance at giving "Dreamcatcher" a home on your bed and help AHC kids find a cure go to:

www.ahckids.org/quiltraffle

Online Raffle Ticket Sales will close at 3 pm Eastern Time on day of drawing. Tickets cost \$5 per chance. Buy 4 tickets for \$20 and get one free. Each \$20 gets you 5 tickets. (10 for \$40, 15 for \$60, 20 for \$80....) If you have any questions, please contact joshua@ahckids.org.



To volunteer with the AHCF contact Lynn Egan at: lynn@ahckids.org

Check out our website



www.ahckids.org



Thank you to everyone who is helping to achieve our **One Mission to End AHC.**