



STEP UP 4 AHC

FUNDING THE AAV PROJECT AND AHC RELATED RESEARCH

AHC Foundation Newsletter

June 2019

EXCITING CHANGES FOR AHCF AS WE SAY THANK YOU

Board of Directors' Election Seat New Officers to the Board This Month



It has been my honor to serve as president of the AHCF for the last 7 years. But like all good things, they must come to an end. I will be stepping down as president at the end of this month.

I have been involved with the foundation since its beginning in 1993. I have watched it grow to the incredible organization it is today. The AHC community is lucky to have a Board of Directors who will continue to work hard for you.

When my daughter was diagnosed 28 years ago, my first thought (like all of you) was to find out everything and anything about AHC. And of course, this is before the internet. Stanford Medical Library was my second home as I read article after article on AHC, educating myself about the disease. Soon I discovered and connected with other families and that was the lifeline I needed.

I am leaving the foundation in wonderful hands. **Josh Marszalek will step in as President and Meredith Schalick will step in as Vice President.** Vicky Platt will continue as Secretary; Gene Andrasco as Treasurer and Bill Gerber, Shannon Berta, Cate Cohen, Rik Greenwood, April Hawk, Sharon Ciccodicola, Heather Gates, and Renee Hodes will continue to serve as Board of Directors.

I will continue to serve on the board as past president and will work with the foundation and support it where I can. Thank you for your support, input and encouragement over the years. **You are all like family to me.**

OUR THANKS TO AN AMAZING AHC PARENT & VOLUNTEER

Carol Presunka Steps Down from AHCF Board of Directors

It is with much appreciation that the AHCF recognizes the service of Carol Presunka to the AHC community and our Board of Directors.

This month Carol is stepping down from the board after serving the foundation for almost twenty years. Her experience, clever wit, and compassionate demeanor are just a few of her amazing qualities. We wish you well and thank you for your service. **Thanks Carol!!!**



AHCF
Serving the
International
AHC community
since 1993



AHCF
Newsletter

Sharing
information for
advocates,
caregivers,
professionals
and families.



Thank you
Lynn Egan
for your many
years of
dedicated
service to the
foundation.

You helped
start an
organization
that will serve
the AHC
community
for many
years to come.

THANKS!



An Update on the AAV Project Just Released on June 14, 2019

A joint message from AHCF, CureAHC and Hope for Annabel was released this month. It is published in our newsletter for all those who haven't had the chance to read it yet. Enjoy!

As we approach the first anniversary of this ground-breaking collaborative effort between Cure AHC, AHC Foundation and Hope for Annabel, we are pleased to share our latest progress on the AAV gene therapy project. Keeping with our space travel analogy, **we are excited to announce that we now have a clear target for our rocket-ship!** So, we just need a few more tests before we are ready to load up with supplies and start the countdown for the first rocket-ship flight to test our viral vectors on mice with AHC.

Since June 2018, AHC Foundation, Cure AHC and Hope for Annabel have been **collaborating on a gene therapy effort** using Adeno Associated Virus (AAV) as a system to deliver functioning ATP1A3 to compensate for the mutated ATP1A3 associated with AHC. This project will require many phases or rocket-ship flights with several steps in each phase/flight to hopefully get to a clinical trial by 2022. We are just in the first phase of the AAV Project where we are developing a viral vector and testing its effects in mice.



In our last update in March 2019, we celebrated the **incredible fundraising efforts of our global AHC community to help fund this initial phase of developing an effective AAV gene therapy** treatment. We also shared that we needed to do additional "quality control" testing on bio-distribution and potency of our viral vectors to make sure they are as effective as possible before we test them in mice with AHC.

We have a target and modified our supply list: After repeating tests on our vectors, we now have incredibly important information about how best to use our vectors to get optimal bio-distribution in mice brains. Based upon this new information, we made some changes to the vector design to also increase potency. We are still waiting for a few more test results but generally we are seeing positive news about the vectors. We are in the process of producing new batch of these modified vectors that should be ready to begin testing on mice with AHC in the next 2-3 months.



We have expanded our crew: We are fortunate to be working with amazing scientists, universities and research labs around the world. Since our last update, **we expanded our "crew" to include another lab to increase capacity and maintain an aggressive timeline for testing our vectors and performing experiments on mice with AHC.** The scientists and labs have shared information and materials so that we can develop an AHC mouse colony for these experiments as quickly and inexpensively as possible.

We took steps to ensure a safe and affordable landing: One of our biggest concerns is that if we **continued on next page...**



**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 thank
you for your
hard work
and
dedication
to AHC
research.**

AHCF 2019 Directors:

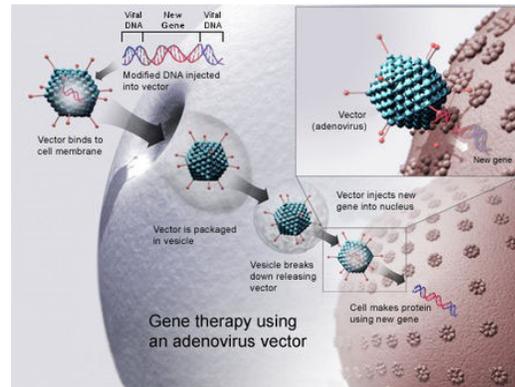
Gene Andrasco
Sharon
Ciccodicola
Cate Cohen
Lynn Egan
Heather Gates
Bill Gerber
Rik Greenwood
April Hawk
Renee Hodes
Shannon Leigh
Vicky Platt
Josh Marszalek
Meredith
Schalick

AAV Project Update Continued ...

are able to develop an effective treatment using AAV gene therapy, we want to make sure that the treatment is affordable for all families. **Due to the free legal services donated by the Cozen O'Conner law firm, we were able to file a patent in the US and internationally with ownership assigned to AHC Foundation, Cure AHC and Hope for Annabel to try to protect the intellectual property rights of this collaborative research effort.** By protecting the science, we are in a stronger position to negotiate with industry to keep the cost of any treatment as low as possible. While we will have filing fees and other costs associated with these patents, we believe that the investment is necessary to make the "landing" or treatment affordable for everyone.

Although we are ecstatic about the recent development of effective gene therapy treatments for the SMA rare disease community, we also see why it is important to protect our intellectual property rights to avoid the astronomical costs often associated with industry development of treatments for rare diseases. The new SMA gene therapy treatments cost per patient between \$2.1 million for a one-time treatment or \$4 million for treatment over 10 years.

Keep supporting our AAV gene therapy mission: As a community, we need to continue to raise money so that we can fund and control the development of a possible effective AAV gene therapy on our terms to try to ensure access for all families. **The more money we raise as a community, the more leverage we have in negotiating terms as we move towards clinical trial by 2022.**



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 please contact:

Lynn Egan (lynn@ahckids.org)

Josh Marszalek
(joshua@ahckids.org)

Meredith Schalick
(meredith@ahckids.org)



It is time to join the movement.

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

Contact Lynn Egan for more info
lynn@ahckids.org



DONATE



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.

A GREAT LOSS TO THE AHC COMMUNITY

Dr. Fred Andermann Passed Away this Month

It is with a heavy heart that we announce the passing of Dr. Fred Andermann, Montreal, Canada. **He was an amazing, thoughtful, compassionate gentle soul, who always had time to listen and offer suggestions when it came to AHC.**

He became involved with the AHC Foundation in 1995. He attended numerous family meeting and was instrumental in co-hosting the International AHC Symposium in 2006. Dr. Andermann was a leading expert in AHC and saw many families from the AHC community during his career as a neurologist at the Montreal Institute and Hospital.

In addition to his work with AHC, Dr. Andermann **was a giant in the epilepsy community with contributions in numerous areas of clinical epilepsy.** He also trained and inspired a large number of fellows who have become recognized leaders all over the world. He is a past vice-president of the ILAE and was involved in many activities of the League. He was awarded ILAE's Lifetime Achievement Award in 2015.

In 2016, the AHCF awarded Dr. Andermann with Medical Advisory Board Member Emeritus for his service to the AHC community. Lynn Egan had this to say about Dr. Andermann,

“he was one of the first neurologists that I spoke to when my daughter was diagnosed in 1991. **While he was very honest about AHC, he also offered me some hope.** He gave me his personal phone number to his country home in the event I needed to talk with him about my daughter. It was comforting to know that I had someone so amazing and willing to help me navigate AHC in those early days.”

Fred is survived by his wife and lifelong collaborator Eva Andermann, his children Lisa, Anne and Mark, and his six grandchildren, Hanna (12), Lara (11), Jamie (9), Leila (9), Benjamin (7) and Kaya (7). **He will be lovingly remembered by his family and his many friends and colleagues.**

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 FAMILY MEETING 2020

Did you just get excited about the next meeting be set for June 25-28?

Did you just get really excited that we'll be in the Los Angeles area in 2020?

Well, if you did, maybe you should consider serving on the planning committee for our next meeting.

We're starting the planning process for our next meeting in June 2020 and can use your input.

If you'd like to help us plan the meeting, Contact Sharon at sharon@ahckids



BIG FUNDRAISING OPPORTUNITY FOR AAV PROJECT BEGINS
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 for more information.



Check out our website



www.ahckids.org

The AHCF wishes all of our AHC Dads a very **Happy Father's Day.**

You are an important part of the AHC community and we hope you enjoy your special day.



Do you have questions about AHC? Are you looking for people who understand life with AHC?

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