



STEP UP 4 AHC

FUNDING THE AAV PROJECT AND AHC RELATED RESEARCH

AHC Foundation Newsletter March 2019



An Update on the AAV Project Just Released on March 5, 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!

To pick an analogy for this second AAV Project update, we would say that it is like preparing for space travel. We have the captain and crew, but before we start the countdown, we need to make sure our rocket has a clear target, is thoroughly tested, and is fully loaded with fuel and supplies.

Since June 2018, Cure AHC, AHC Foundation 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 with several steps in each phase to eventually get to a clinical trial. 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 on January 20th, we shared that the three foundations had funded over \$225,000 of preliminary research and development in preparation for the experiments on mice scheduled to start on April 1st. We shared that this next step of mice experiments would cost approximately \$500,000.

Our rocket is fueled: Due to the phenomenal fundraising efforts by families, friends and strangers, we are thrilled to share that the 3 foundations have \$436,000 in the bank to dedicate to the next steps of the AAV Project! In addition, our international partners from France, Iceland, Ireland, Netherlands, Spain and the United Kingdom have pledged over \$110,000 towards the AAV Project. We are absolutely in awe of the support of this AAV Project by the AHC community and beyond!



While we have been able to pool resources to meet and exceed the \$500,000 goal for the mouse experiments, please don't stop your fundraising efforts! The AAV Project is a multi-phased initiative. Our gene therapy project is already attractive to industry and institutions who are offering financial support, and the more we can raise as a community the more we can attract the right institutions and the right capital partners. The goal is to keep our therapy affordable or ...

AHCF
Serving the
International
AHC community
since 1993



AHCF
Newsletter

Sharing
information for
advocates,
caregivers,
professionals
and families.

It will take
more than luck
to End AHC.

It will take the
work of our
entire AHC
community.

And we're
lucky you're
here to help.



AAV Project Update Continued ...

or free for patients and families, and there is much more that we as a community will need to do and fund before we can accomplish that.

Redundancy plans, safety checks, and rocket-supplies: Our therapy is performing very well in initial experiments, but we have concluded that further testing is necessary before we launch our rocket-ship. We are determined to proceed carefully and methodically. Thoroughness is even more important than speed.

There are still two fundamental questions we need to answer before our rocket has “lift-off”: (1) can our rocket go far enough (biodistribution), and (2) is our payload potent enough (viral potency). As a result, we are doing another round of quality-control testing, and we are building the same set of viral vectors using a different production technique as a redundancy plan.

To avoid lift-off too early, we need to make sure our therapy will deliver ATP1A3 to the right parts of the brain in exactly the right amounts. We estimate these additional tests will take approximately three months. In addition, the development of the mouse colony for experiments is also seeing some delays. We need the mice to breed quickly to create a large enough colony for our planned tests but breeding mice with AHC is challenging. The upside is that by doing our additional quality control testing, the mice colony will have time to grow as well.

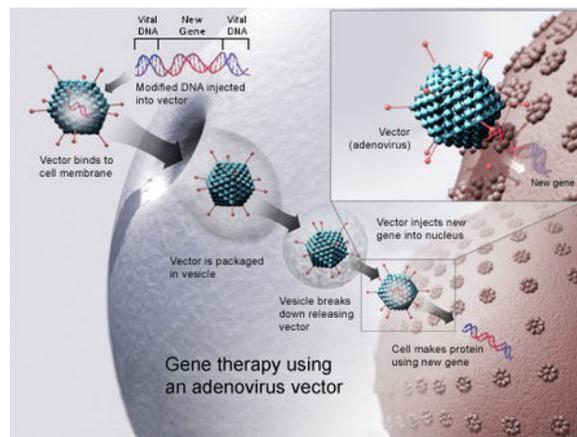
We want to make sure that the families who have embraced this effort are not disheartened. Timeline delays are commonplace for meticulous scientists. We have a rocket-ship that is fully fueled, with a captain and a crew. Take-off is slightly delayed since we have chosen to implement additional tests. **We feel an intense responsibility to be excellent stewards of your fundraising efforts to ensure we get the best therapeutic results for our kids.**

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) or 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.

FAMILY FROM MINNESOTA CRUSHES FUNDRAISING EFFORT

Meet the Matuska's and Learn from Their Amazing Success



It is impossible not to oohh and aahh over this adorable picture of Lainey. She is not only cute as a button, but is also the daughter of Stacie and Rob Matuska and the little sister of Calvin Matuska.

Lainey was diagnosed with AHC at 5.5 months of age (epilepsy too). That is when her parents refused to sit around and let these diseases strip her of a full life and began raising funds for the AAV Project.

The Matuska's fundraising efforts started by setting up an honorarium page for Lainey on the AHCF website. Lainey's page tells her AHC story and lets friends and family donate directly to her cause. It was easy to do and share right away via email and Facebook.

Next came an incredible amount of variety in fundraising efforts. Efforts that were directed towards their family by educating them about AHC. Efforts that were supported by their circle of friends with big and small fundraisers. And efforts that were seen by their local community using social media.

The Matuska's also received publicity on the noon television broadcast of WCCO, a Minnesota CBS news station. They were able to chat about Lainey and the fundraising events that were taking place to raise money for the AAV Project. It was a 3 minute segment and a big opportunity for a family new to the AHC community.



Taking on the challenge of being on a t.v. news segment to benefit the AAV Project was incredibly brave.

The Matuska's have set an incredibly aggressive goal of raising \$100,000 for the AAV Project. In Less than two months, they are well on their way with already raising over \$47,000. An amazing accomplishment indeed.

On Lainey's honorarium page there is a comment from an anonymous friend which says, "While I don't know Lainey personally, her story and courage has touched my heart." The fundraising efforts of the entire Matuska family have now touched the entire AHC community.

We thank Stacie and Rob for stepping up to help fund the AAV Project and work towards our One Mission to End AHC!



AHCF FAMILY MEETING 2020

Did you just get excited?

Did you just wonder if you missed the news about a location?

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, we'd love to hear from you.

Contact Sharon at sharon@ahckids.org

For more info
Thanks!

JAPANESE RESEARCHERS CONNECT ATP1A3 & EPILEPSY

Important New Research Just Published

This month, an article in the journal *Brain & Development* was published by a team of Japanese researchers. The title of the article is, “A Case of Early Onset Life-threatening Epilepsy Associated with a Novel ATP1A3 Gene Variant.”

The subject of the article is a boy who was transferred to the NICU after a normal birth who experienced respiratory failure at 2 days of age.



The researchers reported a pediatric case of catastrophic early life epilepsy, respiratory failure, postnatal microcephaly, and severe developmental disability [Grab your reader’s attention with a great quote from the document or use this space to emphasize a key point. To place this text box anywhere on the page, just drag it.]

associated with a novel heterozygous ATP1A3 mutation. They identified several reported cases with severe symptoms and very early onset epilepsy harboring ATP1 α 3 mutations at structural positions in a protein that differ from that of Phe913. They suggest that further functional studies are required to clarify the relationship between the loss of Phe913 and the

RESEARCHERS I.D. NEW CONDITION RELATED TO AHC

Important New Research Just Published

In the journal, *Neuropediatrics*, a team of researchers in Michigan recently published an article titled, “Homozygous TANGO2 Single Nucleotide Variants Presenting with Additional Manifestations Resembling Alternating Hemiplegia of Childhood—Expanding the Phenotype of a Recently Reported Condition.”

The article was about a 15-year-old Indian girl who presented with epilepsy, developmental delay, neuroregression, and episodes of alternating hemiparesis. Extensive genetic and metabolic work up through the years was unrevealing. Eventually a trio whole exome sequencing (WES) revealed homozygous single nucleotide variants in TANGO2 gene.

They discussed TANGO2 related recurrent metabolic crises with encephalomyopathy and cardiac arrhythmias with only 15 cases being reported in literature at the time of writing. Alternating hemiplegia of childhood which was seen in their patient, has not been described in previous patients with TANGO2 mutation, and thereby expands the emerging phenotypic spectrum of this novel entity. This report also reiterates the utility of WES in diagnosing newly recognized neurogenetic conditions.

To read the article’s abstract, or purchase a copy of the article, go to the publication’s website: <https://www.thieme-connect.com/products/ejournals/abstract/10.1055/s-0038-1677514>

SOMETHING BIG IS COMING APRIL 1ST!



THIS AIN'T NO JOKE

WATCH THE AHC
WEBSITE AND FACEBOOK PAGE
FOR EXCITING NEWS.

YOU WON'T WANT TO MISS IT!
WWW.AHCKIDS.ORG

Check out our
website



www.
ahckids.org

The AHCF extends our condolences to the family of Ashley Piersig, who passed away on January 28. Ashley lived in a group home in Pennsylvania and was a very cheerful person who always liked jokes and pranks. She will be greatly missed.

WHO
WHY
WHAT
WHEN
WHERE

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 COMMUNITY CONNECTIONS

International AHC Foundations Join the AAV Project



It is with great excitement that we share the wonderful news that international AHC foundations are joining in on the effort to raise money for the collaborative AAV Project.

The excitement for creating a gene therapy to End AHC is spreading across the globe. AHC family foundations from around the world are helping to raise funds for the AAV Project.

Raising money for AHC research is challenging. Now add the challenge of transferring funds to the U.S. from different currencies. No matter the difficulty, our fellow family foundations are taking on the added challenges and donating funds to this endeavor.

We encourage you to take a few minutes to get to know our friends from across the ocean and check out their websites. You'll see we have a lot in common. Also, please join us in expressing our gratitude and appreciation to each of these organization and to all families working hard to End AHC.

- 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
 Mario Merida
 Carol Presunka
 Josh Marszalek
 Meredith Schalick



IRELAND: The Alternating Hemiplegia of Childhood Ireland is a non-profit group set up and run by parents of children with AHC. You can check out their website at: www.ahci.ie.

THE UNITED KINGDOM: The Alternating Hemiplegia of Childhood UK support group has been helping patients with AHC for many years. Check out their website at: <http://www.ahcuk.co.uk/>



THE NETHERLANDS: IAHC Vereniging Nederland is the national patient organization for people with AHC. You can check out their website (with translation) at: <https://www.ahckids.nl/>

SPAIN: The Asociacion Espanola Del Sindrome De La Hemiplejia Alternante is the AHC family foundation in Spain. They just held a symposium in February and can be reached at: www.aesha.org.



Iceland: The AHC Association of Iceland also produced the AHC documentary, "Human Timebombs." Check out their site: www.ahc.is



The AHCF is pleased to welcome families to the AHC community from:

- Oklahoma
- North Carolina
- Texas
- Washington
- Bulgaria
- France
- Spain
- United Kingdom

You are among friends and we are here to help.

ahckids.org



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