



AHCF  
Serving the  
International  
AHC community  
since 1993



## AHC Foundation Newsletter

February 2019



### 2019 – There's a lot to Love About it and the Future of AHC

The **biggest fundraising campaign** undertaken by AHCF is continuing with great success! As we prepare to make the largest investment ever made in AHC research, you'll be amazed at what is now within reach for our AHC community.

**Look what some great folks in Indiana just did earlier this month! Way to Go Hoosiers!**

Kathryn Faith Marszalek, 7, daughter of Josh and Dr. Laura Marszalek, began showing symptoms of the condition at just a few months of age. The family received a diagnosis of AHC at Children's Hospital of Cincinnati.

The parish community got behind the family by creating Warriors 4 Kathryn in 2017. That year's fundraiser raised some \$17,000. The second year, the effort brought in more than \$19,000, all for research.

Organizer Carrie Pfeiffer said Warriors 4 Kathryn included a 4K run and a 1-mile run for children in kindergarten through sixth grade. They chose 4K to match the name of the event.

The fundraiser this year included the same activities as last year, ice cream social for kids, Texas Hold'em tournament, wine bingo, basket raffle, raffle for a quilt made by Kathryn's grandmother, 50/50 drawing and a barbecue dinner.



"During each of the last events, it's just humbling to watch the community come out and support something like this," Marszalek said. "Fayette County is economically depressed, but people have come out with \$10s and \$20s. We raised \$19,000 last year but only a handful of the donations were more than \$500. That blows my mind." This year, the event raised over \$26,000. **Amazing work.**

Thanks to this wonderful community support, the funds to meet the financial challenges of the AAV Project are coming together. We thank everyone who organized and participated in this event for helping to meet our **One Mission: End AHC**.



AHCF  
Newsletter

Sharing  
information  
for advocates,  
caregivers,  
professionals  
and families.



Thanks to  
Heather Gates  
for  
coordinating  
another  
successful  
Valentine  
exchange this  
year.



## The Cold Hard Facts about the AAV Project

With funds raised from the AHC Foundation, we hope to develop a cure for all of our AHC families suffering from AHC. We believe a path has been identified to a viable permanent treatment: gene therapy.

### Gene Therapy

The gene therapy we propose will involve inserting a functional copy of the ATP1A3 gene into a virus (an adeno-associated virus or AAV) that has been stripped of its harmful payload and its ability to reproduce. The virus will then be injected into the cerebrospinal fluid and will carry the functional gene to the cells in the brain that are failing. By delivering additional functional copies of the gene, we should be able to rescue those cells, and treat the disease permanently. Gene therapy has been successful before in AHC in mouse embryos, and we now need to prove that it can rescue symptoms in live mice with AHC.

### Remarkable Scientists

Leading scientists from several top universities and scientific institutions have agreed to participate in a collaborative international scientific effort to pursue the delivery of gene therapy for AHC. We have gathered and engaged the most experienced scientists in the fields of mouse viral therapy design and production, preclinical mouse research, toxicology studies, human viral therapy design and production, and human clinical trials. All of these scientists have dedicated their lives and careers to finding cures for children with life-threatening disorders. They are true heroes.

### Applications for Other Rare Diseases

The implications extend well beyond AHC. Ten different diseases are caused by mutations in the same gene that causes AHC and we might be able to treat all of those diseases with the same approach. This technique could also deliver genes to rescue other channelopathies, epilepsies and neurological disorders, with implications for hundreds of thousands of people living with related genetic diseases.

### Speed Matters

If we can get this project to clinical trials quickly, it may be possible to treat our children before it is too late. As patients get older, the long-term effects of brain atrophy may not be reversible. For all of the children who now suffer with AHC, it is imperative that we move fast, and we need your help.

**Gene therapy provides a path to a cure for AHC.** A similar approach has already been successful in many diseases like Batten's disease, Friedreich's Ataxia, Huntington's disease, Niemann-Pick disease, Parkinson's disease, Rett Syndrome, Giant Axon Neuropathy, Multiple Sclerosis, Spinal Muscular Atrophy, and multiple retinal disorders. **This is not only cutting-edge science, but also a proven, effective, and permanent fix of the genetic code.** It will be delivered by a one-time injection into the cerebrospinal fluid and is designed to permanently correct the dysfunction caused by a mutated gene. Unlike drugs, gene therapy targets the source of the problem: DNA. In many diseases it permanently corrects the dysfunction caused by a problematic gene. **We call that a cure.**

Join us in making the AAV Project a reality for the entire AHC Community.



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](#)



Don't Flake Out and Forget to Donate

Support the AAV Project Today

[www.ahckids.org](http://www.ahckids.org)



Rare Disease Day takes place on **the last day of February each year**. The main objective of Rare Disease Day is to **raise awareness** amongst the general public and decision-makers about **rare diseases and their impact on patients' lives**.

The campaign targets primarily the general public and also seeks to raise awareness amongst policy makers, public authorities, industry representatives, researchers, health professionals and anyone who has a genuine interest in rare diseases.



**The campaign started as a European event and has progressively become a world phenomenon, with the USA joining in 2009 and participation in over 90 countries all over the world in both 2017 and 2018.**

Hundreds of cities continue to take part in Rare Disease Day and hopefully even more will join in 2019. On [rarediseaseday.org](http://rarediseaseday.org) you can find information about the thousands of events happening around the world on February 28<sup>th</sup>.

### **WHAT'S GOING ON WHILE YOU SLEEP**

#### **Important New Research Just Published**

On January 25<sup>th</sup>, an article was published in the Journal of Clinical Sleep Medicine titled, "Polysomnography Findings and Sleep Disorders in Children with AHC."

In the article, **the research team at Duke University** analyzed nocturnal PSG and clinical sleep evaluation results of a group of 22 consecutive pediatric patients with AHC who were seen in their our AHC multidisciplinary clinic and who underwent evaluations according to their comprehensive AHC clinical pathway. This pathway includes, regardless of presenting symptoms, baseline PSG and evaluation by a board-certified pediatric sleep specialist.

They found that **out of 22 patients, 20 had at least one type of sleep problem**. Six had obstructive sleep apnea as documented on polysomnogram. Based on sleep history, 16 patients had difficulty falling asleep, staying asleep, or both; 9 had behavioral insomnia of childhood; and 2 had delayed sleep-wake phase syndrome.

**The article concludes that sleep dysfunction is common among children with AHC.** Physicians should routinely screen for sleep pathology, with a low threshold to obtain a nocturnal PSG.



#### **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.

**Check out our website**



**www.  
ahckids.org**



**Do you have questions about AHC?**

**Are you looking for people who understand life with AHC?**

**We're here to help.**  
[www.ahckids.org](http://www.ahckids.org)

## AHC COMMUNITY CONNECTIONS

### Check Out the Epilepsy Foundation's Speak Up, Speak Out Portal on CBD Oil



The Epilepsy Foundation has created a web portal for information about their advocacy work related to medical cannabis and cannabidiol (CBD). If you are looking for credible information on this topic, you may want to check it out.

The Epilepsy Foundation is committed to supporting physician-directed care, and to exploring and advocating for all potential treatment options for epilepsy, including cannabidiol (CBD) oil and medical cannabis. They support safe, legal access to medical cannabis and CBD if a patient and their health care team feel that the potential benefits of medical cannabis or CBD for uncontrolled epilepsy outweigh the risks.

While not everyone with epilepsy should or would consider medical cannabis or CBD as a treatment option, some people living with uncontrolled seizures have reported beneficial effects and reduced seizure activity when using medical cannabis, especially strains rich in CBD.

Further research is needed on the effects of medical cannabis on epilepsy, but when recommended by a treating physician, medical cannabis may be the best alternative for some individuals living with drug-resistant epilepsy and uncontrolled seizures. Legal access to cannabis will support increased research efforts and allow individuals who have failed to gain seizure control an option for treatment.

**Check Out:**  
<http://advocacy.epilepsy.com/medical-cannabis>

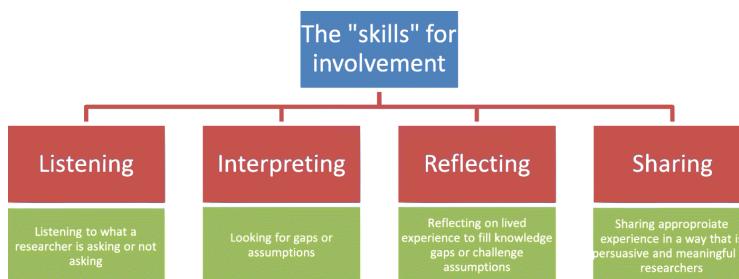
## AHC COMMUNITY CONNECTIONS

### What Can I do to Help Researchers?

An article was published this month which provides insight on how patients and the public can work collaboratively with researchers. The authors explore the expertise and other skills that patients/the public bring to process.



**Patients and members of the public have an important role to play** in helping to answer some questions for researchers. The article concluded that all parties will benefit from further consideration of how it is most appropriate to be involved and why. Addressing this issue could enhance the impact of involvement and ensure a better quality learning experience for all involved.



**Check out the full text of the article at:**

<https://researchinvolvement.biomedcentral.com/track/pdf/10.1186/s40900-019-0144-4>

**AHCF 2019 Directors:**  
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**"All who have accomplished great things have had a great aim, have fixed their gaze on a goal which was high, one which sometimes seemed impossible."**

**Orison Swett Marden.**

**Make the success of the AAV Project part of your aim in 2019 and target the End of AHC!**