STEP UP 4 AHC

FUNDING THE AAV PROJECT AND RELATED RESEARCH

AHC Foundation Newsletter

January 2020

EXCITING NEWS FROM OUR PRESIDENT
– Josh Marszalek

In 2019, the AHC community came together to meet not one, but two matching grants. Through your hard work, double the donations were made to help fund important research. In 2020, we have another opportunity to double our fundraising efforts through our largest challenge grant yet. The first $100,000 of 2020 will be matched by a generous donor.

The Poarch Band of Creek Indians of Alabama have awarded us an opportunity to make a huge impact on the future of our children. Their generosity to this rare disease community exemplifies their community mantra: Many People. One Purpose. We are honored to be their partners in our mission to END AHC.

We thank the Poarch Band of Creek Indians of Alabama you for giving us the opportunity to do what we have a passion for by supporting the foundation in this generous way. To learn a little bit more about our generous partners, read more about them on page 2.

MATCH YOUR DONATION AT WWW.AHCKIDS.ORG

2020 QUILT RAFFLE OPEN TO ALL
"Hope Rises" is Made by AHC Grandparent

We are pleased to share with our entire AHC community, a wonderful opportunity to win an amazing piece of art. Christine Marszalek created a beautiful quilt to be raffled off in honor of her granddaughter, Kathryn Marszalek.

The quilt top pattern is called Vintage Rose and has over 2,200 individual pieces! It is made using a mix of Timeless Treasures Forever patterns and mixed batiks and is 100% cotton.

Raffle tickets are $5 or 5 tickets for $20 and will be pulled at the Warriors for Kathryn event on February 15, 2020. Winner need not be present to win.

To purchase raffle tickets simply go to:
www.ahckids.org/quiltraffle.

Christine, thank you for sharing her talent with us to **help End AHC.**

AHCF
Serving the International AHC community since 1993

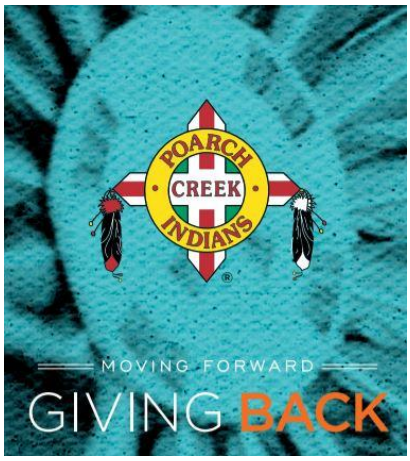
AHCF Newsletter

Sharing information for advocates, caregivers, professionals and families.

Thank you to all of our kind and **generous donors** who made end-of-year donations to the foundation.

You make all things possible for the foundation through your generosity.

Our Friends - Poarch Band of Creek Indians



As descendants of a segment of the original Creek Nation, **the Poarch Band of Creek Indians have a rich heritage.** This nation once covered most of Alabama and Georgia, and can trace its roots to the Paleo-Indian period. Their ancestors lived along the Alabama, Coosa and Tallapoosa Rivers, including areas from Wetumpka south to the Tensaw settlement.

The Treaty of Fort Jackson, in 1814, forever changed the face of the great Creek Nation, as more than 21 million acres of Creek Indian land were ceded to the United States.

In 1830, the historic Indian Removal Act imposed the resettlement of the Creek Nation from the remaining lands to present-day Oklahoma; this journey is known as The Trail of Tears. However, several Creek leaders and their families, because of their service in protecting Creek and White settlers, received **land grants, in 1814 and again in 1836, which became the home of today's Poarch Band of Creek Indians in southern Alabama**.

Through the turn of the 20th century, the Poarch settlement was largely ignored and increasingly impoverished. **In the 1940s, Tribal leaders took action to improve community conditions and educational opportunities,** and, in 1950, more formal leadership was re-established, with a dedicated leader of a formal governing body.

Currently, there are more than 2,900 members of the Tribe who possess at least ¼ Poarch Creek blood quantum.

The Reservation is located eight miles northwest of Atmore, Alabama, in rural Escambia County, and 57 miles northeast of Mobile. **The Poarch Band of Creek Indians has fought hard to preserve its proud heritage while moving forward into the mainstream of today's modern society.** The Tribe's determination to maintain both its identity and inherent right to self-government is evident by its continued efforts to preserve its Tribal culture and improve its community.

To learn more about their story, visit www.poarchneighbors.com.

SOCIAL MEDIA FUNDRAISING
Easy to Do and Successful Too

The AHCF wishes to thank everyone who hosted a birthday or special occasion fundraiser on Facebook in 2019. Social media fundraising in 2019 resulted in over $80,000 in revenue (a 23% increase over 2018).

Shannon Leigh had a positive experience using Facebook to fundraise and had this to say, "Facebook has allowed me to reach more people than I would have been able to without it." If you'd like to try it out, reach out to Shannon and she can help you get started. Shannon can be reached at shannon@ahckids.org

WATCH FOR information on registering for the June 2020 FAMILY MEETING In Los Angeles California

New programs and new sessions to learn about AHC for all AHC families and friends.

AHCF 2020 Directors:
Gene Andrasco
Sharon Ciccodicola
Cate Cohen
Lynn Egan
Heather Gates
Bill Gerber
April Hawk
Renee Hodes
Shannon Leigh
Vicky Platt
Josh Marszalek
Meredith Schalick

PUBLISHED RESEARCH SHEDS LIGHT ON AHC

Three New Papers Offer Important Insights

New research relating to AHC is being published every moth. It is with great excitement that we share with you a short abstract of three papers published in January. A link to the full text article is provided when available.

ATP1A3-related Epilepsy: Report of Seven Cases and Literature-based Analysis of Treatment Response. On January 17th, in the *Journal of Clinical Neuroscience*, an international collaboration of researchers published this interesting article.

The aim of the study was to assess the prevalence and efficacy of commonly used anti-epileptic-drugs (AEDs) in patients with ATP1A3 related seizures. They performed a retrospective study of patients in combination with a systematic literature-based review. Inclusion criteria were: verified ATP1A3 mutation, seizures and information about AED treatment. The literature review yielded records for 188 epileptic ATP1A3 patients. For 14/188 cases, information about anti-epileptic treatment (AED) was available.

Most used AED were levetiracetam (n = 9), phenobarbital (n = 8), valproic acid (n = 7), and topiramate (n = 5). Seizure reduction was reported for 57% of patients (n = 12). No individual AEDs used (either alone or combined) had a success rate over 50%. There was no significant difference in the response rate between various AEDs. Ketogenic diet was effective in 2/4 patients. 43% of patients (n = 9) did not show any seizure relief. Even though Epilepsy is a significant clinical issue in ATP1A3 patients, only a minority of publications provide any information about patients' anti-epileptic treatment. The findings of treatment effectiveness in only 57% (or lower) of patients, and the non-existence of a clear first-line AED in ATP1A3 related epilepsy stresses the need for further research.

White Matter and Cerebellar Involvement in Alternating Hemiplegia of Childhood. On January 16th, in the *Journal of Neurology*, 16 researchers from Italy collaborated on this paper. Their objective was To determine whether brain volumetric and white matter microstructural changes are present and correlate with neurological impairment in subjects with alternating hemiplegia of childhood (AHC). 12 AHC subjects (mean age 22.9 years) and 24 controls were studied with 3DT1-weighted MR imaging and high angular resolution diffusion imaging.

AHC subjects showed prevalent white matter involvement, with reduced volume in several cerebral and cerebellar regions associated with widespread microstructural changes reflecting secondary myelin injury rather than axonal loss. Conversely, no specific pattern of grey matter atrophy emerged. Lower cerebellar volumes, correlating with severity of neurological manifestations, seems related to disrupted developmental rather than neurodegenerative processes.

Clinical and Genetic Spectrum of ATP1A3-Related Disorders in a Korean Pediatric Population. On January 16th, in the *Journal of Neurology*, 10 Korean researchers collaborated on this paper. The aim of this study was to expand the understanding of the genotype-phenotype spectrum of ATP1A3-related disorders and to evaluate the therapeutic effect of a ketogenic diet in patients with alternating hemiplegia of childhood (AHC).

The clinical information of 13 patients with ATP1A3 mutations was analyzed by performing retrospective chart reviews. Patients with the AHC phenotype who consented to ketogenic diet were included in the trial. Ten patients presented with the clinical phenotype of AHC, two patients presented with rapid-onset dystonia parkinsonism, and one patient presented with CAPOS. The conclusion of the research was that a ketogenic diet showed no clear benefit for the patients with AHC.

AHC COMMUNITY IN MOURNING ONCE AGAIN

Sad News to Share

It is with great sadness that that we share news of the passing of a young woman, Allyssa Akins, who was almost 20 years old. Her grandmother notified us that Allyssa passed away this week. Allyssa's passing is likely due to complications of AHC and mitochondrial disease.

Please keep Allyssa and her family in your prayers during this difficult time.

LOOKING TO HEAR YOUR OPINIONS

AHCF Newsletter Needs You

Everything needs a little "spring cleaning" every now-and-then. And so, it is time for the newsletter to get a little attention and sprucing up. If you've had some thoughts about what you'd like to see in our newsletter, now is your time.

We're looking for some volunteers to share their opinions on a few topics. Do you have…

- Marketing skills
- Design skills
- Editing skills
- Social media skills
- Writing content skills

Sharing your talents on the newsletter is a great way to volunteer with the foundation. We'll work via emails and on your own time. Let's see what we can do create a visually compelling and informative newsletter for the entire AHC community to share.

If you're able to help, please email Vicky Platt at vicky@ahckids.org

LOOKING AT AHC THROUGH A DIFFERENT LENSE

Siblings and AHC – Let's Celebrate Them Too

On Saturday, February 29th, we will observe Rare Disease Day. It was created to raise awareness for rare disease and improve access to treatment and medical representation for individuals with rare diseases and their families.

Families caring for AHC patients manage considerable social and financial impacts associated with a rare disease. Siblings of AHC patients can also end up coping with stressful and difficult situations as part of the family. There can be reduced leisure time, isolation, emotional stress and physical fatigue. However, it can also be rewarding.

Siblings are an important support for our AHC kiddos. They provide a level of "normalcy" by being that big brother or little sister. Siblings most often understand at a young age how to watch out for their brother or sister and get help when needed.

Siblings hold fundraisers, man childcare rooms at conferences, provide entertainment to check unwanted behaviors, and show great compassion to those in need. So, next month, on Rare Disease Day, let's celebrate our AHC siblings and all that they do to make living with a rare disease a little bit better every day.