ahcf | alternating hemiplegia of childhood foundation

April 2024 Newsletter

Thank you to all who help support, volunteer, and research for our AHC Heroes!

New Gene Implicated in AHC (SCN2A)

Since over 85% of individuals with AHC have a de novo missense variant in ATP1A3 encoding the catalytic α3 subunit of neuronal Na+/K+ ATPases, the remainder of the patients are genetically unexplained. We are excited to share that there is some important news about the remaining 15% of AHC cases.

In February, an important article was published in the *European Journal of Genetics* and is titled, "Exome Sequencing of ATP1A3-negative Cases of Alternating Hemiplegia of Childhood Reveals SCN2A as a Novel Causative Gene."

One of the notable authors of this article is Dr. Erin Heinzen from the University of North Carolina at Chapel Hill. Dr. Heinzen graciously summarized the findings for us.

> "More than 10 years ago the first AHC gene, ATP1A3, was found. Genetic variants in ATP1A3 are the most common reason children get AHC, but there are still about 15% of children where we do not know the cause.
>
> In this study, we looked deeper into the genes of people with AHC who don't have an ATP1A3 variant. We found that three out of 28 children studied had a change in another gene, **SCN2A**. This gene codes for a protein that helps brain cells send signals and has been linked to seizures and learning and walking difficulties.
>
> **We also found that some children had changes in other genes (CHD2, ANK3, ATP1A2, or RHOBTB2) known to cause similar brain disorders.**
>
> This tells us that more thorough genetic testing should be done for people with AHC, especially if they don't have the ATP1A3 variant. This could help doctors better understand and treat their condition."

Thank you Dr. Heinzen for your summary and continued work with AHC.

Read Full Article

PubMed

National
Library
of Medicine NLM

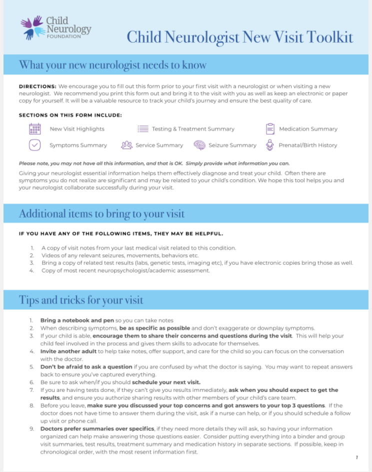
Research

UAB Dept of Neurology Research

Want to help increase knowledge about rapid-onset dystonia-parkinsonism (RDP) and other ATP1A3- related movement disorders? University of Buffalo researchers, under the direction of Dr Allison Brashear, are looking for study volunteers! Click here for more info:

UAB Dept of Neurology

Family Resources



Child Neurology FOUNDATION

Child Neurologist New Visit Toolkit

What your new neurologist needs to know

DIRECTIONS: We encourage you to fill out this form prior to your first visit with a neurologist or when visiting a new neurologist. We recommend you print this form out and bring it to the visit with you as well as keep an electronic or paper copy for yourself. It will be a valuable resource to track your child's journey and ensure the best quality of care.

SECTIONS ON THIS FORM INCLUDE:

- New Visit Highlights
- Testing & Treatment Summary
- Medication Summary
- Symptoms Summary
- Service Summary
- Seizure Summary
- Prenatal/Birth History

Please note, you may not have all this information, and that is OK. Simply provide what information you can.

Giving your neurologist essential information helps them effectively diagnose and treat your child. Often there are symptoms you do not realize are significant and may be related to your child's condition. We hope this tool helps you and your neurologist collaborate successfully during your visit.

Additional items to bring to your visit

IF YOU HAVE ANY OF THE FOLLOWING ITEMS, THEY MAY BE HELPFUL.

1. A copy of visit notes from your last medical visit related to this condition.
2. Videos of any relevant seizures, movements, behaviors etc.
3. Bring a copy of related test results (labs, genetic tests, imaging etc), if you have electronic copies bring those as well.
4. Copy of most recent neuropsychologist/academic assessment.

Tips and tricks for your visit

1. **Bring a notebook and pen** so you can take notes
2. When describing symptoms, **be as specific as possible** and don't exaggerate or downplay symptoms.
3. If your child is able, **encourage them to share their concerns and questions during the visit**. This will help your child feel involved in the process and gives them skills to advocate for themselves.
4. **Invite another adult** to help take notes, offer support, and care for the child so you can focus on the conversation with the doctor.
5. **Don't be afraid to ask a question** if you are confused by what the doctor is saying. You may want to repeat answers back to ensure you've captured everything.
6. Be sure to ask when/if you should **schedule your next visit.**
7. If you are having tests done, if they can't give you results immediately, **ask when you should expect to get the results**, and ensure you authorize sharing results with other members of your child's care team.
8. Before you leave, **make sure you discussed your top concerns and got answers to your top 3 questions**. If the doctor does not have time to answer them during the visit, ask if a nurse can help, or if you should schedule a follow up visit or phone call.
9. **Doctors prefer summaries over specifics**, if they need more details they will ask, so having your information organized can help make answering those questions easier. Consider putting everything into a binder and group visit summaries, test results, treatment summary and medication history in separate sections. If possible, keep in chronological order, with the most resent information first.

Child Neurologist New Visit Tool-Kit

Having trouble getting organized for your first visit to the neurologist? Child Neurology Foundation shares this printable form to take to your next appointment. Keep a paper or an electronic copy for yourself.

Download PDF

Adaptive Equipment Resource

Bathing your child can be a challenge. Episodes may start before, during, or after. Here are some helpful equipment options for those days!

ETAC DME

Adaptive Mall DME

Foundation Announcements

Seal of Transparency Award

We recently earned a Candid Silver Seal of Transparency! Check out our nonprofit profile to keep up to date with our impact. Keep this link handy; we want to make sure you always have the information you need to support our work with trust and confidence.

See Our Profile!

Silver
Transparency
2024

Candid.

New Year Brings New Officers and Directors

Thank you, Josh!

At the beginning of February, Josh Marszalek stepped down as president of the foundation. Josh left having contributed to the foundation in countless ways. His tenure will long be remembered for leading a re-branding initiative and total overall of the AHCF website. The board wishes to thank Josh for his dedication to the foundation, his passion for the community and his generosity of heart.

Vicky Platt - President

As the vice president of research, Vicky steps into the role with extensive AHCF experience. In addition to serving as the secretary, finance chair, and v.p. of research, Vicky helped raise money for the foundation as a co-host of the Chicago Walk for over 15 years. As the parent of a 22-year old AHC daughter (Emma), Vicky is eager to, "apply my knowledge and experience of AHC in a new and exciting role." Please join us in welcoming Vicky to this new role.

Heather Gates - Vice President of Education & Family Support

The board is honored to announce that Heather Gates was elected Vice President of Education and Family Support in January. Heather's experience and caring heart is an amazing fit for this new role. Heather's creativity, having started the Valentine's Day AHC Exchange, and compassion for all AHC families will be an added benefit to the entire AHC community. Heather is already busy coordinating amazing programs this year and off to a record start. Congrats Heather.

Cate Cohen - Secretary & Communication Chair

Cate has served the foundation for many years as a director. Last month, Cate was elected as secretary of the foundation as well as our communication chair. Cate's experience as site chair for three family meetings not only sets her up for success as secretary, it highlights her generosity of spirit and willingness to step up. Cate's professional background in communication will be a wonderful addition to the foundation. Congrats on the election Cate.

Lynn Egan - Director

With the greatest of fanfare, we are thrilled to announce the return of Lynn Egan to the AHCF Board of Directors. Lynn's contributions to the AHC community are unmatched. To know Lynn is to be impressed by her compassion for AHC families and wowed by her knowledge of AHC. When Lynn stepped off the board a few years back, she continued to serve the community on the Family Support Committee and in planning the 2023 AHCF Family Meeting. Her knowledge and insight of all things AHC are a welcome addition(again) to the AHC community. Welcome Back Lynn!

Kathy Sharo - Director

Kathy has volunteered with the foundation for many years. Initially, helping with fundraising efforts at the Chicago Walk and more recently, helping to jump start the website reinvention. Kathy explains her interest in serving the AHC community as two-fold. "Kiley (her niece), of course, has my heart and I'm eager to contribute to support her, families needing support, and finding a cure. The other driver for me is giving back by offering my business and marketing experience as a VP of Marketing." Kathy's professional experience and AHC understanding is a huge benefit to the AHC community. Welcome Kathy.

Put in your calendars!
Announcing Family Meeting 2025

The AHC Foundation is excited to announce our next Family Meeting! With the ever-changing dynamics taking place in our AHC community, we wanted to shift focus on what really matters. We always say, "the entire family has AHC". It's true. That said, what often gets missed is how incredible our AHC Heroes and their families, truly are. Over the years, we have spoken of the hope we have for their futures, the joy we experience as our AHC kids accomplish more than we thought possible and yes, their never-ending strength and resilience in the face of so many obstacles. More than anything else, we wanted the message of our next meeting to be a positive one, reflecting the incredible families, the kids and adults living with AHC and all they CAN do. Without further ado…

"A One in a Million Life! Hope- Joy- Resilience" is the theme of our next Family Meeting, which will take place **June 20th- June 22nd in Cincinnati, Ohio**. Please save the date as this is certain to be our best Family Meeting yet!

ahcf | alternating hemiplegia of childhood foundation