



10 years
2009 - 2019 | **Hope**
Hypothalamic
Hamartomas
Giving Hope. Getting Answers.

IMPACT REPORT
10TH ANNIVERSARY (2019)

HOPE is wishing something would happen.
FAITH is believing something will happen.
COURAGE is making something happen.

OUR MISSION

Hope for Hypothalamic Hamartomas (hopeforhh.org) provides information, support and community to HH patients, caregivers and healthcare providers. We promote research into early detection, improved treatments, living with HH as a complex medical syndrome and a cure.

WELCOME

Founded in November 2009, Hope for Hypothalamic Hamartomas (www.hopeforhh.org) recently celebrated **10 years of service** to HH patient and professional communities. Hope for HH's creation as a 501c3 non-profit organization stemmed from each funding Board member's struggle to obtain timely diagnosis, medically accurate information and access to expert advice concerning prognosis, treatment and comorbidities. Bound by this rare brain tumor and complex epilepsy syndrome, the founding volunteers were determined to ensure that other families, newly diagnosed or managing HH over their lifetime would have a trustworthy, safe and welcoming place to turn for information and support.

In the space of a decade considerable progress has been made; the landscape for those now diagnosed is significantly more positive, with increased knowledge among medical experts, a greater number of expert centers experienced in and able to treat and manage HH throughout the world, and more – and more refined – surgical options. Yet the need for Hope for HH remains. Individual diagnoses are still delayed, or misdiagnosed. Best practice in identifying and treating HH varies widely from country to country, and from hospital to hospital.

While the range of surgical options has increased, along with the number of centers and surgeons able to perform these complex operations, not all treatment options are universally available, not all patients are good candidates for the available options, and research which could determine the long-term efficacy of the different surgical interventions is largely non-existent. We still do not understand what causes HH, how the various symptoms inter-relate with the HH itself, or how to prevent it.

We **appreciate all the patients and families** who make up our community and who have shared their journeys which are often heart-breaking, always inspiring, and which drive us on to do more. We are **indebted to the medical professionals** who dedicate their practice to caring for HH patients and who volunteer their time, skill and resources to improving understanding, treatment and symptom management. We admire and thank the internationally dispersed experts from many institutions who have made understanding HH a priority, and who provide a model for collaboration and partnership across systems and jurisdictions. The following pages highlight accomplishments as testament to what can be achieved by a small but highly-motivated group of skilled, passionate and thoughtful partners.

As we look to the future, we hope that understanding Hope for HH's mission, history, accomplishments and goals will **inspire and empower new members to get involved**. Sustaining this organisation – this valuable community resource – requires more enthusiastic volunteers and more dedicated advocates.

We hope you will **help us commemorate Hope for HH's ten-year anniversary** by contributing however you can – involving your family, your friends, your community and your professional networks. Whether you support other families touched by HH, attend a family or professional conference, organise a fundraiser, make a financial donation, volunteer your time and talent, or participate in a survey, your participation is key to our continued success.

Thank you for putting the **HOPE** in Hope for HH!
The Board of Directors

WHAT IS HYPOTHALAMIC HAMARTOMA (HH)?¹

A hypothalamic hamartoma is a tumor-like, abnormal mass of tissue adjacent to the hypothalamus in the brain that develops at the fetal stage and is present at birth. It is a non-progressive lesion – it does not expand, spread or metastasize to other locations. It grows proportionately to brain growth. The size and form of an HH can vary greatly from patient to patient as can the type and severity of its symptoms. In the majority of cases symptoms are apparent during early childhood. There are two recognised clinical phenotypes of HH: (i) central precocious puberty, and (ii) epilepsy and related neurobehavioral symptoms including developmental delay, cognitive impairment, and impulsive outbursts often described as rage behaviours. Approximately 40% of patients with epilepsy also have central precocious puberty.

HH IS RARE

It is currently estimated that symptomatic HH occurs in 1 in 200,000 children. This estimate is based on a prevalence study carried out in Sweden in 2004. In the intervening 15 years MRI imaging has improved and medical awareness of HH has broadened. Therefore, we suspect that symptomatic HH, while still rare, may be more common than is currently believed. A new prevalence study has been commissioned in the UK, managed by the Chair of Hope for HH's Medical Advisory Board Professor Helen Cross. For each of the next three years every paediatrician in the UK will be sent information about HH and will be required to identify to Professor Cross any HH patients they may have. Since there is no known geographical concentration of HH cases and all ethnic groups are believed to carry the same risk, the UK prevalence study should provide an up-to-date figure for HH occurrence within the population.

THE CAUSE REMAINS UNKNOWN

The underlying cause of HH remains unknown. A review of medical literature suggests that over 95% of cases are sporadic, with no prior known family history and where the identified patient remains the only affected individual within the family. A defect in factors that regulate fetal development of the hypothalamus is thought to be the most likely cause. Less than 5% of cases are attributed to Pallister-Hall syndrome, a genetic abnormality that can also cause extra-fingers and toes, bifid epiglottis, imperforate anus and kidney abnormalities. A research project initiated through Hope for HH is currently examining HH tissue for genetic abnormalities. Early results are promising and suggest that a genetic cause of HH may be present in a significantly higher percentage of patients than current thinking supposes.

SEIZURES & OTHER CO-MORBIDITIES

Gelastic seizures are the hallmark of HH. Superficially they resemble laughter, but often appear subtly different from true laughter such that family members may distinguish between the two. They can be quite subtle and, particularly during infancy, may be mistaken for other conditions including colic and reflux. They can vary in frequency, with the most severely afflicted patients suffering many gelastic seizures per day and even per hour. It has been reported that approximately 75% of those with gelastic seizures in early childhood will go on to exhibit other seizure forms, focal and generalised, most commonly between 4 and 7 years of age. Cognitive deficits are evident in the majority of patients with HH and epilepsy, most commonly difficulties with processing speed and short-term memory. 50% have intellectual disability (full scale IQ of less than 70). A further characteristic symptom of epileptic HH patients is 'rage behaviour' which may be associated with poor frustration tolerance or lack of impulse control. A working group of expert neuropsychiatrists and neuropsychologists commissioned by Hope for HH are conducting a research study into this behaviour with a view to better understanding why it occurs and how it might be managed. Psychiatric symptoms are reported in more than 80% of children and adults with HH and epilepsy. There are a wide range of diagnoses including oppositional defiant disorder (83%), ADD/ADHD (75%) and mood disorders (17%).

DIAGNOSIS & TREATMENT REQUIRES AN EXPERIENCED MULTI-DISCIPLINARY TEAM

MRI using the correct sequencing protocol is the single most important diagnostic test. Experience has taught us that the experience and eyes of the radiologist are as important as the application of the correct MRI protocol: many HHs are shown on MRI scan images but are missed at first instance, later being identified by medical teams with more HH experience.

HH is sometimes wrongly ruled out diagnostically upon review of normal EEG and VEEG results. However, EEG and VEEG results are normal in more than a third of patients with gelastic epilepsy, likely because the seizures arise in the hamartoma deep in the base of the brain, distant from EEG electrodes on the scalp.

Once diagnosis has been confirmed, baseline neuropsychological and baseline endocrine evaluations should be carried out. These are critical to define the pattern of functioning, to monitor any suspected decline in functioning and can help to measure surgical and pharmaceutical benefits when repeated over the medium and long term.

Gelastic seizures do not usually respond to AEDs. Our current understanding of epilepsy caused by HH argues for early surgical intervention if the patient is a good candidate for surgery. It appears that some patients undergo secondary epileptogenesis, a process in which uncontrollable seizures from the original location provoke seizures arising from a second location elsewhere in the brain. Where this has occurred, surgery removing or disconnecting the HH may be less successful in stopping or controlling seizures.

In the 21st century different surgical interventions have been developed and refined to the point that they are recognised as effective and safe, but not without risk. Each patient is different and the appropriate surgical approach for one will not be the same as for another. Each patient's clinical course and symptoms must be minutely assessed, as must the precise anatomy and location of their HH. Consequently, consultation at a referral centre that specialises in the treatment of HH is highly recommended. Doctors from Barrow Neurological Institute and who serve on Hope for HH Medical Advisory Board developed an 'HH Treatment Algorithm' which sets out a clear pathway for the diagnosing neurologist to approach management and treatment according to best practice. It has been approved and adopted by the other Medical Advisory Board members and their expert centers, and is available on our website. Efficacy of surgery has proved variable. While some patients have been cured of seizures and co-morbidities, others may experience seizure cessation or reduction but experience debilitating surgical side-effects. In many cases seizure freedom or significant reduction seems to have been achieved, only for the seizures to return months or even years later. Moreover, not all HH patients are good candidates for surgery.

REMEMBERING THOSE WE HAVE LOST

In the ten years since Hope for HH began we have celebrated many success stories – we are uplifted by the children and adults for whom surgery has provided a cure, or else a demonstrable benefit to their quality of life, and to their family life. Sadly, we also mourn the loss of members of our small community; we remember those who have passed away through SUDEP and due to other causes. The loss of these loved ones and the courage, dignity and strength of their families inspire and motivate us to do more and to do it better.



CELEBRATING TEN YEARS OF GROWTH

Hope for HH incorporated as a nonprofit organization in 2009 and has operated for a decade, run exclusively by a small group of highly dedicated volunteers. Highlights through the years follow.

2009
Hope for HH is founded and incorporates as 501c3 nonprofit organization.

2010
Website and Facebook page are launched. Patient Support begins fielding phone calls, emails and texts from patients and caregivers. Medical Advisory Board launches with inaugural chair, Dr. Kerrigan and Drs. Cross, Harvey, Arzimanoglou, Regis and Rekate. Hope works with MAB to develop international approved definition of HH as a complex syndrome.

2011

Electronic newsletter is launched. Hope joins other epilepsy and rare stakeholder coalitions including Vision 20/20 and National Organization of Rare Diseases (NORD) to raise awareness of HH and rare epilepsies. Hope For HH Board Members testify at the Institute of Medicine (IOM) Epilepsy Across the Spectrum conference. Drs. Rekate and Harvey of Hope's MAB created a podcast and published ten HH articles in *Neurological Focus* a special online supplement of the *Journal of Neurosurgery* edited by Dr. Rekate.

2009

2012
Blog is launched. First Family Forum is organized in Phoenix, AZ. Drs. Curry, Rosenfeld and Fulton join the Medical Advisory Board. The first "M-o-o-ving Through the Mud" fundraiser raises funds and friends.

2013
Dr. Harold Rekate becomes MAB Chair. First research grant is funded. Organized the 2nd International HH Research Symposium in Marseille, France. The first international Family Forum is convened in Marseille.

2014
Joined the Rare Epilepsy Network (REN) along with 10 other rare epilepsy organizations to create a first of its kind rare epilepsy registry funded by a \$1M grant from Patient Centered Outcomes Research Institute (PCORI). Epilepsia journal published Hope for HH - Co-Founder Stories. Funded a second research grantee. An international multi-institutional genetic sequencing project is seeded.

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2015
Dr. Helen Cross becomes MAB Chair. 3rd Family Forum is organized in Houston, TX. Hope for HH-UK established as a charitable trust in the UK. Hope Board Member invited to speak at the twentieth European Gamma Knife Society Annual Congress in France.

2016
Website is relaunched as well as Instagram and Twitter channels. Organized the 3rd International HH Research Symposium in London, UK. Increased participation in national advocacy campaigns including National Epilepsy Awareness Month.

2017
Drs. Gaillard, Berkovic, Oatman, Shirozu, and Wujin Hope's MAB. Award 2 research grants. Organized 4th Family Forum in Washington, DC. Launched Surgical Comfort Care Bag programs. Organized a Facebook Live and Webinar. Special supplement on HH published in *Epilepsia* comprising eleven peer-reviewed articles following 2013 and 2016 symposia. International multi-institutional Comparative Surgery pilot research study is launched. Hope Board Member invited to join UK National Policy Group addressing NHS funding of laser thermoablation for HH.

2018
Launched volunteer program and onboarded new volunteers. Produced educational brochures on Myths and Facts, Caregivers Guide, and Understanding HH. Grantees complete grants and report out findings. FMRI study published in *Epilepsia*. Personalized Facebook fundraising campaigns take off. Year-round awareness campaigns across social media channels flourished. Hope for HH-UK joined EpicARE for European advocacy and collaboration.

2019

Celebrated 10 Year Anniversary. Launched the HH Comorbidity patient/caregiver survey gathering insights from over 250 families about their HH side effects. Expanded outreach to Russia and surrounds through International Outreach Committee and volunteer Roza Wu. Awarded our first Rosenfeld Rekate Pioneer Awards to Dr. Jeffrey Rosenfeld, Dr. Harold Rekate and HH co-founder and advocate Lisa Soeby. Relaunched HH Surgical Comfort Care Bags with distribution at 2 hospitals.

Delivered information packets to all HH US families. Added 5 new MAB members including Drs. Boer, Boerwinkle, Hildebrand, Jacobs-Levan and Bonhage. Organized 4th International Symposium for HH in Washington, DC. Hope Board member spoke by invitation at an international medical symposium held in Marseille, France.

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Until then, the organization works to realize five goals:

- Provide **INFORMATION** to HH patients, caregivers, and healthcare providers
- **SUPPORT** HH patients and caregivers
- Promote **RESEARCH** toward early detection, improved treatments, living with HH and cure
- Increase **AWARENESS** of HH, seizures, co-morbidities and Hope for HH
- Operate **ALL VOLUNTEER ORGANIZATION** (AVO) 501c3 virtually, legally, and efficiently

GOAL #1: PROVIDE INFORMATION TO HH PATIENTS, CAREGIVERS, AND HEALTHCARE PROVIDERS

PROVIDING HIGH QUALITY DIGITAL & PRINT CONTENT. Early on Hope for HH launched digital and print resources designed to keep patients and professionals apprised of HH treatments, news, findings and discoveries, and to build community. The organization also partners with other patient advocacy groups to provide information on hot topics relevant to our families.



- 1500 monthly **Website** visitors
- Weekly **Blogs**
- Daily **Facebook, Twitter, Instagram** posts
- Quarterly **Electronic Newsletter**
- **YouTube** videos on demand
- **Facebook Lives & Webinars** available across time zones and geographic boundaries
- **Multi-lingual Educational Brochures:** Myths and Facts, Caregivers Guide, and Understanding HH
- **Print Guides:** Questions to Ask Neurologists, Neurosurgeons, About Gamma Knife Surgery, About Laser Surgery



STAFFING PATIENT SUPPORT.

Dedicated volunteers provide one-on-one support via phone, email, text, Facebook posts and messenger to newly diagnosed patients and their families, as well as to families confronting new issues as patients mature. Our volunteers help answer questions, suggest referrals and resources, and lend a listening ear to patients and their caregivers around the world.



GOAL #2: SUPPORT HH PATIENTS AND CAREGIVERS

BUILDING COMMUNITY. Most HH families never meet another HH family face-to-face given how rare the disease is and how physically dispersed families touched by it are. To build community, Hope for HH launched a Family Forum program, planning long weekends filled with information, education, and bonding. Bringing together HH families has created life-long friendships and support networks among people that truly understand the HH journey.



4 Family Forums
Phoenix, AZ (2012)
Marseille, FR (2013)
Houston, TX (2015)
Washington, DC (2017)

Another Family Forum Coming 2021!



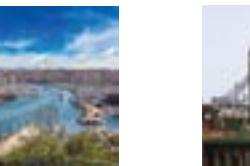
GOAL #3: PROMOTE RESEARCH TOWARD EARLY DETECTION, IMPROVED TREATMENTS, LIVING WITH HH AND CURE

CONVENING MEDICAL ADVISORY BOARD. Hope for HH is guided by a Medical Advisory Board comprising internationally-renowned experts and thought-leaders in their fields and in the fields of epilepsy and HH. All medical content on our website is overseen and approved by MAB members, who also direct our research strategies and programs. Neurologist and epileptologist Dr. John (Jack) Kerrigan of the Barrow Neurological Institute was Hope for HH's inaugural MAB Chairman, followed by neurosurgeon Dr. Harold Rekate, also of BNI, and then Dr. Helen Cross OBE, Prince of Wales Chair of Childhood Epilepsy at NICH and UCL London. Under Dr. Cross' continued leadership the MAB spearheads initiatives to adopt a worldwide consistent definition of HH and its recognition as a complex epilepsy syndrome, to create a paradigm for best practice in diagnosis and treatment, and through international cooperation and exchange to maximise research opportunities and programs.

FUNDING & SEEDING HH RESEARCH. Hope for HH has funded grants to support researchers, lab equipment, International Symposiums, and scientific conferences. Recent grants focused on functional Magnetic Resonance Imaging (fMRI) for surgical planning and the development of an animal model for HH. Hope for HH is currently working with 16 international institutes on a comparative effective pilot to evaluate the efficacy of different types of surgery on seizures and other co-morbidities (side effects) in both the short and long term. Hope for HH also partners with American Epilepsy Society (AES) to identify innovative young scientific investigators and new proposals to advance our understanding of HH. Hope for HH has helped seed partnerships with Duke University, Barrow Neurological Institute and University of Melbourne on genetic sequencing.



CONNECTING RESEARCHERS & CLINICIANS ACROSS BORDERS & DISCIPLINES. Every three years Hope co-organizes an International Research Symposium with a local host to gather the world's foremost HH neurosurgeons, neurologists, epileptologists, geneticists, psychiatrists, radiologists, neuropsychologists, endocrinologists and basic scientists. Each meeting focuses on a different theme. However, they are all highly praised for the opportunity to connect HH researchers and clinicians to exchange information and transfer knowledge. Symposia have increased our understanding of HH, disseminated best practices across borders, and helped identify new research challenges and opportunities.



Marseille France
(2013)



London, UK
(2016)

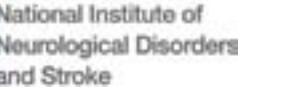
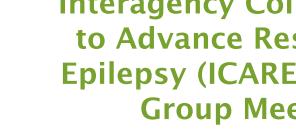
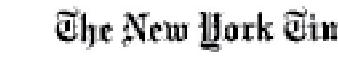
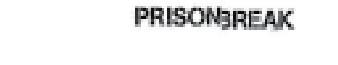


Washington, DC
(2019)

LEVERAGING PARTNERSHIPS TO INCREASE HH UNDERSTANDING. Hope for HH joined the Epilepsy Organization and 30+ other rare epilepsy organizations in receipt of a \$1M Patient-Centered Outcomes Research (PCORI) grant to build a rare epilepsy registry. Hope recruited nearly 100 HH participants and gained valuable insights about the prevalence of HH comorbidities, the usage of anti-epilepsy medications, and similarities HH shares with other rare epilepsies including a proclivity for rage and other behaviors.

GOAL #4: INCREASE AWARENESS OF HH, SEIZURES, CO-MORBIDITIES AND HOPE FOR HH

As a rare epilepsy, Hope relies on partnerships to increase awareness and improve policies for our families. Hope volunteers assume leadership roles in coalitions, partner with key stakeholders on policy, testify at key conferences, and promote HH awareness in popular media.

Leadership Roles Worldwide	Partnerships with Key Epilepsy Stakeholders	Testimony & Presentations at Key Meetings	Stories Shared on Prime Time TV & Cable
Curing Epilepsy 2020	          	     	      

GOAL #5: OPERATE ALL VOLUNTEER ORGANIZATION (AVO) 501C3

Hope for HH is led by a volunteer Board of Directors comprised of five parents of HH sufferers. It is guided by a Medical Advisory Board comprised of 15 internationally recognized clinicians and researchers representing multiple institutions from around the world and disciplines in neurosurgery, neurology, genetics, neuropsychology, endocrinology and basic science. Efforts are supported by the generous donation of time and talent from exceptionally committed volunteers. Many caregivers and professionals have walked along side us over the past decade and we appreciate their contributions. Hope for HH operates virtually without incurring the infrastructure costs of bricks and mortar. Funding raised supports information, education, and research.

BOARD OF DIRECTORS



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President, Co-Founder
Director of Information



Lisa Soeby
Vice-President, Co-Founder
Director of Operations



Kimberly Ranson
Treasurer



Emma Nott
Secretary, Hope For HH UK Affiliate Advisor
Director of Research and Awareness



Kathy Jenson
Director of Support



MEDICAL ADVISORY BOARD (MAB)



J. Helen Cross, MD
(MAB Board Chair, 2015-Present)
Great Ormond Street Hospital (UK)



Daniel Curry, MD
Texas Children's Hospital (US)



Oliver Oitmar, DO
Phoenix Children's Hospital (US)



Alexis Arzimanoglou, MD
University Hospital, Lyon (FR)



William Davis Gaillard, MD
Children's National Hospital (US)



Jean Regis, MD
Timone University Hospital (FR)



Sam Berkovic, MD
University of Melbourne (AUS)



Michael Hildebrand, Ph.D.
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Andreas Schulze-Bonhag, MD
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Madison M. Berl, PhD, ABPP
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Julia Jacobs-Levan, MD
Alberta Children's Hospital (CAN)



Hiroshi Shirouzu, MD, PhD
Nishi-Niigata Chuo National Hospital (JP)



Varina Boerwinkle, MD
Phoenix Children's Hospital (US)



John F. Kerrigan, MD
(MAB Chair, 2009-2012)
Barrow Neurological Institute (BNI)
at Phoenix Children's Hospital (US)



Jie Wu, MD, PhD
Barrow Neurological Institute (US)

EMERITUS

Harold Rekate, MD, (MAB Chair, 2013-2014)
Jeffrey V. Rosenfeld, AM, Monash University (MAB)
A. Simon Harvey, MD, Royal Children's Hospital Melbourne

PRESENT VOLUNTEERS

Bradford Davis, Database Administrator
Mike Conroy, Digital and Print Design
Roza Wu, International Outreach Chair
Angela Donn, Patient Support & Awareness
Traci Flanagan, Patient Support & Awareness
Lauren Mellon, Writer, News Research, & Resources
Lawrence Prosen & Kilpatrick Townsend, Pro Bono Legal Advice
Susanna Kartye, Surgical Support Bags

AFFILIATE-UK

Emma Nott
Carrie Fletcher
Sue Jackson

PAST VOLUNTEERS

Margorie Busby (Volunteer)
Dawn Cerven (Board of Directors)
John Fulton, PhD, University of Utah
Maya Ledis, MD, NIH
Ilene Miller (Board of Directors)
Julie Robinson (Volunteer)
Wendi Tipp (Board of Directors)
Diane West (Volunteer)

IT TAKES A GLOBAL COMMUNITY RAISING FRIENDS & FUNDS

Hope's programs and services are ambitious and require both friends and fundraising. To that end, Hope has partnered with an affiliate in the United Kingdom. The UK Affiliate, founded by patients and caregivers, has spearheaded professional education and outreach, research Symposium planning, and advocacy for access to laser ablation and other novel interventions. Hope for HH actively collaborates with groups of HH patients on social media including HH Survivors Facebook, L'ESPOIR: Hamartome Hypothalamic Facebook (France), and Russian online community. Hope seeks to expand its international imprint in the years to come.

Hope income comes from our generous volunteers, donors, sponsors, and in-kind supporters. A special thanks to Wendi Tipps and the Mudrun Committee who launched and organized "M-o-o-ving Thru the Mud with Landon". Their dedication and success inspired other walks and runs including Walk for Colby and OrthoBethesda Chevy Chase (OBCC) Great Strides 5K. Facebook fundraising campaigns in memory of individuals we have lost and in honor of birthdays, anniversaries and other milestones provide critical support to continue the work of the organization.



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HOPE