CURRENT EVENTS

See what is happening in the SynGap World!

4TH ANNUAL SYNGAP1 GOLF CLASSIC
APRIL 2, 2020
THE GOLF CLUB AT CINCO RANCH
KATY, TEXAS

Sponsors and Players Needed!!!
Click on picture for more information.

WWW.BRIDGESYNGAP.ORG
We need your help to DOUBLE OUR MATCH!! Please help us by creating your own fundraising platform for FREE to help us DOUBLE OUR 10K match!!

Which we are $2000 away from our original 10K match!!

$25K will be going to help 25 families come to our International Conference to attend our FDA PFDD meeting!

Help us get our families in front of the U.S. Food and Drug Administration

We now can DOUBLE THAT on #GivingTuesday Our Giving platform “The Mighty Cause” is helping!

#GivingTuesday is one of the biggest days of the year for nonprofits, and we're using the day to invest in the nonprofits on our platform! We're offering $2,500 in prize grants and $9,500 in advanced subscriptions to help set winning nonprofits up for fundraising success through the end of the year and beyond.
Scientific Family Registry Charter Committee

TAKING APPLICATIONS NOW FOR 2020 - 2023

Objective:
To engage SYNGAPI community members that have a scientific, or analytic professional, background by offering the opportunity to be the voice of the family community in regard to registry surveys, informed consent, Internal Review Board (IRB), Patient Engagement feedback, and direction in research projects via service on the SYNGAPI (MRDS) Registry and Natural History Study Charter.

Co-Chairs: Joshua Neufleder & Victoria Buchanan

To Request an Application Email
admin@bridgesyngap.org

Aims:
1. Serve as a go-between to scientists, researchers, clinicians, and industry looking for work within families.
2. Create literature and outreach programs to engage the SYNGAPI community in current and forthcoming clinical trials and research projects
3. Provide technical expertise or advice in specific areas such as qualitative and statistical/quantitative analysis, clinical design, drug regulation, industry compliance, scientific research, and professional medical insight.
4. Provide an independent sounding board for the board of directors.
5. Serve as an advocate for the organization's mission and vision.

Qualifications/Obligations:
*Must be a parent, caregiver, or family-member of a child diagnosed with SYNGAPI
*Must be an official member of Bridge the Gap (BTG) Family Directory and support BTG's vision and mission.
*Must have a bachelor's degree, or higher, in a statistical or other science-related field such as applied mathematics, operations research, sociology, psychology, biology, or commensurate experience in the areas of qualitative analysis, experimental/clinical design, drug regulation, industry compliance, scientific research, or the medical field.
*Commitment of a 3-year term to serve the Scientific Registry Charter Committee through attendance at committee meetings, response to email correspondence in a timely manner, and support of the research network. Term may be extended with a formal nomination.
*Report to Chief Scientific Officer/CEO or Registry Charter Chairman/Co with questions or feedback.
*Attend the International SYNGAPI Conference (highly encouraged). (Organization will provide travel stipend)
*Attend 3/4 of all annual called meetings which are typically quarterly, evening meetings.
SYNGAP1 SCIENCE FRONTIER

Latest New on SYNGAP1 Research for Families

The quarterly SYNGAP1 Science Frontier brings you close to the frontiers of breakthrough discoveries in the field of SYNGAP1. Each article features a single study and summarizes its highlights in an easy-to-understand language. Spend 3 minutes to keep yourself informed of the exciting research on SYNGAP1!

About the Author

Shaowen (Sarah) Ju is a researcher of SynGAP at the Huganir Lab of the Johns Hopkins University School of Medicine. In the laboratory, she works on developing treatments for SYNGAP1-related disorders by targeting SYNGAP2, a natural antisense transcript of SYNGAP1 found in human. Shaowen also sees SYNGAP1 patients by shadowing Dr. Constance Smith-Hicks at the Kennedy Krieger Institute. Seeing how keen families are to learn about SYNGAP1, she volunteered to write articles for Bridge the Gap ERF to keep families informed of ongoing research in the field.

Recently, researchers published the first longitudinal study that characterized the range of clinical features associated with the SYNGAP1-related disorder. The study also suggested the possibility of using posterior dominant rhythm on electroencephalogram (EEG) as a potential biomarker to assess disease progression and prognosis. Click here to read a quick summary of the study’s highlights and find out the latest discoveries about SYNGAP1!
Highlights from a Retrospective, Longitudinal Study by Jimenez-Gomez, et al. on Clinical Features of SYNGAP1 and Potential Disease Biomarker

Shaowen Ju

Recently, the first longitudinal study on SYNGAP1-related disorder was published in *Journal of Neurodevelopmental Disorders*. The study characterized the clinical features of 15 patients—eight were male and seven were female. On average, individuals were diagnosed at 65.9 months of age (range 28–174 months) and data were collected over a 6-year period, including:

- Genetic diagnosis
- Clinical history and examinations
- EEG
- MRI
- Behavioral assessments (e.g. Gross motor, fine motor, language skills)

**EEG and Epilepsy**

Epilepsy was diagnosed in all fifteen study subjects and fourteen showed epileptiform discharges on EEG. Generalized discharges were more common than focal and 87% of individuals had slow or absent posterior dominant rhythm (PDR), an EEG pattern that develops between ages 2 months and 3 years and achieves the alpha range (8–12 Hz) by 4–5 years of age. PDR is a feature of normal brain development in children. Thus, the abnormal PDR observed in SYNGAP1 patients suggests possible correlations with abnormal development.

Researchers also examined the seizure semiology—seizure signs and other clinical manifestations—in the patient cohort and found atypical absence to be the most common form. Other types of seizure were also observed, including absence, generalized tonic-clonic, generalized atonic, and focal (Figure
Developmental Progression

To characterize the progression of developmental skills, researchers analyzed results from behavioral assessments of gross motor, visual-perceptive/final motor skill and language development. Because of the retrospective nature of the study, not all fifteen subjects had the appropriate assessments. Among the three developmental domains, language skills were the most impaired and gross motor function was the least affected.

Study subjects were also evaluated for key developmental milestones. On average, patients said their first word at 32 months of age (range 10-95 months), first sat unaided at 10 months of age (range 5-15 months), walked independently at 20 months of age (range 15-25 months), scribbled spontaneously at 40 months of age (range 25-60 months), and used utensils at 40 months of age (range 18-70 months) (Figure 2). Please note that the numbers are rough estimates derived from the figure.

Other Abnormalities

- 53% of the patient cohort had abnormal MRI's
- 73% were diagnosed with autism spectrum disorder
- 60% exhibited aggressive behavior
- 33% exhibited self-injurious behavior (e.g. biting oneself)
- 33% had disruptive hyperactivity
One of the most important findings of this study is the correlation between neurophysiologic data—PDR frequency—and developmental function—gross motor, visual-perceptive/final motor skill and language development. When researchers plotted age equivalents against PDR frequency, they found moderate correlations, as represented by the R2 values, in the cases of fine motor and language development (Figure 3a-c). Furthermore, such correlation was not observed between PDR frequency and chronological age (Figure 3d). Together, these findings suggest that abnormal PDR development might be associated with impairment in fine motor function and language development in SYNGAP1 patients.

However, we should be mindful that these correlations are not conclusive. The study is limited by several factors, such as the size of the patient cohort and incomplete data due to the retrospective nature of the study, to reach statistical significance. However, it provided insight for future studies on the developmental progression of SYNGAP1-related disorder and identified PDR as a potential biomarker for further testing.

Reference:


Remember to update the Patient Registry! Your input will enable scientists make breakthroughs possible and bring us one step closer to finding a cure!
Please register your SynGapian with our registry for the National Organization for Rare Disorders. Our doctors use this information for their research in our fight for a cure! REMEMBER to update every 6 months in order to provide them with the most accurate information.

Click on Picture for Link

Together We Can Help

Help the future of SynGap1 by registering your child in the SynGap1 Patient Registry.

https://syngap1registry.iamrare.org

HELP US SPREAD AWARENESS ABOUT SYNGAP1

STRAIT FROM A CREDIBLE SOURCE, OUR RESEARCHERS!

WE ENCOURAGE YOU TO SHARE OUR NEW AWARENESS VIDEO IN YOUR SOCIAL MEDIA CHANNEL

https://youtu.be/ZPB9zS23cDk
Meet my simply amazing family! My husband Mark, sons Andrew and Tommy and my daughter Anna and our sweet dog, Bella. Tommy is our SynGAP warrior who was finally diagnosed in October 2017 after a very comprehensive and extensive medical workup. He has a splice mutation at Intron 13, epilepsy, very limited expressive language, has bowel issues, and global cognitive developmental delays that were first noticed at 4 months old. We currently have him in a fabulous autism school program, in-home support, and recently added ABA therapy. Tommy has an infectious smile, loves 'warm fuzzy' attention and his big brother (26) is a hands down favorite and their connection is the sweetest ever. Some of his favorite activities are watching and cheering at high school sporting events, Chuck-E-Cheese, bowling in a special league, and going out on our boat hoping to see "ales", also known as dolphins. Our little Maltese-ShihTzu has the patience of a saint and loves to snuggle up next to Tommy. His other favorite is a collection of lawn chairs and I mean about 30. He enjoys looking at his extensive collection of photos taken over his 19 years on his iPad. He is very social and active in our community in Chesapeake, Virginia. From what I know he is the only diagnosed Syngapian locally. He is full of life and brings our family so much joy, despite the challenges. He has taught us patience, compassion and adaptability. I like to say I live with Superman!
The Bridge the Gap - SYNGAP Education & Research Foundation, in conjunction with the American Brain Coalition (ABC), recently convened a meeting of organizations representing patients, clinicians, researchers and payers interested in use of cannabidiol (CBD) and other cannabis-derived products for therapeutic purposes. Participating organizations included the American Academy of Neurology, American Academy of Child & Adolescent Psychiatry, American Psychiatric Association, American Society of Addiction Medicine, BIO, and a number of ABC’s patient organization members.

As many Bridge the Gap families are interested in trying CBD and cannabis-derived products to address seizures and other serious health issues, we’re forced to fly blindly for lack of critical information concerning efficacy, dosing, safety, and quality (i.e., what’s in the bottle). This is unacceptable and places our children at risk of additional harm. Our community’s needs, like those of other stakeholders we’ve met with, demands a federal regulatory approach that results in FDA-approved medicines prescribed and used within the confines of a patient-clinician relationship.

Our multi-stakeholder discussion identified shared goals and objectives to collectively pursue and advocate for as legislation and regulatory efforts go forward to allow for the optimal exploration of the therapeutic potential of CBD and other cannabis-derived products.
These include:

- Eliminating barriers and incentivizing research
- Encouraging development of scientifically rigorous evidence relevant to specific conditions
- Enabling clinical decisions based on informed discussion between patients, caregivers and clinicians
- Discouraging the current, largely unregulated environment’s reliance on information based on hype, misrepresentation & pseudoscience
- Actively monitoring the safety and quality of CBD products and removing bad actors

We’ve committed to participating in a one-day “CBD Safety, Science and Policy Summit,” for stakeholders and policymakers. The Summit is scheduled for January 15, 2020 in Washington, DC and Bridge the Gap members interested in attending should contact admin@bridgesyngap.org for additional information, and Monica Weldon at monicaw@bridgesyngap.org

**LOOKING TOWARDS 2020**

Bridge the Gap – SYNGAP Education and Research Foundation

**MISSION:**

**AWARENESS:** Raising awareness and education about SYNGAP1

**ADVOCACY:** Supporting and Facilitating SYNGAP1 Patient/Family Advocacy

**ACCELERATE:** Collaborating with Clinicians, Researchers, Community Leaders and Industry to accelerate SYNGAP1 Research for new treatments

**ACTION:** We are taking Action to Bridge the Gap to a Cure for SYNGAP1 Patients and Families

Please join us in our new fundraising initiative “Cure SYNGAP1 2020!”
We have exciting events scheduled for 2020! Each event will assist us in accelerating our goal to effectively collaborate with SYNGAP1 families, clinicians, researchers, and government decision makers, patient advocacy organizations, including community and industry partners.

**BRIDGE THE GAP – SYNGAP ERF 2020 EVENTS**

- **UK Family Meet Up**
  June 2020 (exact dates to be announced)
  Edinburgh, Scotland, UK

- **International SYNGAP1 Conference**
  November 17 – 19, 2020
  NIH/NINDS Neurological Center, Rockville, Maryland

- **SYNGAP1 Patient-Focused Drug Development Meeting - SYNGAP1: Patient Voices**
  November 19, 2020
  NIH/NINDS Neurological Center, Rockville, Maryland

**ONGOING PROGRAMS & NEW LAUNCHES FOR 2020**

- **SYNGAP1 (MRD5) Natural History Study an IAMRARE™ Registry Program powered by NORD**
  The Bridge the Gap – SYNGAP Education and Research Foundation International Registry is endorsed by the US FDA (US Food and Drug Administration) and the National Organization for Rare Disorders (NORD)

- **NEW: BTG – Scientific Family Committee**
  Launching in early 2020!

- **BTG – Corporate Advisory Board**
  Launching in early 2020!