

BRIDGE THE GAP- SYNGAP



Baltimore Family Meetup: November 8-9, 2019

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CURRENT EVENTS

See what is happening in
the SynGap World!



EARLY BIRD SPECIAL!
\$10 OFF for BTG Members
ONLY!
September 1 - October 1
Use Promo Code
SYNGAPBALTIMORE

Hosted By



Kennedy Krieger Institute

BALTIMORE FAMILY MEETUP Conference
November 8-9, 2019

Sponsored By



Members Only
Early Bird Registration Opens
September 1, 2019
Till
October 1, 2019
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MORE CURRENT EVENTS



OUR FAMILY ADVISORY COUNCIL IS HEADING UP A CAMPAIGN THAT WILL RAISE FUNDS FOR FOR OUR NEW RESEARCH INITIATIVE THAT WILL PROVIDE TRAVEL STIPENDS TO PATIENTS WHO PARTICIPATE IN THE VALUABLE CLINICAL RESEARCH BEING CONDUCTED AT OUR COE'S!

WE WOULD LIKE TO SHARE YOUR VIDEO STORIES DURING FALL OF 2019 TO CELEBRATE OUR 5 YEAR ANNIVERSARY!

TOPIC: D-DAY (DIAGNOSIS DAY)

SUBMIT A 1-2 MINUTE VIDEO SHARING THE IMPACT OF YOUR DIAGNOSIS DAY, HOW IT AFFECTED YOU AND YOUR FAMILY AND WHY IT IS IMPORTANT TO SUPPORT RESEARCH TO HELP YOUR LOVED ONE.

WE WILL SHARE THESE VIDEOS THROUGHOUT OUR SOCIAL MEDIA CHANNELS AND TO SHARE IN YOUR OWN NETWORK ON THE FB FUNDRAISING PLATFORM.

WE WILL ALSO BE COMPILING THE VIDEOS TO BE SHOWN AT OUR 5 YEAR ANNIVERSARY CELEBRATION NOVEMBER 9, 2019.

POSTING VIDEOS ONLINE WILL BEGIN SEPTEMBER 15, 2019.



D-Day Campaign Video
Request:

1-2 Minute Video Describing
Your Diagnosis Day

What Were Your Feeling?
Where Did You Go For Help
and Resources?

Videos Due September 15th

VIDEO REQUIREMENTS:

*1-2 Minutes in Length

*LANDSCAPE VIEW

DUE DATE FOR ALL VIDEOS WILL BE **SEPTEMBER 1, 2019.**

TO PARTICIPATE IN THIS PROJECT PLEASE SIGN VIDEO/PHOTO RELEASE PHOTO/VIDEO RELEASE.

SEND RELEASE and VIDEOS EITHER IN ATTACHMENT OR DROPBOX LINK

TO JOJO.D@BRIDGESYNGAP.ORG

FOR QUESTIONS PLEASE EMAIL US AT ADMIN@BRIDGESYNGAP.ORG

Bridge The Gap Sibling Facebook Page:
Siblings over 13 years old can join our sibling support page

Click Picture For Link



PATIENT REGISTRY UPDATE

Don't forget to register your SynGapian in our NORD Patient Registry. Scientist need our help! Did you know that the registry currently has...

- 223 out of 300 plus known cases worldwide are registered
- 728 surveys have been complete...remember to update every 6 months!
- 169 Consented
- 216 Respondents
- 65 Genetic Reports
- 122 Started Surveys
- 10 Updated Surveys
- 34 Drafted Surveys
- 36 Retaken Surveys
- 1,200 Plus Pages of Patient Records



Click On Picture For Registry

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!

This edition features an article on
NORD describing the SYNGAP1
Mutation. Click on picture for link.



NORD[®]
National Organization for Rare Disorders

ABOUT THE AUTHOR

Shaowen (Sarah) Ju is a researcher of SynGAP at the Haganir 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.

There are studies going on that your child may benefit from as well as further research for our scientists. As always, we are asking for SYNGAP1 (MRD5) Registry and Natural History Study. There are other clinical research opportunities for you to join.

[Click here to find out what more you can do!](#)

FAMILY STORY TIME

What is it like to have a child with SYNGAP? While everyone's experience is unique, I can tell you one thing that is the same, it's not easy and pretty much unpredictable. I could have never predicted receiving a call from the school principal that my 7 year old sweet boy is throwing furniture across the room and threatening to blow up the school. I couldn't imagine how painful it could be to watch my child in complete meltdown in the middle of Chucky Cheese while his classmates enjoyed birthday cake and ice cream and parents tried not to stare. I never thought all my vacation days would be spent in hospitals, doctors offices, therapy sessions and IEPs. When I heard the words IQ of 80, Autism, Emotionally disturbed and Intellectual Disability my world felt like it had been turned upside down. It was natural to question why. He hit nearly all of his developmental milestones. He was such a happy healthy boy. Did I do something during my pregnancy to cause this? Was I exposed to something while working as a nurse in the ICU? What about the delivery? Did he have brain damage after the failed induction, vacuum and forceps? Even after the visit with the developmental pediatrician, MRI and blood work we still didn't have a diagnosis, but suspected he was "somewhere on the spectrum". Then came the genetic test results. We were instructed to come back in for parental testing. Could one of us have unknowingly passed on a bad gene? Could our younger son also have the same thing? Finally, we received the results: de novo del 6(p22.1-21.3). All we knew was this was a random mutation on chromosome number 6.

Fast forward to December 11, 2013. Kyle was now in the full throws of puberty, or what I like to call the years of hell. I could not have ever imagined the horror of my 14 year old son jumping out of my vehicle as we went down the road. I certainly never thought we would be pleading with the Harris county DA to drop charges of assault on a police officer while my son was in a complete psychotic episode. We were now faced with new diagnoses: Bipolar, Schizoaffective disorder and Suicidal Ideations. I no longer had to rely on the GPS to guide me to and from the psychiatric hospital, but the drive was often blurred by uncontrollable tears. Our family was in complete crisis mode. We stopped visiting family in Oklahoma and Missouri to save any PTO for the new "expected emergencies". My marriage was on life support. I didn't know if I was going to be able to continue working. I became an expert at completing FMLA paperwork. I learned how to be an advocate for my son at IEP meetings and demanded home based instruction while he was adjusting to new medications and in and out of the hospital. My younger son adapted to the reality that mom and dad may not be able to come to parents night or the spring concert. My mother moved in to help out wherever she could. We started adjusting our flexible spending and HSA account to plan on

hitting the max out of pocket every year just to pay for medications. At one point I started seeing a psychologist and started an antidepressant. Out of desperation, I began searching the internet for anyone who was going through the same struggles we were, and I stumbled on the Chromosome 6 Facebook group. Just a few days later I received a Facebook message from Monica Weldon and discovered Kyle had the SYNGAP mutation just like her son. For the first time since our genetic test 7 years prior, I finally felt like we were not alone.

We now know that most of Kyle's symptoms are directly related to his genetic deletion. After years of therapy, inclusive classrooms, specialists, hospitalizations, too many medication changes and tests to count, we are finally nearing the official end of childhood. In a few short months Kyle will be 18; officially an adult, but we all know that he will require some degree of assistance and chronic medical care for the rest of his life. I never imagined Kyle would be able to attend mainstream high school, make friends, cook breakfast or fill his med box. I definitely was not sure if he would graduate high school...but he surprises us every single day. He is a thoughtful, funny, happy, smart young man. He loves technology and his dream job is to work at Apple. He starts each day by telling everyone "I'm going to have a great day today!" While Kyle has come a long way, he still struggles with anxiety, social settings and various medical problems. He currently takes several medications every day. Without these medications he would likely begin to have mood swings, depression, headaches, anxiety, dizziness and impaired concentration. He may not ever get his drivers license. He has exactly one friend. He still hits himself when he gets agitated, although the frequency of episodes has decreased from daily to maybe once or twice a month. He has to be reminded to tie his shoes, clean his glasses and put the milk back in the fridge. But these are all little things. We celebrate the victories, like going to Worlds of Fun and riding Patriot with a friend, taking his school picture, passing all his classes and even earning some A's. So what is it really like to have a child with SYNGAP? It's a blessing!

Victoria Buchanan, RN, BSN, CCM, MBA

