



THE VOICE

Quarterly Newsletter

Welcome

We want this newsletter to be valuable for you so please, share your feedback and suggestions to help us improve. We hope you don't but at any time you can simply click the unsubscribe link.

Do you know someone who may like to read our newsletter? Please share!

Sincerely,
The NR2F1 Foundation Board



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NR2F1 Foundation Board Members



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Jennifer Higgins
Secretary



Jeff Thelen
Treasurer



Jennifer Ferrebee
Director



Melissa Thelen
Director

Meredith Howell Steps Down as Board V.P.



It is with heavy hearts that we bid farewell to Meredith who Co-Founded and served as Vice President of the Foundation for over 2 years.

In the very beginning of BBSOAS Meredith connected many of us via her blog, Say Hola Lola. Families no longer felt alone after reading about Lola's (and the family's) journey living with BBSOAS. Some even cried in relief; feeling for the first time a connection in the otherwise extremely isolated world of rare NR2F1 mutations. That small group of parents kept growing and long story short, the NR2F1 Foundation was born.

Meredith has been pivotal in getting the Foundation off the ground and running. We will miss her expertise and positive attitude. Luckily she has graciously volunteered to remain a Foundation consultant.

A HUGE thank you to her husband Rob who designed our beautiful [website](#), with Meredith providing the content.

Meredith continues to devote her career to helping other families in Indianapolis as Regional Director of [Visually Impaired Preschool Services \(VIPS\)](#).

Meet the Foundation's newest Board Member

Erin stepped up to join team NR2F1 and we are thrilled to have her on board!



Erin Camper

Director

After a long road trying to find answers to her son Colton's many challenges and diagnoses Erin received a call from a neurologist at the Children's Hospital of Colorado on a cold January day while she was watching her daughter play indoor soccer. The neurologist had been conducting research into genetic causes of a catastrophic type of epilepsy known as Infantile Spasms that Colton had thankfully been successfully treated for at 7 months of age. Colton had a mutation in the NR2F1 gene that explained everything. Full exome sequencing as a part of the research study had finally found the answer, about two years after the initial blood samples had been taken, and almost 5 years into Colton's life. For Erin, the information was a relief, an explanation, and a door into a community of families who were facing the same challenges. For Erin, finding the NR2F1 Foundation and BBSOAS family group were life changing. Meeting other BBSOAS families felt like reconnecting with old friends. When a position on the board opened Erin knew she had to be involved with this small but mighty group who is always working to make life better for families impacted by this rare disease.

Erin has a B.S. in Biology and a B.S. in Nursing and has been a registered nurse for nearly fifteen years. She has worked in the Pediatric ICU, Neonatal ICU, as a school nurse, and at a pediatric urgent care. Currently, she is a home care nurse in addition to caring for Colton and advocating for him every

day to be the very best he can be. Erin also loves to watch Colton's sisters do what they love, play soccer (Addy) and compete at gymnastics (Stella).

Erin lives in Timnath, CO with her husband Matt, her daughters Stella (12.5), Addy (11) and of course Colton (6). Erin tries to squeeze in some reading, breakfast with friends, pedis with her daughters and streaming favorite shows with her husband once Colton is in bed to take care of herself a bit!

SPOTLIGHT

BBSOAS KIDS



Meet Three-Year-Old Emma!

How did you choose the name Emma?

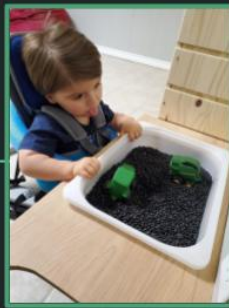
Her first name was chosen before we even started the process to have her. Emma is Diana's grandmother's name and she had always wanted to give that name to her daughter. Luckily for her, I too love the name so that was the easy one. Her middle name, Leimomi, was my grandmother's middle name; it is Hawaiian and has a few different meanings, literal being Strand of Pearls, but it also means Blessed Child. In the Hawaiian culture, the grandparents are responsible for helping chose the Hawaiian name, so my

mom and I had many conversations about which name to go with and since we were going with Diana's grandmother's name, it only made sense to choose my grandmother's name for her middle name.

Three words that describe Emma:

[Read more about Emma](#)

A Day in The Life of Henry



Henry is 2 years old, and has a 5q14.3-5q15 deletion including the NR2F1, MEF2C, and ADGRV1 genes.

12:30 am: Loud stomping and joyful squeals come from Henry's room as he wakes up full of energy. Henry has always loved the early morning hours, and spends this time babbling and scooting around the crib.

2:30 am: Henry's squeals get quieter and further apart.

3:00 am: There is silence in Henry's room. I look at the video baby monitor to confirm that Henry wore himself out with all his kicking and scooting and has fallen back to sleep.

[Read More About Henry](#)

ANNOUNCEMENTS



The 2020 Family and Scientific Conference has been postponed....again. The Foundation has decided, with feedback from the families, that 2022 would be a better time to consider a conference. There are still too many unknowns with the risks still outweighing the benefits.

To be clear, this is a postponement, NOT a cancellation. We WILL have a conference in the future. We are now considering the Spring of 2022 (same location; Orlando, FL) and we look forward to the unity, information and advocacy that it will bring to all BBSOAS families!



In 2018, families who have loved ones with Bosch-Boonstra-Schaaf optic atrophy syndrome (BBSOAS) met in Houston, Texas for the first-ever BBSOAS Family & Scientific Conference. This commemorative video shows just how special it was!



BBSOAS families - What a great Zoom group gathering we had, hosted by board member Jen Ferreebee. How enjoyable it was to share and listen to each other's stories of pre and post diagnosis. What a wonderful and supportive way to celebrate each other and Rare Disease Day!

The next Parent Support Zoom call will be

Sunday, March 28th, 3-4:30p EST

We'll post the link in our private FB page.

Looking forward to it!

P is for Patient Registry

A Patient Registry is an organized system to collect information about a group of patients who share the same disease/condition.

A Patient-Powered Registry (PPR) is when patients & family members "power" the registry by managing the collection of data & controlling the research agenda.

(Patient-Led Research)



The Foundation continues to work hard behind the scenes at developing the first-ever Patient Registry!

Why?

1. Research The collective voice of the patients paves a path showing what is happening to those living with BBSOAS, how many patients there are world-wide, symptoms, medications, the type of mutation....all of this data informs the questions which leads to research which leads to answers which leads to a better life via treatments and a potential cure.

2. Convenience No more carrying around large and bulky binders . All of your and/or your child's health info including docs you can upload like an I.E.P in a comprehensive, portable and multilingual app.

Although we've experienced some set-backs beyond our control, we are determined to get this off the ground and running. Please stay tuned for exciting(!) announcements on our various social media platforms and quarterly newsletter.

THE GIVING VILLAGE

NR2F1 PROJECT PATIENT VOICE

We're raising money for the
NR2F1 Project Patient Voice at
mudlove.com/thegivingvillage!



50% of your purchase goes
toward our goal!

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 **NR2F1**
FOUNDATION

Please help the NR2F1 Foundation in making history with the first-ever BBSOAS Patient Registry!



Buy Now For a Limited Time!

Countdown to Rare Disease Day
with
the ABC's of NR2F1!



Countdown to Rare Disease Day with the ABC's of NR2F1!

— Rare Disease Day 2021! —

Z is for Zebra



The Zebra is the mascot for rare diseases because of their uniqueness and an old story that when you hear hoofbeats, don't assume it's a horse...it could be a Zebra!

This is why we say #showyourstripes! Share this post with a photo of you wearing your stripes today in support of rare diseases!



Our community of friends, family and supporters learned a little about NR2F1 and BBSOAS each day for a month leading up to Rare Disease Day 2021 on February 28th.

Missed it?

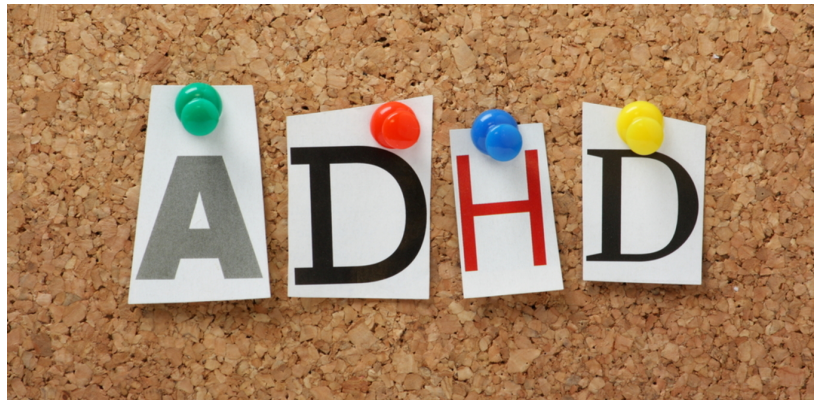
Follow us on social media!



EDUCATIONAL SPOTLIGHT

What is Attention Deficit Hyperactive Disorder (ADHD)?

ADHD is one of the most common neurodevelopmental disorders of childhood, often lasts well into adulthood, and sometimes



is not even diagnosed until adulthood. The disorder is characterized by an inability to focus, acting impulsively, and/or being overly active. While all children may exhibit some of these traits from time to time, when a child or adult has ADHD these traits are more severe and can cause difficulty for the individual at home, at work or school and with friends.

Characteristics of ADHD: (may vary between children and adults)

- Impulsiveness
- Disorganization and problems prioritizing
- Poor time management skills
- Trouble focusing/inattentiveness
- Difficulty with multitasking
- Excessive activity or restlessness
- Poor planning
- Low frustration tolerance
- Frequent mood swings



- Problems following through and completing tasks
- Short tempered/explosive personality
- Trouble coping with stress

Do all people with BBSOAS have ADHD?

ADHD is just one of a handful of diagnoses that can affect behavior in individuals with BBSOAS. According to the research study “Phenotypic expansion of Bosch-Boonstra-Schaaf optic atrophy syndrome and further evidence for genotype phenotype Correlations” (Rech, Et al. 2020) 37/46 individuals were diagnosed with an Autism Spectrum Disorder (ASD) or displayed features of ASD, and 36/46 individuals displayed cognitive/behavioral anomalies such as OCD and ADHD. Furthermore, based on data provided by families, it is estimated that approximately 20% of those with BBSOAS have a concurrent diagnosis of ADHD. It is important to note that behavioral diagnoses such as ASD and ADHD can have overlapping characteristics which can be further confounded by behaviors that can be related to vision impairment including head-banging and the need for movement to help the individual see (a characteristic of Cortical Visual Impairment.) Sensory processing disorders (SPD) and issues with proprioception can also sometimes look like ASD and or ADHD. SPD is frequently misdiagnosed as ADHD. Individuals with BBSOAS are incredibly complex and may present with a myriad of concurrent diagnoses which can make it difficult to determine the exact cause of some of the symptoms. It is critical to work with physicians and other experts to best determine the root cause(s) of various behaviors in order to create the best plan to help the child or adult manage the symptoms in order to be as successful as possible at school (or work), home, and in relationships with family and friends.



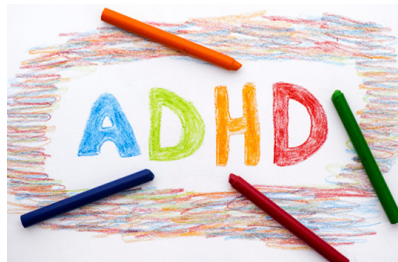
Treatments/Therapies for ADHD:

- Behavior therapy (to include training for parents)
- Medications.

For children under 6 years of age, the American Academy of Pediatrics recommends behavior training for parents. For children over the age of 6 the recommendation is behavior training/therapy combined with medication as well as behavior interventions at school and including the school team in the plan. This type of therapy can be provided by psychologists and behavior experts within school districts. There are other types of therapies which are, at present, not well-researched but can sometimes prove helpful for individuals with ADHD, such as Applied Behavioral Analysis (ABA) which is much more commonly used for children

with Autism.

Medication is also an option, and a very personal decision, that must be thoroughly discussed and decided upon with an individual's medical team. Often, a primary care doctor can prescribe medication, but they may seek input from specialists such as neurologists, psychologists, psychiatrists etc. Some primary care doctors do not have the knowledge or experience to prescribe behavior medications and may refer that decision and management to a psychiatrist. Psychiatrists are physicians that are specifically trained in mental health and are highly knowledgeable regarding medications for behavior and other mental health issues.



There are two main categories of medication for ADHD:

- **Stimulants** are the most widely used and are fast acting.
- **Nonstimulants** are a newer approach, first approved for use in 2003, and are not as fast acting, but may be more appropriate for some children.

Some research indicates that stimulant medications may aggravate seizures or worsen EEG findings in individuals with epilepsy. Other research articles have not noted an increase in seizures, particularly with methylphenidate. Therefore, without clear evidence in favor of stimulants, nonstimulants may be a safer choice for individuals with BBSOAS who have a history of epilepsy. Anyone who falls into this category should involve their neurologist in the decision on which medications are the safest option.

Individuals with BBSOAS face many challenges every day. Some of them are faced with the additional challenges that come with ADHD, but there are strategies to help them and the more we know the more we can help them to be the best they can be!

Resources:

[CHADD - Improving the lives of people affected by ADHD](#)

<https://www.mayoclinic.org/diseases-conditions/adult-adhd>

<https://www.additudemag.com/slideshows/signs-of-sensory-processing-disorder/>

<https://www.cdc.gov/ncbddd/adhd/facts.html>

<https://onlinelibrary.wiley.com/doi/10.1002/ajmg.a.61580>

<https://pubmed.ncbi.nlm.nih.gov/11275457/>

RESEARCH CORNER



PROJECT NR2F1

Project NR2F1 launched this Fall!

It was an inter-university competition for designing the best research roadmap for BBSOAS. We targeted the top 20 neuroscience programs in the country offering a \$2,000 prize for the best proposal.

The students demonstrated a thorough understanding of the existing research into BBSOAS and unique perspectives on the areas of focus with the most potential to result in treatments quickly. The NR2F1 Foundation voted the team from University of Michigan: Luke Man, Kayvon Sanjasaz and Max Wagner as the winners, and they very generously donated the prize money back to the Foundation to pursue future research initiatives!



While the BBSOAS community gained so much from their expertise, the winning team stated that they also gained a better understanding of the challenges faced by the families of an ultra-rare disorder and are grateful for the perspective this will allow for them to have as they pursue their medical careers and research going forward. Equally as exciting is the work happening in Dr. Schaaf's lab at Heidelberg University. Magdalena Laugsch, one of the research investigators actively working with the BBSOAS induced pluripotent stem cells, has been awarded a very generous grant by the DFG (German equivalent of the NIH) for advancing her research.

**A BIG
Thank You
to
MYZEBBZ**



For donating \$890!!

**Thank you to Mandy for your continued support for the
Foundation and for raising BBSOAS awareness!**

Every Zebbz product is made with exquisite fabrics and each unique pattern is named after a person living with BBSOAS, honoring their uniqueness. You'll find bandeaus, pillows, tablecloths, blankets and even phone purses. Zebbz is based in the Netherlands and will ship anywhere in the world.

A generous portion of all sales go to the NR2F1 Foundation.



Euclid Ave. Designs is a husband and wife duo who enjoy working together and making handcrafted wood items for the home and family. They are excited to be supporting the NR2F1 Foundation with **50% of their profits** from the month of **April!**

Be sure to check out their beautiful, custom-made products with several new items launching in April.

Thank you Euclid Ave. Designs!

Selling for a Cause

Do you have a business you would like to share with the BBSOAS community and beyond? Would you like to support the NR2F1 Foundation in empowering those affected by rare Nr2f1 mutations?

Let's Talk!

Send us a [message via Facebook](#) or email [Carlie via our website](#) or email directly at: carlie.monnier@nr2f1.org



Dr. Christian Schaaf presented a 'TED-style' talk in Charlotte, North Carolina at the 2018 annual meeting of the American College of Medical Genetics and Genomics (ACMG), a professional society of biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other health care professionals who are committed to the practice of medical genetics.

Dr. Schaaf's talk titled, "The Power of Patient No. 1" described the puzzling case of Ms. Peggy Fahed. He explained how solving her medical mystery led the physician-scientist to discover a rare new

neurological condition called the Bosch-Boonstra-Schaaf optic atrophy syndrome (BBSOAS).



Help lead the way to answers!

Help fund NR2F1 research and awareness initiatives

What does the future look like for those who have BBSOAS? There is still so much to discover, but families need hope. We need your help to learn more about NR2F1 mutations and how it affects the ones we love. Your tax-deductible donation will support research that could lead to life-enhancing treatments and interventions for those affected by this rare genetic mutation, a biennial family conference and outreach efforts. Please make a donation today!

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