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THE VOICE

Quarterly Newsletter

Welcome To Our First Issue!

We are excited to launch our first newsletter to inform and inspire you on a quarterly basis with all things BBSOAS (Bosch-Boonstra-Schaaf optic atrophy syndrome). Parents, families, friends, doctors, scientists, educators, and supporters interested in learning about the rare world of NR2F1 and BBSOAS--we welcome you.

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 have family, friends or colleagues who may benefit from the information in our newsletter? Please share!

Sincerely,
The NR2F1 Foundation Board

NR2F1 Foundation Board Members

Meet the board members of the NR2F1 Foundation



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SPOTLIGHT

BBSOAS KIDS



Meet Three-Year-Old Aydn!

How did you choose the name Aydn?

We chose the name Aydn due to a bet between my husband and I. We made a deal that if I were to birth a girl, I would name her. If I were to have a boy, he would name the baby. Although I dreamed I would have a daughter and had expectations of the same, he won the bet. Aydn was one of seven names on his list, and one of the few with which we agreed. His name is spelled slightly different than people typically expect. I wanted to spell his name A-y-d-e-n which is Irish and means “little fire”. I think this is fairly accurate given his personality.

Three words that describe Aydn:

He is loving, independent, and resilient.

How and when was Aydn diagnosed with BBSOAS?

Aydn was 2 years old when he was diagnosed with BBSOAS. This diagnosis came after a couple of years of trying to convince various medical professionals that my motherly instincts were telling me something wasn't right with his development. We went through seizures, constipation, failure to thrive, aspiration, a G-tube placement, lots of physical, occupational, and speech therapy as well as a significant trip to ICU. During this trip, Aydn fell into deep sleep, would not wake up and his vitals began to decline. Each time he would go to the hospital, for the most part, his blood work and

vitals were mostly normal. I could not dodge the feeling that there was something significant that wasn't showing up on the typical tests. We spent many nights in the hospital, and he endured innumerable amounts of testing until it was finally recommended that we take a genetics test. After we met with genetics, he was diagnosed with BBSOAS.

Does he have any siblings?

Aydn has a very sweet and helpful older brother named Owen, who is seven years old.

What are Aydn's favorite things to do?

Aydn's three favorite things to do are to listen to Cocomelon videos on Youtube, play with his brother, and to go outside on nice, breezy days to enjoy the sunlight and surroundings.

If eating solid foods, what are his favorite things to eat?

Aydn is currently fed via Gtube, so he receives all of his nutrition via a Kangaroo pump. However, when he was able to eat a little by mouth, he enjoyed sweet things such as popsicles.

What are Aydn's strengths?

One of Aydn's strengths is that he is resilient. He has a high pain tolerance which I am thankful for since he has been poked and prodded more in his "almost" three years of life than I have in my thirty-one years. He also has a sense of humor even while being non-verbal.

What has he taught you?

He has taught me that God has control over His life and not me. I have to release the desire to control every aspect of the present and future and enjoy the moment. He has also taught me to be grateful in ALL things because he has fought through so much just like other children with BBSOAS, yet he rarely complains by the way of crying. Usually if he does show discomfort, he just wants a hug or to be cuddled.

What is the hope you have for Aydn's future?

My hope is that Aydn will be as independent as possible so that both he and his brother can enjoy each other's company and be successful when my husband and I are no longer here. My biggest immediate hopes are that he walks and talks.

What are the most challenging and rewarding aspects of raising a child with BBSOAS?

The most challenging aspects of raising a child with BBSOAS are finding others who are in a similar situation with which to discuss the complications that can arise from the disorder. This is especially the case since this disease is so rare. I was told by the geneticist that we, the parents, are really the experts as we are experiencing the challenges in real time. I am VERY thankful for the BBSOAS parent support group on Facebook and the NR2F1 foundation as both of these have been instrumental with helping me to cope with each new symptom or change in behavior.

The most rewarding is that I have learned how to truly take one day at a time. Although I still have dreams and goals, I know that the best moment is the present, as that is the only moment you are sure that you have. Also, it has been rewarding to be a part of a community of parents from all over the world who offer hope, suggestions, and support when things get rough. Each parent of a child with a rare disease such as BBSOAS are strong warriors as we navigate this battlefield together.

If you could go back in time and give yourself one piece of advice on the day Aydn was diagnosed with BBSOAS, what would it be?

I would say find an outlet to deal with the grief process and use it consistently. Many times people associate grief with death, but I do think there are some stages that we go through when we get a diagnosis. For instance, with Aydn, I always thought he would walk, talk, etc, but as he kept having seizures and had significant developmental delays, I had to come to terms with the fact that I don't know what the future holds for him with such a rare disorder. I didn't know if he would survive to see the age of two, so I had to find a way to deal with those conflicting feelings that I went through daily. My outlet is writing, so I started a blog called epi-mom.com to help guide other parents through this journey. Some of you may paint, or crochet, or hang with friends. Either way, have a plan for dealing with your feelings and emotions through the ebb and flow of the disorder.



Meet Four-Year-Old Ebony!

How did you choose the name Ebony?

We had a list of names that we liked but we couldn't decide so we waited until she was born to see what name suited her character.

Three words that describe Ebony:

Adventurous, curious, cute

How and when was Ebony diagnosed with BBSOAS?

She was diagnosed in late 2018 after a long battle with the doctors. We knew that something was wrong from when she was first born. She was born with severe hip dysplasia, Oromotor dysfunction and hypotonia. Ebony was in a full harness for six months then double nappies for nine months after that. Ebony couldn't breast feed at all so we had to go for bottle fed from birth. She was always very floppy as well. She had her first tonic clonic seizure at twelve months old in which she stopped breathing and I had to perform CPR on her until she started breathing on her own again. It was very scary but I'm glad that I knew how and what to do. We pushed and pushed the doctors and pediatricians both public and private because we new that there was an issue here and we wanted answers so we could give Ebony the best chance of a good life. The doctors finally came up with doing a genetics test. We had to wait for about four months for the results. In late December Ebony had another seizure which resulted in

another hospital visit and overnight stay and that was when we found out the results. We were dealt a double blow that day as we found out that not only Ebony had this syndrome but her mom Kristy also had this syndrome as well for her whole entire life without knowing what it was. We were happy to finally find out what it was because we could finally start giving Ebony the best care but at the same time absolutely terrified because it is such an unknown syndrome with lots of ailments.

Does she have any siblings?

Ebony has an older brother named Robert and he is seven years old. He is the best big brother anyone could ask for. Always protecting and loving his little sister. He understands what she wants better than us sometimes. He has since been tested and the result came back with no deletion or mutations which was good.

What are Ebony's favorite things to do?

Ebony loves to look at picture books and rip the pages out. She loves playing with matchbox cars and she loves playing doctors and most of all she loves to watch her iPad.

If eating solid foods, what are her favorite things to eat?

Ebony loves to eat everything she can find in the fridge and cupboard. She loves fruit the most. We thought we were going to have a real issue with her having oromotor dysfunction (the inability to swallow properly and chew for long periods of time), but she has proven us wrong. Now we have to do our best to stop her from eating too much as she will graze all day if you let her. When she gets sick is when we worry. If Ebony is too tired due to being sick she won't eat at all.

What are Ebony's strengths?

She is headstrong, kind, loving and cute.

What has she taught you?

Ebony has taught us patience as we always need to be with her all the times. She has taught us to be alert at all times, as at any point in time she could be getting into something she shouldn't be into or doing something dangerous because she has no understanding of fear. Above all Ebony has taught us to love and accept people more from all walks of life.

What are the most challenging and rewarding aspects of raising a child with BBSOAS?

The most challenging aspects of looking after a child with BBSOAS is that you have to watch them 24/7 due to them having so many issues. For example with Ebony she has learned to open doors now so if you leave the door unlocked and you take your eye off her for one second, she will be out the door and down the road before you know it and she won't come back if you call. If you forget to lock the cupboard Ebony will get into whatever is in the cupboard and that could be dangerous. When we are out for a walk we have to hold on to her hand because if we don't she will run out onto the road and she won't stop if you scream for her. She can and does have seizures at any time day or night without notice. She has a habit of licking everything no matter what it is and when she hurts herself, if you don't see her do it you don't know that she has hurt herself until you see the blood, scratch, bruise burn or cut as she doesn't scream or cry when it happens. It is rewarding when Ebony just surprises you by saying something that she has never been able to say before or doing something that she has never been able to do before right out of the blue. The inchstone accomplishments that pop out of nowhere. It is the most rewarding when Ebony acknowledges us by giving us a hug or a kiss and saying "Love you". That is a rare occurrence.

If you could go back in time and give yourself one piece of advice on the day Ebony was diagnosed with BBSOAS, what would it be?

The only advice I could give myself if I could go back to the day that we got the results about Ebony and Kristy would be to not give up the fight. There are kind people out there that are willing to help but they won't come to you. You have to go out and find them.

What is the hope you have for Ebony's future?

We hope that Ebony has a happy and healthy future with lots of love from all the people that she meets along the way and we will always be there for her just like everyone does for their children. We know she has a hard and bumpy road ahead. That's why it is so important to spread awareness about BBSOAS. The more people know and understand about BBSOAS and the people that have been diagnosed with this syndrome the better it will be for everyone.

ANNOUNCEMENTS

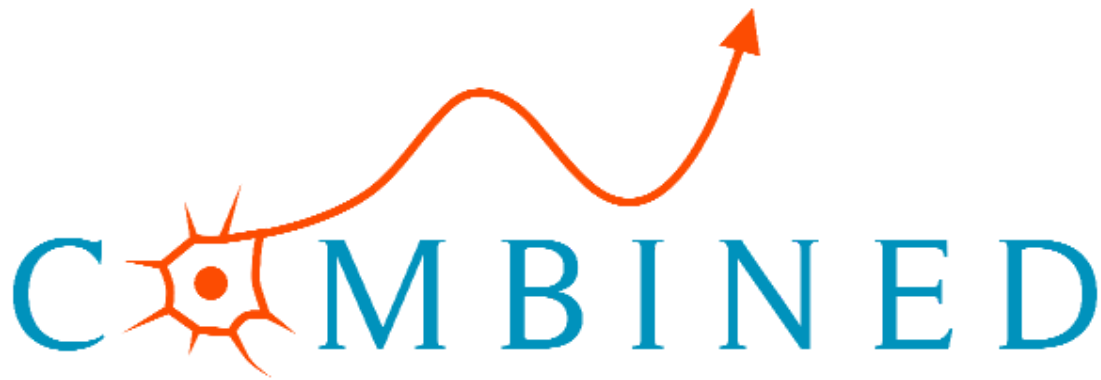


It is with a heavy heart that the NR2F1 Foundation decided to postpone the 2020 Family and Scientific Conference in October. There are several reasons for the decision but the two most important are:

1. The unpredictability of COVID-19 with regards to a potential surge in the Fall due to the eventual lax in social-distancing.
2. Many of our families, including the Foundation, have been and will continue to be affected by this pandemic financially, not leaving any room for travel expenses.

To be clear, this is a postponement, NOT a cancellation. We WILL have a conference in the future, potentially in the Fall of 2021 and we look forward to the unity, information and advocacy that it will bring to all BBSOAS families!

[Listen to board president, Carlie, share the news](#)



The NR2F1 Foundation is excited to announce that we have recently joined forces with COMBINEDBrain! COMBINEDBrain is a consortium led by patient advocacy foundations, working with the clinicians, researchers and pharmaceutical firms that are developing treatments for the disorders they represent.

<https://www.combinedbrain.org/>

The mission is devoted to speeding the path to clinical treatments for people with severe rare genetic non-verbal neurodevelopmental disorders by pooling efforts, studies and data. Drs Christian Schaaf and Jane Edmond have graciously accepted our invitation to join the Scientific and Medical Advisory Board of COMBINEDBrain on behalf of the NR2F1 Foundation. A crucial next-step we are taking is developing a patient registry. One step closer to fast-tracking BBSOAS research!

[Listen to a Facebook Live interview Carlie has with the founder of COMBINEDBrain, Terry Jo Bichell](#)

EDUCATIONAL SPOTLIGHT

What is Cortical Visual Impairment?

CVI stands for Cortical Visual Impairment. It is a brain-based, not vision-based, condition. Children with CVI can receive visual input, but their brain can't make sense of that information. As a result, children can appear to see some things but not others.

The ten characteristics of CVI (as identified by Dr. Roman-Lantzy) are the following:

1. Preference for color
2. Need for movement
3. Visual latency (delayed response when looking at objects)
4. Visual field preferences
5. Difficulties with complexity
6. Need for light
7. Difficulty with distance viewing
8. Atypical visual reflexes (visual blink reflex/visual threat response)
9. Difficulties with visual novelty
10. Absence of visually guided reach

Do all children with BBSOAS have CVI?

CVI is just one of the visual impairments that can be a characteristic of BBSOAS. Per the most recent research publication, "Vision impairment continues to be a primary characteristic of BBSOAS, evident in 47/52 (90%) of individuals. Though the full vision phenotype will be reported separately, it is important to note that vision challenges extend beyond optic atrophy (OA), as the syndrome name would suggest, to also include optic nerve hypoplasia (ONH) as well as CVI. Though 41/50 individuals (82%) have OA or optic nerve pallor, 19/39 (49%) have ONH or small optic discs, and (68%)

have CVI, with many individuals exhibiting more than one of the above." Rech ME, McCarthy JM, Chen C-A, et al. Phenotypic expansion of Bosch–Boonstra–Schaaf optic atrophy syndrome and further evidence for genotype-phenotype correlations. Am J Med Genet Part A. 2020;1–12.

[Read the Full Article](#)



How can you support the visual learning of your child with CVI?

- Introduce objects with bold colors and use simple pictures
- Reduce distractions and visual clutter (ex: place objects in front of an all-black background)
- Ensure your child is properly positioned to accommodate for central field loss
- Provide needed physical support (ex: hold up the child's head)
- Use familiar and real objects rather than abstract objects to encourage visual attention
- Check out this link from Paths to Literacy. It has several activities to try at home that are designed by Dr. Roman-Lantzy. The activities are even broken down by what phase your child is in. [Click Here](#)
- Check out these books that have been adapted for CVI

1. [Sunflower House Under Construction](#)
2. [I am Batman](#)
3. [Dragons Love Tacos](#)
4. [Brown Bear Brown Bear What Do You See?](#)
5. [Pete the Cat- I Love My White Shoes](#)

6. [Pete the Cat and His Four Groovy Buttons](#)

7. [The Very Hungry Caterpillar](#)

How can CVI affect behavior?

- CVI can impact behavior in many ways.
- Some of these include...
- Not recognizing people and appearing to ignore them
- Not reading facial expressions and appearing to lack empathy
- Bumping into people or objects
- Engaging in a "CVI Meltdown"
 - [Read more about CVI meltdowns](#)
- [Read more information on how CVI can impact behavior](#)

More Resources for learning more about CVI

- [Perkins eLearning Videos](#)
 - Access a collection of videos aimed at teaching about CVI
- [Kaleidoscope: The Cortical Visual Impairment Podcast](#)
 - A podcast dedicated to all things CVI
- [Paths to Literacy](#)
 - Promotes literacy for children who are blind, with specific resources for kids with CVI

Do you have any CVI strategies that have worked with your child? We'd love to hear from you!

[Send us an email!](#)

2018 BBSOAS Family and Scientific Conference





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!



Dr. Bosch and Dr. Schaaf presenting at the conference.
Click the picture above to view the actual slides from their 2018 presentation!

RESEARCH CORNER

Dr. Schaaf's latest publication on BBSOAS, co-authored by Megan E. Rech

"Phenotypic expansion of Bosch–Boonstra–Schaaf optic atrophy syndrome and further evidence for genotype–phenotype correlations"

<https://doi.org/10.1002/ajmg.a.61580>

Ongoing BBSOAS research

In May 2019, and in less than a week, BBSOAS families, friends and supporters raised over \$21,000 to fund the iPSC's project.

Dr. Schaaf writes:

"Induced pluripotent stem cells, generated from seven individuals with BBSOAS, were transferred at a temperature of -190 degrees Celsius in liquid nitrogen tanks from Houston to Dr. Schaaf's laboratory at the University of Heidelberg. There, cells are safely stored in liquid nitrogen. First batches of samples have been thawed and taken into culture, growing and dividing, and now waiting to be differentiated into various neuronal cell types. This work promises to increase our understanding of why deletion mutations are less severe than point mutations in the DNA binding domain of the gene. It will also inform us whether there are critical time points in development when mutation in the *NR2F1* gene causes detrimental alterations in the function of those neurons and the developing brain."

Stem cells could also be used as a resource, to be shared with other research groups interested in NR2F1.



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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