Dear

The start of the year has been full, with launching our drug repurposing project, as well as making sure we have enough fundraising activities to support our committed projects, and working on our new website.

April 10 was National Sibling Day. So we turned April into ‘Sibling Month’. To mark this, Brian Read, our Foundation Treasurer, shares his very personal story of being a brother to a sibling with a rare disease. Along with this, Brian has an incredible fundraiser happening - any amount raised during April (“Sibling

Month”) up to $5,000 will be matched by Brian's family in honor of his sister, Stephanie. Read his inspiring story on his campaign page.

Brian and Stephanie

Keep reading to find out about our research, fundraising and board members updates. We have started profiling a BBSOAS family each month,

so check out Susan and Ashlee's stories. If you would like to share your experiences, please get in touch.

Thank you so much for your ongoing support and commitment. Without you, we can't do this!

Best,

Jennifer

NR2F1 Foundation President, and most importantly, Edith's Mum

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Research Corner

Watch the interview with Dr Clement Chow on our drug repurposing project

Dr Elsa Wassmer update

Read Dr Wassmer's short progress update from the Schaaf lab at Heidelberg University here

BBSOAS and Bioinformatics Analysis Discovery

Using a grant issued by the NR2F1 Foundation, Dr. Magdalena Laugsch and her team at the Institute of Human Genetics, Heidelberg University,

have made an important step in biomarker discovery for BBSOAS. Read more here

New research paper The Natural Course of Bosch-Boonstra-Schaaf Optic Atrophy Syndrome (thanks to our wonderful team at Heidelberg University)

Get involved in Research - BBSOAS parents

Observer Reported Toileting Abilities Survey

We have an opportunity with our friends at COMBINEDBrain to better understand BBSOAS and toileting abilities. COMBINEDBrain are conducting a pilot study of the **Observer Reported Toileting Abilities Survey** (ORTAS) for BBSOAS children aged 1 - 6 years old. To participate check my previously sent email, the Facebook group, or email jennifer.coughlin@nr2f1.org for more info

NR2F1 Biorepository 2025 - have your childs genetic sample used in research

We are collecting again in the US and have collection sites in the following states: Tennesse, Colorado, Missouri, Arizona, Maine, Connecticut, Pennsylvania and Georgia. To participate check my previously sent email, the Facebook group, or email jennifer.coughlin@nr2f1.org for more info

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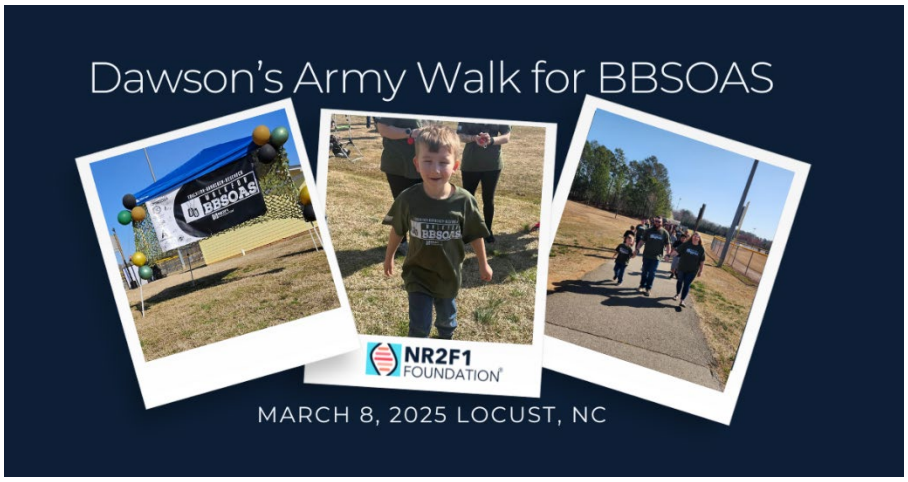
Fundraising Update

Year to date we have raised $40,000 towards our annual target of $205,000 based on our current committed projects.

March was a busy month. On Saturday March 8 we had two fundraisers!

Atlanta, Georgia (US) Mom (*and new board member*), Kari hosted her 4th annual Walk for Vincent. This year was the biggest event yet, with 70 walkers joining. Kari raised over $14,000.

Locust Park, North Carolina (US). Mom, Kaileigh organised her first BBSOAS Dawsons Army walk. In her first ever fundraiser, she managed to raise nearly $4,000. Read more about Kaileighs story in this blog post



April - Sibling month campaign
May 17 - Sarah is making 10 miles meaningful view the campaign here
June 14 - A team of 7 will ride in this years **Million Dollar Bike Ride in Philadelphia.** BBSOAS Leo will be riding again with his parents. Read more about the campaign here. We still have space if you would like to join the team, to ride and fundraise for the Foundation.
July 6 2025 - Devon, UK. BBSOAS Dad, Sean and a team of friends are competing in a Triathlon in honor of son Albert. Read more and support the fundraiser here

To make this years financial commitments, most importantly, paying the second year cost of Dr Elsa Wassmer, we need to continue this momentum of activity in the second half of the year with fundraising activities. If you can help support, no matter how big of small, we would love to hear from you. Drop me an email!

Board Members - the people making it happen

The NR2F1 Foundation only exists because of the incredible work and time the board members commit. Our amazing board members give between 6 - 130 hours each a month to foundation work!

We want to say a huge **THANK YOU to Brigette Hinger.** Brigette has served on the board from 2022-2025 and was the powerhouse behind

organising the 2024 Family and Scientific Conference. Thank you for your all your hard work, Brigette.

Welcome to new our newest board members. We were overwhelmed with the interest we received from our community for our board members opening. So much so that we decided now is the right time to expand our board, as we need additional skills to help us to keep growing and have the best chance to achieve our mission to support families living with BBSOAS, through education, advocacy and research.

We welcome Kim, Kari, Susie and Candice to the NR2F1 Foundation Board.

Kim Ferruzzi

Kari Kruckow

Susie Wong

Candice Conseur

New volunteers

We have onboarded two wonderful new volunteers. After recently organising Dawsons Army fundraiser, Kaileigh is joining to help with our

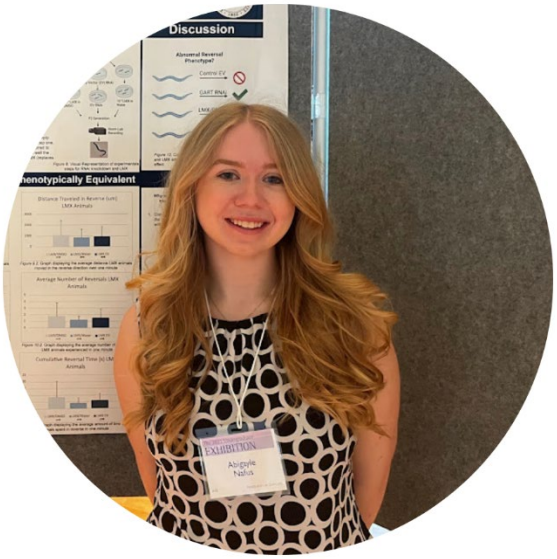
fundraising efforts. Amanda, mom to BBSOAStrong Jacob, has also joined the team and will be getting involved in our Patient Registry projects.

Amanda with her family

Kaileigh with her family

Student volunteer

Meet Abi Nafus! Abi is at Kean University studying a Genetic Counseling Graduate Program. Read all about Abi and the fantastic patient count project she is doing for the Foundation here.



Abigayle Nafus

Meet our BBSOAS families

We have been profiling our BBSOAS families in our blog posts. Read our 2 amazing stories:

“My 56-year-old Daughter Has Just Been Diagnosed” Read Susan's story **here**

Little Susan

“A parents journey of grief and uncertainty” - Mom and NR2F1 Foundation board member, Ashlee Manjon-Stierstorfer shares her feelings **here**

Ashlee, Diana & Emma

2026 NR2F1 Family & Scientific Conference

Save the date for next years conference coming soon…

New NR2F1 Foundation website

We've been working on this behind the scenes project for 10 months and hope to go live this summer. Importantly the website will be available initially in English, French, Spanish and Italian, with other languages following. Our ambition is to improve the newly diagnosed journey for families.

BBSOAS Guides - now available in Spanish/Guías de BBSOAS - ahora disponibles en español

We heard you! You asked for our new guides to be published in Spanish, so we partnered again with UK charity Unique to bring these to you

1. **Full BBSOAS guide/Guía completa de BBSOAS** - a robust guide to BBSOAS/NR2F1 for anyone looking to understand BBSOAS and the NR2F1 gene more comprehensively (click here for English) o aquí para Español
2. **'My Chromosome Story' booklet/Folleto 'Mi historia cromosómica'-** aimed at children. This can be used to start explaining a child's diagnosis to them at a young age. It is also helpful for siblings and classmates (click here for English) o aquí para Español
3. **'Easy Read' guide/Guía de lectura fácil-** aimed at BBSOAS patients who may need a shorter version of the full guide (click here English) o aquí para Español