



So much is happening!

Dear **Samson**,

After a fun few weeks in the summer enjoying some all important time together with our families, September was right back to our desks, and its been non-stop since!

October has been a crazy month of travel, thoughtful discussion and importantly, action. We've held our first [Research Strategy meeting](#) with our board and incredible team of [Scientific Advisors](#) as well as attended both the COMBINEDBrain and NORD conferences, representing the Foundation.

We have also launched [registration for our 2024 BBSOAS Family Conference](#)! It will be in person and virtual, so we really hope to reach our target of increased attendance and most importantly connect with all our families around the world.

Like most charities we are also gearing up for Giving Tuesday on November 28. No surprise you'll be hearing more from us on this, but please save the date and consider the Foundation in your donations. We have come up with a [fantastic research plan](#), but we need to raise (wait for it) **\$365,000 to make it happen!**

On that bombshell scroll down to read the details on exactly what we want to do to help find out more about BBSOAS. Hopefully all this action has peaked your interest and you are wondering how you can get more involved - well good news, [we are hiring!](#) Happy reading!

Best,

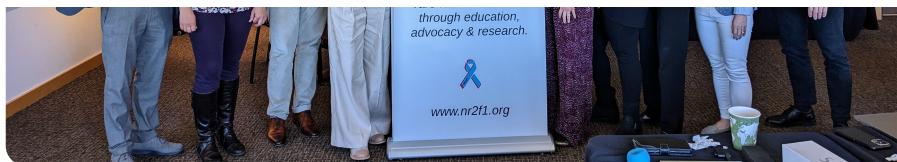
Carlie

Carlie Monnier

President of NR2F1 Foundation and Mom to BBSOAS Sidelle

NR2F1 Research Strategy meeting - October 7, Denver, Colorado





What a weekend! After our Strategic Planning weekend with the board in Atlanta at the start of the year, we were excited to bring together our board and Scientific Advisors to build out our Research Roadmap.

On October 7 we all came together at the University of Denver. We had Dr Schaaf (the 'S' in BBSOAS!), Dr. Henning Fröhlich and Johann Maaß, medical student, all from the University of Heidelberg. Dr. Michele Studer from the University of Nice, France, Dr. Veeral Shah, neuro-ophthamologist from Cincinnati Childrens Hospital and Dr. Anna Pfalzer from COMBINEDBrain.

The day began by hosting local BBSOAS families for 'breakfast with the scientists', a powerful reminder of why we were all there.

The [agenda](#) was then full to bursting! The purpose of the meeting was to develop a research strategy and build a roadmap for moving forward. This is the first [strategic research meeting](#) that the foundation has had that joined all NR2F1 / BBSOAS researchers together in one room to collaborate and brainstorm a research roadmap.

[Read all about it here, and learn about the latest research we have funded with Dr Shah.](#) We will use Giving Tuesday to kick off our fundraising efforts to raise the huge \$365,000 needed to fund some incredible research.

COMBINEDBrain and NORD Conferences

Jen (NR2F1 Foundation Vice President, based in London) and I have been racking up the air miles (and jetlag) this month!

Jen, was fortunate enough to win a grant to attend the NORD conference in Washington DC on October 16 & 17. Our friends at COMBINEDBrain coordinated and held their conference on October 15, and Carlie and Jen were able to attend.

As well as providing fantastic networking opportunities with other Patient Advocacy Groups, and companies focused on rare disease it was a really great reminder of what is the aim of all this - to find thereapeutic treatments for our BBSOASstrong.

Giving Tuesday - Tuesday November 28

To support our 2024 research ambitions, we need to raise an eve

watering \$365,000!

What do we need the money for? We've come up with a 3 year Research Roadmap and a list of initiatives we would like to initiate - which ofcourse comes along with a hefty price tag. We have decided as a Foundation we want to tackle this ambitious goal head on as it's the only way we feel research can really move forward

What will the money raised fund?

1. Fund 3 Post Doctoral researchers for 3 years to work alongside our lead scientists to exclusively research NR2F1 and BBSOAS (*will require year 2 and 3 fundraising in addition*)
2. Create a NR2F1 New Investigator grant to attract junior researchers to research the NR2F1 gene
3. Develop a new mouse model
4. Biomarkers – host a biorepository collection at our 2024 Family Conference and develop a BBSOAS biomarker

Could you support a matching campaign? We have had a family approach the Foundation that is offering a \$12,500 matching offer to help us launch our Giving Tuesday campaign for November 28. Would you like to consider adding to this to help us raise the stakes? This year is the first year that we will have quite the target to achieve and need all the help we can get.

DONATE NOW!

2024 BBSOAS Family Conference - April 3-5, 2024.

Where? [Embassy Suites Orlando Lake Bueno Vista](#) in Orlando, Florida.

Is it in person or virtual? Both! The conference is be available in person and also virtual.

Who is confirmed to speak so far? So far we have confirmed [Dr Christian Schaaf](#) (the 'S' of BBSOAS) Medical Director and Department Chair of the Institute of Human Genetics at Heidelberg University (Germany). [Dr Veeral Shah](#), Pediatric Neuro-Ophthalmology, Division of Pediatric Ophthalmology, Cincinnati Children's Hospital Medical Center and head of the BBSOAS Center of Excellence. [Sarah Poliquin, PhD](#), NR2F1 Scientific Officer and COMBINEDBrain Postdoctoral Fellow, and [Michèle Studer, PhD](#), Research Director at Institute of Biology, Nice, France...and more names will be hitting the line up shortly

including Breakout Sessions!

Help us achieve our goal! Our ambition for 2024 is to increase attendance by 20%, so we hope you can join us for some family fun as well as lots of informative and educational presentations by BBSOAS experts.

Could you be a event Sponsor? Believe it or not, the conference will cost around \$35,000 to host! Registration fees only go a small way to covering those costs. If you would like to donate to covering the costs, or be an event Sponsor [click here](#)

[Register HERE for In-Person or Virtual!](#)

Biorepository on the Road - your child's genetic sample used in research - we need 4 more patient samples to be eligible for a pharma industry biomarker program

All year we have partnered with COMBINEDBrain to collect BBSOAS bio samples (e.g. blood and urine), to be used by researchers in future projects.

There are 4 remaining conferences and opportunities to participate this year.

Please help - we need 4 more patient samples for BBSOAS to be considered, to be part of a pharma industry biomarker program!

In combination with the biorepository we have the chance for BBSOAS to be included in a pharma industry biomarker program. This would be testing and analysis of our BBSOAS patient samples done free of charge to us and an amazing opportunity to engage with the companies who we want to provide treatments to our patients! But for us to be considered eligible, we need 4 more diagnosed BBSOAS samples.

Upcoming dates:

- **LA November 4th 2023**, 9am-5pm (230 South Main St, Suite 210, **Orange County, CA**92868) (basically LA!)
- **Malibu November 5th 2023**, 9am-5pm (27318 Winding Way, Malibu, CA, 90265)
- **Orlando November 30th 2023**, 9am-5pm (9700 International Dr, **Orlando, FL** 32819)
- **Orlando December 1st 2023**, 9am-5pm (8978 International Drive **Orlando, FL**, 32819)

Email Jennifer.Coughlin@nr2f1.org to find out more and register

Newly Diagnosed?

Behind the scenes we have been working hard to update the NR2F1 website and have added a new page '[Newly Diagnosed](#)' with our recommendations on what to do when you receive your diagnosis

Our top recommendation? Register for the [Patient Registry](#).

We are hiring!

One of the priorities from our strategic plan is to ensure long term success of the Foundation through succession planning. So planning ahead we have a few vacancies on our board coming up in 2024.

We are looking for a **Fundraising focused Board Member**, and a **Treasurer**. If you are interested in either of these, [check out our website for more details](#).

We also have some great **volunteering opportunities**, if you are a **Social Media** whiz, or a **Quickbooks Online** pro - get in touch! A few hours a month, go a long way to helping the Foundation operate.

December 7 International BBSOAS Awareness Day

With awareness day not far away, order your T-shirt or sweater now

Click here for [Dec 7 BBSOAS Awareness Day tshirts](#)

Do you want to host a fundraiser for us?

Whether its a bake sale, marathon, bike ride or anything in between, we need your help to raise much needed funds. Just reach out to us and we can help build you a webpage, fundraising link and support your campaign.

As always, please do get in touch with feedback, ideas or suggestions. We love hearing from you.

Yours sincerely,

Carlie Monnier

NR2F1 FOUNDATION President and Mom to BBSOAS Sidelle

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