

Subject: What a Year for the NR2F1 Foundation!

Date: Monday, 2 January 2023 at 02:04:24 Greenwich Mean Time

From: NR2F1 Foundation Board

To: jenniferkcoughlin@gmail.com

[View this email in your browser](#)



2022 in Review

2022 has been a remarkable year of accomplishments for the the NR2F1 Foundation and as this year has drawn to a close, it is worth pausing to reflect.

Of course, we couldn't do any of this without the support of you, our friends, family, donors, and the BBSOAS community. We thank you for for all your continued support!

Sincerely,

The NR2F1 Foundation Board



Originally set for October, 2020 and rescheduled 3 times The NR2F1 Foundation was determined to make this happen. Finally, in April 2022 we hosted the first-ever [Hybrid Family and Scientific Conference in Florida!](#)

At the conference, we launched the first-ever [NR2F1 Foundation Patient Registry/Natural History Study](#) exclusively for the International BBSOAS

[Registry/Natural History Study](#) exclusively for the international BBSOAS community. This is a crucial first step to furthering research and a must-have for any future clinical trials.

Are you a BBSOAS caregiver or patient? Have YOU signed up?

[BBSOAS Patient and/or Caregiver? Sign up HERE](#)

In collaboration with COMBINEDBrain, of which the NR2F1 Foundation is a proud board member of, we were awarded a \$15K grant from the Global Genes Health Equity Grant!

[Read more HERE](#)

We were awarded this [\\$17K grant](#) to support research utilizing mouse models to create new treatments! The grant application was initiated by Dr. Kyle J. Horning and Dr. Terry Jo Bichell, PhD, MPH, of COMBINEDBrain, who successfully assisted the foundation in winning it.

Earlier this year Tim and Jennifer Coughlin received the news that their youngest daughter, Edith, was diagnosed with [BBSOAS](#), a rare neurological mutation of the [NR2F1 gene](#). In May Tim's close friend Chris Schastok decided to run the [2022 Bank of America Chicago Marathon](#) to raise funds for the NR2F1 Foundation. Upon learning this, Tim decided to join him!

With no idea of how much money they would be able to raise, as of race day on October 9, 2022, they had raised over \$130,000, and donations continued to come in. The donations will be used to fund research that can change the lives of those who have been diagnosed with BBSOAS.

"Thanks to their support and all who donated, [current research projects](#) underway can be expanded upon, as well as exploration

of other avenues that were not otherwise financially viable... will yield... a better life and hope on the horizon for children like Edith and her international BBSOAS family. The NR2F1 Foundation Board is grateful beyond words.”
-Carlie Monnier, Board President

[Invest in BBSOAS Research HERE](#)

Speaking of research...

When assessing the communication ability of individuals with neurodevelopmental disorders (NDDs), like those living with BBSOAS, existing clinical outcome assessments (COAs) have significant limitations, including the inability to differentiate among individuals and ***lack of input from parent advocates***.

With a grant from the U.S. Food and Drug Administration (FDA), Dr. Christy Zigler (Principal Investigator) and other CHM investigators will work with [COMBINEDBrain](#), of which the NR2F1 Foundation is a member of, to expand the ORCA measure to a range of neurodevelopmental disorder (NDD) populations.

The team also includes a Stakeholder Engagement Group consisting of 16 leaders in the field ***including parents, patient advocates, and clinicians*** who will provide access to 13 distinct NDD populations, including the NR2F1 Foundation and patients living with BBSOAS.

Speaking of ***MORE*** research...

The NR2F1 Foundation launched the [TRIP BBSOAS](#) (Translational Research Investigating Phenotype of BBSOAS) research fundraiser.

This joint project between Drs. Studer and Schaaf (a ***dream team of collaboration***; one expert in the nr2f1 gene and one expert in BBSOAS) will give us insights into the role of the NR2F1 gene in brain development, and its downstream targets and pathways

brain development, and its downstream targets and pathways. Importantly, it could also be used as the **basis of future therapeutic treatments for BBSOAS patients.**

[Learn More About TRIP BBSOAS](#)

Dr. Schaaf briefly explains TRIP BBSOAS and how important it is for our research road map.

The NR2F1 Foundation welcomed 7 new board members to the team this year!

[Click HERE](#) to learn more about them and the role they play.

The NR2F1 Foundation is deeply grateful to the law firms, [King & Spaulding LLP](#) (Atlanta, GA) and [Coughlin Midlige & Garland LLP](#) (Morristown, NJ) who will be representing the foundation pro bono.

A huge thank you to Patrick of CMG for making all of this happen.

The NR2F1 Foundation is always looking to forge new partnerships and memberships and 2022 was no exception!

We amped up our [Candid membership](#) from [Bronze to Gold status](#).

We joined the [Global Genes Global Advocacy Alliance](#).

We joined the [National Organization for Rare Disease](#) as a [Platinum member](#).

As of November, 2022 shoppers have raised \$1,662.26 for the NR2F1 Foundation this year!! By turning on [AmazonSmile](#) to NR2F1 Foundation, 0.5% of all eligible purchases is automatically

donated to our foundation!

The great news is that [AmazonSmile](#) is now supported on the Amazon shopping app on iOS and Android mobile phones! Simply follow these instructions to turn on AmazonSmile and start generating donations.

1. Open the Amazon Shopping app on your device
2. Go into the main menu of the Amazon Shopping app and tap into 'Settings'
3. Tap 'AmazonSmile' and follow the on-screen instructions to complete the process

We celebrated the 2nd annual [International BBSOAS Awareness Day](#) this year!

All across the world, families, friends and supporters wore their stripes to help celebrate our #BBSOAStrong.

A private BBSOAS Grandparents support group has arrived! Thanks to two wonderful BBSOAS grandmothers who moderate the group.

Are you a #BBSOAStrong Grandparent?

[Join here](#)

[Join HERE if you are a BBSOAS Grandparent](#)

The NR2F1 Foundation strives to provide education and resources for the BBSOAS community it serves.

One particular resource we are proud to offer this year is a [document curated by Dr. Kyle Horning](#) exclusively for the BBSOAS community which can be found on our website.

Trying to understand the scientific jargon in a genetic report can be overwhelming and defeating to say the least.

Now families have a way to easily break it down and most importantly, become empowered with the knowledge they need to advocate for themselves and/or their child.

(And coming soon, this document will be available in French, Italian, German and Spanish!)

Check your inbox this week for the next installment: 2023 Plans for the NR2F1 Foundation

Happy New Year!

You shop. Amazon donates to NR2F1.

You can support the NR2F1 Foundation every time you shop on Amazon! [Visit Amazon Smile](#) and select the NR2F1 Foundation as your charity of choice. With every purchase you make, Amazon will donate 0.5% to the NR2F1 Foundation.



Want to change how you receive these emails?
You can [update your preferences](#) or [unsubscribe from this list](#).