a guide for

Newly Diagnosed GRIN2B Families

GRIN2B Foundation

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The creation of this booklet was made possible thanks to a grant from **GRIN Therapeutics.**

GRIN THERAPEUTICS
A Neurvati Neurosciences Company

Started in 2017 by a group of GRIN2B parents.

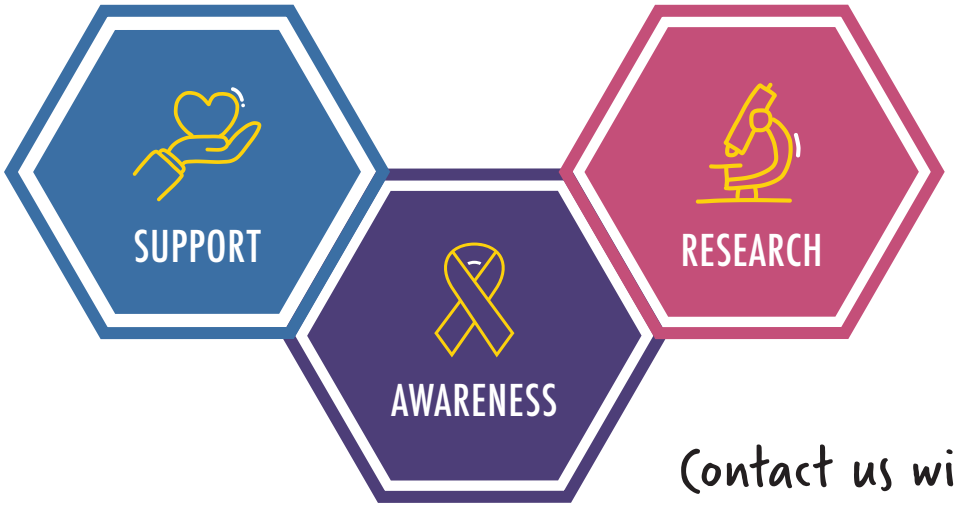
Mission Statement

GRIN2B Foundation is a parent-run organization dedicated to furthering research on the GRIN2B gene and providing support and education to the small, but growing community of individuals and families impacted by a GRIN2B diagnosis.

Our Vision

Our vision is a world where every family impacted with a GRIN2B diagnosis feels empowered and supported.

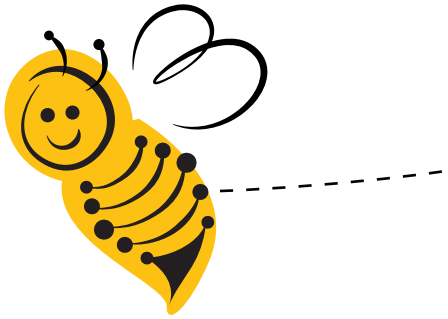
Our Strategy



Contact us with any questions!

info@grin2b.com


Our journey has had its good days and bad days, but I 100% know it has been made easier by the support of the Foundation, as well as the parents involved. We are forever grateful.
-GRIN2B Parent



Hello there, friend.

I bet you can't believe you're reading a booklet about a rare genetic disorder which someone you love so dearly has just been diagnosed with. You have no doubt already been down a Google rabbit hole and have not stopped thinking about your child's future and what therapy or medication you need to start tomorrow to give them their best shot. Your heart might be aching with the shock of it, or maybe you're feeling a little bit relieved to have an answer. Or maybe both. There's no right or wrong way to feel right now.

The coming days, weeks and months might feel like a blur. You might feel like a different person in a different world, or you might be so busy, exhausted and sleep-deprived that you feel numb. Please know that when you're ready, we're here for you. We know what you're going through, and we're all muddling through it as best we can, supporting each other. I wish I had known back then in the early days that everything would be OK. Because it is OK, and my GRIN2B daughter is not just OK, she's totally awesome! I have been exposed to and involved with an amazing community, and I have learned so much about myself and am so proud to be a disability ally. That's not to say that it's been an easy path to walk. Some days I despair at how exhausting and boring it is to parent a child with a disability, while simultaneously being in awe at just how perfect and wonderful she is. I hope you find some comfort knowing you are not alone and that there is an incredibly supportive world out there of therapists, clinicians and researchers who care for and are dedicated to improving the quality of life for our children.

Lauren Hookings

Board Member
GRIN2B Foundation

Welcome to GRIN2B Foundation,

The idea for this foundation came about after my then-18 month old daughter was diagnosed with a GRIN2B variation in 2014, and I was frustrated by the lack of information and resources available to me.

The original goal was to offer support and share research to the small but growing community of families whose children have been diagnosed with alterations on their GRIN2B gene. We formed our Board of Directors early in 2017 and, after a lengthy application process, finally received our 501(c)(3) tax-exempt status by the United States government by the end of that year.

We know you may be feeling confused and overwhelmed. Keep in mind that everyone processes the diagnosis differently. Some may want to dive into the research, while others may be understandably overwhelmed by all the complex medical terms. It is perfectly acceptable to give yourself some time to come out of the initial fog of diagnosis before you attempt to read through all the research. Please continue to check our website and social media pages for updates as the research on GRIN2B is constantly changing and emerging.

I have seen firsthand that no matter how attentive and skilled the doctor is, no one will care as much about understanding how GRIN2B affects our children as we parents will. It is our job to stick together and drive the ongoing research so we can better understand what our children are facing and how we can best help them. Though each child is impacted slightly differently by their specific GRIN2B change, they are all deeply loved and adorable. They deserve to be seen as more than just “puzzles” or “mysteries.” Let’s work together to help unravel their mysteries and change the narratives surrounding our kids.

Liz Marfia-Ash
President
GRIN2B Foundation

Read Liz’s first **blog post** about her daughter’s GRIN2B diagnosis.

about GRIN2B

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GRIN2B is not the name of a disease or disorder, but is the name of a gene located on the 12th chromosome. Its function is to encode a protein that forms a receptor responsible for sending chemical messages to the brain. A variation on the GRIN2B gene means that some portion of this specific genetic code either got deleted, duplicated or rearranged. As of 2018, changes to the GRIN2B gene are now being referred to as **GRIN2B-Related Neurodevelopmental Disorder.**

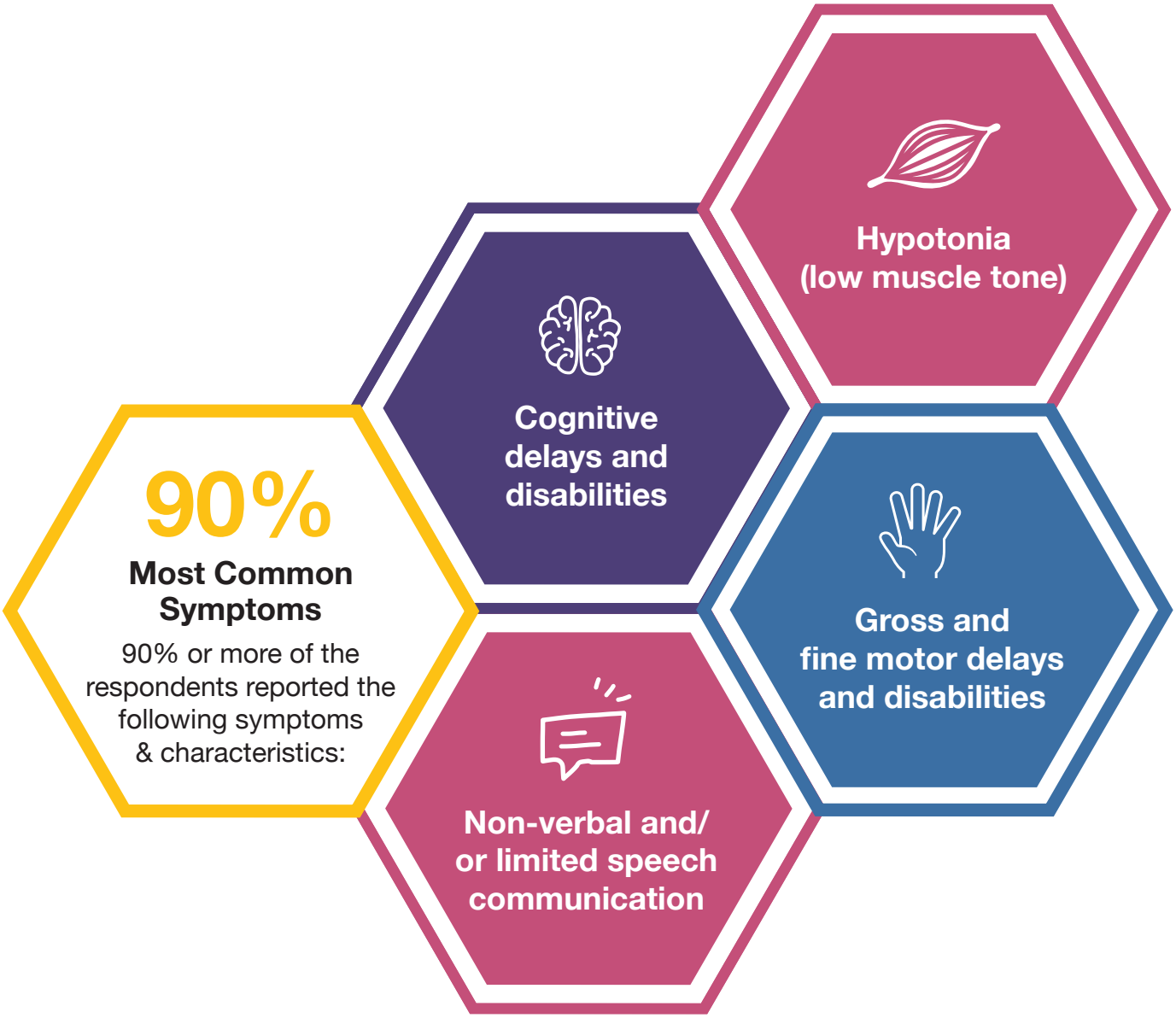
GRIN2B stands for: Glutamate Receptor, Ionotropic, N-Methyl D-Aspartate 2B

GRIN2B is part of a larger "GRIN" gene family. There are seven* of these genes, four of which (GRIN1, GRIN2A, GRIN2B, and GRIN2D) are currently associated with syndromes. *Not much is known about the other 3 genes yet (GRIN2C, GRIN3A, GRIN3B), though that will likely change over time.

Additional disorders that are associated with GRIN2B are on the GRIA, GRID and GRIK genes.

Please refer to the following website pages for more information on understanding GRIN2B.

Common symptoms



See our complete symptoms infographic **here.**

PLEASE NOTE - this infographic was completed in 2019 and it may not reflect all symptoms. For example, movement disorders (such as dystonia) and paroxysmal sympathetic hyperactivity (psh or dysautonomia) were not commonly discussed back in 2019 and therefore did not make it onto our symptoms survey. However, as of 2023, these are considered common symptoms of some of our patient population.

A note about symptoms from GF President, Liz Marfia-Ash:

When you join our GRIN2B Parent Support Group on Facebook, you're going to encounter GRIN2B kids with all different abilities. Our children are a very diverse bunch, and the range of symptoms for GRIN2B is much wider than it is for the other GRIN genes. The truth is…every single individual in our GRIN2B community has value and is exactly who they're supposed to be, no matter what milestones they achieve, or how "functioning" they are perceived to be. Functioning labels are far too limiting and have very little meaning. It's always bothered me that we only ever talk about high or low functioning. That's such a narrow way to define our kids or even anyone.

It is important to always presume competence/potential when interacting with our GRIN2B family members with limited speech. "Presume competence and potential" means to assume an individual is aware and able to understand you even though they cannot show this to you in any obvious way. Talk to them as you would any other typical, same-age child or person. Doing so respects the value of human diversity.

> Not being able to speak is not the same as not having anything to say.
>
> -Rosemary Crossley

What are the treatments?

While there is presently no cure for a GRIN2B change, physical, occupational, and speech therapies can help manage symptoms in children and may help them reach developmental milestones in their own time. Epilepsy, if present, is treated by a specialist. There are currently no FDA-approved medications on the market to specifically alleviate the symptoms associated with GRIN2B, though clinical trials are in the developmental phase.

A note about therapies...

There is no right or wrong amount of therapy for an individual with GRIN2B. The need for therapy will be highly specific to each individual's needs. There are many other types of therapy that children from GRIN2B have benefited from. Please visit our website or check our Private Facebook Group for Parents for more information. Keep in mind that what works for one child may not work for another. Also, there may be limitations on services depending on the region you live in, what your health insurance provider will cover and many other factors.

For help finding local resources, we suggest asking your doctor(s), therapist(s), teacher(s), etc. You can also search for a Facebook support group for parents in your area.

Please refer to our website for more treatment options.

Treatments

Clinical Trials

you are not alone

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It takes a while to learn how to cope as a parent of a child with a disability.

We have been there and are happy to share what we've learned along our journeys. Here are five tips that have helped us deal with the stress, fear and uncertainty of this unknown diagnosis.

FIVE Tips for Coping

1. **Pace yourself.** You don't need to teach yourself genetics overnight (unless you really want to, of course). It's a lot of information to process, especially if you don't have a scientific or medical background. Take as much time as you need.

2. **Find people who get it.** Lean on your fellow GRIN2B parents, but also look for a local support group for parents of disabled children. It will be important for you to have a safe space to regularly share your feelings, whether it's a support group or a therapist.

3. **Grief is a normal part of the diagnosis.** Grief is a natural starting point because there's just no way to prepare yourself for this diagnosis. But try not to linger too long in that grief stage and, most importantly, don't do it in front of your child. Our children know and understand much more than they let on, and they will know if we are grieving them. Channel your grief away from your child and towards external sources, such as how society is structured against disability or how some people will never understand our lives.

4. **Live in the present.** It may take years to get FDA-approved treatments and even longer for a "cure," however that may look. Learn to be happy with your situation today while you hope for better treatments and resources for our GRIN2B loved ones.

5. **Take some time to learn about disability from those who are actually disabled.** Learn about ableism. This may involve some serious self-reflection on your part if you've not had much exposure to the disabled community before. Disability is a normal part of life. Do what you have to do to find acceptance. We promise you, this mindset will make a world of difference towards your mental health.

(download a print out here)

Two ideas that really stuck with me after the GRIN2B conference is the understanding that disability is a natural part of the human experience and Disability Pride is accepting and honoring each person's uniqueness and seeing it as a natural and beautiful part of human diversity.

Please read that again...

A natural and beautiful part
of human diversity.

Turning that over in my mind and
in my heart has really shifted something
inside me that feels less like sadness
and more like solace.

-Brittaney Crider, GRIN2B Parent

GRIN2B foundation support programs

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Supporting our GRIN2B community is one of the key pillars of our mission. We are pleased to offer the following Support programs.

Patient Assistance Grant Program:

Provides funds for families in need of specific, therapeutic equipment that is not otherwise covered by insurance or government funding. Conditions apply.

Care packages:

Please let us know if your GRIN2B loved one is in the hospital for an extended period of time, and we will send a care item to brighten their day. *Email* **info@grin2b.com**.

Regional Meetups:

We are happy to support a limited amount of **regional family meetups** per year. Meetups may be hosted by the Foundation or hosted by families. If you are interested in hosting, we can help connect you to families in your area. See our website for more information.

Private Facebook Parent Support Group:

This private Facebook group is for parents and primary caregivers only. After you request to join, you will be asked a series of questions to confirm that you are the parent or caregiver of a person with a GRIN2B variation. One of these questions will ask if you've completed our **Family Contact Registry** on our website. You must complete this registry to be approved to join the group. If you have questions about this process, please email **brittaney.crider@grin2b.com**. Once you have been approved, if you feel comfortable, please post an introduction detailing where you're from and a little bit about your child.

A tip about finding local support:

Our online support group is a wonderful resource for parents looking for help navigating the GRIN2B diagnosis. However, it's likely you'll need help from families in your hometown to help you navigate local resources. There may already be a Facebook group local to you that supports families of kids with disabilities. Try searching your town, city, county, country or area and the words "special needs", "developmental delays" or "developmental differences or disabilities". Or ask all the families, doctors and therapists you know. Sometimes, these groups are secret and cannot be found by searching, and you will need to be added by a member.

Thank you to the Grin2B Foundation for your support of Alex and our family. Alex had a successful admission to the Epilepsy Monitoring Unit at British Columbia Children's Hospital and the Grin2B Patient Assistance Grant Program helped alleviate a lot of financial stress associated with travel.

-Kelly Caleb,
Grin2B Parent

Awareness Resources

Awareness Cards

Awareness Apparel

Awareness Presentation [Template](#) for Schools

[School Info Sheet](#)
Can be customized with your child's info. Email lauren.hookings@grin2b.com

Awareness Week Facebook Profile Frame

specialists to see:

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Is there a type of doctor that specializes in GRIN2B?

There are doctors around the world that currently see GRIN2B patients, but not enough. We are proud to fund the first **North American Center of Excellence for GRI-Related Disorders.**

Depending on the individual's specific symptoms, GRIN2B patients may be best served seeing neurologists, geneticists, general pediatricians, developmental pediatricians, ophthalmologists, or various other specialists. Because GRIN2B diagnoses are still so rare, you will want to find a local doctor willing to listen and learn about GRIN2B along with your family.

For a list of doctors recommended by other families,

Please see these links that our friends at CureGRIN Foundation put together.

International Doctors

United States Doctors

Helpful tips for GRIN2B families navigating medical appointments

Keep an electronic copy of your child's genetic report on your phone and computer. This will come in handy when you're participating in research studies/clinical trials.

We recommend using an app such as **MyMejo** to track all of your child's medical and therapeutic history.

Bring a copy of the **Symptoms Infographic** with you to appointments.

Child Neurology Foundation
New Visit **Toolkit**

MyMejo App for tracking

GRI Center of Excellence

GRIN2B Foundation partnered with **Children's Hospital Colorado** in 2022 to create the first North American GRI Center of Excellence.

The GRI Center of Excellence will work to continually advance our understanding of all GRI-gene Related Disorders (GRIA, GRID, GRIK & GRIN). Over time, our experts will research how these conditions unfold so they can develop standards of care for all those affected.

For information on how to schedule an appointment as well as details on additional United States locations, please check our **website.**

PLEASE NOTE - the Center of Excellence is not intended to replace your primary care doctor. Rather, they can provide ongoing expert consultation.

Children's Hospital Colorado

patient registries

GRIN2B Foundation

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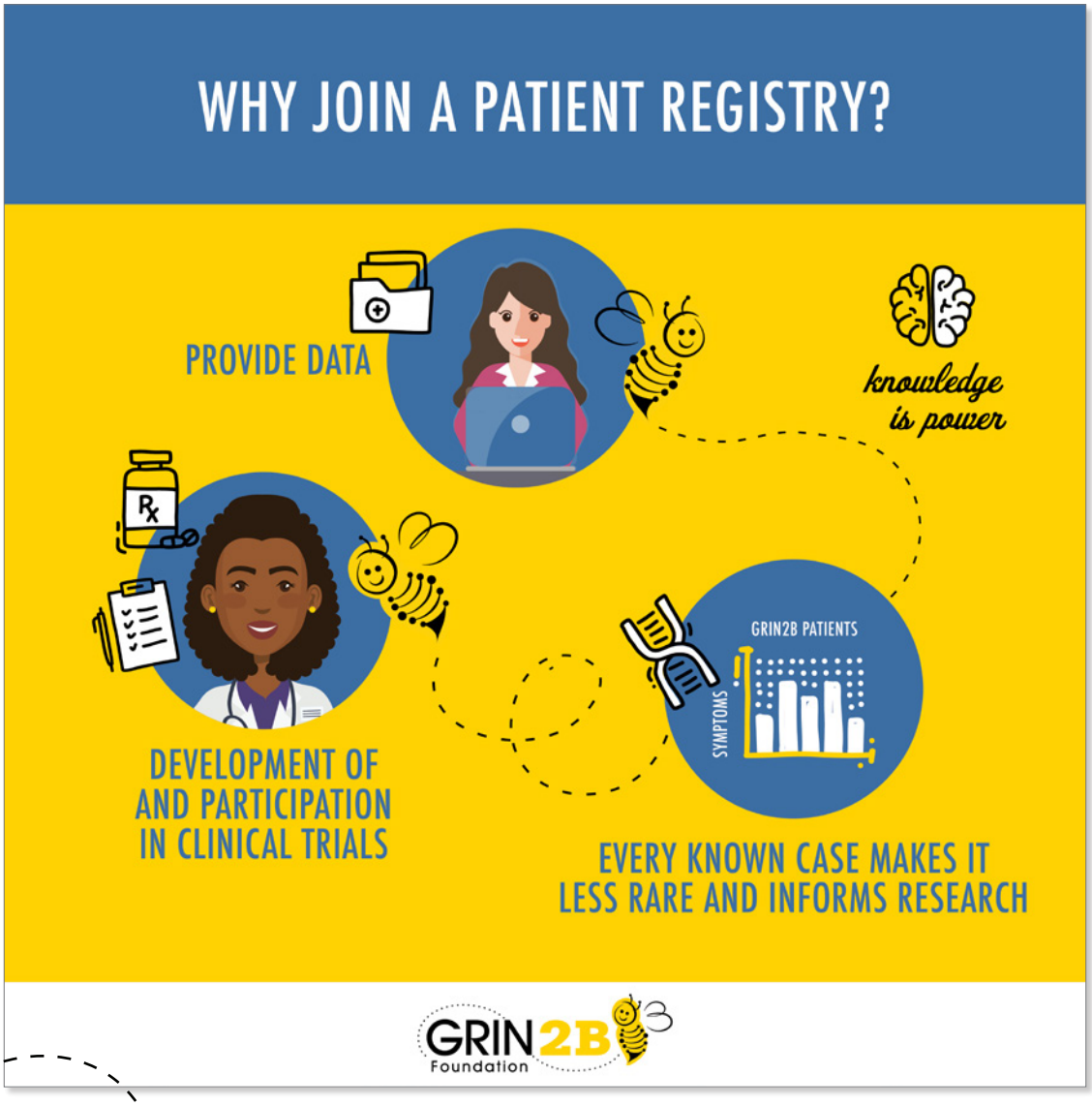
Why should I sign my child up for a patient registry?

This is a question we get asked often. Patient data is the foundation needed towards developing standards of care, and, ultimately, treatments and a cure. With increased data, comes increased knowledge. This is why we need each and every family to share their child's data.



There are 3 registries we ask you to join.

GRIN2B Foundation's Family Contact Registry.
This puts you on our mailing list and gives us a general idea of how many families we're supporting and where everyone is located.

join here: http://grin2b.com/register/

GRIN Variant Registry.
A joint effort between the University of Colorado and the University of Leipzig. This will support a comprehensive understanding of all different GRIN variants. The data from the registry is needed in order to inform the broader community, especially industry and the FDA, in order to design and implement successful clinical trials in the years to come.

enroll here: https://grin-portal.broadinstitute.org/

Simons Searchlight.
This registry studies genes that cause rare neurodevelopmental disorders, including GRIN2B.

join here: https://www.simonssearchlight.org/

SIMONS SEARCHLIGHT

Please print!

Please print out our **GRIN2B Registry Checklist** to help you keep track of when you have completed enrollment in each registry.

REGISTRIES TO JOIN

GRIN2B Foundation Contact Registry:
grin2b.com/register

What is it?
GRIN2B Foundation's internal contact registry for families. This database gives us a better understanding of how many families we represent and where they're located.

How does this help my family now?
You'll receive our mailings, newsletters, event and research announcements and you'll gain access to our Closed Facebook Parent Support Group.

Questions? Contact *brittaney.crider@grin2b.com*

If you access this registry using Google Chrome, you can use Google Translate.

Date registration completed:

_____

UNIVERSITY OF COLORADO *and* **UNIVERSITY OF LEIPZIG**

GRIN Variant Registry:
grin-portal.broadinstitute.org

What is it?
This is the primary patient registry to support a comprehensive understanding of all different GRIN variants. The data from the registry is needed in order to inform the broader community, especially industry and the FDA, in order to design and implement successful clinical trials in the years to come.

How does this help my family now?
Once enrolled, your child's variant will be added to the queue to have functional analysis performed. The results of this analysis help researchers understand how your child's variant functions and will inform what types of medications and clinical trials may best help your child.

Questions?
If you are from North or South America or Australia, contact *GRINResearch@cuanschutz.edu*

If you are from Europe, Asia, Africa or other, contact: *GRIN@medizin.uni-leipzig.de*

Date registration completed:

_____

SIMONS SEARCHLIGHT

Simons Searchlight GRIN2B Registry:
www.simonssearchlight.org

What is it?
Simons Searchlight is an international research program with the goal of accelerating science and improving lives for people with rare genetic neurodevelopmental disorders, including GRIN2B.

They collect different information than the GRIN Variant Registry so it is important to enroll in both.

How does this help my family now?
You'll receive quarterly reports with registry data and Amazon gift cards for each survey completed.

Questions? Contact *coordinator@simonssearchlight.org*

Registry available in English, Spanish, Dutch and French

Password & user information:

_____

Date registration completed:

_____

research

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GRIN2B Foundation is actively involved in both funding research and collaborating with universities, institutions, pharmaceutical companies and additional GRIN or GRI nonprofit organizations around the world.

Read about our funded research **here.**

PubMed **search** for GRIN2B research articles.

Summaries of GRIN2B research articles, by **Simons** Searchlight

If you are interested in learning more about the drug development process, you can take NORD's **Rare Disease Drug Development Course.**

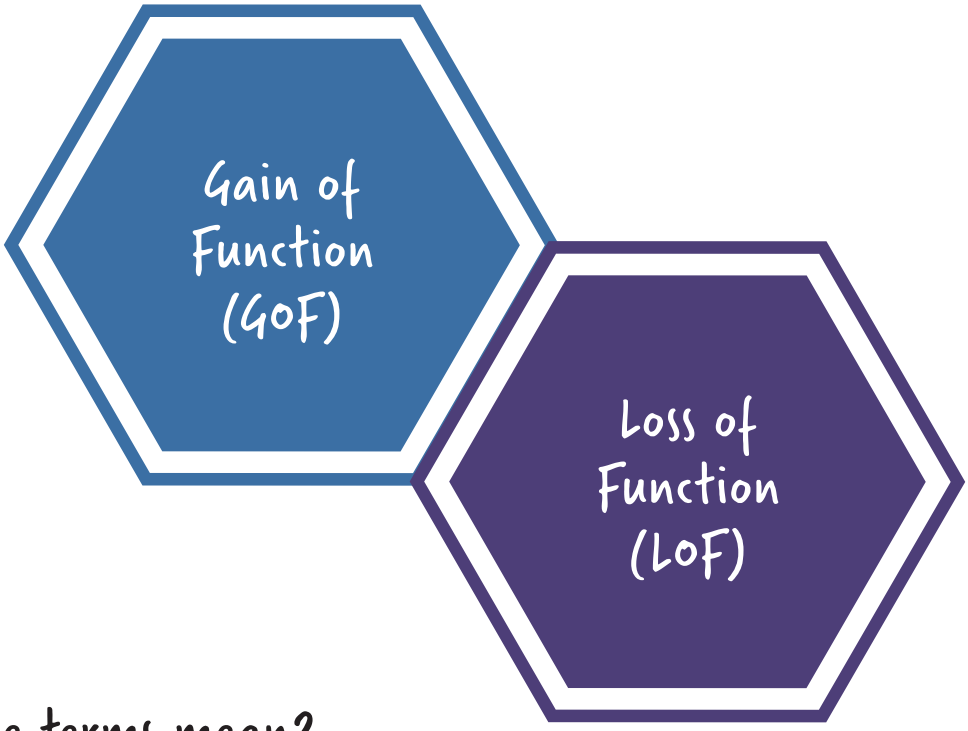
Please participate in Research!

As our community is so small, it is critically important that as many of us as possible participate in research studies when possible. Even simple data about your affected loved one's symptoms can make a big difference to our understanding of GRIN2B.

The best way to keep up to date with all ongoing research studies and clinical trials available to our community is to enroll in our **Contact Registry** and follow us on social media. We regularly email families regarding any research study opportunities.

For info on GRIN2B clinical trials, search **clinicaltrials.gov**

Two important terms to know:



What do these terms mean?

Where there is a change in the GRIN2B gene, it will be classified as either a gain of function or loss of function variant.

Gain of Function (GOF)

In a gain of function variant, the receptor is more active than normal. This means it is turned on too much of the time, or that the signals are too strong.

Loss of Function (LOF)

In a loss of function variant, the receptor is less active than normal. This means that it is turned off too much of the time, or that the signal is too weak.

LoF can also occur if a variant gene is totally or partially deleted or the protein product of the variant gene is shorter than expected *(this is the case with a so-called truncated variant or nonsense variant).*

Why is this important to know?

- Gain of function variants typically have a more severe phenotype (symptoms).
- Though there are no current fda-approved treatments, different medications will be used to treat gain vs loss of function.

For more information on GOF & LOF, check out the following resources:

Webinar: [Gain of Function vs Loss of Function Variants]

[GRIN Europe information on GOF & LOF]

Infographic: [Understanding Gain & Loss of Function]

Functional Analysis

It is important to understand whether your child's variation is a Gain or Loss of Function as some treatments will only be relevant and safe for the specific functional status. A **functional analysis** is a series of tests done in a lab (Emory University) which determines if the variant is gain, loss or complex (meaning, that due to current limitations in science, the function cannot yet be determined).

How do I get the functional analysis testing done for my child?

If multiple patients share the same variant as your child, then it is possible the analysis has already been completed. The **GRIN Portal** is an interactive website for families, clinicians and researchers looking to understand GRIN-related disorders. You can search specific variants and obtain further information such as how many people are registered with that variant and whether or not the functional analysis has been completed.

If a functional analysis has not been performed on your child's variant, you must first enroll in the **GRIN Variant Registry**. Once enrollment is complete, your child's variant information will be sent to Emory University for the functional analysis*.

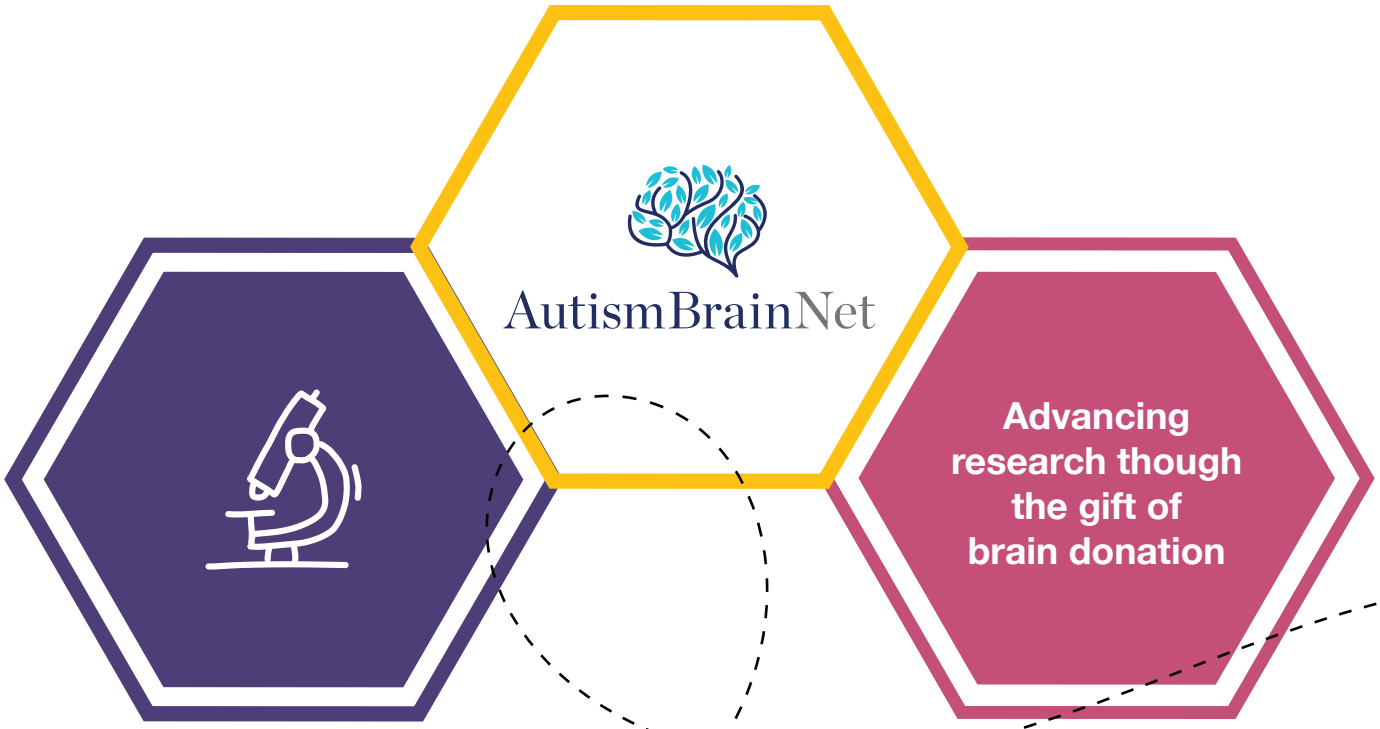
PLEASE NOTE - The functional analysis tests are both time-consuming and costly to the team at Emory University. This process can take 6 months or more. Our organization is in regular contact with the team at Emory as to how we can assist in overcoming these barriers in the future.

Emory will only release the functional analysis results to a physician. Please have your child's doctor contact the team at Emory to find out if the results have been completed.

What does this diagnosis mean for my child's future?

This disorder does not appear to be progressive, meaning it does not worsen over time. But keep in mind, there is likely a larger population out there with undiagnosed GRIN2B changes, so the phenotype and prognosis will continue to evolve. Unfortunately, we do not have a crystal ball, and no one can predict how your child will be impacted.

Though deaths in our community are incredibly rare, GRIN2B Foundation has a partnership with Autism BrainNet to help collect postmortem brain tissue from people who have GRIN2B-Related Neurodevelopmental Disorder. Please visit our **website** to learn more.



other resources and recommended reading

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Further Reading

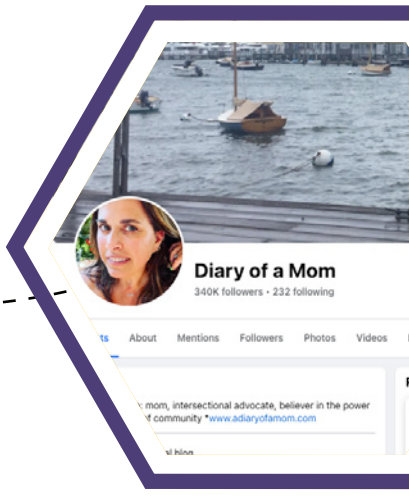
First and foremost, please check out our website and have a look through it at your own pace. It has a wealth of knowledge and links. Here are our top recommendations for further reading if and when you're ready.

Facebook Pages

- Diary of a Mom - she has an adult daughter with Autism, ID and epilepsy.

Articles

- Accepting our New Normal, by Liz Marfia-Ash (GRIN2B mom)
- Why I Let Go of Comparing My Child With Developmental Delays to Others, by Brittaney Crider (GRIN2B mom)
- Welcome to Holland, by Emily Perl Kingsley
- On Radical Acceptance (& Not Fixing Your Kid) by Heather Lanier



Books for Adults

- Raising a Rare Girl: A Memoir by Heather Lanier – A memoir about raising a child with a rare syndrome.
- Special by Melanie Dimmitt – Special is an uplifting, candid companion for those in the early stages of navigating a child's disability.
- The Reason I Jump: The Inner Voice of a 13-Year-Old Boy with Autism by Naoki Higashida
- Demystifying Disability: What to Know, What to Say, and How to Be an Ally by Emily Ladau
- Everything No One Tells You About Parenting a Disabled Child by Kelly Coleman

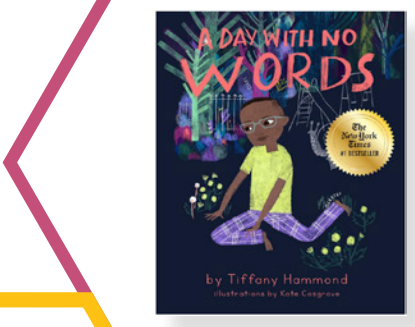
Books for Young Children

These books help to normalize disability and can be read by younger children with GRIN2B and their siblings.

- Some Brains by Nelly Thomas
- Just Right For You, A Story About Autism by Melanie Heyworth
- A Day With No Words by Tiffany Hammond

Books from a Sibling Point of View

- My Brother Otto by Meg Raby
- Just Because by Rebecca Elliot



Podcasts

- Celebrating Rare: A GRIN2B Podcast by GRIN2B parent, Phil Ash
- The Rare Life
- The Parenting Spectrum

Videos/Webinars

- GRIN2B Foundation YouTube Channel
- GRIN2B Symptoms Video #1
- GRIN2B Symptoms Video #2
- GRIN2B Parent Panel - It Gets Better
- Heather Lanier Ted Talk - “Good” and “bad” are incomplete stories we tell ourselves
- Rachel Callander Ted Talk - Superpower Baby Project

Miscellaneous Resources

- State Insurance Guide - from Angelman Syndrome Foundation
- Respite & Caregiver Support - A Mother’s Rest, Angel Aid
- Patient Assistance - Angel Flights, Miracle Flights & National Organization for Rare Disorders
- Seizures - Seizure Action Plan
- IEP resources
- Financial Planning Information
- Librarey - curated resources for rare disease families
- Transitioning to Adulthood

events

Annual

- Rare Disease Day - Last day of February
- GRIN2B Awareness Week - March 12 - 18
- Bee Active for GRIN2B Walk, Run & Roll Fundraiser - Choose any date to participate in September
- Giving Tuesday / Giving Season Fundraiser — Nov-Dec, including Giving Tuesday (Tuesday after Thanksgiving)

Every other Summer (even years)

- **GRIN2B Family Weekend** - Check our website for information on our next Family and Research Conference

Ongoing Events

- **Bee Connected Zoom Meetups for GRIN2B families** - general support and specific topics
- **Educational Webinars**

The GRIN2B Family Weekend exceeded my expectations in every way. Not only was I able to connect with other parents moving through the same journey as our family, but I was able to hear firsthand from many of the doctors and professionals working on GRIN2B. The weekend was perfectly balanced between scientific, research discussions and the social, emotional support I needed as a newly diagnosed family with GRIN2B.

-GRIN2B Parent

how family & friends can help

How can your family and friends support you during this time?*

Our friends and family WANT to help but don't always know how or what to do. It can sometimes feel like one extra burden on our shoulders when we have to make the call, send the text or even think of the thing that would help us, so we scrap it all together and live in overwhelm. This checklist of things can ease the mounds of tasks and give others direct actions that would make a big impact in your day to day. Sharing the following page with your family and friends will give them direct actions to make a big impact in your day-to-day life.

How family and friends can help

(download a print out here)

1 home

- Leave dinner at the door
- Fold laundry
- Put away monthly supplies
- Grocery shop
- Take my car for a wash

2 support

- Sleepover for a night
- Come to my house and send me for a walk
- Get to know my kid, like really know him/her
- Check in on the siblings and take them for an activity
- Bring over yoga mats and stretch with me

3 advocate

- Take an interest in learning about GRIN2B
- Ask for books and podcasts to learn more
- Volunteer and engage at my events
- Teach your Neurotypical kids that disability is a normal part of human diversity

4 hang in

- Continue to invite me
- Understand I am living in crisis mode
- Check in regularly - send a text and be ok without a response
- Keep offering to help even if it's years after the diagnosis and it "appears" that we're fine
- Treat me the way you did before

*Credited to Facebook post by Once Upon a Gene Podcast on 27th July 2022, with additional ideas from the GRIN2B Parent Support Group

stay connected

- **Read our emails.**
- **Make sure our emails aren't caught in your spam folder.**
- **Join our private GRIN2B Parent Support Group on Facebook.** We've got an incredibly welcoming community! (Be sure to answer our questions when you request to join or we cannot approve you). Search for GRIN2B Parent Support Group on Facebook.
- **Follow/Like us on Facebook*, Instagram, X (Twitter), LinkedIn and YouTube.** Just search for GRIN2B Foundation. *Facebook's algorithm is notoriously picky and does not prioritize the posts of smaller nonprofits. The more you comment or like our Facebook posts, the more likely Facebook is to continue showing you our posts in your feed.
- **Add** your family's location to our interactive **map.** *(optional)*
- **Check out the GRIN Portal.** This is an interactive website for families, clinicians and researchers looking to understand GRIN-related disorders. The variant analysis is based on the GRIN Variant Registry above. You can search specific variants and obtain further information such as how many people are registered with that variant.

Visit the **GRIN Portal**

get involved

When you're ready, help us "bee" the change.

- Participate in research studies if you can.
- Sign up for all the patient registries.
- Plan an Awareness Week event.
- Let us know if you have a special skill.
- Join a committee.
- Sign up for our **Bee Active for GRIN2B Walk, Run & Roll fundraiser** in September.
- More ways to volunteer and **get involved.**

home >

appendix

GRIN2B Foundation Resources

GRIN Therapeutics
https://grintherapeutics.com/

GRIN2B Foundation Mission

Mission Statement
https://grin2b.com/who-we-are/

Contact us with questions
https://grin2b.com/contact/

Welcome To GRIN2B

Liz's Blog Post: To the Person Who Just Googled 'GRIN2B' for the First Time
https://themighty.com/topic/grin2b/to-the-person-who-just-googled-grin2b-for-the-first-time/

Understanding GRIN2B - Related Neurodevelopment Disorder

GRIN2B-Related Neurodevelopmental Disorder (National Library of Medicine)
https://www.ncbi.nlm.nih.gov/books/NBK501979/

Common Questions
https://grin2b.com/common-questions/

About GRIN2B
https://grin2b.com/about-grin2b/

Genetics 101
https://grin2b.com/understanding-genetics/

Genetics and GRIN2B
https://grin2b.com/genetics-and-grin2b/

GRIN2B Gene Review
https://www.ncbi.nlm.nih.gov/books/NBK501979/

Common Symptoms Infographic

https://grin2b.com/wp-content/uploads/2019/10/GRIN2B_infographic2.pdf

Treatment Options

Treatments Page
https://grin2b.com/treatments/

Clinical Trials for GRIN2B
https://grin2b.com/clinical-trials/

Five Tips for Coping (Printable)

https://grin2b.com/wp-content/uploads/2025/02/Grin2B_WelcomeBookletProject_5CopingTips.pdf

Support Programs

Support Assistant Grant Program
https://grin2b.com/resources/patient-assistance-grant/

Regional Meetups
https://grin2b.com/regional-family-meet-ups/

Private Facebook Parent Support Group
https://www.facebook.com/groups/grin2bfamilysupport

Family Contact Registry
https://grin2b.com/register/

Awareness Resources

Awareness cards
https://grin2b.com/awareness-cards/

Awareness Presentation for Schools
https://grin2b.com/awareness-for-schools/

Awareness Apparel
https://www.bonfire.com/store/grin2b-foundation-store/

School Info Sheet
https://grin2b.com/wp-content/uploads/2023/05/GRIN2B-Info-Sheet-for-Schools-3.pdf

Recommended Specialists

Recommended doctors - United States
https://bit.ly/CureGRIN-InternationalDocs

Recommended doctors - International
https://bit.ly/CureGRIN-USDoctors

Navigating Medical Appointment Resources

Download MyMejo
https://www.mymejo.com/

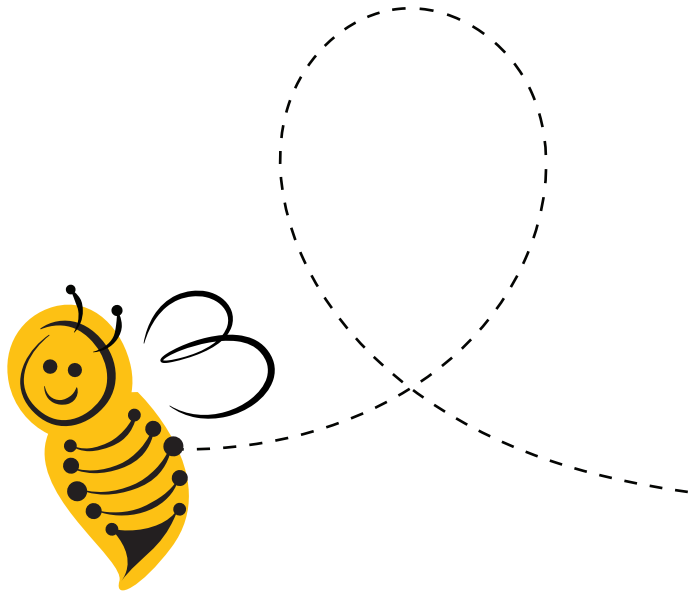
Common Symptoms Infographic
https://grin2b.com/wp-content/uploads/2019/10/GRIN2B_infographic2.pdf

Child Neurology Foundation Toolkit
https://www.childneurologyfoundation.org/child-neurologist-new-visit-toolkit/

GRI Center of Excellence

Children's Hospital Colorado
https://www.childrenscolorado.org/

Center of Excellence Info
https://grin2b.com/center-of-excellence/



Registries to Join

Family Contact Registry
https://grin2b.com/register/

GRIN Variant Registry
https://grin-portal.broadinstitute.org/

Simons Searchlight
https://www.simonssearchlight.org/

Registry Checklist
https://grin2b.com/wp-content/uploads/2023/06/Registry-Information_Sheet_v4-1.pdf

Research

Funded Research by GRIN2B Foundation
https://grin2b.com/awarded-grants/

PubMed to search for GRIN2B Research articles
https://pubmed.ncbi.nlm.nih.gov/?term=GRIN2B

Summaries of GRIN2B articles by Simons Searchlight
https://www.simonssearchlight.org/research/what-we-study/grin2b/

Rare Disease Drug Development Course
https://learn.rarediseases.org/rare-disease-drug-development-series/

Contact Registry
https://grin2b.com/register/

Important Terms to Know: GOF vs LOF

Webinar on GOF vs. LOF Variants
https://www.youtube.com/watch?v=rpkIOLWfpzc

GRIN Europe Info on GOF & LOF
https://www.grineurope.org/what-are-gof-and-lof/

GOF and LOF Infographic
https://grin2b.com/wp-content/uploads/2023/06/GOF-LOF-infographic.pdf

Functional Analysis Tests

Functional Analysis by Emory University
https://academic.oup.com/hmg/article/32/19/2857/7208877

GRIN Portal
https://grin-portal.broadinstitute.org/

GRIN Variant Registry
https://grin2b.com/wp-content/uploads/2023/06/GOF-LOF-infographic.pdf

Autism BrainNet

https://grin2b.com/autism-brain-net/

Blogs and Articles

Diary of a Mom (Facebook page about her adult daughter with Autism, ID and epilepsy)
https://www.facebook.com/adiaryofamom

Accepting our New Normal, by Liz Marfia-Ash (GRIN2B Mom)
https://grin2b.com/2017/04/16/accepting-new-normal/

Why I Let Go of Comparing My Child With Developmental Delays to Others,
by Brittaney Crider (GRIN2B Mom)
https://themighty.com/topic/grin2b/letting-go-comparison-child-developmental-delays/

Welcome to Holland, by Emily Perl Kingsley
https://www.emilyperlkingsley.com/welcome-to-holland

On Radical Acceptance (& Not Fixing Your Kid) by Heather Lanier
https://starinhereye.wordpress.com/2016/07/05/on-radical-acceptance-not-fixing-your-kid/

Books for Adults

Raising a Rare Girl: A Memoir by Heather Lanier
https://a.co/d/bkspV3N

Special by Melanie Dimmit
https://a.co/d/bypEV4o

The Reason I Jump: The Inner Voice of a 13-Year-Old Boy with Autism by Naoki Higashida
https://a.co/d/5KoAQb0

Demystifying Disability: What to Know, What to Say, and How to Be an Ally by Emily Ladua
https://a.co/d/gpNXNET

Everything No One Tells You About Parenting a Disabled Child by Kelly Coleman
https://a.co/d/ijf8rBx

Books for Young Children

Some Brains by Nelly Thomas
https://a.co/d/j6QF3Cq

Just Right For You, A Story About Autism by Melanie Heyworth
https://a.co/d/2CE6O5t

A Day With No Words by Tiffany Hammond
https://a.co/d/cSKbekZ

Books from a Sibling Point of View

My Brother Otto by Meg Raby
https://a.co/d/4hQWgYQ

Just Because by Rebecca Elliot
https://a.co/d/1Gp1r63

Podcasts

Celebrating Rare: A GRIN2B Podcast by Phil Ash (GRIN2B Dad)
https://www.podomatic.com/podcasts/grin2b

The Rare Life
https://therarelife.org/

The Parenting Spectrum
https://www.abc.net.au/listen/programs/the-parenting-spectrum

Videos/Webinars

GRIN2B Foundation YouTube Channel
https://www.youtube.com/channel/UCFx6ZmVpzF1VMGTlYvhdrtg

GRIN2B Symptoms Video #1
https://www.youtube.com/watch?v=LbOjTSSbjvA

GRIN2B Symptoms Video #2
https://www.youtube.com/watch?v=Fc4HEz_p9oc

GRIN2B Parent Panel- It Gets Better
https://www.youtube.com/watch?v=kAmALVH56Xc

Heather Lanier TedTalk: “Good” and “Bad” are incomplete stories we tell ourselves
https://www.ted.com/talks/heather_lanier_good_and_bad_are_incomplete_stories_we_tell_ourselves?subtitle=en

Rachel Callander Ted Talk: Superpower Baby Project
https://www.youtube.com/watch?v=wUYBpsujxdw

Other Resources

State Insurance Guide (from Angelman Syndrome Foundation)
https://9d869b08-6fd3-4df5-bded-e31a2fb4491a.usrfiles.com/ugd/9d869b_12d6571f6d3c43cdb2de9991876e2582.pdf

A Mother’s Rest, Angel Aid (Respite & Caregiver Support)
https://www.amothersrest.org/

Angel Flights, Miracle Flights (Patient Assistance)
https://miracleflights.org/

National Organization for Rare Disorders (Patient Assistance)
https://rarediseases.org/patient-assistance-programs/

Seizures Actions Plan
https://seizureactionplans.org/for-families/

IEP Resources
https://adayinourshoes.com/

Financial Planning Information
https://www.youtube.com/watch?v=UzbOCXdJOQI

Librarey (curated for rare disease families)
https://www.librarey.com/

Transitioning to Adulthood
https://www.viprarecare.com/

Events

Rare Disease Day (Last Day of February)
https://www.rarediseaseday.org/

GRIN2B Awareness Week (March 12-18)
https://grin2b.com/awareness/

Bee Active for GRIN2B Walk, Run & Roll Fundraiser
(choose any date in September to participate)
https://grin2b.com/bee-active-walk-run-fundraiser/

Giving Tuesday (November-December, including the Tuesday after Thanksgiving)
https://www.givingtuesday.org/

GRIN2B Family Weekend (every other summer, even years)
https://grin2b.com/family-weekend-2024/

How Family and Friends Can Help (Printable)

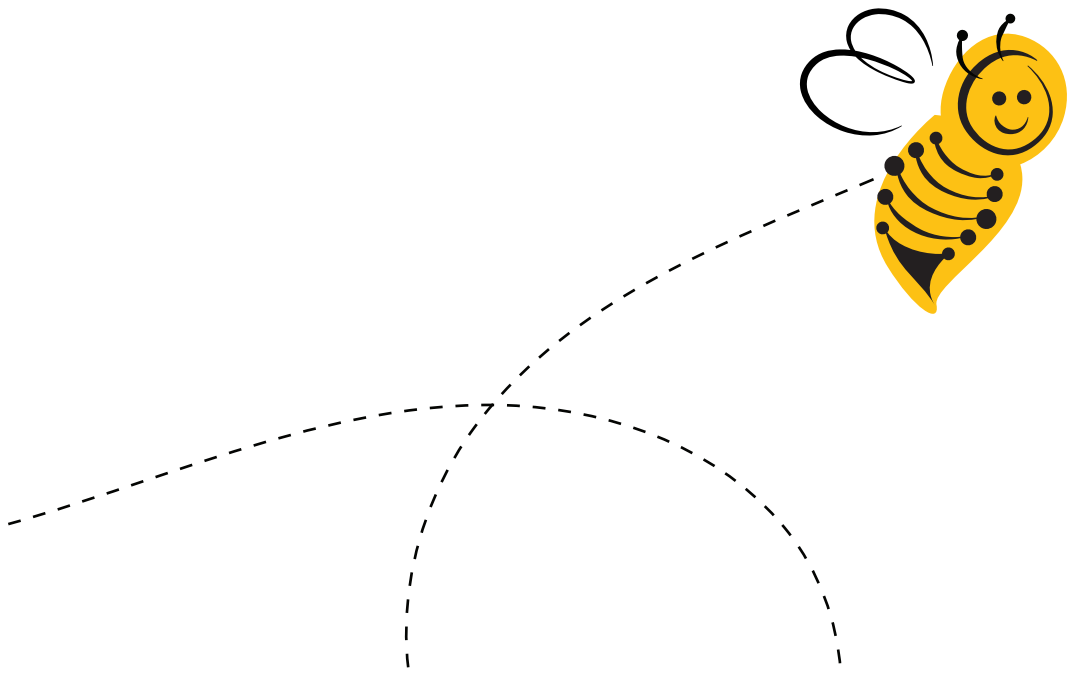
https://grin2b.com/wp-content/uploads/2025/02/Grin2B_WelcomeBookletProject_FamilyFriends.pdf

Stay Connected and Get Involved

Add your Family Location to our Interactive Map (optional)
https://grin2b.com/grin2b-map/

Sign up for the Bee Active GRIN2B Walk, Run, and Roll Fundraiser (September)
https://grin2b.com/bee-active-walk-run-fundraiser/

Volunteer or Get Involved
https://grin2b.com/get-involved-with-the-grin2b-foundation/



May we not just grieve, but give.
May we not just ache, but act.

-Amanda Gorman

GRIN2B Foundation