



Quarterly Newsletter

| January 2021 | Yellow Brick Road Project HNRNPH2 Q4 2020 News

Latest News

Fundraising efforts for Giving Tuesday raised an amazing **\$33,000** towards research into treatment!

Research into potential treatments continues at Andlit Therapeutics and at St Jude Children’s Research Hospital.

The latest publication on Dr Jennifer Bain’s natural history study will appear in the peer-reviewed journal *Neurology Genetics* in the new year.

Merry Christmas & Happy Holidays!

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PROJECT

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or our Facebook page at

www.facebook.com/yellowbrickroadproject

Featured Family: Meet Amber

It was a true diagnostic odyssey but in January of 2020, just one month shy of her 24th birthday, Amber's updated genetic testing revealed an HNRNPH2 mutation giving us the true explanation of her developmental disabilities.



Amber was born healthy after an uneventful pregnancy but we quickly noticed that she had extremely low muscle tone, and she wasn't meeting any of her milestones. Our search for answers began with her first MRI at only five months of age. Genetic and metabolic testing would go on for several years as we looked for specific conditions. Some of the characteristics of both Rett Syndrome and Angelman Syndrome became apparent as she got older including wide spaced teeth and an unusual wide-spaced gait. An atypical Rett Syndrome diagnosis was made and it did seem to fit best despite negative genetic results. Amber had stereotypical hand movements including hands to the mouth, and she also had a hallmark symptom of hyperventilation. However, Amber never regressed in her development. She continued to make steady, albeit, very slow progress over the years.

Amber took her first independent steps just before her fifth birthday, and she began to acquire verbal language between the ages of six and eleven. Her verbal language began through humming the tunes to songs, and with music integrated into all of her therapies, Amber was able to form words and finally, full sentences. She continues to make progress with her verbal communication even today.

As she entered her early 20's, we learned about updated genetic testing. We knew that Rett

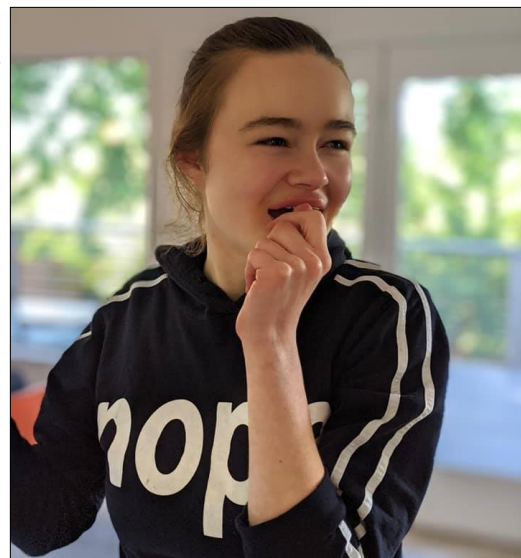
Syndrome was not her true diagnosis, and we also knew that science was moving at the speed of light and if she was going to be eligible for any treatments or interventions that could improve her health, development, or even independence, we would need to know the true cause.

After a two year wait to see the geneticist, we finally had an answer and a new family of parents and children who have the same diagnosis.

We have learned so much in such a short amount of time about the similarities that are incredibly uncanny in so many of the children (and adults) who have an HNRNPH2 mutation. Amber's feet – their shape, the severe pronation, the loose ligaments, etc., were always quite unique to her. Suddenly, we see that this is a hallmark feature in those with HNRNPH2 mutations. Furthermore, the role that music plays on the development of these individuals is astounding! Facial characteristics, gait patterns, visual issues, anxiety, feeding issues, sensory issues, and even positioning when holding a tablet or music device are all common among them.

Since January, Amber has been fitted with glasses. We've modified her environment for cortical visual impairment, and thanks to this group and the collective knowledge and sharing, we are learning new ways to help Amber to make progress in her learning and development. We also know that research that will lead to those important treatments and interventions is within reach and that Amber may also benefit even in adulthood.

We are so grateful that we pursued updated genetics testing and we're thrilled to see the progress that will be made for all who are living with an HNRNPH2 mutation.



Spotlight On: Creating a Plan for Independent Living

For many of us, thinking about independent living for our children is overwhelming and even impossible to imagine. However, understanding what independent living means can help us make better decisions. In defining independent living, we must examine our culture, our family values, our country and/or state's laws or rules, and more. In the United States, children who have disabilities and who receive special education services in public schools are required to have an Individualized Education Plan (IEP) that includes transition to adulthood goals by the age of 16. These goals must be focused on "post-secondary education, employment, and independent living when possible." It's the "when possible" section that isn't defined and may even create confusion for families and youth with disabilities. When we consider independent living as a philosophy, we can begin to understand that it is possible for everyone. After all, most of us are dependent on others throughout our lives. Very few of us live in complete isolation, and those who do, do so by choice which brings us to the definition or philosophy of independent living – it is all about choice. When we can make our own choices and those choices are honored by those who support us, we are exercising our independence. Making an assumption that one must live completely on their own, working a traditional 9:00-5:00 job, communicating verbally, and managing all aspects of self-care is what prevents people from realizing they can achieve independent living. Ensuring an individual has the supports and accommodations they need in adulthood is critical to their independence no matter what their living arrangement may be. With all of this in mind, a plan can be made for a healthy, happy, and productive adult life with as much independence as possible.

When planning, if your child is still very young, it is important to determine what is important to you for their adult life. Determine what you hope for their future and at the same time determine what it is you do NOT want for them. As they get older, you may find these things may change or evolve, and you may include them in this decision-making process.

To begin that process, be aware of terminology, programs, and services for individuals with disabilities. Each of these focus on the strengths and needs of an individual as they transition to adulthood.

Some of these include:

- Person Centered Planning
- MAPS (Making Action Plans)
- Supported Decision Making
- Guardianship and Alternatives to Guardianship

Furthermore, be aware of your country's or your state's laws when it comes to saving money for individuals with disabilities, developing budgets that may be paid for by the government or yourself when that is not an option, and be sure you understand what housing may look like for people with disabilities.

Consider the following scenarios as far as housing/living arrangements go:

- He or she would like to stay with us in our home and we like that too.
- We may need additional supports in our home.
- We may have access to funds that can help us care for our adult child.
- We may have access to day programs to allow for social opportunities and/or supported meaningful employment.
- He or she would like to live outside of our home.
- We might consider if we have the ability to add on to create a separate space.
- We might consider how renting from an outside source along with roommates and additional supports.
- We might determine that a group home or other setting is most appropriate.
- Other options?

In the US, there are laws that impact people with disabilities and their ability to access services and supports throughout their lives. There are also similar laws in countries around the world.

- Americans with Disabilities Act
- The Rehabilitation Act of 1973
- The Individuals with Disabilities Education Act
- The Workforce Innovation Opportunity Act

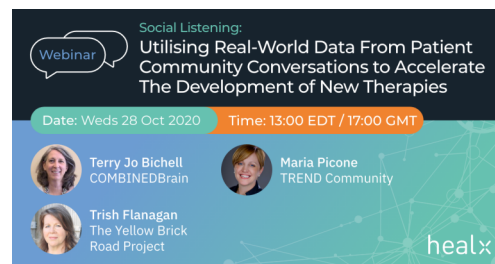
The Disability Rights Education and Defense Fund (DREDF) has compiled a comprehensive list for international laws: <https://dredf.org/legal-advocacy/international-disability-rights/international-laws/>

To learn more about any of these laws, resources, programs, etc., please feel free to contact Angela Lindig, Amber's mom, at angela@ipulidaho.org

Conferences and Webinars

As COVID-19 continues to affect all aspects of our lives, the Board and Committee members of the YBRP have adapted to attending and presenting at conferences and webinars online. These events are an important way for us to keep up-to-date on the latest developments in genetic research and drug development, as well as a way to continue to spread awareness of HNRNPH2 mutations and the work of the YBRP. Just a few of the events we have participated in over the past few months include:

- ◆ World Orphan Drug Congress USA
- ◆ NORD Rare Summit
- ◆ World Orphan Drug Congress Europe
- ◆ Healx Webinar (Trish Flanagan co-presented on *Utilising Real-World Data from Patient Community Conversations*)
- ◆ Virtual Rare Disease Showcase (Stacy Paddon took part in a panel on *Building Patient Communities in an Isolated World*)
- ◆ Northwest Rett Syndrome Association (Angela Lindig presented the webinar *When It's Not Rett Syndrome After All*)
- ◆ RareFEST 2020 (*Quest to a Cure* video showcased)



RARE Champion of Hope Award Nominee

The Yellow Brick Road Project, parent led patient advocacy organization for HNRNPH2 neurodevelopmental disorder, was honored to be recognized in this Global Genes nomination as "groundbreakers, leaders and advocates who inspire and catalyze change in rare disease." Congratulations to the other nominees and winners. All boats rise, and we are proud of your advocacy and work that informs and furthers our own. <https://globalgenes.org/rare-champion-of-hope-award-nominees>



NORD Member Social Media Contest

YBRP has entered a video in NORD's 2021 Member Social Media Show Your Stripes Contest! Submissions will be showcased on NORD's facebook page thru Rare Disease Day at the end of February. The winner will be decided based on likes, comments, and shares, so when you see YBRP's Show Your Stripes video, be sure to do all of the above and help us bring home the grand prize! Click here to go to [NORD's Facebook page](#) and give them a follow so you don't miss it!



Fundraising Update

The grant that the Yellow Brick Road Project has awarded to Andlit Therapeutics is to provide \$420,000 for research into HNRNP2 mutations and potential treatments.

In July 2020, the first payment of \$150,000 was issued from funds that were raised in previous years. Since then, through the efforts of our wonderful parents and partners, we had succeeded in raising an additional \$110,000.

We still need to raise an additional \$160,000 by July 2021 in order to continue funding this important work.

There are many ways to get involved and contribute to our fundraising efforts. Please visit our website at www.ybrp.org for all the details or read on to learn more about two of these efforts.

Bricklayer Program

In our last newsletter, we introduced you to the Bricklayer Program, YBRP's new corporate sponsorship program. We are delighted to announce our first two sponsors under this program:

RUBY LEVEL: Haywood EMC

GOLD LEVEL: Mager Law Group

For more information about our sponsors and about the program in general, including how to get involved, please visit <https://yellowbrickroadproject.org/pages/bricklayer-program>

Corporate Matching

Another important way we can encourage corporate donations is to look for matching programs at the companies our donors work for. Many businesses offer to match donations made by their employees to charitable organizations such as the Yellow Brick Road Project. Usually this is on a 1:1 basis to a maximum amount, ie. if the employee donates \$500, the company will also donate \$500, but sometimes the program can be even more generous.

So far in 2020 alone, we have received three corporate match donations worth more than \$5000.

This is an easy and effective way of increasing the impact of your donation! If you are considering making a donation to YBRP, please check with your company's HR department to see if a matching program is available. YBRP is more than happy to provide any assistance needed with completing forms or providing any information that's needed.

Amazon Smile

Don't forget that using AmazonSmile is also a great way to increase donations to the YBRP, at no additional cost to you! Any purchases made through AmazonSmile (which has all the same products and prices at Amazon.com) come with a donation of 0.5% of the purchase price to the charity of your choice.

On your first visit to AmazonSmile - smile.amazon.com - you'll be asked to select a charitable organization. Please search for "YBRP Inc".



Giving Tuesday

Giving Tuesday 2020 took place on Tuesday, December 1, and it was an amazing success!

Thanks to the dedication and generosity of our families and donors and their extended support network, we raised an amazing **\$33,000** towards our goal of fully funding the research project at Andlit Therapeutics.

Nearly 20 individual fundraisers were set up through Facebook with many additional donations coming directly through the YBRP website.

This shattered our previous Giving Tuesday fundraising record of \$16,000 which was set in 2019.

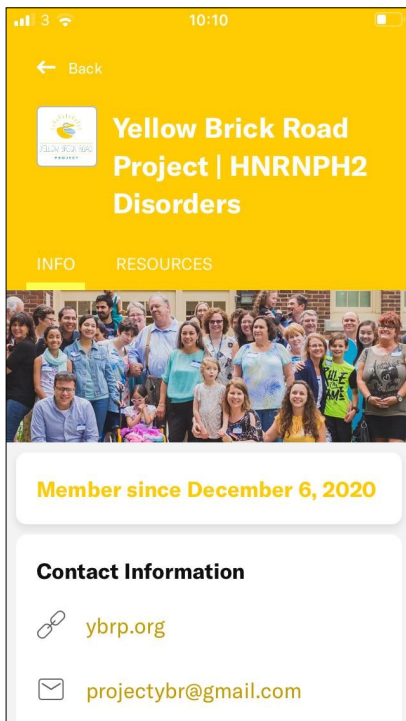
We really couldn't do any of the work that the YBRP does without all of your support.

Our next major fundraising drive will take place in conjunction with **Rare Disease Day** on **February 28, 2021**. Please watch the YBRP website and Facebook page for all the details on how you can get involved.



Backpack Patient Registry

Earlier this year, we shared with you our plans to begin collecting patient data through a company called **Backpack**. The official Yellow Brick Road Project / HNRNPH2 Disorders patient group was launched on Backpack in December 2020, and we have already signed up more than 10% of our patients through a soft launch which took place that month.



A full roll-out to the remaining patient families will be taking place in the early part of 2021, so if you have not yet signed up, don't worry! YBRP will provide full details on how to get your data shared very soon.

Building our own patient registry with information on exactly how the disorder affects our patients will be vital as we continue to move forward with research towards the very exciting goal of conducting clinical trials on potential treatments. In order to understand how effective treatment is, we need to be able to accurately measure the impact on our patients against a baseline of how they were affected prior to treatment.

Backpack is free to use, available through an app or through their website, and can be accessed in English, Spanish, French, Portuguese, German and Italian.

All information shared with YBRP through Backpack is **de-identified**, meaning that no information can be linked to any specific individual. Its purpose is to provide a better understanding of the disorder overall.

Natural History Study Update

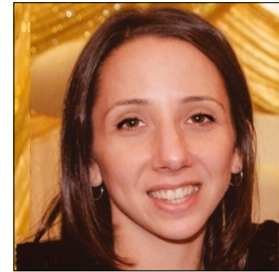
The newsletter will contain a regular update from the research team conducting the HNRNPH2 Natural History Study. Dr Jennifer Bain is the principal investigator and Olivia Thornburg is her Research Assistant. They are based at Columbia University in New York City, USA.



Dr Jennifer Bain

What is a natural history study?

A natural history study follows a group of people over time who have a specific medical condition or disease. The study collects health information in order to understand how the condition develops and how to treat it. It can be used as a baseline to determine if proposed treatments are effective.



Olivia Thornburg

Since the initial 6 individuals were identified, we have enrolled 50 additional cases of individuals with variants in hnRNP H2, although many more individuals have been identified. The majority of individuals affected are girls, although we have confirmed variants in 5 boys. Our research has employed the use of a prospective testing battery where we administer assessments remotely to test for various neurologic, behavioral, psychiatric, developmental, and social factors in this genetic group. We have also gathered information from former genetic reports and medical records. Our virtual family meeting this year was incredible. We were so ecstatic by the turnout. Of the 41 families who attended the meeting, we had the opportunity to conduct physical and neurological exams on 25 individuals, occupational therapy assessments on 20 individuals, physical therapy assessments on 14 individuals, and gait analysis on 11 individuals. These testing measures revealed similar clinical symptoms to that of the original cohort, including atypical facial features, neurologic abnormalities, atypical gait, and abnormal muscle tone, among other clinical findings. Our research manuscript was recently accepted to the journal of Neurology Genetics, and will soon be publicly available! We are immensely grateful for your contribution and hope to give back what you have so generously given to us.

We invite families with hnRNP H2 variants to help us improve our understanding by contacting us and partnering. We need to better understand why changes in hnRNP H2 causes problems with brain function so we can develop new treatments. Please see our posting on [clinicaltrials.gov](https://clinicaltrials.gov/ct2/show/NCT03492060?cond=hnrnp&rank=1) for more information on our study: <https://clinicaltrials.gov/ct2/show/NCT03492060?cond=hnrnp&rank=1> or reach out to us at hnrnp@columbia.edu.