The HCU *Herald*

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HCU HERO: GRAYSON FROM COLORADO

My name is Brittany, and I live with my husband, Robert, and three children in Colorado. My family's journey into the rare disease community began suddenly and without warning almost nine years ago. We were living on the East Coast and had just given birth to our second child and first son.

Drew was born on a beautiful spring day, May 18, 2011. We welcomed him into our lives with all the love a child could possibly experience. To us he was perfect. However, as the days and weeks progressed, we began to grow concerned about certain things we noticed in his development. We frequently shared these concerns with our pediatrician only to be told that our concerns were invalid and that there was nothing wrong with our son.

Our whole world changed just a few days after his 2 month well child visit when Drew had his first life-threatening seizure. This episode caused him to stop breathing altogether due to the stress on his little body. I performed CPR on him in a desperate attempt to keep him alive until the ambulance could arrive. He was rushed to the emergency room of the local hospital, but once again, our concerns were dismissed and the doctors told us that he must have simply had acid reflux.

Less than a week after being discharged, Drew suffered another seizure again requiring CPR to stay alive until an ambulance could arrive. Unbelievably, and in spite of our adamant insistence to the contrary, doctors continued to believe that acid reflux was the cause. As a result, they kept him overnight only for observation but did not insert an IV, a choice that almost cost him his life. The following morning Drew experienced yet another seizure, but because he didn't have an IV, there was no way for doctors to administer life-saving medication to quickly control his seizure. It took a team of doctors to resuscitate my son. Finally, the doctors began to listen to our concerns.

What ensued thereafter was three long weeks in the Pediatric ICU involving MRI's, EEG's, NG and G tubes, multiple spinal cord taps, countless blood draws and eventually having to be placed on a ventilator to stay alive. Despite all of these efforts, the stress on his little body became too much. Drew passed away on August 27th 2011 at the age of 3 ½ months. He had suffered severe brain atrophy caused by an undiagnosed ultra-rare metabolic disorder. His life lasted for 100 days... 2,402 hours and then he was gone.

Nearly two full years after Drew's death, and after multiple genetic tests and thousands of dollars in testing costs, we finally discovered that our son had been affected by a disorder known as Homocystinuria caused by a Cobalamin defect. However, we still had no way of knowing what sub-type of this disorder had taken his life.

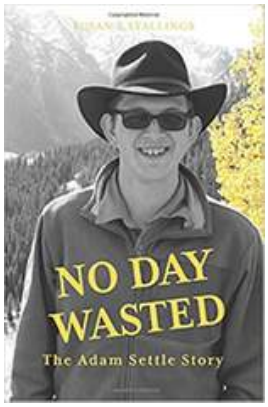
It wasn't until seven years later in March of 2019 that we were finally given a definitive diagnosis: Homocystinuria with a Cobalamin G defect. We received this knowledge because we gave birth to our fourth child and second son, Grayson, who suffers from the same disorder. As a result of thoughtful doctors who listened to our concerns during pregnancy and thanks to improved technology, immediately upon Grayson's birth we were included in a research study at Children's Hospital Colorado that shortened the genetic testing turnaround time from twelve weeks to three days and provided the results that we had sought for so long. Finally we were given answers that could explain why our son had died eight years earlier. These same answers provided a roadmap for how to treat Grayson so his outcome would be better than Drew's. While we had previous knowledge of a genetic mutation for CblG that I was a recessive carrier for, we were told that the mutation that my husband carries is a novel mutation (which is why earlier testing did not detect it). Dr. Rosenblatt in Canada confirmed the diagnosis just a few months after Grayson was born.

Homocystinuria is a genetic metabolic disorder that causes toxic levels of the amino acid Homocysteine to build up in the blood and urine. When Homocystinuria is caused by a cobalamin defect, the body cannot properly metabolize cobalamin, more commonly known as vitamin b12, leading to elevated levels of Homocysteine and abnormally low levels of Methionine, both of which have serious health implications. Without early treatment, Homocystinuria can cause serious, life threatening issues. Even with treatment, individuals living with this disorder may experience vision problems, seizures, developmental delays, anemia, heart disease or blood clots as well as feeding and growth problems. Early detection and treatment are critical to success in treating this disorder.

Grayson is now a year old and he has endured aweeklong stay in the NICU, an MRI, EEG, regular visits to metabolic specialists, pediatric ophthalmologists and neurologists, weekly blood draws to check levels of amino acids, daily injections of a specialized form of B12, and three different medications and supplements taken at each meal. While the medication tastes terrible, gives him an upset stomach and causes bad acid reflux, we have had to be creative in how we administer it. We have currently discovered a miracle elixir, Sunny Delight. While the sugary drink has absolutely no nutritional value, it somehow masks the bitter, disgusting taste of the medication. For that we are grateful because it means no tears or meltdowns at meal times…for Grayson or me.

We count our blessings every day that Grayson continues to grow and develop. He currently has occupational therapy through the early intervention program to monitor his development. He loves to be outside on his little playground and play with his sisters. We are especially grateful to be living in Colorado where we have an excellent team of doctors that monitor his growth and development on a regular basis.

BOOK RELEASE:
NO DAY WASTED: THE ADAM SETTLLE STORY



No Day Wasted: The Adam Settle Story
By Susan F. Stallings

Waste no day to achieve the impossible! When Pennsylvania started testing newborns for metabolic diseases, Adam Settle was one of the first to be diagnosed with Cobalamin-C Deficiency. Legally blind and mentally delayed, Adam never let his limitations hold him back from accomplishing his dreams. His love for people and his desire to help others comes through the pages of this book. Whatever has been holding you back, this book will help you charge through the obstacles you face to achieve the impossible. Adam's story will inspire, encourage, and give you hope.

Interview with Adam Settle

How did the idea of writing *No Day Wasted* get started?
The idea came from encouragement from others to tell my story. After the third person within a week, I decided it was time.

Can you tell us about the process of having No Day Wasted written?
I was at an Engage Conference with 99 Balloons in Arkansas with a friend Ryan. He said to me, Adam, you don't waste a day! And the next thing I knew we decided that should be the name of the book!

What was the hardest part having the book written?
Waiting and deciding how to publish the book!

You have lived a very full and exciting life, and you aren't that old - how did you decide what parts to have written about and what parts to hold back?
The author decided we would start at the beginning of life, which of course was a rocky one! All the way to graduation from high school and all the adventures in between!!

We encourage patients to share their stories, what would you say to patients and caregivers who might be a little bit hesitant to share their/their child's story?
When you share your life story, others know they are not alone! We are all loved and uniquely made. We all need encouragement and hope! I hope my book touches hearts and changes attitudes! Everyone has a story to share!

You can buy your copy now on Amazon (https://www.amazon.com/No-Day-Wasted-Settle-Story/dp/B088GMJZJV/) Or look for HCU Network America's book give away opportunity on Facebook/Instagram/ and Twitter in June

EVENTS

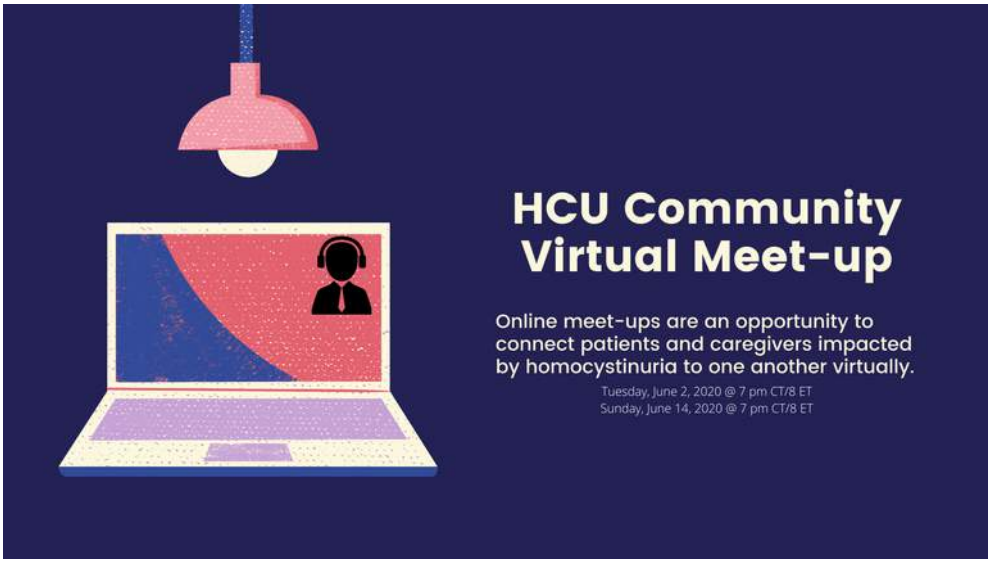
Come check out our Virtual Homocystinuria Meet-ups!

Join our virtual meet-up for a chance to meet, connect, and learn from other patients and caregivers who are facing similar challenges. Whether it's navigating adherence issues, insurance, clinic visit, or life transitions, you are not alone.

Space is limited, so make sure to register early! Once registered, you will receive additional details about accessing the meet-up.

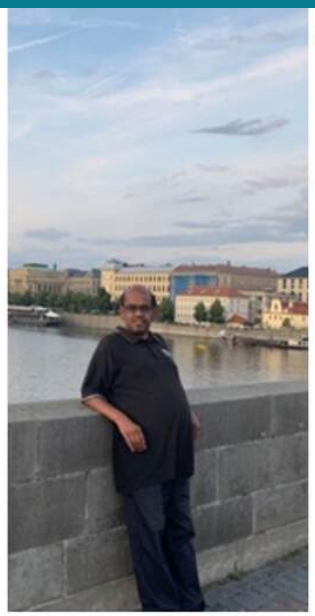
Register now at: https://www.eventbrite.com/o/hcu-network-america-30163980100

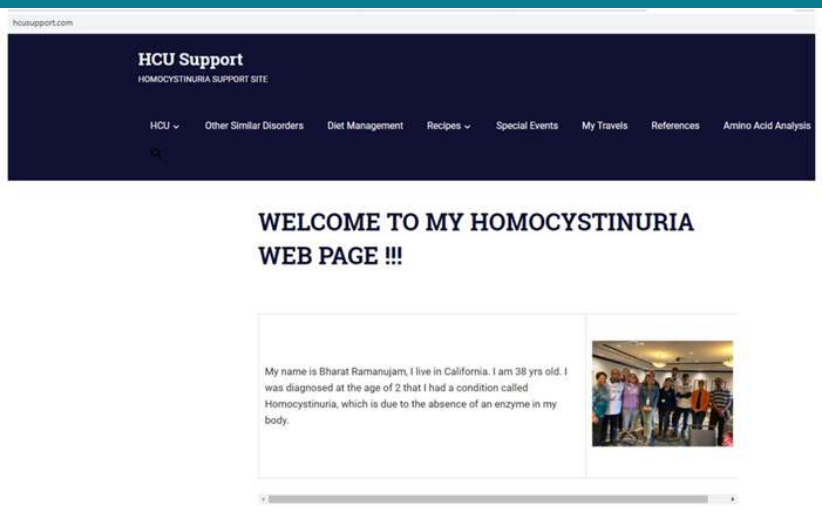
For Classical Homocystinuria



For Methylmalonic Acidemia with Homocystinuria patients and caregivers (different than Classical HCU)

HCU SUPPORT WEBSITE RELAUNCH





What inspired you to create the original HCU Support website?
In 1999 I was extremely sick in intensive care unit with my seizures. Even though my HCU levels were maintained, I have had multiple fractures over the years, with a surgery recently. I have had seizures until 2015 . Dr Koch encouraged our family to do something, sharing our experiences, due to lack of support for HCU. With my father's help my mother created Lowprotein.com and I created hcusupport.com

When you originally created the website what was your hopes and goals for it?
My family and I were hoping to form the Homocystinuria Community. Then it was hard to reach other people because internet was not like it is right now.

What are your goals for your website now?
I am finally able to update my website; it has been transferred to a different platform using WordPress instead of Microsoft Frontpage, which is not used anymore and people from other countries couldn't access the website. You can view the new site at hcusupport.com.

I want my website to be a reference and help for other HCU families throughout the world. When my father finishes his software for nutrition it will be helpful for everyone to manage their.

Before the internet, had you connected with HCU patients? How has your website and other social media platforms like Facebook, Twitter and Instagram changed how you connect with patients?
My family and I attended two conferences where they had an HCU session, and met other families with HCU.

I now have more contact via social media platforms such as Facebook, Instagram, Twitter, LinkedIn and WhatsApp . I also enjoy talking with other HCU people on Zoom.

School's out! The kids are home and it seems they are asking," what's there to eat" every 5 minutes. We have put together a Low Protein Snack Off Challenge. Each week we will be putting out multiple snack face offs for you to vote on. Follow along on Facebook to make your vote count.

We will be sharing links of various low protein products and recipes to go with specific challenge items as well.

Vitaflo's range of medical foods offers a variety of options for Homocystinuria with convenient packaging and 3 versatile formats to fit the lifestyle demands of modern living

Vitaflo®
Innovation in Nutrition
A Nestlé Health Science Company

HCU gel™

Designed to prepare a semi-solid (gel/paste) consistency or low volume drink!

- Suitable from 1 year of age
- 1 packet provides 10 g protein equivalent
- Neutral taste (Unflavored) allowing flexibility in flavoring

HCU express®15
HCU express®20

Provide flexibility to prepare a variety of delicious low volume drink options!

- Suitable from 3 years of age
- Designed to be mixed with approximately 3 fl oz water or other permitted beverages to prepare a low volume drink
- 1 packet provides 15 g or 20 g protein equivalent
- Neutral taste (Unflavored) allowing flexibility in flavoring

HCU cooler®15

Portable ready-to-drink options on-the-go to school, work or travel!

- Suitable from 3 years of age
- 1 pouch provides 15 g protein equivalent
- Available in Orange and Red flavors!

FOR REIMBURSEMENT ASSISTANCE, VISIT:

formula4success™
www.Vitaflo4Success.com

USE UNDER MEDICAL SUPERVISION
Consult your healthcare professional to determine which Vitaflo products are appropriate for your diet.

Visit VitafloUSA.com to request a sample!

*Advertisements are not an endorsement of products advertised

HCU COMMUNITY COOK BOOK

Jalapeno Poppers

Makes 6 servings

Ingredients:

- 84 g Cambrooke Cream Cheese, room temperature
- 1/2 strip(s) MorningStar Veggie Bacon Strips, cooked and cut into pieces
- 20 g Violife Just Like Cheddar Shreds
- 1/2 clove(s) Garlic, minced
- 1/4 tsp Salt
- 1/8 tsp Black Pepper
- 36 g Raw Jalapeno Peppers

Topping:

- 2 TBSP Cambrooke low protein bread crumbs
- 4 g Follow Your Heart Parmesan Style Shredded

Nutritional Information

- Serving size: 1 Popper
- Protein per serving: 0.5 g
- Calories per serving: 50

Directions:

1. In a small bowl add the cream cheese, bacon, cheddar shreds, salt, pepper, and garlic. Gently mix to combine. Cut the tops with the stems off the jalapenos. Then cut in half lengthwise and scrape out the seeds. Fill each jalapeno with cream cheese filling. Place filled jalapenos in the freezer for 10 minutes.
2. Combine the bread crumbs and Parmesan shreds in a small bowl. Set aside. Preheat oven to 375 degrees and line a baking sheet with foil. Gently spray foil with nonstick cooking spray.
3. Remove the jalapenos from the freezer. Dip each filled jalapeno, filling side down, into the topping and gently press to make sure topping sticks. Place onto prepared baking sheet. Bake for 15 minutes. Then turn oven to LOW broil and broil until the topping is golden brown. This should only take about one to two minutes so watch them closely. Allow to cool for 5 minutes before serving.

HCU COMMUNITY COOK BOOK

Cherry Cobbler

Nutritional Information
- Serving size: 145 g
- Protein per serving: 1.1 g
- Calories per serving: 346

Makes 6 servings

Ingredients:
- 4 c Cherries, frozen
- 1/2 c Brown Sugar
- 1 tsp Vanilla Extract
- 1 tsp Almond Extract
- 1/8 tsp Salt
- 2 tsp Cornstarch

Topping
- 1 1/2 c Cambrooke MixQuick Baking Mix, gently packed
- 30 g Instant Vanilla Pudding, dry mix only
- 6 TBSP Butter, cold, sliced thin

Directions:
1. Preheat oven to 350 degrees. In a 10 inch cast iron skillet add all the cherries, brown sugar, vanilla extract, almond extract, and salt. Place skillet over medium heat and cook until heated through and sugar has dissolved. Bring to a boil. Add the cornstarch and stir well. Cook until thickened. Remove from heat.
2. In a medium bowl add the MixQuick and pudding mix and gently mix to combine and break up lumps. Dump the mix over the cherry filling and spread evenly to cover. Place slices of cold butter all over the topping. Bake in oven for 30 minutes. Remove and let cool for 5 minutes. Serve warm with your choice of nondairy ice cream.

CALLING ALL PATIENTS WHO WERE MISSED BY NEWBORN SCREENING AT BIRTH!

WE HAVE AN OPPORTUNITY TO HELP CHANGE THE PROCESS BUT NEED YOUR STORY TO GIVE US THE EVIDENCE TO BUILD OUR CASE

But we have newborn screening For HCU...

According to recent statistics, approximately 25-50% of patients are missed by newborn screening for Homocystinuria. There are multiple factors that can play into these numbers. Currently it is federal mandate that all states screen for Homocystinuria through the newborn screening test, but there are no set standards. Meaning, every state or region can set their own methionine cut offs. A handful of states also do tier two testing—meaning they have a second round of newborn screening, making it more likely for homocystinuria to be picked up. Another factor that plays into the effectiveness of the test, is how elevated the patient's levels are at the time of the test. Patients who are pyridoxine (B6) responsive, or have more functioning CBS enzyme, are less likely to be picked up by the newborn screening.

So how can you help?
If you or your loved one were missed at screening, we need to hear from you ASAP so we have enough evidence to bring about change. Contact Danae if you can help us, and she will lead you through the process that is outlined below.

Talk to your geneticist about the newborn screening survey and urge them to complete it! This will help us build support for changes to the process to increase the likelihood that HCU patients will be diagnosed at birth.

On the following page you will find the letter portion. We ask you to give to your clinic, followed by the survey form:

To Whom this may concern,

I would appreciate your support in answering a brief survey to help support efforts to improve newborn screening for classical homocystinuria.

I have been working with HCU Network America, a patient advocacy and support group for Homocystinuria (HCU), for whom I serve as a medical advisor. One of their key goals is to improve newborn screening for HCU, as it is estimated that over half of patients are missed by the current screening process and often are not diagnosed until they have developed serious clinical symptoms. To build support for an improved process, we are collecting information on patients missed by the current screening process, which we intend to then publish in a consolidated case report.

Could you please support our efforts by completing the attached brief questionnaire, and sending it to me vie-email at: FICICIOGLU@email.chop.edu

Sincerely,
Can Ficicioglu, M.D., Ph. D.
Director of Newborn Metabolic Screening Program, Children's Hospital of Philadelphia

Survey on Classical Homocystinuria (HCU) Patients Missed by Newborn Screening

Do you have any patients with classical HCU missed by NBS and diagnosed later based on symptoms?
( ) Yes ( ) No

If yes, at what age were the patients diagnosed, and what year were they born and in what state?

Age at diagnosis (mos.) _____ Year of birth _____ State born_____
Age at diagnosis (mos.) _____ Year of birth _____ State born_____
Age at diagnosis (mos.) _____ Year of birth _____ State born_____
Age at diagnosis (mos.) _____ Year of birth _____ State born_____

Would you be willing to provide information to contribute to a "Case Report" we plan to publish on patients missed by Newborn Screening?

What is the name and address of your clinic and the best contact person for further information?
Clinic Name:
Clinic address:
Contact Person:

- Name
- E-mail
- Phone

Please send completed survey to Dr. Can Ficicioglu at Ficicioglu@email.chop.edu
Or complete the survey online: https://hcunetworkamerica.org/survey-on-classical-homocystinuria-patients-missed-by-newborn-screening/

HCU NETWORK AMERICA

IS LOOKING FOR STATE AMBASSADORS

Looking for active and outgoing members of the HCU community

What does an ambassador do?

Ambassadors...

- *Connect with local HCU families*
- *Share their story*
- *Advocate and raise awareness for HCU*
- *Amplify and support our mission*
- *Help fund-raise*

Get involved today! Contact Danae'
dbartke@hcunetworkamerica.org

BECOME A STATE AMBASSADOR FOR HCU NETWORK AMERICA

HCU Network America

FUNDRAISING TIP

Every year come May and June we all start to see the neighborhood garage sales pop up! Typically we share how to host a charity garage sale this time of year, but many states are still not allowing these activities to take place, so we thought we'd give you another option – Online Charity Garage Sales!

There are many ways to do this – Facebook Marketplace, Craigslist, Let Go, Offer Up, Varage Sale and those are just to name a few! Each has their own unique set up and audience, but they all allow you to sell items you have and want to part with. Another option is to create a page on Facebook and list all your items with pictures and prices, people can browse your pictures and then tell you what they would like. You can arrange a pick up at your place, a drop off at their place, or a mutual meeting place (we suggest the latter).

Here are some pros to having an Online Garage sale vs an in person one.

- Minimal to no heavy lifting
- No after clean up
- Bad weather resilience
- Opens up a larger audience

How to get started:

- Organize your items (start by the initial organization – donate, throw away, sell. After, organize items into categories, this will be important if you have a lot of items to sell).
- Find a platform
- Write detailed item descriptions
  - People like to know the size, make, model (where appropriate) and condition
- Take good pictures
  - Try to set your items to a white, or plain background
    - Minimize visual distractions. People will more likely pass an item if it's hard to tell what they are looking at.
- Set boundaries
  - People are likely to try and negotiate a price. Know where you want your bottom line to be ahead of time. If things aren't moving maybe reconsider the price
- "Advertise" share your virtual garage sale on your social media platforms and with friends and family

orphan technologies

OrphanTechnologies.com

OT-58

Enzyme Replacement Therapy

Clinical Trial

Orphan Technologies has initiated a first in human study of OT-58, an enzyme replacement therapy that addresses the underlying enzyme deficit for patients living with classical homocystinuria. The goal of this trial is to evaluate the safety and efficacy of OT-58 in patients with classical homocystinuria and identify the appropriate dose. Patients between the ages of 12 and 65 years of age with classical homocystinuria may be eligible to join. For additional information on criteria for eligibility, please go to:

https://clinicaltrials.gov/ct2/show/NCT03406611?cond=Homocystinuria&rank=1

There are four sites in the US currently participating in the trial:

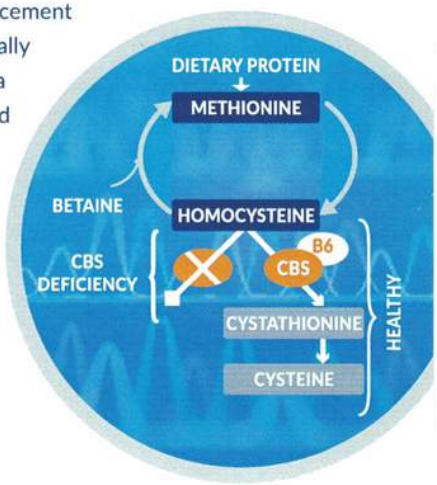
- **Children's Hospital of Philadelphia** – open to patient enrollment
- **Boston Children's Hospital** – open to patient enrollment
- **Indiana University** – open to patient enrollment
- **Children's Hospital Colorado** – open to patient enrollment

Payment for time and travel may be available to patients who participate in this trial.

To inquire about participation into the trial, please email: info@orphantechnologies.com

OT-58 is designed to address the underlying cause of classical homocystinuria which is a deficiency in the naturally occurring enzyme cystathionine beta synthase (CBS).

Reduced or total lack of CBS activity results in the inability to metabolize, or process, homocysteine. This can lead to dangerous elevations of homocysteine. OT-58 is a modified recombinant enzyme replacement therapy that may dramatically decrease tissue and plasma levels of homocysteine, and as a result may prevent, delay, and reverse clinical abnormalities, as well as reduce dietary restrictions.



orphan technologies

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COMMITTED

to improving the quality of life for patients with homocystinuria

GET WELL SOON, JORDAN !

Jordan is 23 years old and from Louisville, Kentucky. He was diagnosed with HCU when he was 3 years old. Recently, Jordan was also diagnosed with Leukemia (ALL) and began his treatment in Louisville. Due to the aggressive nature of leukemia and the impact chemotherapy has on HCU he was transferred to Cincinnati Children's Hospital for more comprehensive care. Overall, Jordan has been in the hospital for 35 days and they're currently estimating another month of being in the hospital. While he's there, our goal is to fill an entire wall of his room with cards. If you click this link and send an e-card the hospital with print it and deliver it to him! Please join us in a good cause!! Don't forget to add your name and the state/country you're sending from.

Jordan Neutz / A507
https://www.cincinnatichildrens.org/patients/resources/support/ecard?

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