The HCU Herald

Featuring...

HCU Hero

Andrea from Texas

HCU NETWORK AMERICA

All things Homocystinuria: patient stories, resources, research, events and more!

HCU HERO: ANDREA FROM TEXAS

The Diagnosis

Andrea was admitted to the hospital when she was just four days old and taken to the neonatal intensive care unit (NICU).

Before that, I already felt something wasn't right. She was very lethargic, slept almost all the time, and had little energy. She couldn't feed well, she would fall asleep every time, whether I tried to breastfeed or give her a bottle. We tried everything to wake her up, to keep her alert, but nothing worked. She also had mild jaundice, though not enough to need treatment, so her pediatrician was checking her daily, but there was no improvement.

The day we decided to take her to the hospital, we had just received an abnormal newborn screening result, specifically from the PKU test. Combined with her symptoms, we rushed her to the emergency room.

When we arrived, the nurses tried to reassure us, saying it might just be a false positive. But when the attending doctor came in, she told us something that changed everything:

"We're going to have to admit her. This is something very rare, something we don't really know. We need to investigate and monitor her closely."

HCU HERO: ANDREA FROM TEXAS

That's how our NICU journey began, two long weeks of uncertainty, fear, and hope.

At first, every doctor and nurse told us the same thing: "It could be a false positive." With IV fluids, Andrea started waking up a bit more and feeding better. I began pumping milk, tracking every ounce, every feeding, every diaper. She seemed to improve, and everyone said we'd probably be going home soon. But that didn't happen.

We were admitted on a Friday, and every shift brought new doctors who repeated the same words: "Maybe it's a false positive." Then Monday came, and the main neonatologist, who had been away for the weekend, finally examined her. Her words were completely different: "You're not going home anytime soon. We need to investigate this thoroughly. It's a very rare condition, and we need to keep your baby here under close observation."

The days that followed were filled with tests and waiting. Eventually, the doctor came back and said: "We think we know what this is, but we can't confirm it yet. We need genetic testing, which may take about a month. The good news is that the condition appears to be treatable."

The next challenge was finding the right medication, which wasn't available. It had to be ordered from Houston.When it finally arrived and Andrea received her first injection,

HCU HERO: ANDREA FROM TEXAS

e immediately noticed a change, she was more awake, more alert, more alive.

Soon after, we were able to go home with medication, daily injections, and a completely new reality. We were trained by the nurses to give her shots ourselves, and we left the hospital filled with hope, fear, and deep love. That was the beginning of our life with **Cobalamin C (CblC)**, a rare genetic disorder and the start of a journey of faith, learning, and resilience.

Learning About Cobalamin C

Andrea was only three weeks old when we met with the genetic doctor. That appointment helped us start to understand her diagnosis and the possible effects, but we had no idea what was still ahead of us. It was devastating to face such a rare condition, something we had never heard of, something that made us feel alone.

We wanted to learn everything we could. We read, we asked questions, and we found support groups for families of children with CblC. Through those groups, we discovered that Andrea was not receiving the correct medication dosage.

Thanks to the guidance of other moms, we were able to connect with nurses at the NIH (National Institutes of Health), who then contacted our

HCU HERO: ANDREA FROM TEXAS

geneticist to explain the importance of using a higher dose of hydroxocobalamin to protect Andrea's brain and vision.

By that time, Andrea was about three to four months old, and she had already started showing some vision problems. We went to a retina specialist who performed tests under anesthesia and confirmed that Andrea had maculopathy. That diagnosis was incredibly painful to hear but through the support groups, we realized we weren't alone. We learned that this condition, while rare and challenging, was part of her CblC journey, and that there were other families walking this same path with strength and love. Shortly after the anesthesia, Andrea developed an aversion to eat. She stopped feeding, and she would cry every time we tried to feed her. It took a long time for her to accept her bottle again. She went through several procedures until we finally found the right formula and she started eating. To this day, she continues with feeding therapy, and with a lot of effort, we were able to avoid the need for a feeding tube.

Andrea Today

Today, Andrea has an amazing team of doctors and therapists who follow her closely. She regularly sees her geneticist, hematologist, gastroenterologist, neurologist and ophthalmologist, usually every three months. Every year she needs to see her cardiologist and retina specialist. She also receives occupational therapy, physical therapy, speech and feeding therapy, vision services, and orientation & mobility lessons,

HCU HERO: ANDREA FROM TEXAS

all through ECI (Early Childhood Intervention) and our local school district.

Andrea is also part of a **clinical trial at the NIH**, where we've felt an incredible amount of support from the doctors and researchers dedicated to studying Cobalamin C. Through them, we've learned so much about the condition and how to care for her better. We're deeply grateful for their kindness, their dedication, and their ongoing efforts to help children like Andrea.

We're also so thankful for Andrea's entire care team - the doctors, nurses, therapists, and teachers who help her thrive. Because of their guidance and care, Andrea keeps improving every day.

She has shown us more strength and resilience than we ever imagined possible. She's taught us patience, courage, and how to be her strongest advocates.

This journey has been incredibly hard, but it has changed our lives in ways we never imagined. Andrea is our motivation, our teacher, and the reason we never give up.

Today, Andrea is a one-year-old full of joy and light. She loves life, her sister, and her family and her smile is the biggest proof that anything is possible. With love, medical care, and faith, she continues to grow and surprise us in every way.

HCU HERO: ANDREA FROM TEXAS

We know the future is uncertain, and that Andrea has her own rhythm to learn and develop but every little milestone is a victory. Every smile, every attempt to communicate, every small step reminds us that miracles do exist.

Andrea shows us that with love, determination, and hope, **anything is possible.**

Today, one year after Andy's diagnosis, we feel truly happy, life has turned colorful again.

Seeing her happy, enjoying every moment, smiling, and doing things we once thought impossible fills our hearts.

If I could speak to the version of myself who was in the NICU, praying and crying for her little baby, I would simply say: trust that everything will get better. One day, you'll wake up, and the storm will have passed. One day, almost without realizing it, life will feel lighter again, you'll smile, you'll breathe deeply, and joy will quietly find its way back. Through Andrea, as we watched her grow stronger, we rediscovered happiness.

We know the road is not done and more challenges are to come, but we will continue fighting for the better days.

Raising Joy Double Good

FUNDRAISER

RESULTS

HCU NETWORK AMERICA | Raising Joy Double Good

Total HCUNA Donation	**$1,106.50**
Supporters	**35**

Thank you to all who participated by hosting a sale or purchasing popcorn!

These funds will be applied towards our End of Year $25K match!

Welcome to Our

New Dietitian Advisor

CLICK HERE!

Emily McDermott is a metabolic RD at Children's Hospital of Richmond at VCU. She completed her undergraduate degrees in Psychology and Human Nutrition, Foods and Exercise at Virginia Tech and her Dietetic Internship at Johns Hopkins Bayview Medical Center. She is a Certified Specialist in Pediatric Nutrition (CSP) and previously worked in pediatric nutrition at Johns Hopkins Children's Center and Children's National Medical Center.

Emily Mcdermott

IN CASE YOU MISSED IT...

The HC&U Podcast is back!!!

HC&U is a podcast about Homocystinuria, sponsored by HCU Network America and hosted by Ben & Lindsey.

Meet your hosts!

The latest episode

Welcome to the HC&U Podcast! We are Ben and Lindsey, your hosts. We are so excited to be starting this as extra resources for the Homocystinuria community. We hope you like our content!

Ben welcomes Tiny to the table!

Tiny, mother of #hcuhero Alexander shares her families story of late diagnosis and how newborn screening factored into this. You won't want to miss this heartfelt conversation.

To Listen:
https://hcunetworkamerica.org/hcu-podcast/
or click below on your favorite option!

LISTEN ON Spotify

Listen on Apple Podcasts

LISTEN ON iHeartRADIO

Listen on amazon music

HOMOCYSTINURIAS DATA COLLECTION PROGRAM

POWERED BY RAREX

HERE ARE THE TOP 5 REMETHYLATION SYMPTOMS

81%

BRAIN/NERVOUS SYSTEM

75%

DEVELOPMENT

56%

DIGESTIVE

62%

COMMUNICATION

56%

MUSCULAR

CANPCU+ CANPKU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RAREX

MEET CLEMENTINE AND EVERETT

a story of hope in the face of a missed diagnosis

“Serious, but treatable”

is a phrase often used to describe conditions like homocystinuria (HCU). When identified early, HCU can be managed with treatment, allowing individuals to live typical lives. Unfortunately, approximately 50% of patients are missed by newborn screening, resulting in delayed diagnoses and sometimes life-threatening consequences. Newborn screening tests for levels of methionine in the blood, which isn’t always elevated in the first 24-48 hours after birth. In addition, not all types of homocystinuria can be detected with elevated methionine.

Today, we want to share a story that illustrates advocacy, collaboration the profound impact of a missed diagnosis — Clementine and Everett’s Story.

HCU NETWORK AMERICA

Clementine was born into a family overjoyed with the birth of their first daughter, the sister of two devoted big brothers. After an uncomplicated pregnancy and birth, Clementine presented as a perfectly healthy and normal baby. When the pediatrician repeated the newborn screening blood spot at her two week check up the family knew the drill and didn’t think much of it.

Settling into a new routine, the family was shocked to receive the call stating her newborn screening was abnormal and they needed to bring her to Children’s Hospital Colorado right away.

They reacted as any parent would, with panic. They began googling what conditions were on the newborn screening and trying to piece together what could be wrong.

The following day, they took her for bloodwork, which revealed a homocysteine level of 153.9 mcmol/L and a methionine level of 736 nml/L. They did't know what that meant other than it was WAY above the normal range.

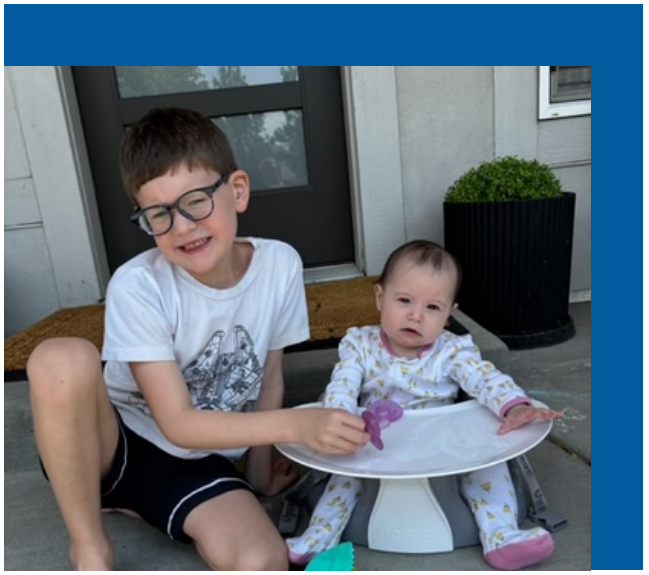
The family was frantic for more information. What did this mean? Was she in immediate danger? Was there treatment? A cure? What do we do? These are all the questions swirling in their minds.

Thankfully, their questions would be answered upon receiving a call from a genetic counselor at the metabolic clinic from Children's Hospital Colorado. She confirmed that Clementine was born with a rare metabolic condition called Homocystinuria. They were able to meet with the metabolic team the following day to discuss the diagnosis and treatment options.

The family learned that each of their other children would have a 25% chance of also having HCU. The family was confused because both their older boys passed newborn screening without issue. They thought they were in the clear. Both siblings had their blood drawn, and it was discovered that one of the other brothers also had homocystinuria with an astounding level of 222.7 mcmol/L.

The family was shocked, devastated, and distraught. How could this have happened? Everett missed the chance to be treated from birth. The family learned that the Colorado State Newborn screening labs' previous threshold for methionine was 100 mcmol/L.

> **Clementine and Everetts story is a testament to the importance of revised newborn screening for HCU patients and the collaboration that takes place between organizations, clinics and labs.**



After attending an event and learning about the issues related to HCU from HCU Network America's President, Margie McGlynn, the Colorado lab director, in collaboration with the metabolic clinic at Children's Colorado, lowered the threshold to 33! Everett was born when the threshold was higher and therefore missed on the screening. Clementine was the first baby born in Colorado to be diagnosed with a lowered threshold.

We are excited to announce that HCU Network America will be participating in the APHL Newborn Screening Symposium in Providence, RI. Our primary objective at this event is to bring awareness to state labs, the importance of lowering the methionine threshold, and educate physicians on recognizing the signs and symptoms of homocystinuria, as well as to provide guidance on patient referrals to genetic experts.

As we approach the end of the year, we kindly request your support to continue our mission. Your generous donations enable us to provide essential resources, support, and hope to families like Clementine and Everett's. Together, we can make a meaningful difference in the lives of those affected by homocystinuria.

THIS HOLIDAY SEASON

Make Hope Possible

FOR PATIENTS LIKE EVERETT AND CLEMENTINE

What is Giving Tuesday?

It's a global generosity movement unleashing the power of radical generosity. GivingTuesday was created in 2012 as a simple idea: a day that encourages people to do good. Since then, it has grown into a year-round global movement that inspires hundreds of millions of people to give, collaborate, and celebrate generosity. *This year, Giving Tuesday will be* ***December 2nd.***

How do I get involved?

We're asking you to assist us in reaching our $10,000 GivingTuesday fundraising goal.

To get started:

- Set up your own GivingTuesday fundraiser on GoFundMe, Facebook, or Instagram!
- Start to reach out to your friends and family in advance and get them to pledge a donation on December 2nd!
- Let them know that by giving to HCU Network America, they are supporting programs and resources that directly benefit the patients and caregivers of our community.
- Remind them that *all donations* are tax-deductible AND will be matched by 3 anonymous donors (up to $25,000!)

Keep an eye on HCU Network Amrica's social media on 12/3!

We will be posting *"12 reasons to give this Giving Tuesday"*, which will feature all 12 of our 2025 #HCUHeroes!

The posts will include a link to our donations page, so if you'd prefer, you can share our posts instead of setting up your own fundraiser.

GIVING TUESDAY

Matching Gift Challenge

Three generous donors have pledged to match EVERY gift up to $25,000 of donations!

$14,917 of $25,000 goal 60%

During the winter holidays, warmth and generosity can be felt near and far. We ask every patient and family to help us raise funds to continue our homocystinuria community outreach, education, and advocacy initiatives to educate providers and be there for families like Everett and Clementine's. Please share our appeal letter with your colleagues, friends, and family.

Three Easy Ways to Donate

Click here to donate

https://bit.ly/hcumatch25

By check
HCU Network America
15 S. Mallory Ave
Batavia, IL 60510

HCU Hero
Andrea from Texas

HCU Hero
Alexander from Belgium

HCU Hero
Zoraiz & Areeba from Virginia

HCU Hero
Everett & Clementine
From Colorado

HCU Hero
Grayson from Colorado

HCU Hero
Olive from the United Kingdom

HCU Hero
Nico from Scotland

HCU Hero
Michael from Australia

CLICK HERE

Giving Tuesday

HCU Hero
Ari from Ny

HCU Hero
Andreana from IL

HCU Hero
Trace from NC

HCU Hero
Enelcita from Ny

EMPLOYEE MATCH PROGRAM

Did you submit for your Employers Corporate Matching Gifts Program?

What are Matching Gifts?
Corporate matching gifts are a type of gift giving in which companies financially match donations that their employees make to nonprofit organizations. When an employee makes a donation, they need to request the matching gift from the employer, who then makes their own donation. Companies usually match at a 1:1 ratio, but some will match 1:2 or 2:1.

Why are Corporate Matching Gifts Valuable?
Corporate matching gifts are valuable because they are free money for your nonprofit of choice, HCU Network America! Your donation has double the power without you having to give double the amount.

Does HCU Network America Really Benefit?
If corporate matching gifts weren't valuable, we wouldn't be sending this letter. In 2020 we received approximately $45,000 in corporate matching gifts!

How Do I Find out if my Employer has a Corporate Matching Gifts Program?
Your company website or websites such as doublethedonation.com make it easy for you to search and see if your employer takes part in corporate matching gifts. If you can't find it online, please consult with your employee handbook, HR rep or manager to find out.

How Do I Request my Donation is Matched by my Employer?

1. The donor completes their donation
2. The donor submits matching gift request
3. Company reviews donation and nonprofit eligibility and reaches out to nonprofit
4. Nonprofit verifies the donation was made
5. If eligible, the nonprofit will receive the matching gifts request!

Top Matching Gift Companies

Company Match Ratio

- General Electric 1:1
- Gap Corporation 1:1
- ExxonMobil 3:1
- Johnson & Johnson 2:1
- Microsoft 1:1
- Pfizer 1:1
- Coca-Cola 2:1
- And many more!

Did you know some companies match retired employees donations?

UPCOMING EVENTS

FUELING A PHYSICALLY ACTIVE LIFESTYLE ON A LOW-PROTEIN DIET

CLICK HERE!

WEBINAR
PRESENTED BY

Are you physically active, or are you thinking about becoming more active? Do you have questions about managing your metabolic condition while exercising or playing sports?

Join us for a special joint session hosted by HCU Network America and the MSUD Family Support Group, featuring Dr. Karen Reznik Dolins, sports nutritionist and Research Lead for the MSUD Family Support Group.

Dr. Karen Dolins
Columbia University

Thursday, December 11, 2025
6:00 - 7:00 pm ET

Register now at: https://bit.ly/lp-nutrition-activity

Thank you to our generous sponsors

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NOVEMBER RECAP

MSUK
Conference Recap

On November 15, Metabolic Support UK, as part of their Community Conference, held an HCU Breakout Session. Our Executive Director, Danae' Bartke, was invited to chair the HCU track. The day included discussions on the E-HOD guidelines for HCU and upcoming therapies, a group discussion on patient needs, an Ask the Expert Panel, and concluded with a brief overview of the HCU Champions program and a panel showcasing our patient and carer experts and their experiences living with HCU and advocating. We are excited to continue our collaboration with Metabolic Support UK and help strengthen our ties with the shared international cohort of HCU patients and carers.

NOVEMBER RECAP

Tour At The Promin Headquarters

Last week, our executive director, Danae' Bartke, had the honor of being invited to tour the Promin headquarters in the UK. She was given a guided tour of their production facility and saw their packaging machine in action. Then she sat down over lunch with them, where she discussed HCU Network America's advocacy efforts, plans for 2026, and they spoke about future collaboration to better serve the HCU and other low-protein communities around the globe.

She wants to express her thanks for this unique opportunity and describes it like visiting Santa's workshop!

INDUSTRY NEWS

Quarter Three Earnings Report

Travere Theraputics

The Company has successfully manufactured the first commercial-scale batches of pegtibatinase and is engaging with the FDA to restart enrollment in the Phase 3 HARMONY Study in 2026.

At the 15th International Congress of Inborn Errors of Metabolism (September 2-6), the Company presented new long-term data from Cohort 6 of the Phase 1/2 COMPOSE open-label extension (OLE) Study, demonstrating that at the target dose of 2.5 mg/kg twice weekly, participants treated with pegtibatinase in the OLE maintained significant reductions in disease-related metabolite levels, including a 53.5% relative reduction in total homocysteine and a 67.1% relative reduction in methionine over 50 weeks of treatment.

CLICK HERE!

WINTER 2025

VIRTUAL MEET UP

CLICK HERE!

SUNDAY 14 DECEMBER

4:30 PM CT/5:30 ET/
3:30 MT

GET READY FOR...

HCU NETWORK AMERICA'S

2026 CONFERENCE

MORE LAUGHS, MORE COMMUNITY, MORE HOPE

It's A FAMILY AFFAIR

In The City of Brotherly Love

July 10-12, 2026
Philadelphia, PA

- Patient Focused Panels
- Breakout Sessions
- KidsZone and Teen Zone
- Community Building Opportunities

- Dedicated Science Day for Families and Professionals
- Scientific Poster Session
- HCU Hero Award Banquet
- Networking Opportunities

Patient & Family Conference

REGISTER NOW

HCU NETWORK AMERICA

Classical HCU | Cobalamin Disorders | Severe MTHFR

FRIDAY AGENDA

- 7:00am Registration & Breakfast
- 8:30am Welcome and Opening Comments

Professional Track

- 9:00am Rare-X for Research & Medical
- 9:45am HCU Network America Research Grant Recipient Presentations
- 12:00pm Lunch
- 1:30pm Biotech Research and Clinical Trial Updates
- 3:15pm Ask the Experts Panel

Family Track

- 9:00am What is Homocystinuria?
- 9:45am Treatment Guidelines
- 10:30am Q&A Panel
- 11:00am What is ERT, Chaperone Therapy, Gene Editing, Gene Therapy
- 12:00pm Lunch
- 1:30pm Classical HCU & Remethylation Research Overview
- 2:45pm Research: What you should know and ways to get involved

Return to Entire Group

- 3:45pm Closing Comments, Optimizing Self Care
- 4:30pm Cocktails, Appetizers and Posters
- 5:30pm Dinner & HCU Hero Award Ceremony

SATURDAY AGENDA

- 7:00am Registration, Breakfast, Vendors open
- 8:30am Welcome and Keynote
- 9:00am Panel: “It’s a Family Affair”
- 10:30am Panel: Family Planning
- 12:00pm Lunch
- 1:30pm Newborn Screening
- 2:30pm Breakout Sessions #1
- 3:30pm Breakout Sessions #2
- 4:15pm Conclusion
- 4:30pm Parents Pick-Up from Kidzone

SUNDAY AGENDA

- 7:00am Breakfast
- 8:30am Community Building Activities
- 12:00pm Lunch
- 12:00pm Conclusion of Conference

CHEMISTRY RX
INNOVATIVE THERAPIES

Experts In Compounding Medications For Metabolic & Mitochondrial Disorders

Chemistry Rx Offerings

- **Oral Suspensions, Capsules, & Powders**
- **60+ Active Ingredients**
- **Insurance Assistance**
- **One-on-One Care with Our Clinical Nurses**
- **Fully Accredited & Licensed Across the USA**

Flavors!

Flavor Options for Patients with Specific Dietary Restrictions. Please Ask Us!

Chemistry Rx Compounding Pharmacy
855-790-0100
info@chemistryrx.com
950 Henderson Blvd, Folcroft, PA 19032

Share Your Story

With Us!

To Join or Learn More:
Email
info@hcunetworkamerica.org

JOIN OUR FUNDRAISING TEAM

Help create, organize, and support new and existing fundraising ideas!

These virtual funds help support our outreach, programs, and research!

To Join or Learn More:
Email
Dbartke@hcunetworkamerica.org

CUSTOMIZE YOUR KIT FOR FREE!

At HCU Network America, we believe that one of the most important steps to empowering patients and caregivers is giving them the support and tools needed to succeed! We know that a new diagnosis can be overwhelming and riddled with concerns and questions. To us, one way to combat those feelings, and give you the confidence you need, is by providing you with one-on-one support, educational resources, and practical tools, such as scales, cooler bags, and more! Our request for a kit survey allows you the opportunity to request a one-on-one introductory call (with more opportunities to connect), and then a customized kit to the patient's needs. Don't want a call or a Zoom? That's fine too - we are happy to send you the customized kit.

Request your kit now - **https://www.surveymonkey.com/r/HCUKitSurvey**

**Kits can only be sent to patients in the continental US. However, we are happy to connect virtually and share the educational materials with you via weblinks*!

Contact Register

What?

- **What** is it?
  - A secure private survey for individuals or families affected by Homocystinuria
- **What** will I share?
  - Patient's birthdate, gender, exact diagnosis, and how they were diagnosed
- **What** will my info be used for?
  - Confidential and will not be shared unless we have permission
  - Helps HCUNA achieve our goals

Why?

- **Why** should I join?
  - Able to find other families and patients in your state and request contact information
  - Access to exclusive materials (ex: we may have a webinar that a presenter doesn't want to share publicly but is okay sharing with just our community)

How?

- **How** do I participate?
  - The form takes 3-5 minutes to complete
  - Visit our website and click on "contact register" tab or...

Click Here

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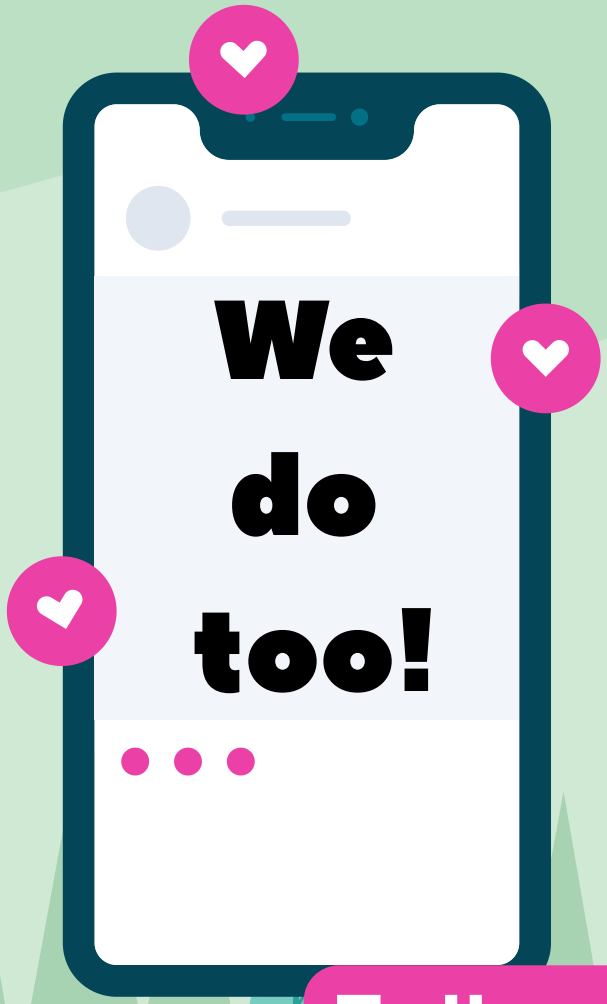
Do you have social media??



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