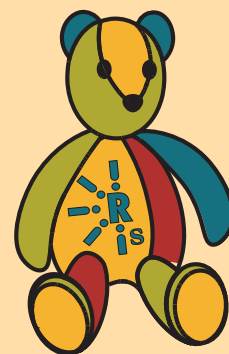


April 2018

RARE Bear Army Newsletter

Spring 2018



Accelerating Cures for RARE Kids



Christina's Corner

To Our Rare Bear Army Members, Volunteers and Supporters,

Spring is here! And we have had some exciting changes and opportunities that have come our way! As many of you may already know, I recently joined the genomics firm WuXi NextCODE as Senior Vice President and General Manager of the Global Rare Disease Program. WuXi NextCODE is one of the leading data and genomics companies in the United States. Their capabilities span study design, sequencing, secondary analysis, storage, interpretation, scalable analytics, AI (artificial intelligence) and deep learning.

In my new role, I work to provide outreach to patients and patient communities, with the hope of connecting them together; similar to our RARE Bear Program which does exactly that!

By partnering with research institutions, healthcare systems and pharmaceutical companies, we

will be able to solve and eventually treat more rare disease cases. This is an exciting next step for RARE Science. I will continue to keep you up to date on how we progress from here. More about that in the coming months!

For those of you who participated in our International Sew-In Day on March 17th! Thank you, Thank you! The day was a huge success. Hundreds of bears were made worldwide and we couldn't be more proud and thrilled to have such an amazing group of volunteers who selflessly give up their time and talent to help us spread the word about RARE Disease and our very special RARE kids. A huge shout out to Spoonflower! Katie Berman and Meredith Feingold spearheaded the event once again at their Greenhouse in Durham, North Carolina. We are so grateful for

their continued support. Mark your calendars for next year! The tentative date for the next International Sew-In Day is Saturday, March 18, 2019!

Look forward to sharing more with you in our next newsletter; but for now . . .

Why we do what we do.



Fondly,

**Christina A Waters, PhD, Founder
and CEO RARE Science, Inc.**

IN THIS ISSUE

Christina's Corner	1
Spotlight on	2
Meet Your Makers	3-4
Fat Quarters	5-6

Forever Bear Friends.	7-9
RARE Science in the News	10-11
RARE Science	12
Bear Necessities	13



SPOTLIGHT on . . .

The RARE Child Behind the RARE Bear Program

In this segment we highlight and share stories of our brave RARE kids and their families, and hope to provide some insight into how families are finding help and hope for a child who battles with rare disease.

I remember sitting in my doctor's office and her asking me about genetic testing. "Why not?" I thought, not even considering the possibility of a worrying result. A few weeks later I learned I was a carrier for the genetic condition Cystic Fibrosis, but my husband would also have to be a carrier for it to affect our baby. He took the test, and he was also a carrier. Still, the baby only had a 25% chance of having Cystic Fibrosis. After an alarming ultrasound, we had the baby tested. At 26 weeks pregnant, I did not know the gender of my baby but I did know the child would live with the life threatening disease, Cystic Fibrosis.

When Julia was born, we knew her life would involve pills, nebulizers, frequent hospital visits, long hospital stays and a life that needed extra protecting. But, we also knew she was just like any other baby and she deserved a happy life.

30,000 people in the US have Cystic Fibrosis and about one in 3,844 births are of a baby with Cystic Fibrosis. It is a genetic mutation that affects the way your mucus is created. For Julia, this means she must take pills at every meal to replace the enzymes her body is not able to move out of her pancreas. She is also more susceptible to lung infections and bacteria and must do nebulizer treatments at least once a day, as well as 15 minutes of physical therapy. She needs to eat foods that are high in salt and fat in order to replace what her body is not able to produce. But other than that, she is developing as a completely normal two year old.

We try to live our life to the fullest and help Julia learn about the world around her. Rather than keeping her inside, we let her explore the world to its fullest while giving her proper treatments and relying on medical precautions to protect her. Whether at home, at Disney World or in Amsterdam, Julia is doing her treatments everyday but also having fun and enjoying all that the wide world has to offer. That balance is

so important to our family. We want her to live a long, healthy life, but we also want that life to be positive and fun.

The positive side of this is that the CF community is filled with hope right now. Treatments are prolonging life, and the life expectancy of someone with the disease is increasing almost every day. For the first time ever, there are just as many people living with this disease over 18 years old as under 18 years old. New drugs, the first of their kind, are coming out that actually treat the source of the disease and not just the symptoms. Even with these new treatments and medicines, there is no cure and people with CF are losing their battle every day. Most adults with CF have or are preparing to have a double lung transplant, and many children are in a similar situation as well. Children with CF spend hours of their time hooked up to machines and many days a year in the hospital. Hope is there, but the reality is that with CF, it can change at any moment.

Julia's story is a story of hope; its a story of how science and hope can change the future. She is our fighter and our hero.



Meet your Makers

A Special Shout Out! To some of our new members and some of our old who go above and beyond to support us in bringing RARE Bears to our RARE Kids!

BEARometer!
5,812 bears
logged
to date!

By May of 2015, the Rare Bear concept had been born and Tonita Waters and I, Monica Scott, have made several bears for Christina Waters to gift to children with rare diseases. Then Chris told us that she wanted to gift 100 bears in the next couple of months. Well, we had already done a number of bears but the idea of doing that many in that short of time was just mind boggling. We needed help if we were going to do 100 bears!

We were trying to figure out who we knew that sewed and might be willing to make bears for us. Well both of us belong to North County Quilter's Association, in Escondido, California, and quilters are sewers. So we went to our President and Board and asked if Chris could present our request to the guild membership. The year was ending with only one meeting left. Our last meeting of the year is always a potluck so we don't have a speaker like we do at our regular meetings. A perfect time for Chris to talk to the group.

Quilters as a whole are generous and giving. Every guild has a charity or two they support. North County Quilter's Association is no different. We support four or five charities every year. Mostly by financial contributions made possible by our Opportunity Quilt we raffle off every year. We make quilts for our local Interfaith Council which helps the homeless, wheelchair quilts for the military at Balboa Naval Hospital and our smaller Neighborhood Group makes quilts for a local dialysis center.

When Chris presented the appeal for Rare Bears, the response was overwhelming. The members were interested and excited about making bears and joining the young Rare Bear Army. We also received great input from the members as we mingled during dinner. As quilters are known to spread the word about things they are passionate about, the word spread quickly. We had no problem giving Chris the number of bears she wanted.

North County Quilter's Association is very proud to be the first guild in the Rare Bear Army. Rare Science is now one of the charities we support.



**Thank
you to our
Sponsors!**

Sewing Partners/Sponsors/Donors

Cosmo
Clover
Kai Scissors
Bernina
Coats and Clark

Fairfield
Man Sewing
Mountain Mist
Quilts Inc.
Quilt Alliance

The Quilt Show
Sulky
Simplicity
Spoonflower

Other Partners/Sponsors/Donors

Sweetwater Capital
CIRM
Cellular Dynamics
University of Houston

Meet your Makers continued

A Special Shout Out! To some of our new members and some of our old who go above and beyond to support us in bringing RARE Bears to our RARE Kids!

Martha and Ben McKillip - Elbert, Colorado

Margaret Paulson - Bly, Oregon

Wanda Mozdy - Sacramento, California

Sharon Carbine and the Red Rose Quilters Guild
(RRQG) - New Holland, Pennsylvania

Cynthia Schmoyer - Anchorage, Alaska

Cherrie Ruben - Oceanside, California

Connie Emmen - Boise, Idaho

Bruce Curl - Phoenix, AZ

THANK YOU, THANK YOU, THANK YOU! TO SWEETWATER CAPITAL PARTNERS!

Ever wonder what happens at a RARE Bear Gifting and where it all takes place?

Well, thanks to James Gammet and the team at Sweetwater Capital, their office suite is opened to us on the third Sunday of each month for our RARE Bear Giftings! During the gifting, volunteers from all over San Diego County (and sometimes beyond) gather to select a RARE bear for a child with a rare disease.

Hundreds of bears are scattered around the office. Volunteers grab a request list for a child and go hunting for just the right bear. The list contains the child's age, gender, favorite colors and favorite things. The volunteers work tirelessly to find the perfect bear. The bear is then checked for appropriate stuffing weight, scanned for pins, and given a final quality check for any

open seams or construction issues. Our MASH unit is on hand to help!

After receiving the green light, the bear is taken to our shipping team who packages the bear, gets the label on the bag and sends the bear on its way! The day can be long and tiring. Over the last two years we have gifted over 5,800 bears!

Thank you, James and all of the folks at Sweetwater Capital for allowing our RARE Bear Army to commandeer your offices once a month! We are BEARY grateful for your support!



SWEETWATER
CAPITAL PARTNERS

Planning is underway for a RARE Bear retreat !

Who: All Rare Bear Army Members

What: A weekend retreat of making bears, meeting fellow bear members, etc.

When: To be announced

Where: Vina De Lestonnac (tentative location) Temecula, California
<http://www.lestonnacretreatcenters.com/>

If you are interested in learning more about the event as plans come together, please reach out to Kris Groscup bgroscup@earthlink.net or Tonita Waters tonitawaters@gmail.com for additional information!

Fat Quarters



Highlights from Our Past Quarter!

January 6 Girl Scout Training day at Sweetwater! We now have our GS leaders trained to host their own sew-in stuff-ins around town!

January 13 THANK YOU to Kris Groscup for hosting a beautiful Volunteer Appreciation Luncheon! Those who were able to attend had a marvelous time.

January 21 RARE Bear Matching Day! 450 Bears sent to their new homes! A HUGE THANK YOU to UCSD Philanthropy Group Alpha Lambda Chapter, Delta Epsilon Mu; Alpha Phi Omega from SDSU, Giselle's Pledge and all of the other volunteers who took time out of their busy schedules to help out! We are so BEARY grateful!!

February 11 Giselle's Pledge Stuff-in/Sew-in! 52 Bears Stuffed, sewn and "faced." 17 more bears stuffed and ready to roll. What a fantastic day!! AND what an amazing group of young women. Thank you GP for your incredible support and dedication.

<https://gisellespledge.org/>

February 13 Sponsored by Women in Bio, Chris Waters kicked off Rare Disease Awareness for the month by presenting: "Unlocking the Secrets of Genetics and How it Relates to Disease: Rare Disease Global Communities are Paving the Way"

February 18 RARE Bear Matching Day! Sponsoring Alagille Syndrome Alliance

March 1 Sew and Stich Sew Expo in Puyallup, WA check out the link:

<https://sewexpo.com/classesevents/events/charityeventsrarebearshumanesociety/>

Follow all the exciting things we are doing and keep up-to-date with all the new happenings!

Facebook: [@RAREScience](#)

Twitter: [@RARE_Science](#)

Instagram: [RARE_Science](#)



March 7-10 Re (ACT) International Congress On Research Of Rare And Orphan Diseases

<https://www.reactcongress.org/>

March 12-14 Chris Waters Presentation! BioData West <http://www.terrapinn.com/conference/biodatawest/ourstory>

Join the RARE Bear Army!

- Support life-changing research
- Make a special friend
- Bring a Smile to a RARE Child

Fat Quarters continued

WE HAD SEW MUCH FUN at the Sewing and Stitchery Expo at the Washington State Fair Grounds in Puyallup! We have been invited back next year! If you are interested in volunteering for next year's event, please let us know!

Dates will be February 28-March 3, 2019. info@rarscience.org



2018 RARE Science & RARE Bear Army Calendar

Below are some of the events and activities planned for RARE Science and RARE Bear Army in the coming months.

April 14 12th Annual National Walk for Epilepsy Saturday, 7:00AM-Noon; National Mall, Washington, DC

April 15 RARE Bear Matching Day! Sponsoring PKU Phenylketonuria

April 26, 27 World Orphan Drug Congress USA Conference: focuses on the most pressing challenges and opportunities to bring rare disease therapies to patients faster.
<http://www.terrapinn.com/conference/world-orphandrugcongressusa/index.stm>

May 12 Saturday Open Community Service Day 10:00AM-2: 00PM

May 20 RARE Bear Matching Day!

June 9 Saturday Open Community Service Day 10:00AM-2: 00PM

June 17 RARE Bear Matching Day

July 14 Saturday Open Community Service Day 10:00AM-2: 00PM

July 15 RARE Bear Matching Day Sponsoring Moebius Syndrome Foundation

October 21 RARE Bear Matching Day Sponsoring HCU Homocystinuria

November 5-7 Epilepsy Awareness Day at Disneyland!

The third Sunday of the month we meet at Sweetwater Capital for RARE Bear Giftings

Location:
662 Encinitas Blvd. Suite 230
Encinitas, CA 92024
10:00AM to 2:00PM

Bring a friend!

Forever **BEAR** Friends



A huge thank you from Vincent from Dublin who received his rare bear today. It was a surprise so he was chuffed with himself and such beautiful detail in it. Thank you again.



Thank you so much for the wonderful RARE bear for my sweet little 2 yr old, Mia. The past couple of weeks have been challenging for a variety of reasons but receiving this in the mail today lifts up my spirits, likely even more than it does for Mia at this point! I'm sure it will grow to be a great reminder that she is special and loved, even by wonderful strangers that have never met her.

Many thanks for all that you do.



Thank you Rare Bear so very much for making Jayden's day today!! He loves it!

Travis was so excited to receive his Rare Bear in the mail today!

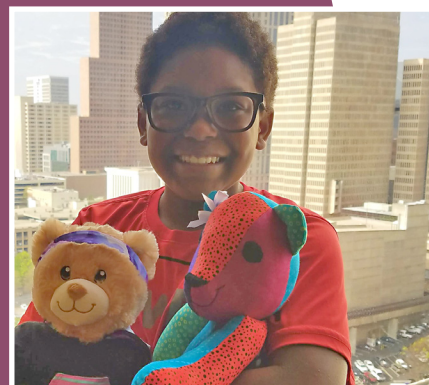
He said "for me?"

Melts my heart!!!!!!

Thank you a million times over!!!!



Ricardo was sooooo excited to wake up and find he had a special delivery!!! He loves his fire truck rare bear! Thanks so much!!!



Thank you so much for such a beautiful bear. It couldn't be more perfect. My son loves it! It arrived just before our trip to Atlanta, so of course Luna (the bears name) had to go everywhere with us ;)



Check out our Facebook page. See more smiling faces of the kids with their own unique RARE Bear. If you're not a RARE Bear Army member, join us! This is why we do what we do.

<https://www.facebook.com/RAREScience>

Forever **BEAR** Friends continued

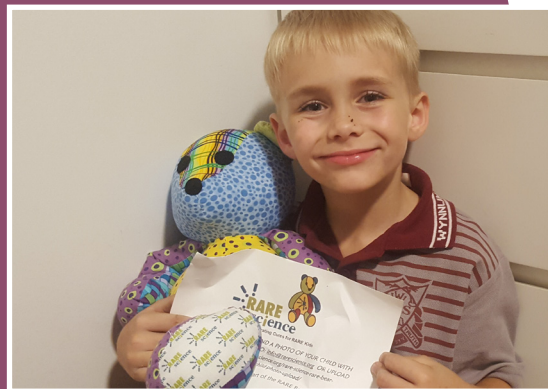


We received my daughter Anna's rare bear yesterday. She is 16 months old and has type 3 osteogenesis imperfecta. So far she has had 10 breaks. When I opened the bear yesterday she got so excited."



Thank you so much for sending our sweet Ava Grace a rare bear from Rare Science. She absolutely loves it!!! She will always cherish this bear!! Thank you for all you do for the rare diseases and rare disorders and for coming up with such a neat idea!

This is Breyleigh with her RARE bear. Breyleigh is almost 9 months out from a heart transplant. She loves her bear. Thank y'all so much.



Thank you so much for our Rare bear for our son Aiden Prior, he is very loved. My son was very excited to receive him.
Thank you again.



Thank you for sending a bear as beautiful and unique as Yalina is. She pushed all her other babies and bears off her bed as soon as she received this bear and now exclusively sleeps with only this one. She's named her bear "Happy Bear". Thank you for taking the time and effort to make and send this to her. You are appreciated more than you know

Need more feet and tags?
<http://www.rarescience.org/kits> or
email your order request to info@rarescience.org

Register to be a Rare Bear Army Member <http://www.rarescience.org/rare-bear-army-signup/>

Become a Volunteer! <http://www.rarescience.org/sign-up-to-volunteer/>

Know a Rare Child who would like a Rare Bear? Please visit <http://www.rarescience.org/rare-science-rare-bear-request/>

For any information, inquiries or questions, please contact us at info@rarescience.org

Forever BEAR Friends continued



Thank you so much for the beautiful bear. The Cat pattern is just perfect. Matthew Jr suffers from Coffin Siris Syndrome. A SmarCA4 mutation. He smiled so big when he got your package. Thank you again.



Hi,
I want to thank you for that you sent this special bear to Poland. My son is very happy:)

Bobby and his Rare Bear have been traveling the last 2 weeks. Last week we drove from Atlanta to Cincinnati for 3 days of appointments at Cincinnati Children's and this week we were off to Gainesville Florida for Bobby's HUB visit for his drug trial. Luckily Bobby and Rare Bear like to travel!



Dr. Christina Waters outlines her vision for sharing global genomic data to better diagnose and treat rare diseases.

Rare disease touches families all over the world. We aim to overcome geographic boundaries, lower the barrier of access to genomics, shorten diagnostic odysseys, and accelerate new treatments for #EveryRare disease.

“Kids with rare diseases urgently need an ‘end to end’ solution to find out what’s causing their condition and if there are any options to treat it,” says Christina Waters, PhD.

Many advocacy groups are funding research, raising awareness, and pursuing important new options for their specific diseases. But a true systematic approach that can solve more rare disease cases more quickly and understand the biology underlying these diseases, requires lots of global collaboration, data, and the technology to pull it all together and put it to use.

The foundation of understanding rare disease needs to be genomics, our DNA. And we need to link families who have the same rare disease but are scattered across the globe. That will let us identify what is similar and different, genetically, and uncover biological causes. Rare disease knows no geographic boundaries, and no matter where you are in the world or what language you speak, our DNA provides the common language needed to decipher biological causes and lead to new and improved treatments for rare diseases.

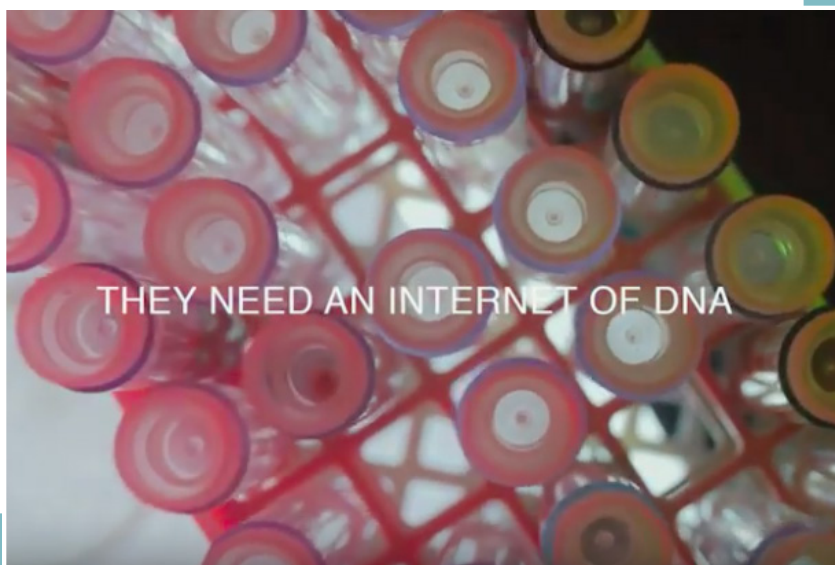
As someone with extensive experience studying the biological

basis of complex genetics, Waters has found that by sequencing each rare individual, we can understand the biological pathways and possibly even repurpose approved drugs, for example, which can greatly accelerate access to new and better therapies.

But to have broadest impact, these approaches must be based on a standard holistic, global platform that integrates all kinds of data linking genetics to observable traits of rare diseases.

“Nobody else was doing that, so it became my calling,” says Waters, who founded the non-profit RARE Science in 2013. With RARE Science she has succeeded in creating a framework to power rare disease research and bring together families across 38 countries who share one of the almost 400 conditions the organization is working on.

Waters recently joined Wuxi NextCODE as senior vice president heading the company’s rare disease program. “By working at WuXi NextCODE, I can take the approach we developed at RARE Science to a level I never dreamed would be possible,” she says.



That's because WuXi NextCODE is building the leading global platform for storing, sharing, and interpreting massive sets of genomic data. "This platform uses DNA sequencing, functional and clinical data and artificial intelligence (AI) to answer important questions about biological targets," she says. These tools make it possible to start solving rare diseases. They allow Waters to take her non-profit's goals to scale and help more patients with rare diseases receive definitive diagnoses and more effective treatments.



"The reality of rare disease means we must work globally if we are going to have enough patients in any of our studies to have an impact," she explains.

Rare conditions are, after all, much harder to study because in any given place fewer people have them. For some conditions, the number of known patients are fewer than 100 worldwide. Still, overall, there are many rare conditions – about 7,000 and affecting hundreds of millions of people world-wide, over half of whom are children, 30% of which

will die before their fifth birthday.

As someone with extensive experience studying the biological basis of complex genetics, Waters knew creating a solution for kids with rare disease was going to be a challenge. But she has worked in the Life Sciences for over 25 years, in both large pharma, biotech, non-profits and consulting for large life sciences firms to accelerate the translation of discovery to clinical impact. She knows what is available to patients now, and it is not enough.

"Kids are our future, and to address the urgent needs of children with undiagnosed and rare diseases, we should be sequencing every one of these children wherever they are in the world," she says.

"For some, sequencing would lead to a really easy fix, like changes in diet. For others, it is going to take more time and more work. But if we cast our net wide enough, by working globally and united, we can take steps to improving their lives too and leverage what we learn from one child to another."

Some patients, she explains, have conditions that have never been seen before, which makes diagnosis harder. For others, the problem is purely practical. They can't afford to get sequencing. "We need to provide sequencing to anyone who needs it, anywhere in the world," says Waters.

"There is tremendous excitement about new technologies such as gene therapy, stem cells and gene editing [CRISPR]," says Waters. These tools have great potential to uncover crucial biology and point to cures. "But you have to write the book of the genome before you can edit it."

Waters now aims to write as much of that book as possible, thanks in part to WuXi NextCODE's global genomics platform.

May 20th 2018 PKU (Phenylketonuria)**What is PKU?**

Phenylketonuria is an autosomal recessive genetic metabolic disorder that greatly reduces the ability of those affected to metabolize phenylalanine, one of the amino acids in protein. A buildup of phenylalanine (phe) in the blood can cause profound damage to the central nervous system in children and adults. If untreated in newborns, PKU can cause irreversible brain damage within months. Lifelong treatment is required to prevent neurological and physical deterioration.

Learn more at www.pkunews.org

July 16th 2018 Moebius Syndrome**What is Moebius Syndrome?**

Moebius syndrome is a nonprogressive craniofacial/neurological disorder that manifests itself primarily in facial paralysis. Individuals with Moebius syndrome cannot smile or frown, and do not have lateral eye movements. Many individuals with Moebius syndrome also have skeletal involvement with limb abnormalities or Poland syndrome. People with Moebius syndrome can have respiratory problems, speech and swallowing disorders, visual impairments, sensory integration dysfunction, sleep disorders, weak upper body strength and autism spectrum disorders.

Learn more at www.moebiussyndrome.org

October 21st 2018 HCU (Homocystinuria)**What is Homocystinuria?**

Homocystinuria is elevation of the amino acid, homocysteine (protein building block) in urine. Homocysteine can also be elevated in blood. People with Homocystinuria (HCU) cannot digest the amino acid called Methionine. This leads to accumulation of Homocyst(e)ine, which is toxic. This can lead to stroke, eye lens dislocation, mental retardation, seizures and many other problems.

Learn more at www.hcunetworkamerica.org



Bear Necessity Tips

Traveling Bearskins

- If you are sending one skin, feel free to simply use a 6" x 9" envelope. If folded flat, the bear skin fits perfectly and it is the least expensive way to mail your skin!
- Also, no need to use padded envelopes (they cost more!) and there is no need to wrap the bear skin in tissue or place in a Ziploc bag. The envelope is plenty of protection
- Likewise, no need to send back the plastic kit bags – feel free to recycle them!



Torn to "Pattern" Pieces

- Why would you ever buy more than one pattern? We know that those pieces get tattered and ruined fairly quickly but you can make a template of your pattern on poster board or card board so that you can use it again and again!
- Some of our bear makers even laminate the original pieces so that the directions, seams and notches can still be viewed.
- AND...we know you are hooked when you have wooden templates cut and sanded to perfection...yup, imagine that!

Do you have a tip that you would like to share? LET US KNOW!

Email us at:

info@rarescience.org with your tips, ideas or suggestions!

Helpful HINTS

- Be BEARY Careful-- use little to no heat on the ribbon tag! There is a serial number on each tag and the serial number is transferred onto the ribbon tag using transfer paper. If the tag gets too hot, the serial number will melt off or will transfer onto the fabric on your bear.
- **BOY OH BOY!** We could sure use some RARE BOY BEARS! Sports fabrics, Star Wars, Super Heros, Lego, Minecraft, video games, outdoors, animals, camouflage, you name it!
- You can now order additional bear sole fabric and serial numbered tag kits through our website! Just visit: <https://www.rarescience.org/rare-bear-program/rare-bear-army-signup/kits/>
- OUCH! Watch out for pins! Please make sure your bears are pin free when they come back to us. Hugging a bear with a pin can hurt! If you use pins, please be extra vigilant about making sure they are all out!

For help or additional information, please reach out to our Technical Support Team!

Tonita Waters
tonitawaters@gmail.com

Ariana Nesser
aria.kyo@gmail.com

Monica Scott
monicascott@cox.net