

# RTS Newsletter

To inform, educate and empower all those whose lives are touched by RTS

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[www.rtsplace.org](http://www.rtsplace.org)

## RTS Foundation on track to fund record amount to support RTS related research in 2022 ... more funding needed

The RTS board has been very active in its fundraising efforts in the first half of this year. Especially our vice-chair Laurie Sperou and board member Staci Hanson, who have done the lion's share of this challenging work. The overall fundraising efforts resulted in the RTS Foundation being able to allocate a record amount of funds for research. In the first half of this year, the RTS Foundation was able to fund \$50,000 to support the RTS and related research of Dr. Lisa Wang, Texas Children's Hospital. Additional funding is planned for the fourth quarter of this year.

It is imperative that the RTS foundation continue to provide funding for research, given that access to Governmental and institutional funding has diminished greatly, or in some cases, dried up completely.

The RTS board believes that research is crucial to our understanding of the molecular basis of RTS and that research, ultimately, holds the potential key for future treatments and, one day, a possible cure for RTS.

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- RTS Board Members

We are grateful to our RTS families and friends for their ongoing support of our various fundraising endeavors throughout the year. Without you, we would not have been able to reach this research funding milestone. Thank you!



## RTS Registry – Participate in research to find effective solutions to manage RTS symptoms

Are you a participant in the Rothmund-Thomson Syndrome (RTS) Registry? If you answered no, we are looking for **YOU!**



### What is the RTS research Registry?

The RTS Registry at Texas Children's Hospital/Baylor College of Medicine in Houston, Texas is the largest group of RTS patients and family members worldwide (yes.... WORLDWIDE!). This registry, developed and overseen by Dr. Lisa L. Wang, focuses on the molecular basis, clinical features, and natural history of RTS. So far there are approximately 150 patients and over 275 family members participating from all around the world, including Europe, Asia, South America, and North America.

### Who is eligible to participate?

- Patients with a confirmed or clinical diagnosis of RTS
- Parents and full siblings of a person with RTS



### What does participation involve? It's **EASY!**

- Completing some paperwork at the time of initial enrollment. The initial paperwork provides your basic medical information and takes less than an hour to complete.
- Providing medical and health updates once a year (which is typically one sheet, takes less than 10 minutes to complete and can be returned via email)
- Providing biologic samples (for example, swabs from inside the cheek or a tube of blood) one time during enrollment - these are optional and can be done any time.

### Why should I sign up?

- **To learn more about RTS and to help others with RTS!** There is so much about RTS that is still unknown. Participating in the Registry can help researchers answer questions about the natural history of the disorder and find better ways to effectively manage symptoms related to RTS.
- **To connect and communicate!** Being in the Registry is a great way to stay in touch with other RTS patients and families worldwide.
- **To help your doctors take care of you!** Participation in the Registry allows your local treating physician to connect directly with Dr. Wang to discuss RTS related-questions and symptom management.

### How can I sign up?

Contact Dr. Wang's research staff via telephone or email to request an enrollment packet:  
Ta-Tara Rideau, Clinical Research Coordinator  
Email: [trideau@texaschildrens.org](mailto:trideau@texaschildrens.org)  
Phone: 1-832-824-4224





## Breaking News! Shauna, the face of RTS, turns 18!

If you've heard of RTS, most likely you have seen or heard of Shauna Sperou. With her 18<sup>th</sup> birthday coming up we wanted to give her a little tribute and share a little about her!

### Some fun facts you may not know!

1. Has had soup for breakfast or lunch (Progresso Italian Wedding) pretty much every day since she was 4.
2. Fractured her bones over 50 times before she was 16.
3. Had a weather blog on Facebook with over 1000 followers for many years.
4. Loves roller coasters...the bigger the better. Plans to jump off the Stratosphere in Las Vegas and talks about sky diving.
5. Loves MATH.
6. In 8<sup>th</sup> grade gave a "compliment note" to her Math teacher every day.

7. Constantly mistaken to be much younger and is ALWAYS being given the Kids Menu at restaurants.
8. Loves Boston sports and has been on the Jumbotron more times than we can count...
9. Will ALWAYS have your back!
10. Brings out the best in everyone around her!

We surveyed 100 people and the top 6 answers to the question. Name two words that come to mind when you think of Shauna:

1. Inspirational
2. Smart
3. Funny
4. Strong
5. Thoughtful
6. Compassionate



## New children's book inspired by father's train ride experiences with son in Colorado - What a story!

Hello RTS community! My name is David Ramirez. You may remember my wife Melissa and son Eli from our Washington, DC, visit about 10 years ago. We haven't seen you in person since then, but we keep up on Facebook and love you all.

I wrote a children's book called "Daisy and Jacob Ride the Train" (ages 4-8) about two siblings who take a steam train into the Colorado mountains and see the sights. The book's illustrator is my daughter Sophia, a student at Wichita State University, studying digital arts. The character Jacob is based on Eli, who is now 15 and attending high school as a sophomore. Eli and I spent two weeks riding 14 trains around Colorado in 2017, and the book is a reminder of that wonderful time.

Sometimes we parents feel conflicted about what to do when people stare at our kids in public, unless you're my wife who knows exactly what to say to them. Eli is especially bright and personable, and it would be simple for him to educate others on his condition and why it's impolite to stare. Of

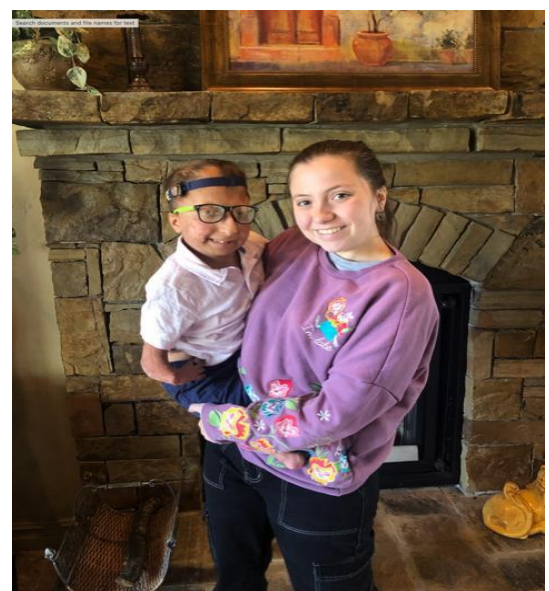


course, staring happens so regularly, there aren't enough hours in the day to talk to all of them. I often think, "I wish they'd go read a book about special needs kids and learn they aren't so different." So, I wrote one! If the book does well, I plan to write future volumes about Daisy meeting children with other physical, cognitive, and emotional disabilities.

We may even see Jacob again. "Daisy and Jacob Ride the Train" is now available on Amazon, Barnes & Noble, iTunes and Google Play.

The notes in the back tell a little about Eli and include a link to the RTS Place website [www.rtsplace.org](http://www.rtsplace.org)

I'm excited to share it with you!



Eli and Sophia Ramirez



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## Live RTS Sharing and Caring Conference set for Maine, 2023



After a three-year absence of live RTS conferences, we are back on track for 2023. With the gracious help from board member Staci Hanson, a Maine resident, we were able to secure a venue that meets our needs. The live Sharing and Caring conference will be held from 4/28-4/30/2023. We will send out the conference itinerary as we get closer to the conference date. The RTS board looks forward to seeing and connecting with RTS families, which is always the best part of the conference experience.

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“Seeing and connecting with RTS families is always the best part of the conference experience.”

- RTS Board Members

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## The “I love Lucy RTS Fun Run”

*Get on your mark, get set... Go!!!*

The “I Love Lucy RTS Fun Run” is a virtual 5k, half marathon or kids sprint taking place on **September 18<sup>th</sup>, 2022**. This event is a great way to get your family and friends together to support the RTS community, the endeavors of the foundation and to get your body moving! Lucy Hanson is a free spirited four-year-old that loves her community, Disney princess, animals, having books read to her and playing outside. She is the inspiration behind the event. To date we have raised \$6,121 and have 94 people signed up to participate. The kids will all be putting on their free RTS superhero capes on and sprinting their hearts out. Please click the link below to sign up and/or considering donating today.

<https://runsignup.com/Race/ME/Westbrook/ILoveLucyRTSFunRun>

## Volunteers needed - RTS needs your special talents!

Are you a good writer, social media wiz, event organizer or want to be considered for a position as a RTS board member? Reach out to us and we'll be happy to explore with you where your skills and talents might be a good fit with the needs of the RTS Foundation.



Remember, everyone on the board or involved with RTS is a volunteer. While we all come from different backgrounds and life experiences, we share a common goal when it comes to RTS. The work can be challenging, at times, but it's also rewarding and fun working together with a group of people who share a common goal. And if that's enough to convince you that volunteering is a terrific way to spend some of your free time, then consider the well documented psychological and physical benefit of volunteering. According to HealthGuide.org, “volunteering provides many benefits to both mental and physical health... helps to counteract the effects of stress and anxiety.” How many other outlets can make that claim?

“Volunteering provides many benefits to both mental health and physical health... helps to counteract the effects of stress and anxiety.” [HelpGuild.org](https://www.healthguide.org/)

Please contact us at: [support@rtsplace.org](mailto:support@rtsplace.org)



## Parenting a child with RTS: From birth to young adulthood

Sheila Townsend – RTS Board Member

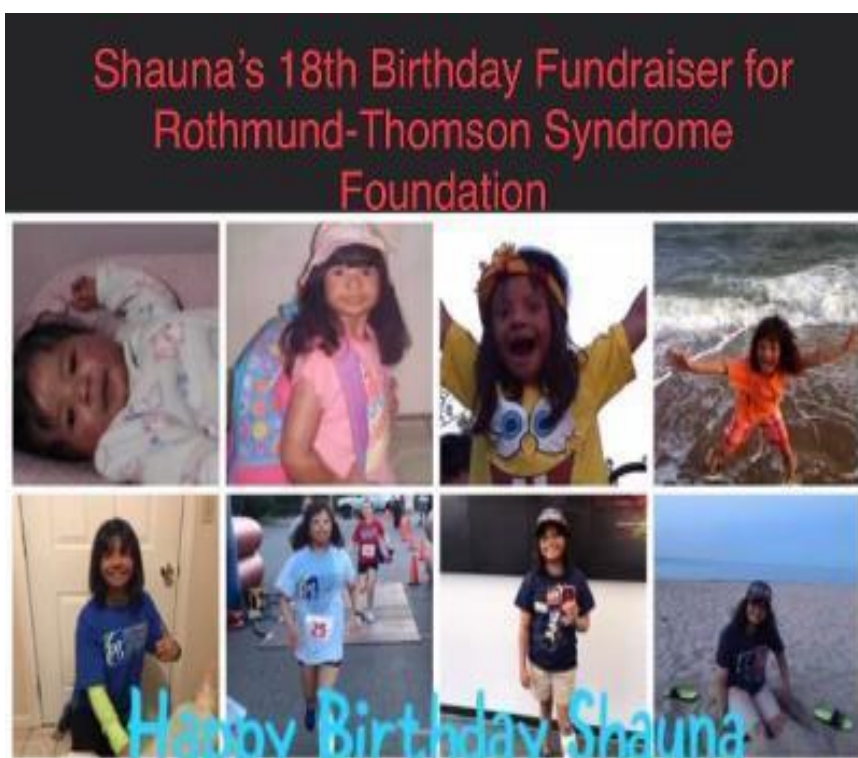


Even though this all happened over 18 years, when he became an adult, it felt sudden and a little scary!! The control and influence we once had to help keep him safe and protected was no longer in our hands. He was an adult who could make his own decisions and choices, even if those choices seemed counter to what we believed was best for his health!! I am sure most parents have this experience where their son or daughter becomes an adult and independent. It is a moment where, as a parent, you are both proud you have raised your child to the point of being a well-adjusted adult ready to face the world and terrified that you are sending your baby out into the world on their own! I think this experience is amplified for a parent with a child with RTS. Not only do you have all the same emotions most parents have but also have the added worries of all concerns that come with RTS and little to no influence to ensure your child stays protected! The best you can hope for is that you have educated your child to now take the lead in their own care, and that they will stay the course on all the precautions you had in place for the first 18 years.

To repeat the question I started with, what is it like to have an adult child with RTS? It is like every other parent who has an adult child. You hope they are happy and well adjusted, that they find a career they love, have great friends, and find a soul mate to share their life with and of course, give us grandbabies! In the end, like all parents, you must let go and allow them to find their own way in the world and with RTS.

In our personal experience, Michael is well on his way.

What's it like to be a parent of an adult with Rothmund-Thomson Syndrome (RTS)? Let me give you a little background on how we got to this point in our lives. Michael is soon to be 26 and was diagnosed with RTS in 1997 when he was 6 months old. This is when we learned that people with RTS have a higher likelihood of developing skin cancer, juvenile cataracts, and of course, RTS related alopecia and poikiloderma. For the next several years we had regular appointments with ophthalmology, genetics, and dermatology and became experts in sunscreen, sunglasses, and protective skin coverings (hats and swimwear). Michael was 7 years old when we heard about Dr. Wang's study searching for a possible link between RTS and an increased risk of osteosarcoma (bone cancer). We went to Houston, met Dr. Wang, and became part of her study that led to the discovery of the RECQL4 mutation. We now know that he does not have the RECQL4 mutation and is considered to have "Type 1" RTS. Throughout the rest of his childhood, the list of specialists Michael saw expanded to include nephrology, cardiology, neurology, and infectious disease after having some mysterious medical issues. These involved years of appointments and several surgeries, including cataracts as a teenager. As he got older, we were able to slowly transition away from most of the specialists.



We are honoring Shauna's birthday by sharing her story, year by year! Each day you can read a new update, told by her mom, on Shauna's 18th Birthday Fundraiser for RTS Foundation on Facebook. We started our goal at \$5000 but it's been doing so well, we're hoping to double that. If you want to help us celebrate Shauna's 18th birthday you can go to the Facebook page (2) Facebook or if you don't have Facebook you can donate via the website Donate ([paypal.com](https://www.paypal.com)) Just remember to put that it's for Shauna's Birthday Fundraiser. This will run until September 8th! The day after she becomes an adult!

The Rothmund-Thomson Syndrome Foundation is a **501(c)(3) nonprofit organization**

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