

RTS Newsletter

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

March 2023

Spring Issue
No. 1



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RTS Research Update - This issue highlights recent studies published by Dr. Lisa Wang and her colleagues as well as an upcoming study focusing on RTS related hearing loss and skin cancer.



Key Publications:

- Publication describing the discovery of the first gene, ANAPC1, to be associated with Type 1 RTS and is also associated with cataracts.

Ajeawung NF, Nguyen TM, Lu L, Kucharski TJ, Rousseau J, Molidperee S, Atienza J, Gamache I, Jin W, Plon SE, Lee BH, Teodoro JG, Wang LL, Campeau PM* (*co-senior authors). Mutations in ANAPC1, encoding a scaffold subunit of the anaphase promoting complex, cause Rothmund-Thomson syndrome Type 1. [Am J Hum Genet](#). 2019 Sep 5;105(3):625-630*

- Publication that describes a new function for RECQL4 discovered in the laboratory.

This study used induced pluripotent stem cells that were generated from samples from RTS patients enrolled in the RTS research study at Baylor College of Medicine.

Jewell BE, Xu A, Zhu D, Huang MF, Lu L, Liu M, Underwood EL, Park JH, Fan H, Gingold JA, Zhou R, Tu J, Huo Z, Liu Y, Jin W, Chen YH, Xu Y, Chen SH, Rainusso N, Berg NK, Bazer DA, Vellano C, Jones P, Eltzhig HK, Zhao Z, Kaiparettu BA, Zhao R, Wang LL, Lee DF* (*co-senior authors). Patient-derived iPSCs link elevated mitochondrial respiratory complex I function to osteosarcoma in Rothmund-Thomson syndrome. [PLoS Genet](#) 2021 Dec 29;17(12):e1009971*

- Publication to determine whether carriers of RECQL4 mutations - for example, a parent of a Type 2 RTS patient - has an increased rate of cancer. Conclusion: There is no increased cancer in RECQL4 heterozygous carriers.

Martin-Giacalone BA, Rideau T, Scheurer ME, Lupo PJ, Wang LL* (*co-senior authors). Cancer risk among RECQL4 heterozygotes. [Cancer Genet](#). 2022 Apr;262-263:107-110.*

Ongoing Projects:

- Collaboration with Chenghang Zong, PhD, Department of Molecular and Human Genetics, Baylor College of Medicine, to investigate a new mechanism for osteosarcoma development ("high ploidy status") which may be important in RTS. *Grant submitted to Rally Foundation January 6, 2023. Results in April 2023.*
- Collaboration with two international research groups (Germany and Brazil) investigating two new genes that may be responsible for RTS (Type 1). Publications have been submitted to journals.

**New Project:

- New study being conducted by a high school student in the Gifted and Talented Mentorship Program in Fort Bend ISD in Texas. She is being mentored by Dr. Wang and will determine the rate of **hearing loss** and **skin cancers** in RTS, topics that have not been formally studied before. In order to accomplish this, she will be sending out questionnaires/surveys to all families enrolled in our research study at BCM and populating the RTS Registry.
 - Because this is only a 4-month project, we need you to respond quickly. We encourage all RTS families to **please respond as soon as you receive the survey**. It will only take a few minutes of your time but will be so important for the RTS community. Surveys will be sent out both by mail and electronic link. If you have not been in contact with Dr. Wang's group recently, please reach out to them at: trideau@texaschildrens.org or **832-824-4224** to make sure they have your contact information.

Lucy's Trash Service

At the beginning of January 2023 Lucy Hanson, age 4, decided to start a neighborhood service of bringing the trash and recycling bins up the neighbors' driveways for a small fee of \$1 per week. At this moment, Lucy has five families participating within our small neighborhood here in Westbrook, Maine. Some of her "customers" are paying her weekly while others have prepaid for her services. Do you have a little one that may be up for the same task in your area? One thing I have come to conclude when it comes to fundraising for the RTS foundation is that people love to donate. We have tried a multitude of avenues to raise awareness and donations from selling T-shirts on [Bonfire.com](https://www.bonfire.com), Lucy's Lemonade Stand to her annual "I Love Lucy Fun Run". So far, my favorite fundraisers have been the ones that have brought our family, friends, and community together to raise awareness for RTS. Lucy's lemonade stand had many people asking more questions about the condition, and nothing makes me smile more than when I see people wearing our t-shirt designs. What I love about Lucy's new trash can collecting adventure is it gets her actively involved in doing good for others while raising money and understanding the importance of a good cause! Knowing how difficult it is to secure funding for future research, keeping the registry alive makes each of these ventures so incredibly important. What kind of fun and unique fundraising opportunities are out there for your family and friends.



Last Call - Live 2023 RTS Sharing and Caring Conference

The RTS Sharing and Caring Conference is now only a few weeks away. We have had a good response from RTS families with many who have confirmed their attendance at the conference. As of this writing, there are a couple of open slots available for interested RTS families. The conference presents a wonderful opportunity to connect with RTS families for either the first time or to reacquaint yourself with families that you may not have seen for some time.

The conference itinerary has been finalized with presentations focusing on, the status of research, medical and clinical challenges of RTS, integration of patients, doctor, and researchers using patient registries, management of dermatological concerns and the psychosocial aspects of living with RTS and its impact on the family. As in prior years, all attendees can participate in the Sharing and Caring part of the conference. This is a unique opportunity for families to share their lived RTS experiences, provide feedback and find mutual support.

Due to the limited accommodation at the Home2 Suites Hotel in Maine, please contact us via support@rtsplace.org as soon as possible, if you are interested in attending the conference. We look forward to seeing you at the conference.



Here is the pertinent information about the 2023 RTS conference:

Location:
Home2 Suites by Hilton Portland Airport
50 Maine Mall Road
South Portland, ME 04106
Phone: 207-517-3636

Date(s): Friday April 28 to Sunday April 30, 2023

Friday - 4/28	5pm - 8pm Welcome mixer for all conference attendees
Saturday - 4/29	8am - 5pm Full day conference with lunch break
Sunday - 4/30	8am - 10:30am Presentation with conference ending remarks

Kara Hanson Shares Her Experience as an RTS Board Member and Aunt to Her Niece, Princess Lucy.

I am writing to share the story of my dear niece, Lucy, who was diagnosed with Rothmund-Thomson Syndrome (RTS) at just one year old. Lucy's parents first noticed something was not quite right when she was just a few months old. She had persistent eczema and an unusual skin condition that did not seem to respond to treatment. As Lucy grew, it became clear that she was small for her age, and her skin remained thin, and fragile. It was not until she was one year old that a dermatologist finally made the diagnosis of RTS.

The news was a shock to our family, as we had never heard of this condition before. It was hard to understand how something so rare could affect one of our own. But as we learned more about RTS, all of Lucy's symptoms were ringing true. We were determined to do everything we could to help Lucy and her parents manage this news.

I have been deeply involved in supporting Lucy and my family since her diagnosis. I have participated in fundraisers and joined the board of directors of the RTS Foundation to continue to learn and to help advance research for this condition. It is my hope that through our efforts, we can find new treatments and eventually a cure for RTS.

Lucy is now four years old, and despite the challenges she may face, she is a happy, spunky, and determined little girl. She is an inspiration to us all, and I am grateful to be her auntie. I hope that by sharing her story, we can raise awareness of RTS and help others who may be affected by this condition. Princess Lucy, you are very loved.



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

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