March 2023

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

www.rtsplace.org

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 neighborhood here in Westbrook, Maine.

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: lhideau@texaschildrens.org or 832-824-4224 to make sure they have your contact information.

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, Roussau J, Molinel Perez S, Atienza J, Ganuche I, Jin W, Phou 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 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-Gucalemente BA, Rideau T, Scheurer ME, Lupo PJ*. Wang LL* (*co-senior authors). Cancer risk among RECQL4 heterozygotes. Cancer Genet. 2022 Apr;262:107-110.

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

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