Wolfram Research Clinic –
The Work Continues...

While we wait for the final determination of funding we are hard at work improving our methods and techniques for collecting data. Our Wolfram Clinic Data Manager, Tasha Doty has taken a moment to describe what she has been working on and how it will benefit patients and families at future clinics.

Collecting questionnaire data from patients and their parents is an extremely valuable method of obtaining information to better understand Wolfram Syndrome. From these questionnaires, we learn how symptoms of Wolfram Syndrome may impact patients’ daily life, and how symptoms might change over time from a scientifically validated measure.

We also know that filling out these questionnaires can sometimes take a lot of time, and feel a little bit repetitive at times. Over the course of this study we have been able to determine what questionnaires are the most useful and sensitive to the changes that can occur in Wolfram Syndrome. To make completing the questionnaires easier for patients and families, we are working to make the questionnaire experience better by:

1) Eliminating the health and medical history sections from the questionnaires so these items can simply be updated as needed when meeting with the endocrinologist at the clinic,
2) Removing some of the questionnaires, and
3) Shortening some of the questionnaires.

We hope this will make filling out the study questionnaires a little bit less demanding. We thank you for providing us this valuable information every year!

Thank you,
Tasha Doty, MA

RESEARCH UPDATE

Dear Research Clinic Families:

As you now have heard from Samantha, I have determined that the Wolfram Research Clinic that was tentatively scheduled for July 2018 will not be able to happen. This was a very difficult decision, but ultimately we felt it was the most ethical choice. Due to delays in NIH’s funding decision and its impact on our ability to prepare, we just could not provide the kind of experience you deserve and that the research demands. We felt that having a clinic under those circumstances would be a disservice to us all.

Please know that we care deeply about you and this research and will start planning with enthusiasm once we get our funding notification. I’m assured by NIH that it will come soon, but there are many bureaucratic hurdles that they have to overcome due to their backlog.

While we are very disappointed that the clinic will not happen as we had originally imagined for 2018, we already have several ideas of what we could do to make future clinics even better, such as Tasha’s work on the questionnaires, holding mini clinics throughout the year, and adding some testing of siblings without Wolfram Syndrome.

We also continue to work towards analyzing and publishing the data that have already been collected, thus providing other researchers and clinicians with important information.

We appreciate your understanding and apologize for the uncertainty that the funding situation has caused. We will keep you informed of any new information. Please feel free to call me personally with any questions.

Sincerely,

Tamara Hershey, PhD (tammy@wustl.edu; 314-362-5593)
Scientific Director and Principal Investigator
WU Wolfram Syndrome Research Clinic
Suite 2203, East Bldg, 4525 Scott Avenue. St. Louis MO
**CLINICAL CARE UPDATE**

Dear Wolfram families,

All of the Wolfram team is sad that we will not be able to see all of you in July. Please know that you are still a very high priority and that this bump will not derail the work at Washington University. We will not allow that to happen! You likely all saw the update from Dr. Barrett in the UK that his intervention trial is not yet underway as they also work through issues, but that it is making progress. Dr. Urano’s dantrolene study is moving along and he will be updating you on those results soon.

The Association du Syndrome de Wolfram meeting is coming up in June and Drs. Hershey and Urano and I will be going to hear updates from the other groups working on the syndrome alongside us, so we will update you in the next newsletter.

Some of the information you all have contributed by participating in the TRACK study was used to develop a paper led by Dr. Barrett’s group: Monogenic diabetes syndromes: Locus-specific databases for Alstrom, Wolfram, and Thiamine-responsive megaloblastic anemia. Human Mutation. 38(7):764-777, 2017 Jul.

This paper analyzes the specific gene changes in 309 people with WFS1 gene alterations in order to determine which changes are likely to cause a particular presentation in a person – for example, some genetic changes cause full-blown Wolfram Syndrome, which others cause diabetes mellitus without other features, others cause hearing loss without other features, etc. This will be very helpful information for patients at the time of diagnosis, getting their genetic testing results and wondering what to expect for their health.

As always, please get in touch if you need assistance with your health or with letters to insurance, etc.

All the best,

Bess Marshall, MD ([Marshall@kids.wustl.edu](mailto:Marshall@kids.wustl.edu))
Pediatric Endocrinologist
Medical Director, WU Wolfram Syndrome Research Clinic

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**Special Thanks**

Even though funding had not been confirmed, many of our Wolfram Clinic partners who understand and appreciate the complexity of the planning process go above and beyond for us year round. We’d like to give a special shout out to our team members.

Ann Doyle, Fadra Nance, Libby Beach, and the PCRU staff are critical in getting our space reserved for testing, meals and gathering. They start reserving space almost a year in advance.

Heather Lugar and the imaging team work year round developing novel and advanced methods for analyzing the brain imaging data. Based on all that has been learned over the years, combined with significant technical advancements in the field, they are currently focused on developing new imaging techniques that will improve their ability to identify and track brain vulnerabilities in Wolfram Syndrome.

Special thanks to the large group of Wolfram team members who meet monthly year round to discuss all aspects of Wolfram Syndrome research from mouse models to drug trials to everything in between.

Thank you to our examiners and physicians who work hard to clear their schedules months in advance to be available to volunteer their time to the clinic.

Space and time in all shapes and sizes is critical to the clinic. Space in the PCRU and at the hotel, time in the CCIR, audiology, ophthalmology, are all critical. Thank you to all of you who help us to reserve these times and work with us to balance these with the needs of the clinics and other research studies.

**There are so many more who do so much and we are so very grateful!**

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**Need Help?** For questions or requests regarding the Wolfram Syndrome Research Clinic please contact the WFS Research Clinic Coord., Samantha Ranck, MSW at 314.362.6514 or [rancks@npg.wustl.edu](mailto:rancks@npg.wustl.edu)