



WU Wolfram
Research Clinic

Washington University School of Medicine

Wolfram Research Clinic Newsletter

May, 2017 Vol.7

WU 2017 Wolfram Research Clinic

Planning for the 2017 Wolfram Syndrome Research Clinic is in full swing. This year we'll have 9 families attending: 6 returning families and 3 new families. Since the clinic is much smaller than in previous years we're doing things a bit differently. The clinic will begin with an orientation open house the evening of Sunday, 7/9/17 and run thru the end of the day on Wednesday, 7/12/17.

Support for New Families

As mentioned above, we currently have 3 new families joining the clinic this year. In the scheme of things 3 new patients doesn't sound like much but considering that this year's clinic is smaller, these families make up 1/3 of clinic attendees! As we all know, the clinic can feel a bit overwhelming initially. Past experience and comments made by families regarding when they were new to the group indicate that being able to speak with another family prior to attending the clinic can be very helpful. Families offer a kind of support and information that is not covered in the more formal communications coming from the research staff. If you are willing to make your name and contact information available to a new family giving them the option of speaking with you, please e-mail Samantha and let her know. We would be thrilled to offer this valuable resources to

Medical Records

Please remember that even if you aren't attending the clinic this year we would still like to add results from any new audiology, ophthalmology, sleep or endocrinology testing to our data set. Please send test results / updated medical records to Samantha.



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

RESEARCH UPDATE

Dear Research Clinic Families:

I hope you all had a good spring and are planning some fun family time this summer. I know many of you have been busy with your involvement in the Dantrolene safety study, and that everyone is eagerly awaiting the outcome of this trial to know whether it will proceed with an efficacy study.

As Samantha notes, the Research Clinic will proceed this summer according to the plans we discussed with you all in person and in the last newsletter. By the time this summer's clinic is over, we should also have some information from the National Institutes of Health about the likelihood of our grant being funded again. Assuming that it will be funded, we are working on plans to integrate the natural history study with any efficacy trial to benefit both efforts and ensure that everyone eligible can participate in one or the other.

By the time of the next newsletter, I hope to have much more concrete information for you!

Finally, I wanted to let you know about a paper that is in the process of being published on "The effect of disease-related symptoms on daily function in Wolfram Syndrome" in the journal Translational Science of Rare Diseases. This paper was based on some of the questionnaires that you all generously filled out last year. We hope that the information in the paper will help physicians better understand the complexity of symptoms in Wolfram Syndrome and to know which ones matter most to your daily lives.

I look forward to seeing those of you coming to St. Louis for the Research Clinic or the Safety Study. Please feel free to call, email or stop in anytime!

Best

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CLINICAL CARE UPDATE

Hello to everyone from St. Louis,

I have a few updates for you since the last newsletter. Our abstract that was submitted to the 10th International Meeting of Pediatric Endocrinology is still being reviewed. We should hear something soon. Dr. Barrett has had a major manuscript on Wolfram accepted for publication in "Human Mutation" that is titled, "Monogenic diabetes syndromes: Locus-specific databases for Alström, Wolfram and Thiamine-responsive megaloblastic anaemia." That paper was a huge amount of work on the genetics of Wolfram Syndrome.

You may have heard in the news that a group in Texas has cured Type 1 diabetes. This development is making a big splash in the press and is exciting. But, the news doesn't always point out that the cure was in mice, so there is a ways to go before it will be tested in people. The group is headed by Dr. Ralph DeFronzo at the University of Texas in San Antonio, who has worked on diabetes research for many years and is very well respected. They have cured diabetes in mice by making a virus that is designed to convert cells in the pancreas that don't normally make insulin into cells that can make insulin. The virus is assembled out of a lot of parts of other things. The basic virus is Lentivirus (a big group of viruses that are able to put some of their virus RNA into an animal's DNA so that it is a permanent part of the cell's DNA). They have added a mouse glucokinase gene (glucokinase is the enzyme that starts the breakdown of glucose once it gets into a beta cell), a part of the cytomegalovirus (that's a virus that can cause hepatitis and other illnesses if you have the whole virus), and a part of the woodchuck hepatitis virus, a really exotic sounding bunch of parts! They added a gene that normally allows things like insulin to increase the number of beta cells in the pancreas, and a third gene involved in normal development of beta cells. When they give the mouse that concoction, some cells in the mouse's pancreas were transformed into cells that can make insulin like a beta cell. Very cool, but also quite complicated.

Big unanswered questions include: will it work in people and will it work in Wolfram? Only time will tell, but if it can work in humans, it should help in Wolfram, at least temporarily.

CLINICAL CARE UPDATE cont.

I also have an update for you on the insulin pump that is paired with a continuous glucose monitor that I mentioned in the February 2017 newsletter. I called Medtronic and learned that the company is still expecting their 670G hybrid closed loop pump to become available in a few offices this spring and then be generally available later this summer. I have not heard any further updates on how well it works, so we will have to wait a little longer to see if it will be a benefit for people with Wolfram and diabetes mellitus.

Please continue to feel free to ask for letters for insurance, disability, school accommodations, scholarships, etc. Please contact Samantha Ranck to make your request and I'll work with her to get your requested letter to you as soon as possible.

All the best,

Bess Marshall, MD (Marshall@kids.wustl.edu)

Pediatric Endocrinologist & Research Clinic Medical Director



Scientific Director and PI Tamara Hershey, PhD and Study Coord. Samantha Ranck, MSW reviewing the 2016 clinic schedule.

RESOURCES

*For more information about Wolfram Syndrome and the WU Wolfram Research Clinic go to:

hersheylab.wustl.edu

*Additional information about the WU Wolfram Research Clinic study can be found at <http://ClinicalTrials.gov> ; *WU IRB approval #: 201301004

*One way to interact with other Wolfram patients and their families, to learn more about fundraising activities and scientific developments in the US and UK as well as, links to helpful resources you can follow the "Wolfram Syndrome" page on Facebook (www.facebook.com)