**WU 2018 Wolfram Research Clinic Thoughts and Ideas**

Even though we are still waiting for the final confirmation of funding for the 2018 Wolfram Research Clinic, that has not stopped us from beginning the planning process. We anticipate the 2018 clinic to be our largest clinic yet! In order to accommodate an increase in the number of participants, we will be spreading the clinic over more days. The current dates under consideration for the 2018 clinic are Monday, 7/9/18 thru Wednesday, 7/18/17. The idea would be to bring patients in for 3-4 days of appointments at some point during that time. Of course it is very early and we will know more in the coming months. I would like any feedback you may have regarding this plan. If you have any preference or comments please e-mail me at rancks@npg.wustl.edu. I would love to hear your thoughts!

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**Updates**

Dr. Hershey was recently contacted by the National Organization of Rare Disorders (NORD) with a request to provide an updated report on Wolfram Syndrome. NORD maintains a database of reports on rare diseases written for patients and families. This database is often quoted in national, state and local media stories and is made available to students and others through subscribing medical schools, universities and public libraries. About 90% of the nearly one million visits made to NORD’s website each month go first to rare disease reports. You can view the full report written by Dr. Hershey and Dr. Marshall at: [https://rarediseases.org/rare-diseases/wolfram-syndrome/](https://rarediseases.org/rare-diseases/wolfram-syndrome/)

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**RESEARCH UPDATE**

Dear Research Clinic Families:

I hope you are having a busy and productive fall! In the lab, we are working to describe how symptoms change over time in Wolfram Syndrome. This information is very helpful to physicians, families and in the planning of clinical trials, particularly after it has undergone peer review at a scientific journal. After review and acceptance, the data have been thoroughly vetted by other scientists and can be found by anyone interested in Wolfram Syndrome. As one example, we recently published a paper on quality of life in Wolfram Syndrome (Doty, Foster, Marshall, Ranck and Hershey; The effects of disease-related symptoms on daily function in Wolfram Syndrome. Translational Science of Rare Diseases 2 (2017) 89–100).

Wolfram Syndrome affects many different physical and emotional functions. Our team wanted to know how these symptoms could affect individuals’ quality of life (QoL) and participation in activities they like to do.

As you may know, each year, study participants and parents in our Natural History Study fill out numerous questionnaires. One specifically asks about different symptoms that can impact QoL. Our analyses show that lower QoL is associated with greater severity of symptoms, in particular urinary problems and temperature regulation. We also found, surprisingly, that diabetes, hearing and vision loss, some of the most common symptoms in Wolfram Syndrome, did not relate to QoL.

In another questionnaire, we asked about participation in different activities. Participation in daily activities is important because it leads to more independence, productivity and life satisfaction. We found that both children and adults with Wolfram Syndrome are likely to restrict their activities because of performance difficulties associated with their symptoms.

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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
Dear Wolfram families,

We are excited to be planning the clinic for next summer. We have written a few letters for insurance, etc., and I hope they succeeded in getting what was needed! Please keep letting us know if you need letters or information about Wolfram for school, work, disability, insurance, etc.

I noticed an interesting article a few months ago from Dr. Andrew Hattersley’s group in Exeter, England, reporting a new syndrome caused by mutations in the Wolfram gene, WFS1. These mutations are dominant and the kids who have them develop diabetes in infancy and are born with deafness and cataracts. They found these mutations by sequencing all the genes in children who developed diabetes before age 6 months in their huge diabetes database in Exeter. Dr. Hattersley is an expert on diabetes in infancy. Our own Dr. Urano helped by doing studies to see what the mutations do to the protein made by the WFS1 gene. We have not really sorted out yet exactly why there is so much variation in what different body functions those WFS1 mutations affect, but information like this from Exeter added to all of Dr. Barrett’s and our information should eventually help figure that out.

All the best,

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Social functioning and physical activities were particularly challenging. We found that 50% of adults acknowledged that driving and being in a long term relationship are difficult for them, while 20% identified exercise/fitness outdoors as difficult. For children, we found that over 30% reported difficulties with sport-related activities.

It is important to understand how Wolfram Syndrome affects people physically, as well as emotionally and functionally. Understanding which symptoms interfere most with which activities can help guide the development of interventions towards targets that are most meaningful for individuals.

One approach that may be helpful for managing the impact of symptoms on QoL is Occupational Therapy (OT). OT may help people learn methods to manage symptoms and improve their level of engagement in activities that they care about. For example, OT can help people manage fatigue, use adaptive equipment (like canes or walkers), and adapt school and work environment to the person’s needs.

We thank our participants and families for completing these long questionnaires! The information provided is very valuable guiding our and other labs’ future research.

Sincerely,

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*Thanks to Marisol Ponton, Tasha Doty and Samantha Ranck for writing the QoL summary.