Wilson Disease Registry Study.

Since the initiation of the Wilson Disease Registry in December of 2017 we have enrolled 122 subjects, including 81 adult and 31 children, and we have completed 217 study visits. Despite delays in enrollment and follow-up visits due to the COVID-19 pandemic, study activities have gradually resumed, and we are currently active at 6 study sites in the US, UK and Germany. (https://clinicaltrials.gov/ct2/show/NCT03334292?term=schilsky&cond=wilson&draw=2&rank=3)

In 2020, our collaborator on this project, Dr. Chris Harrington in Surrey, UK was senior author on a publication describing a novel technique for accurately measuring copper in proteins from human specimens. (Harrington et al., Biomedical copper speciation in relation to Wilson's disease using strong anion exchange chromatography coupled to triple quadrupole inductively coupled plasma mass spectrometry. Anal Chim Acta. 2020 Feb 15;1098:27-36. doi: 10.1016/j.aca.2019.11.033. Epub 2019 Nov 15. PMID: 31948584.) This work has important implications for monitoring treatment in patients with Wilson disease. Recently, the Harrington lab welcomed a senior health scientist and bio-inorganic chemist Dr. James Coverdale to their research team to help advance techniques for copper measurement in specimens from our registry biorepository.

Samples from the Registry biorepository were also used by our collaborator Prof. Sihoun Hahn from Seattle Children's Hospital to develop a novel test for measuring ATP7B (the protein affected in Wilson disease) in blood spot samples. Results of this work was published in the journal Gastroenterology ,and further testing of this assay is ongoing under his direction .Christopher J. Collins, Fan Yi, Remwilyn Dayuha, Phi Duong, Simon Horslen, Michelle Camarata, Ayse K. Coskun, Roderick H.J. Houwen, Tudor L. Pop, Heinz Zoller, Han-wook Yoo, Sung Won Jung, Karl H. Weiss, Michael L. Schilsky, Peter Ferenci, Si Houn Hahn. Direct Measurement of ATP7B Peptides Is Highly Effective in the Diagnosis of Wilson Disease, Gastroenterology, Volume 160, Issue 7, 2021, Pages 2367-2382.e1, ISSN 0016-5085, https://doi.org/10.1053/j.gastro.2021.02.052. (https://www.sciencedirect.com/science/article/pii/S0016508521004571)

Recently, the Journal of the Academy of Consultation-Liaison Psychiatry accepted for publication a manuscript describing results of work by our Registry research group. This work, led by senior author Dr. Paula Zimbrean focuses on the effect of liver disease, neurological disease and mental health issues on the quality of life in patients with Wilson Disease. (Michelle A. Camarata, Aftab Ala, Ayse K. Coskun, Yanhong Deng, Regino P. Gonzalez-Peralta, Kaitlin R. Maciejewski, Amar Patel, Susan Rubman, Uyen To, Ricarda Tomlin, Michael L. Schilsky, Paula C. Zimbrean, The Effect of Mental Health, Neurological Disease, and Liver Disease on Quality of Life in Patients With Wilson Disease, Journal of the Academy of Consultation-Liaison Psychiatry, 2021, ISSN 2667-2960, https://doi.org/10.1016/j.jaclp.2021.04.004. (https://www.sciencedirect.com/science/article/pii/S2667296021000781)

Dr. Zimbrean and our study team are also in the process of preparing another manuscript focusing on Major Depressive disorder in patients with Wilson disease. Our research found a significant incidence of Major Depressive Disorder and bipolar disorder in patients with Wilson Disease. This finding suggests that standardized mental health instruments used to detect mental health problems should be incorporated into the routine care of Wilson Disease patients.

Our ongoing research also highlighted the urgent need for development of effective psychiatric and neurological assessment tools for adults and children with Wilson Disease. Dr. Patel, the lead adult neurologist for the Registry at the Yale Site, in collaboration with 5 expert neurologists from the US and Europe, is working to validate a modified standard neurological assessment of Wilson disease patients to establish a more effective and time-efficient neurological assessment tool. Meantime, with our growing pediatric cohort, we are hoping to contribute the development and validation of standardized assessment tools for children as well.

Recently results of ongoing work analyzing aspects of liver disease in registry subjects was submitted for consideration for presentation at the upcoming American Association for the Study of Liver Disease (AASLD) meeting planned in November 2021. We will share the highlights of this abstract with you in the near future.

We want to extend our thanks to the Wilson Disease Association for their continuing support for this effort, and to the many adult and pediatric patients and their families for taking the time to be part of the Registry project. With your support, we are hoping to continue this project and make major contributions to our understanding of the natural history of Wilson Disease and the care of our patients with this disorder.

This is an interesting article. For the electronic version of newsletter, how about doing links to the articles? I've checked them all out:

1. Sends you to correct article
2. Suggest using this link since the existing one takes you to a list https://pubmed.ncbi.nlm.nih.gov/31948584/
3. https://www.gastrojournal.org/article/S0016-5085(21)00457-1/fulltext
4. https://www.sciencedirect.com/science/article/pii/S2667296021000781?via%3Dihub

For the hard copy you could create footnotes.

Jamie add this for a closing paragraph:

The Wilson Disease Association relies on fundraising campaigns and individual donations to support the important work of the Patient Registry Study. If you'd like to support the ongoing study, please click here https://www.wilsonsdisease.org/ways-you-can-help/donateThe first WD patients who enrolled in the study are now participating in the fourth year of their five-year commitment. New patients are still being enrolled.

Jamie

For the email version we can use the links in Blue instead of the footnotes. For the newsletter we should put the footnotes at the end.