Registry Study-
How we get scientific answers
by Rhonda Rowland

I’ve read and reported on scientific studies for my entire career, and now I’m finally getting my chance to be a part of one. It’s a patient registry study for Wilson disease! Medical studies yield the information doctors rely on to diagnose our diseases. They guide them towards a best course of treatment. If there is a potential new treatment, well-executed experiments evaluate its risks and whether it really works. So without carefully planned scientific studies, the practice of medicine would not advance safely.

And behind all those statistics are the people with the diseases. People who volunteer their time and share about their disease experience. Sometimes they even take personal risks in order to help others.

Registry studies: why we need them
In 2018, the Wilson Disease Association announced that our disease now has its own registry study. For someone like me, who has been living with the diagnosis of Wilson disease for more than 35 years, this was exciting news! While medicine has made progress in detecting it early – so that people can begin treatment before debilitating symptoms set in – far too often doctors fail to make the diagnosis in time, before Wilson’s effects become irreversible or even fatal. Findings from a registry study could help solve that devastating problem.

What is a patient registry study?
It’s a systematic collection of as much data as possible from people who have a particular disease. That includes: signs and symptoms leading up to the diagnosis, how a diagnosis was made, which treatment they used, how well the treatment worked, what day-to-day life is like living with the disease.

Becoming a part of the WD registry study is easy. The announcement I read said to email Dr. Michelle Camarata at the Yale Wilson Disease Center of Excellence. She wrote back asking when I wanted to come, and I suggested July. She got back to me with a couple dates to choose from. I asked if I should bring copies of my medical records; she said that would be great.

continued on page 5

A bittersweet gift

Last year we had a short article in the Copper Connection with a gentle ask to have the WDA remembered in estate plans. We would rather have all our loved ones with us forever but if there is one constant, it’s that everyone eventually leaves this life. This month, we received a check for $7500.00. The deceased died in January of 2019 and under his estate plan, he left the Wilson Disease Association that gift. His law firm in Maryland sent us a check with that explanation.

We wanted to share this story with all of you because it was a beautiful gesture and the deceased is not here to thank. We had no idea we were in an estate plan or that his death occurred. This was the first gift from an estate plan and it made a big impact on us. We are asking again, if when doing estate planning, would you also consider the Wilson Disease Association and the work it does?
# Table of Contents

- Registry Study: How we get scientific answers .............................................. 1
- A Bittersweet gift ................................................................................................. 1
- Letter from the Executive Director .................................................................... 3
- In memory of Jack Levin .................................................................................... 4
- The Wilson Disease patient registry: Creating our legacy .............................. 6-7
- 2020 WDA Conference ....................................................................................... 8
- The Big WOW 2019 ........................................................................................... 8
- Patient privacy warning ...................................................................................... 9
- Wilson Disease treatment options in UK ......................................................... 9
- Thanks for your support ..................................................................................... 10
- Membership form ............................................................................................... Inset

**NEWSLETTER DEADLINE:**
If you would like to submit an article to be published in the next printed edition of The Copper Connection, the deadline for submission is January 31st, 2020. Please e-mail your article to the WDA office at info@wilsonsdisease.org

**The Wilson Disease Association**

The Wilson Disease Association is a nonprofit 501 (c)(3) organization. The Copper Connection is a semi-annual newsletter published by the Association that informs members of findings in the area of Wilson Disease. There is no copyright. Newsletters and other publications can disseminate any information in The Copper Connection. Please cite attribution to the Association and the author.

**The Copper Connection**

1732 1st Ave #20043
New York, New York 10128
414-961-0533 • Toll Free: 866-961-0533 • Fax: 414-962-3886
info@wilsonsdisease.org
www.wilsonsdisease.org

**Our Mission Statement**
The Wilson Disease Association funds research and facilitates and promotes the identification, education, treatment and support of patients and other individuals affected by Wilson Disease.

---

**Letter from the Executive Director**

Dear Copper Connection Readers,

I recently sent out an appeal by mail and email about the Patient Registry. You hear about it often because it is the largest undertaking we have ever started and perhaps the most important. It is so important the WDA has to mention it in almost everything we do. In the center of this Copper Connection is a special section illustrating what has been discovered and what plans for the Patient Registry project are in the future.

It is worth repeating that for years, a medical practitioner’s only goal for the treatment of a Wilson Disease patient was the survival of the patient. Survival! That’s all. If you are the patient you probably wondered, “Is that all there is?” “What about my quality of life?” “Can’t you help with my depression?” “Don’t I have more treatment options?” You already know that here have been too few options and too few answers. This project is providing the researchers with answers to their questions and enlightening them about other things to consider. We support it knowing that they will experience those epiphanies of understanding or “Eureka” moments!

We finished the first two years of research and are building momentum. We are entering year three. We are concerned because even though some people have been extremely generous our donations have been down in 2019. As you read this Copper connection and look at everything the WDA is trying to do, know that we can’t do it without a lot of support. Please give now and keep this project going. Would you risk not knowing what might be discovered in years three, four and five? We have only met half of our financial obligation and the project gets more expensive as Registry sites are added. Our financial obligation for year three is more than $800,000.

Please give now and keep this project going. Would you risk not knowing what might be discovered in years three, four and five? We have only met half of our financial obligation and the project gets more expensive as Registry sites are added. Our financial obligation for year three is more than $800,000.

Respectfully
Judi Keller
Executive Director
IN MEMORY OF JACK LEE LEVIN
July 11, 1932 - October 6, 2019
Jack L. Levin, was born July 11, 1932, and died peacefully in his sleep on the morning of Oct. 6, 2019. He was 87 years old.

Jack was born in Burlington, NC to parents Stella and Sol Levin and was the baby brother to Seymour and Ruth. He graduated from Georgia Tech and served in the US Air Force as a Lieutenant. After serving his country, he returned to North Carolina with wife, Mimi, and daughter, Jana and settled in Greensboro. He began work in his family's business in Burlington, Levin Brothers Inc. and had two more daughters, Linda and Karen. He and his brother took over the business when Sol and Stella retired and were very successful. Jack became involved in industry trade groups such as the Institute of Scrap Iron and Steel and the success and reputation of his business was very important to him.

Jack was a very artistic person and loved taking photos of beautiful scenes across the USA. He traveled widely and spent many hours immersed in this wonderful hobby. His photos pleased both himself and many others. Whomever has one of his photographs considers themselves in a lucky club. Besides loving photography, Jack spent many hours writing poems, stories, and cards, usually for special occasions. He would become very involved in these projects, forgetting to eat or feed others who were with him. He had the drain of a businessman but the soul of an artist.

In his life, Jack had to overcome some serious health issues. Diagnosed as a young man with Wilson’s Disease, he spent a great deal of time raising money and awareness for this rare disorder. In his 70’s, he fought back from a terrible home accident, a scare to those who knew and loved him. After the accident, his health curtailed his various movements ranging from smiling to drawing. Thank goodness I had just gotten a pedicure, because my feet were ready for their video moment when they needed me barefoot to test my balance!

In his life, Jack had to overcome some serious health issues. Diagnosed as a young man with Wilson’s Disease, he spent a great deal of time raising money and awareness for this rare disorder. In his 70’s, he fought back from a terrible home accident, a scare to those who knew and loved him. After the accident, his health curtailed his various movements ranging from smiling to drawing. Thank goodness I had just gotten a pedicure, because my feet were ready for their video moment when they needed me barefoot to test my balance!

Jack’s generosity didn’t stop with family and friends. He gave to many charities both in the Jewish and general community, including programs he established at Wake Forest including the Jack Levin Professorship in Gastroenterology and Hepatology, The Stella P Levin Lectureship in Hepatology, and the Vardaman M Buckalew, Jr Fund in Nephrology. At Georgia Tech, the Jack L. Levin Scholarship Endowment Fund supports NC students who attend Georgia Tech.

Jack is predeceased by his parents Stella and Sol Levin. He is survived by his brother Seymour (Carol) Levin of Greensboro, and sister Ruth Geisenheimer of Chicago, his daughters and their husbands: Jana (Andy) Fields of Greensboro, Linda (Sick) Hixon of Tallahassee FL, Karen (Kevin) Bauerstedt of Greensboro, and his former wife, Mimi Levin. Jack is also survived by his loving grandchildren: Dana Fields (Joseph Streeter), Stacy Fields, Sam Hixon, Nathan Hixon, Sophie Little (Jason), and Lindsey Ansel. He loved them each with a ferocious protectionism. They could do no harm in his eyes. He gave them each the gift of higher education and much more.

Registry study cont.

What patients do in a registry study
On the morning of my appointment, Dr. Camarata led me to a large exam room. Since I’m so interested in Wilson’s that I’m writing a book about it, I had a lot of questions! I found out that she’s completing her fellowship training in hepatology and Wilson disease and will soon be working with Wilson’s patients in England. (Dr. Camarata now practices as a hepatologist at the Royal Surrey County Hospital, which is about an hour outside of London.) She told me this study may help answer an unsolved mystery: Why does our disease look so different in different people? Why do some have primarily liver, others neurological, and others psychiatric symptoms? So far, genetic mutations don’t explain it. The cause of the variations could be environmental, dietary, or some other factor.

Convincing people to volunteer for studies can be a challenge — but not this one. Dr. Camarata said she has enthusiastic volunteers. However, since our condition is so rare and each participant is so valuable, more are still needed. And by the end of 2019 the registry study should be up and running at the WD Centers not only at Yale but in Florida, Texas, England, and Germany.

That’s excellent news, since registry studies can also help identify people who may be good candidates for other studies, like studies testing new drugs.

Three specialists collect data
After Dr. Camarata asked me the details about my health, both leading up to and after my Wilson’s diagnosis; she gave me a brief physical exam – listening to my heart and examining my abdomen near my liver. Then Dr. Michael Schlisky, who heads Yale’s WD Center stopped by to ask some additional questions.

Next, a neurologist came in. He set up an iPhone tripod to capture video of his part of the exam; asking me to make various movements ranging from smiling to drawing. Thank goodness I had just gotten a pedicure, because my feet were ready for their video moment when they needed me barefoot to test my balance!

The final specialist I met with was a psychologist. She tested my memory, asking me to remember 5 things that I’d be asked to repeat at varying times (I’m still wondering if I got them right!). The rest of the exam was yes-or-no questions about my thoughts and behaviors. From what I’m leaning in research interviews for my book on Wilson’s, I believe this part of the study may provide the most important information about living with this disease. It will reveal the depth and extent of the disease’s psychological burden – on area where we need to know more.

You’ll need to give blood for this study. But those of us with Wilson’s are used to that. And this time we may be given the genetic information that will help unlock a newborn screening test, and help with future gene therapy studies!

In total, my appointment for the registry study took about three hours.

Registry study commitment: 5 years
That left plenty of time for me to take the train from New Haven to Stanford to visit my high school BFF. Ironically, she’s in a study too, but hers is to compare three treatments for advanced ovarian cancer. Somehow we found ourselves laughing, if ruefully, at where we are 40 years after we met – me with a rare disease and her with a tough cancer to beat.

Before leaving, I told my friend that I’d be returning to Yale once a year for 5 years for the study, so we have those visits to look forward to. “If it’s still here,” she said. I told her I had faith that she would be. That’s why we do these studies. It’s a chance to do something practical towards the hope that these diseases will become more and more treatable.
What we have accomplished so far

DATA FROM YEARS 1 & 2 HAVE ALREADY YIELDED INFORMATION THAT WAS PREVIOUSLY UNKNOWN.

We have a current enrollment of 75 patients: 52 adult and 19 pediatric. They range in age from 2.5 to 74 years old with an average age of 36.

From this population we have learned that 50% of the adults have a major depressive disorder. This indicates that we need to evaluate depression and anxiety in the pediatric population as well.

This illustrates the valuable progress that has been made in two short years with only a small cohort of patients at one center. Years 3 – 5, which will include a larger (>225 patients), a more diverse cohort of patients from other registries will yield information that is even more significant.

Moving forward

YEARS 3 – 5 WILL BE CRITICAL IN EVALUATING THE FOLLOWING STANDARD TESTS USED TO MONITOR FOR GOOD COPPER CONTROL. They will be carefully studied and conclusions will be made about what is the best way to treat and monitor WD patients. Improved rating scales could be developed using this information. Clinical trials may also be designed to determine the best practices that will benefit current and future patients.

- GENETIC- Molecular analysis of the ATP7B gene, will be done on each patient enrolled. The analyses will all be done by Dr. Siouan Hahn at Seattle Children’s Hospital in order to produce consistent results.

- NEUROLOGIC EVALUATION- All enrollees will be evaluated for neurological symptoms using a testing instrument called the Unified Wilson Disease Rating Scale (UWDRS). They will also be videotaped (with consent) to capture subtle differences that may not be picked up by the UWDRS.

- COPPER METABOLISM- As previously stated, ceruloplasmin and serum copper will continue to be tested and level of “free copper” determined. 24 hour urine copper for copper content will also be analyzed. Urine zinc levels for patients on zinc treatment will also be measured.

FAST FACTS

- It is the first of its kind in the world.
- Another study was conducted using data from the registry.
- A multi-center registry enables us to collect data and bio-specimens (blood, urine) from a diverse population of patients, both adult and pediatric.
- It is a study that will require at least five years.
- It has a cost of approximately $3 million dollars as it is currently structured.
- We receive no government funding for it.

WATRS IS THE PATIENT REGISTRY & WHY IS IT SIGNIFICANT?

The Wilson Disease Patient Registry (WDPR) is a natural history study. Patients, medical practitioners and caregivers have waited years for this type of study in order to address unmet needs and unanswered questions about diagnosis, treatment, and monitoring. This type of study follows a group of people over a period of time who have a specific medical condition or disease. The study collects health information in order to better understand how the disease develops and how to treat it. The data collected will also be used to evaluate new therapies. In addition to data, the WDPR is also collecting and storing bio-specimens from each patient enrolled. All specimens are analyzed by the same laboratory in the UK to ensure consistency and stored in a central facility at Yale University.

AIM OF THE WDPR STUDY

By analyzing patient data and bio-specimens, the goal is to determine the ideal tests needed for diagnosis and the best monitoring practices. When compiled and analyzed by all study sites investigators, this information will reveal the proper use of current treatments and guide the development of future therapies. These data may also help to define ideal patient outcomes.
Welcome patients, caregivers, family and friends to our next annual conference to be held in Downtown Denver. The Convention and Visitors Bureau provided us with some wonderful videos about the area and taking the new high speed rail from inside the airport terminal to Downtown. We have the links on our website and suggest you take a moment to view them. Transportation from the airport to the hotel is very simple and inexpensive when using the train.

The hotel gave the group rate for three days prior to our dates and extended them to three days after our conference for those of you who might want to stay and vacation in the area. We are staying at the Grand Hyatt Hotel Downtown Denver.

The hotel built a special link for reservations for our event that is on the conference page of our website. It already provides the discount code. https://www.hyatt.com/en-US/group-booking/DENRD/G-WILO

We are working on a program and do not have conference ticketing information at this time. When we do, it will be on our website, sent out in emails, appearing in Facebook and in the next Copper Connection. We believe it will be ready late in January.

The program is typically a reception for everyone attending to mingle and socialize on Friday evening. The conference check in is Saturday morning. After the conference there is a sit down banquet on Saturday evening. No times are set yet. Times, menus, tickets will all follow.

The Big WOW | 2019

Our volunteers raised a slightly over $50,000 in the 2019 Big Wow. When one considers that it is random volunteers step up and organize a walk, this is something short of a miracle. This $50,000 is pledged to the Patient Registry.

These wonderful volunteers not only plan a walk and promote their event among family and friends but they solicit donations. These donors are recognized in the donation section of our Copper Connection. All of these volunteers step out of their comfort zone to do and ask. The results are remarkable. The WDA is very grateful and is recognizing their efforts here.

VOLUNTEERS

This year approximately $50,000 was raised to fund the patient registry.

Wilson disease treatment options expanded in the UK

Professor Aftab Ala, MD, who is the WDA Center of Excellence Director of the Royal Surrey Hospital in Guildford and Co-Investigator of the WD Patient Registry there, is now able to provide care to more WD patients. He received an official appointment to the faculty of King’s College London. Below is a description of King’s Wilson disease services and the team members.

Adult Wilson Program: Director: Professor Aftab Ala MB BS MD PhD FRCP, Consultant Hepatologist, Institute of Liver Studies, Kings College Hospital (KCH), London UK. We accept National and International referrals. Email aftab.alal@nhs.net

Team Members: Professor Aftab Ala and Dr Adrian Bomford - Hepatology, Institute of Liver Studies, KCH; Transplant team, Institute of Liver Studies, KCH; Dr Michael Samuel, Consultant Neurologist with specialist interest in movement disorders, botulin toxin, deep brain stimulation movement disorders, KCH; Professor Richard Thompson Consultant Paediatric Hepatologist and specialist interest in Genetics, KCH; Research and Development and Clinical Trials; Psychiatry and Psychology; Ophthalmology.

Patients privacy warning: We know you want to help, but know this first!

We love that you are so willing to help when called upon but please be very cautious about completing surveys or providing your personal information to pharmaceutical companies, researchers, public relations firms, marketing representatives, or other entities who may contact you through inspire or social media, or even meet you at a WDA conference. These people may promise you money or other things that never occur or are not true. If there is no contract, there is nothing guaranteeing that you will be paid. Recently, we have become aware of such people contacting WD patients directly with surveys or requests for interviews. It is a company well known for promising stipends and not paying. The WDA will be issuing a written statement to pharmaceutical companies that may need patient information indicating that we will help those who can document a legitimate need for such information and that they will protect any patient information they collect. That means we will help companies to promote legitimate patient surveys, or we will tell patients about clinical studies. Hopefully, that should eliminate the need for them to hire strangers to stalk you on Facebook, inspire and even at our conferences. So, if you are approached by someone asking you to respond to a patient survey or be interviewed, and you haven’t heard anything about that particular effort from the WDA, please contact us at info@wilsonsdisease.org before responding to them.

The WDA spends a lot of time researching and talking to pharmaceutical companies. We treat them with courtesy if they treat us with courtesy and respect our patients. We know who is new on the scene and who is about to start clinical trials. We do whatever we can to advance new treatments and research for you.

You all know that there have been many complaints about the high cost of drugs or insurance companies that won’t cover the prescription costs. A flurry of pharmaceutical companies has jumped into the possible development of Wilson Disease treatments because they are aware of the profits that can be made. They are businesses and not philanthropists, so their first priority is to their business and shareholders. Some are more ethical than others. Others are so new that sometimes they are hiring marketing companies to do research even though these marketers do not have any background in the disease at all. WDA will always announce legitimate clinical trials and surveys on our website, Facebook and emails. If you have any questions about any entity asking you for information you should contact us and we will verify them.
Wilson Disease Awareness Month is March

Facebook users should watch for activities created especially for awareness month. We are working on plans for the month right now and there will be more news to follow. Watch for the awareness month logo.