Newsletter

Legacy News Vol 9 | November 2023

GENETIC ALS & FTD: END THE LEGACY

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A Note from the Chair, Jean Swidler

I am writing this from Basel, Switzerland, where the International Alliance of MND /ALS Associations just closed its annual meeting. Our Vice Chair Cassandra and I have been overwhelmed with the gracious welcome from the Alliance team and many across the global spectrum of organizations and professionals caring for the ALS community. Our presentation on the support pillar of our mission was well received, and we have been able in both the large room discussion and informal chats to advance the interests of our community. Earlier in November, we sat in on the National Institute of Health's Alzheimer's Disease and Related Dementias Roundtable with a different field of committed individuals. The team of people impacted by Genetic FTD who were able to be present ensured our community was not forgotten in that conversation. With the interests of our families being up for debate in so many places with so many different conveners, we are so thankful to our donors and sponsors who make the ability to participate in the face-to-face conversation possible.

We are so heartened to come close to the end of our first year in operation with the initiatives and passion of our volunteers and advocates not letting up for an instant.

Finally, we cheer our teammate Wanda Smith, who received a well-deserved nomination for the Everylife Foundation for Rare Disease RareVoice award. Read more about it here.

END THE LEGACY
GENETIC ALS & FTD

Inside this edition:

2023 ALS & MND Alliance Basel, Switzerland

Vice Chair Cassandra Haddad and Jean presented our support efforts at the 2023 ALS & MND Alliance Meeting in Basel, Switzerland. The talk was well received, and the meeting provided many opportunities for connection and discussion on topics important to our community. You can see the slides here.

NIH Alzheimer's Disease and Related Dementias Meeting

Four members of the genetic ALS & FTD community attended this exciting meeting in Bethesda near the NIH campus. Cheers to Wanda, Betsy, Linde, and Jean! Each contributed important points to the discussion, and we learned exciting news about possible government-supported trials in FTD. Wanda pushed for broader genetic testing in dementia, Betsy shared that the ability to participate in prevention trials while remaining blinded to one's genetic status is desirable, Linde pleaded for earlier diagnosis, and Jean pushed the prioritization of TDP-43 not only for ALS and FTD, but also for people impacted by LATE, or TDP-43 predominant late-life memory impaired dementia.

RESEARCH NEWS

ALS ONE Research Symposium

There were fascinating commentaries on the possibilities of early initiation of trials or therapies in the at-risk population from Drs Miller and Schnieder at the ALS ONE Research Symposium. While neither offered definitive stances, we appreciate their reasoned exploration of these ideas. We so appreciate them and the other researchers who participated, and ALS ONE for making the event and recordings accessible to all. Watch Dr Miller's talk here, and see the panel discussion where Dr Schnieder discussed early treatment with ASO's at about the 25-minute mark here.

6th Annual ALS ONE Research Symposium

Timothy Miller, MD, PhD.

Neil Schneider, MD, PhD.

Signs of Pre-clinical Disease

A new study from researchers in the Netherlands showed carriers of MAPT and C9ORF72 displayed brain volume size in specific regions below the 5th percentile compared to healthy controls at age 45 without evidence of cognitive impairment. This is another corroboration of a long pre-clinical disease period in our families.

PATIENT-LED RESEARCH

NIH Alzheimer's Disease and Related Dementias Meeting

Our presenters for the inaugural workshop on care and advice for the at-risk genetic ALS and FTD community in September submitted their summary section for inclusion in the upcoming meeting white paper. Great work, Cassandra, Linde, and Jean!

Announcing Upcoming Genetic Research Education Series

We are excited to announce we will be providing a webinar series intended to explain to specific genetic communities what research people with their mutations may be eligible to participate in. The presentations will follow a similar format, including discussions on genetic counseling, the experience of people with that gene participating in research, and an expert overview of what research is available to volunteer for.

Genetic Research Education Series Dates

SOD1 on Tuesday, January 23rd

C9orf72 on Tuesday, January 30th

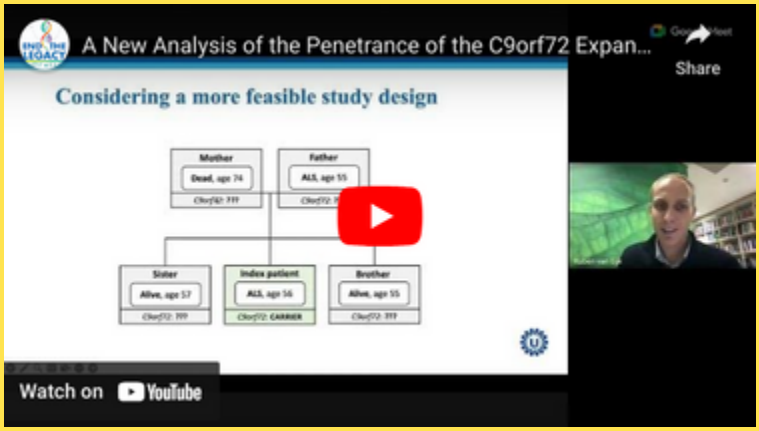
GRN on Tuesday, February 6th

MAPT on a date TBD

If you would like to attend or learn more, please email us at info@endthelegacy.org.

More information will be shared in the new year.

Webinar: A New Analysis of the Penetrance of the C9orf72 Expansion



We were pleased to host the first public discussion of the new analysis of the penetrance of the C9orf72 mutation. We thank Professor Ruben van Eijk for providing his time and for the talk and discussion. You can watch it on our YouTube here.

Peer Support Hour

We continue to hold our Peer Support Hour on the 3rd Wednesday of the month, with the next installment coming up on December 20th at 7 pm Eastern. The compassionate facilitation of our Board member Linde Jacobs makes this a welcoming space for anyone in our community who wants connection.

Sign up here.

Genetic ALS & FTD: End the Legacy Team Meetings

Our weekly community meeting can also be a space for connection and support even where the subjects are more action orientated, and we detailed this connection in our presentation at the Alliance meeting - if you are not on our weekly email list and would like to be, please reach out at info@endthelegacy.org.

END THE LEGACY
GENETIC ALS & FTD

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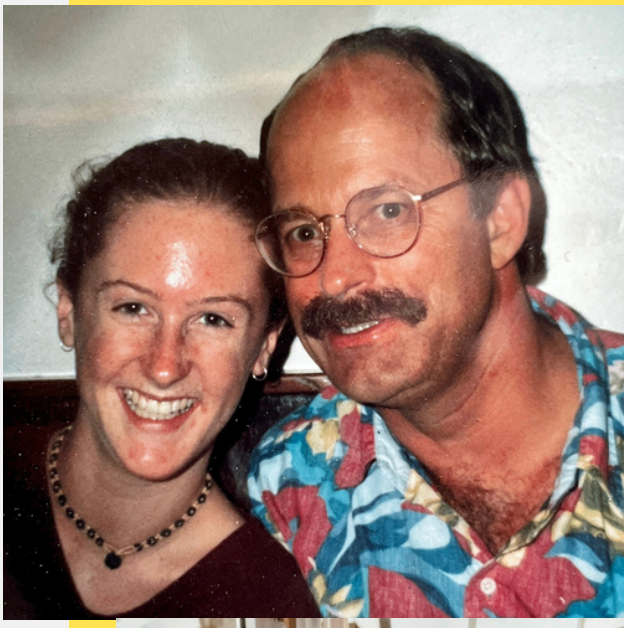
MEMBER SPOTLIGHT

Jesse Travis, Archive Team

Interviewed by Mindy Uhrlaub



Can you please tell us something about the gene mutation that runs in your family?

For a long time, I didn't know which gene mutation ran in our family. My father died of ALS in 1999 and was not tested, though we knew it was genetic because his father had died of ALS in 1957, his father's sister in the mid-80s, and then later one of her four children in the 2000s. We weren't really in contact with my great aunt's descendants, until a few years ago when my brother and I reached out to them and learned that the relative who had died in the 2000s had been tested, and they had learned the variant was SOD1.
It turned out that it is one of the fast progressing variants that qualifies for the ATLAS trial. After taking necessary steps, I decided to get tested in March of this year through ATLAS, and found out that I did not inherit the mutation.

Short of a cure, what is your greatest wish for people with genetic mutations for ALS/FTD?

That they have access to quality mental health care, and to support from a community like End the Legacy, where so many of us have found agency and purpose in working together to advocate for ourselves, our loved ones, and all people who carry or potentially carry genetic mutations for ALS/FTD.

Tell us something about yourself that we wouldn't already know.