Genetic ALS & FTD: End the Legacy

April 2023 Newsletter

Julie Granning, Editor

A Note from the Chair

Tofersen, now Qalsody, has been approved by the FDA for treatment of SOD1 ALS.

On April 25, the FDA granted accelerated approval for Qalsody (Tofersen). This approval is big news for our community as it is the first genetically targeted treatment for ALS and also recognizes Neurofilament (NFL) as a biomarker for neurodegeneration. This is a monumental development and heralds a new era for our community—but only if we capitalize on it. We will do everything possible to ensure no SOD1 carrier must suffer any loss of function before having the chance to access this treatment.

Recognizing there are huge issues necessitating an organized community response for any issues that may arise about Qalsody or the presymptomatic Atlas Study, we have formed a Qalsody Committee. If you are interested in being a part of this team, email our vice chair Cassandra.

Finally, knowing that this is a huge achievement that so many of us have participated in as trial participants, relatives of trial participants, or just strongly advocating for it, we are hosting a celebration!

If you are excited about Qalsody being approved, come to our Celebratory Hangout on Tuesday, May 9, at 8:00 p.m. (EST)! meet.google.com/jbb-yjbj-pjh

Jean Swidler, Chair

Advocate **Support** **Educate** **Research**

Stay Updated

We have lots of ways to keep up with us, like our new Instagram account, thanks to Amy Edelstein, MD, a member of our new Social Media Committee. Check out our recent video shorts in reaction to the great Qalsody (Tofersen) news and be sure to like, follow, and subscribe!

Follow us Instagram
Tweet us @End_The_Legacy
Join us Facebook
Watch us Youtube

Mission Statement

The Genetic ALS & FTD community is large and growing. ALS & FTD are terminal conditions, and being at a heightened risk for them can have profound impacts on people and families. We organized Genetic ALS & FTD: End the Legacy to provide educational and support resources to, encourage and promote research about, and advocate for the genetic ALS & FTD community.

ADVOCACY

A Legislative Update from Bill Musick

"Discrimination based on genetic information is alive and well in most states, including South Carolina. Citizens with ALS (and other) genes have been denied the purchase of life and LTC (long-term care) policies that would help keep them off of the government coffers if they became affected by ALS.

The South Carolina legislature has introduced legislation to prevent genetic discrimination that some experience. The Honorable Tommy Pope, Speaker of the House, introduced H 4218 - Genetic Protection to prevent discrimination based on genetic information. The bill is in the Committee on Labor, Commerce and Industry and time is running out for the 2023 session that adjourns around May 12, 2023. If we can get this bill out of committee and onto the floor, it may pass this year. But time is of the essence.

If you or family have a residence in South Carolina, please reach out to committee members and encourage them to support this legislation. This same legislation passed in the Florida House 110–0 and 35–3 in the Senate and governor DeSantis signed."

More Legislative Updates

If you have connections in New York, have them reach out to their state legislature about this bill on genetic protections in insurance: https://nyassembly.gov/leg/?bn=a2083a

If you want to make sure the Genetic ALS & FTD community is included in any efforts you are involved in, please reach out to us! Email us at geneticalsftd@gmail.com.

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SUPPORT

We have several members attending the AFTD Education Conference on May 5 in St. Louis, where they hope to be present with others in the FTD community and ensure people don't feel alone.

EDUCATE

Professor Guillaume Hautbergue is an expert in RNA biology and he and his lab recently published a paper on a peptide that was able to prevent the death of nerve cells and protect them from degeneration in C9orf72–ALS/FTD animal models. He also recently reported a gene therapy approach targeting the same biological process. He joined us recently for a talk: Watch here.

Upcoming Educational Webinars

Watch our social media for links to our upcoming events, including a healthcare worker panel discussion and a webinar on cardiovascular health and disease progression in genetic FTD carriers on May 25.

If you are interested in presenting to the Genetic ALS and FTD community, please reach out!
Email us at geneticalsftd@gmail.com.

RESEARCH

"AMP" ALS

The National Institutes of Health (NIH) recently announced a new public-private partnership created in response to Act for ALS: a new "Accelerating Medicines Partnership" administered by the Foundation for the NIH (FNIH). They held an initial planning meeting was held, in which our chair Jean, policy lead Karen, and member Wanda were able to secure last-minute invitations to. Thankfully, as the only people speaking for the needs of our community, we were able to raise the possibility of pre-symptomatic treatment for the genetic community today, and the need for pre-symptomatic carriers to receive private clinical care rather than merely be enrolled in research studies. After the meeting, the FNIH informed us that they were instituting a payment for participation rule for patient advocates. The amount requested is currently beyond the reach of our small non-profit. We are evaluating our options.

Baricitinib Trial

We have received news that the MGH (Massachusetts General Hospital) Baricitinib trial for pre-symptomatic C9orf72 carriers has increased the number they are enrolling from two to five, and one of our members appears likely to be accepted! We cheer all who participate in research! More details Here.

Synapticure

Synapticure has announced a program for the presymptomatic ALS community. The program focuses on the drug Riluzole - which is currently FDA-approved to treat ALS. See the program and enrollment details here.

Our website has a research page, including links to curent, genetic-focused research that you may be interested in: End the Legacy Research.

Member Spotlight

Wanda Smith, Interviewed by Mindy Uhrlaub

I understand you come from a family that carries the progranulin gene that causes FTD. Do you think it's important for people in families that carry genetic mutations such as progranulin to get genetic testing and why?

We recently learned that TMEM 106B is a protector gene for those who test positive for progranulin. It protects genetic carriers from disease. The Progranulin protein is comprised of seven granulins. Current trials seek to increase production of the entire protein.

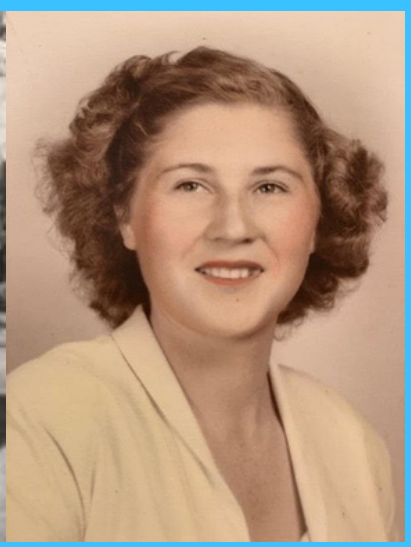
A study released Friday shows that repairing granulins #2 and/or #4 may be a potential treatment option instead of repairing the entire protein. This is terrific news as it is a better understanding of the cellular mechanisms. Progranulin may play a larger role in other neurodegenerative diseases too.

Can you speak a little about your family's experience in clinical trials and "expanded access" to clinical trials?

Family members can participate in many of the clinical trials once they are known as progranulin positive carriers. They describe to me the trials as challenging, as the travel is demanding and the cognitive assessment criteria are often unpredictable. Sometimes they can pass the cognitive tests and sometimes they cannot. Clinical trials are exhausting for the caregivers and vary among family members. Once a genetic status is identified they can enroll in clinical trials. Most people enrolling in clinical trials will have some symptoms, however, progranulin trials will enroll asymptomatic family members as well.

Will you share a fun fact about yourself?

I love to fly kites!



Advocate Support Educate Research

From Founding Member Daniel Barvin

I'm excited to share with you that I'll be riding in Unbound Gravel, a 200-mile gravel race in Emporia, Kansas, on June 3, but my journey goes beyond the race itself. As a carrier of C9orf72, a genetic mutation that causes ALS/FTD, I have a duty to do everything I can to empower my community to take action to better their future.

I'll be racing to raise awareness and funds for Genetic ALS & FTD: End the Legacy. My goal is to raise $100,000 to support research, care, and advocacy for pre-symptomatic carriers at risk of ALS and FTD and their families.

I've lost my uncle, aunt and father to ALS and FTD. My future is at stake and I'm determined to be part of the solution. Your support can make a real difference in the lives of those at risk of these diseases.

I invite you to join me as I embark on this journey by donating to my fundraising campaign. Together, we can help make a positive impact in the fight against ALS and FTD. Every dollar counts, and your support means the world to me.

Donate Here: https://givebutter.com/EndTheLegacy

Thank you for your generosity and for being part of this important cause. Let's make a difference together!

A huge shout-out to Tyler Curtis and his wife's non-profit, Emporia Imaginarium, for hosting me during my stay in Emporia!

Another huge shout-out to LifeTime and Visit Emporia for hooking me up with a race pass! This campaign and journey wouldn't have been possible without you.

With heartfelt appreciation,
Daniel Barvin

Other News

The ALS Therapy Development Institute podcast "Endpoints" interviewed Jean: listen here.

The FTD story podcast "Remember Me" just released "Allison, Pt. 1" featuring our member Linde, telling the story of her mother. Listen on apple and Spotify.

The ALS/MND Alliance held a webinar on genetic testing in ALS, featuring Jean. Watch it here.

Press Release from the FDA approving Qalsody
News Release from Qalsody maker Biogen
Biospace Article On Qalsody

Let's keep our growing community strong and informed!

Visit us: https://www.alshf.org/end-the-legacy
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