Genetic ALS & FTD: End the Legacy Newsletter #1

Introduction

Welcome to our first newsletter! As the first patient-led organization dedicated solely to the needs of the genetic ALS and FTD community, we have much to accomplish and are proud to share updates on our initiatives and activities with you. Additionally, each month we will strive to share stories from the community—both those at risk and those who are working with us.

We Are Getting Organized

Our grassroots community group, which began on Facebook, has morphed into Genetic ALS & FTD: End the Legacy, a small-but-mighty non-profit. Jean Swidler is the founding Chair, and further structure is being established. We have formed an Archival Team, and have an incredible mentor in Dr. Terry Heiman-Patterson, Professor of Neurology at Temple University, and founder of the ALS Hope Foundation.

Our Founding 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.

Our newsletter will update you about our progress with each of the following priorities.

Progress Toward Our Goal

Support

Our most recent Friday meeting was attended by ALS Hope's executive director, Jamey Piggott, and Mary Holt-Paolone, MSRN, its clinical nurse counselor and nurse coordinator, who both expressed commitment and support to our group.

Mary will offer her expertise in setting up professional online support group sessions for us, navigating topics such as being new to our at-risk community; family planning and IVF treatment; and support for spouses and non at-risk family members.

Education

We are hosting researcher Dr. Matt Disney on Wednesday, February 22, at 7:00 p.m. (EST), to share more on his recent discovery of a potential treatment for C9orf72. Register to attend today.

We are planning more educational events. If you are interested in presenting to the genetic ALS and FTD community, please reach out! Email us at geneticalsftd@gmail.com.

Research

In addition to our continued participation as subjects in many genetic ALS studies, team members attended external meetings, including the California ALS Research Summit and the most recent NEAL's Clinical Research Learning Institute. We are grateful to the organizers for including us!

Patient-Led Research

Last fall, we concluded a survey about gene carriers' desire for presymptomatic treatment and the willingness to take risks, which had 174 eligible respondents. We are just now concluding drafting (and revising!) submissions for publication in the coming months. Beyond ensuring this valuable information is available in the literature, we were informed by the FDA they want this type of data, and we know it is important for government regulators to have citable references. Many thanks to Paul Wicks, Ph.D., for his assistance in preparing these submissions.

Patient-Requested Research

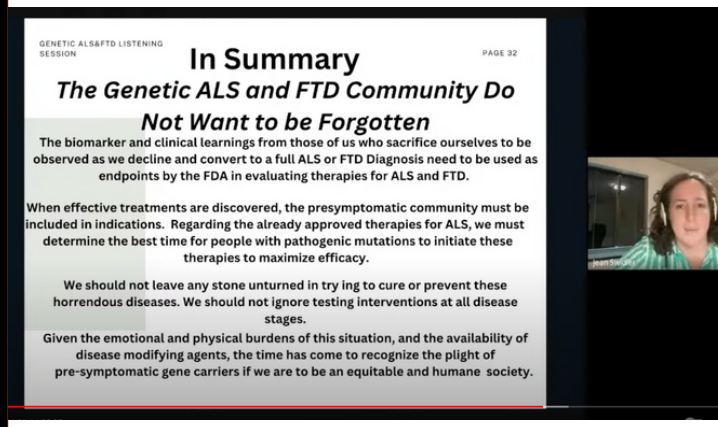
As urged by our community, Dr. Heiman-Patterson and ALS Hope are planning the first-ever expert workshop to establish what care is appropriate for those at risk of genetic ALS and FTD. More news to come on this exciting initiative as it develops!

Advocacy

FDA Listening Session

Over 40 FDA members, representing multiple FDA centers, attended our patient listening session for the pre-diagnosis genetic ALS and FTD community, featuring eight amazing speakers who shared their stories of how their lives have been impacted by genetic ALS and FTD genes C9orf72, Sod1, GRN, and Tardpb. A full written summary, approved by the FDA and linked to on their website, will be released in coming weeks.

So many people were interested in attending the private listening session, we hosted a redo of it for the public: watch it now.



A still from our redo of our FDA presentation , available in full on youtube.

Member Spotlight

For our initial member spotlight, here is a short excerpt from an interview with Jean in the ALS Hope Foundation's January Newsletter.

What is your story from the eyes of a caregiver to someone you loved?

The first thing my Mom, Kathy, told me when she was diagnosed with ALS, in the fall of 2017, was, "All I can think about is my mother Lillian hanging in a lift, over a commode at the breakfast table in my parent's house while your Dad and I were there, and my father Bernie saying 'Oh look, she smiled' as he wiped drool from her mouth—I don't want that to happen to me." I realized this was likely the image in her mind for the years she spent expressing anxiety about developing ALS, but it only came out after she was diagnosed. I think the perceived loss of dignity and independence was haunting her. So as her symptoms progressed, I tried to help maintain normalcy in whatever way I could. I would try and help get her to stores or have quiet moments together to delay that feeling as long as possible.

In coming months, watch for our team member Mindy's interviews!

Sign up for ALS Hope's newsletter on their website.

A Final Word

Let's keep our growing community strong and informed. Get involved—email us at geneticalsftd@gmail.com to join our efforts, or give us feedback (about anything—including how we could improve this monthly newsletter).

If you would like to support our efforts financially, please contribute via the ALS Hope Foundation. In the message section, note that your donation is intended for Genetic ALS & FTD: End the Legacy.

END THE LEGACY
GENETIC ALS & FTD