

ALS: Creating a Better Future for the Patient Community



A Series by:

Real World Health Care
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ALS is a progressive neurodegenerative disease that affects nerve cells in the brain and the spinal cord. It progressively affects voluntary muscle action, leading to the potential of total paralysis in patients at the later stages of the disease.

ALS: Creating a Better Future for the Patient Community is a recently published series of articles that spotlights the organizations and efforts dedicated to ALS research, treatment and patient services. Please accept this complimentary copy as our way of thanking you for your commitment to this community and advocating for healthier futures.

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What is ALS?

May is ALS Awareness Month, and Real World Health Care wants to support the ALS patient community by bringing awareness to this disease. The following “primer” provides need-to-know information about Amyotrophic Lateral Sclerosis (ALS), also known as Lou Gehrig’s disease.

ALS Defined

ALS is one of a group of conditions known as motor neuron diseases (MNDs) and accounts for 85 percent of MND cases.

ALS affects nerve cells in the brain and spinal cord that make muscles work in both the upper and lower parts of the body, making them stop working and eventually die. The nerves lose their ability to trigger specific muscles—such as those used for chewing, walking and talking—which causes the muscles to become weak, start to twitch and waste away. It is a progressive disease, with symptoms worsening over time, and eventually leads to paralysis and the inability to breathe.

A number of inherited factors have been found to cause familial ALS, although the cause of most other cases of ALS is unknown. Scientists are studying potential factors including heredity, environmental exposures, diet and injury.

ALS Symptoms and Diagnosis

The onset of ALS can be subtle, with the following symptoms gradually developing over time:

- Muscle twitches in the arm, leg, shoulder or tongue
- Muscle cramps
- Tight and stiff muscles
- Muscle weakness affecting an arm, leg, neck or diaphragm
- Slurred and nasal speech
- Difficulty chewing or swallowing

Eventually, people with ALS will not be able to stand or walk, get in or out of bed on their own, or use their hands and arms. They will have difficulty breathing as the muscles of the respiratory system weaken and will eventually need a ventilator to breathe.

ALS is primarily diagnosed based on detailed history of the symptoms and signs observed by a physician during physical examination, along with a series of tests to rule out other mimicking diseases. No one test can provide a definitive diagnosis, and it typically takes about a year before a final ALS diagnosis is made.

How Many People Have ALS?

Estimates suggest that anywhere from 15,000 to up to 30,000 people in the U.S. have ALS, with about 5,000 new cases diagnosed each year. The disease is more common in men than women, and most people are diagnosed between the ages of 55 and 75. About 5-10 percent of cases occur within families. In those families, there is a 50 percent chance each offspring will inherit the gene mutation and may develop the disease. Military veterans are about twice as likely to develop ALS as other population groups.

Most people with ALS live an average of two to five years after symptoms develop. People who are younger when the illness starts live slightly longer. About 10 percent survive 10 or more years. People with familial ALS usually do not fare as well as people with ALS who are not related, living only one or two years after symptoms appear.

There is no cure for ALS and no effective treatment to halt or reverse the progression of the disease. However, there are treatments that can help control symptoms, prevent unnecessary complications and make living with the disease easier. Supportive care includes drug treatments, therapy and special equipment to keep people as mobile, comfortable and independent as possible.

Creating a Better Future for People with ALS

The CDC has established the [National ALS Registry](#) to help researchers find answers to important questions about the disease. The Registry collects and analyzes patient data to help researchers look for disease pattern changes over time and identify whether there are common risk factors among individuals with ALS. By joining the Registry and taking the risk factor surveys, people with ALS will help provide a better picture of who gets ALS and risk factors for the disease. [Join the Registry](#).

The National Institutes of Health (NIH) has a list of federally and privately sponsored [ALS clinical trials](#). To find clinical trials on ALS, visit www.clinicaltrials.gov and enter the terms “amyotrophic lateral sclerosis” or “ALS” AND (your state). NIH also supports a [NeuroBioBank](#) that supplies scientific investigators with tissue from people with neurological and other disorders. Tissue from people with ALS is needed to enable scientists to study the disorder more intensely. Prospective donors can learn more at <https://neurobiobank.nih.gov/donors-how-become-donor/>.

The [ALS Association](#) has support groups for people living with ALS and their families and loved ones. Support groups offer a forum to share information and practical experience, a safe place to show emotions, and a gathering spot for speakers and caregivers to address topics of interest. [Find a support group in your state](#). The ALS Association also has [resources for caregivers](#).

There can be significant costs for medical care, equipment and home health care during later stages of the disease. People with ALS should become knowledgeable about their health plan coverage and other programs for which they may be eligible including Social Security Disability Insurance, Medicare, Medicaid and Veterans Affairs benefits.

Real World Health Care blog sponsor, the HealthWell Foundation, provides financial assistance to people living with ALS. Through the fund, HealthWell will provide up to \$15,000 in copayment or premium assistance to eligible patients who have annual household incomes up to 500 percent of the federal poverty level. To learn more, visit HealthWell's [Amyotrophic Lateral Sclerosis Fund](#) page.

Resources

[ALS Association](#)

[Centers for Disease Control and Prevention](#)

[National Institutes of Health: National Institute of Neurological Disorders and Stroke](#)

ALS and Precision Medicine: Learning from Patients Worldwide

The [ALS Therapy Development Institute](#) (ALS TDI) is a comprehensive drug discovery lab focused solely on amyotrophic lateral sclerosis (ALS). ALS is a progressive neurodegenerative disease that attacks the nerves responsible for voluntary movement. People with ALS can ultimately become completely paralyzed and the average life expectancy is usually between 3 and 5 years after diagnosis.

At ALS TDI, researchers believe that learning about ALS from people who have ALS is a key to discovering and developing treatments. ALS TDI is able to achieve this through their [Precision Medicine Program](#) (PMP), the most comprehensive and longest running translational research study in ALS. Largely a telehealth initiative, the PMP enables ALS TDI to learn from people with ALS around the world, often without ever asking them to leave their homes.

Partnering with the ALS Patient Community

The PMP was developed in 2014 after the ALS TDI science team recognized that they needed to partner directly with people with ALS to better understand the mechanisms of how ALS affects the body. This was enabled by the emergence of new technologies in the fields of cell biology and biometrics relating to body movement, voice, and potential to detect markers of disease in blood.

Through the PMP, researchers at ALS TDI partner with people living with ALS to share and gather data about voice, movement, lifestyle, medical and family histories, genetics, biomarkers, and patient cell biology to better understand the disease. The goal is to use these data to aid the discovery of new targeted treatments for ALS and make clinical trials faster and more efficient, all while empowering people living with ALS by giving them access to their personal data.

“The Precision Medicine Program is unique in multiple ways,” said Fernando Vieira, MD, ALS TDI chief scientific officer. “ALS is a heterogenous disorder, meaning it can affect each patient differently. The PMP acknowledges that the person with ALS is the expert of their experience with the disease. We partner with them to learn from their experiences and couple that essential information to the data generated by our scientists and technicians in the lab in Cambridge. We believe that relating these data can ultimately demystify the processes that drive ALS and reveal pathways for the efficient development of helpful medicines to treat ALS.”



Fernando Vieira, MD, ALS TDI Chief Scientific Officer

Inviting ALS Patient Participation

Participation in the PMP is free, and anyone with ALS can add their data to PMP—regardless of where they live or how far their symptoms have progressed. Participants, who [sign up](#) on ALS TDI’s website, are asked to provide several different categories of data about themselves and their condition. At the outset of their participation, and every three months thereafter, they complete a personal background survey covering things like lifestyle, medical history, occupation, and ALS symptoms. Each month, they are asked to

complete the ALS Functional Rating Scale (ALSFRS-R), a widely used survey for tracking ALS disease progression.

What truly sets the PMP apart, however, is the wealth of quantitative data collected in addition to these subjective questionnaires. Participants who meet certain criteria are provided with accelerometers – devices that can be worn on the wrists and ankles to track the movements of each limb. Worn for three days each month while participants perform a directed series of movements, these accelerometers provide precise data about how each participant’s ALS progression is affecting their motor function. Similarly, participants can track how the disease is affecting their speech. They are asked to record a series of phrases every month over phone. These recordings are then analyzed through techniques developed in collaboration with [Google artificial intelligence researchers](#).

In-Home Blood Collection

Additionally, participants can provide biological data by submitting blood and cell samples. To alleviate the burden of coming into the ALS TDI’s Cambridge, Mass., laboratory for blood collection, the institute has established a first-in-kind [in-home blood collection program](#). Participants, who often have limited mobility, are visited in their homes by mobile phlebotomists contracted by ALS TDI, and the samples are sent to the lab through the mail. ALS TDI utilizes these samples for projects including harvesting cells for drug testing and searching for potential biomarkers that could be used to provide a biological measure for tracking ALS disease progression.

Since its launch in 2014, more than 600 people living with ALS from all over the world have enrolled in the PMP. These volunteers have provided more than 20,000 survey responses, 13,500 voice recordings, and 15,000 accelerometer data sets. In addition to helping support ALS TDI’s research to find effective treatments for ALS, the program also provides these participants with access to their own individual data. Participants can use ALS TDI’s secure online portal to view their data and track disease progression, providing them with valuable information to make more informed decisions about their treatment and care.

“We hope that the information we share back to the participants about their own symptom progression informs them about how their activities and interventions might be influencing their outcomes,” added Dr. Vieira. “We’ve received a lot of feedback that the information can be empowering to a person living with ALS.”

For more information about the PMP, including how to enroll and participate, [visit ALS TDI](#).

A Message from Our Sponsor

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