University of Texas Southwestern

Consent (Permission) to Participate in a Clinical Research Study

Study Title:	PHENOTYPE, GENOTYPE, AND BIOMARKERS (PGB) IN ALS AND RELATED DISORDERS
Study Sponsor:	National Institutes of Health (NIH)
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You are being asked to take part in a clinical research study. This consent form contains important information, so that you can decide if you wish to take part in this study. If you have any questions that remain unanswered, please ask the study doctor or one of the research study personnel before signing this form.

PURPOSE OF THE STUDY

The purpose of this study is to learn more about amyotrophic lateral sclerosis (ALS) and other related neurodegenerative diseases, including frontotemporal dementia (FTD), primary lateral sclerosis (PLS), hereditary spastic paraplegia (HSP), progressive muscular atrophy (PMA) and multisystem proteinopathy (MSP). More precisely, we want to identify the links that exist between the disease phenotype (*phenotype* refers to observable signs and symptoms) and the disease genotype (*genotype* refers to your genetic information). We also want to identify biomarkers of ALS and related diseases. A *biomarker* is an indicator of the disease that can easily be measured. Our long-term goal is to use this information to advance the development of therapies for this group of neurodegenerative disorders.

You are eligible to participate in the study for one of the following reasons: 1) you have ALS or a related disease, or 2) you have a family member affected with ALS or a related disease who is already taking part in the study.

NUMBER OF PARTICIPANTS

A total of approximately 1400 people will take part in this study at 13 institutions. Of these 1400 people, approximately 700 will be primary participants – being evaluated on multiple occasions at one of the study centers. Approximately 700 will be secondary participants – i.e. family members of the primary participants. These secondary participants will be evaluated only once.

STUDY VISITS

Patients with ALS or a Related Disease

If you agree to participate in this study and have signed the informed consent, you will be asked to complete both in-person and remote (from your home) visits.

Table 1. Frequency of study visits for patients with ALS or a related disease.

Diagnosis	In-Person Visits						Remote Visits
	Baseline	Month 3	Month 6	Month 12	Month 18	Month 24	Annually from last in-person visit
ALS	X	X	X	X	X		X
PMA	X	X	X	X	X		X
HSP	X		X	X	X	X	X
PLS	X		X	X	X	X	X
MSP	X		X	X	X	X	X

In-Person Visits

For the in-person visits, you will have to travel to the research site to undergo a number of study procedures. You will be asked to complete a total of 5 in-person visits. If neuropsychological testing is required based on the result of the ECAS screening tool (description on page 7), you may be asked to visit the clinic one additional time. There will be 3 or 6 month intervals between visits. The time interval between your in-person visits is determined by your diagnosis, as you can see in the table above. Wherever possible, we will try to coordinate the timing of your in-person visits with your clinic appointments in order to minimize the travel burden.

If you become physically unable to travel to the research site for some of your in-person visits, you may be asked to complete study procedures at your home while you are on the phone with a member of the research team. From the time that you can no longer travel to the research site for in-person visits, phone calls will replace any remaining in-person visits and they will take place at intervals that correspond to the schedule of in-person visits above.

Remote Visits

For the remote visits, you will be contacted by phone by a member of the research team and you will be asked to complete some study procedures at your home. Your first remote visit will take place about 1 year (12 months) after your last in-person visit, and there will be about 1 year (12 months) between each of your remote visits. These annual remote visits will take place for as long as you are willing and physically able to complete them.

Family Members of Patients with ALS or a Related Disease

If you agree to participate in this study and have signed the informed consent document, you will be asked to complete a single visit to undergo some study procedures. This visit can be done either in-person at the research site or remotely (from your home).

STUDY PROCEDURES

The table below lists all study procedures to be completed at each visit for patients with ALS or a related disease and their family members.

Table 2. Schedule of Study Procedures

Procedure	Patients with ALS or a Related Disease	Family Members

	Baseline Visit	Follow-up In-Person Visits	Annual Visits	
Measurement of your vital signs	X	X		
Collect pedigree information	X	X		X
Collection of biological samples including blood and urine	X	X		X
Genetic counseling for release of your genetic test results		X		
Completion of questionnaires about your symptoms and diagnosis	X	X	X	X
Completion of questionnaires about your daily activities and quality of life	X	X	X	
Examination of your neurological function	X	X		
Measurement of strength of your breathing muscles	X	X	X	
Screening of your neuropsychological function	X	X		
Extended evaluation of your neuropsychological function	X	X		
Evaluation of your disease severity and progression	X	X	X	
Lumbar puncture/collection of CSF (Optional Procedure)	X	X		
Update of information about your health, current medications and possible side effects related to your participation	X	X	X	
Assessment of your survival	X	X	X	

Below is a detailed description of each study procedure performed in this study:

Vital Signs
A member of the research team will measure your height and weight.

Risks Associated with the Procedure
- There are no risks associated with this procedure.

Genetic Testing and Genetic Counseling
Genes control heredity from parents to children. Research on genes is called genetic research. Genetic research is necessary for many important reasons including: 1) it may help us understand the cause(s) of ALS and related diseases, and 2) it may help us develop new tests and treatments.

We will be conducting genetic testing as part of this study. With your permission, we will look for changes (mutations) in your genes that are known to cause ALS or a related disease. In some circumstances, we may provide you with the results of genetic testing (see below for details). The genetic test results will not be included in your medical records unless you communicate the results to your own health care provider. With genetic testing, we may find information about your genes that applies to members of your family. We will ask for your permission to share the results of your genetic testing with your family members.

Blood Collection for Genetic Testing

A trained professional will collect a blood sample from a vein in your forearm when you come to the research site. A standard sterile technique will be used during sample collection. We may collect up to 72 ml (about 14 teaspoons) of your blood. If you cannot come to the site or if you are a family member and you are completing your visit remotely (at your home), we may send you a kit in the mail for the remote collection of a sample of your blood. The kit will include the necessary supplies for a health-care professional to collect your blood sample. We will also send you a special kit for shipping your blood sample back to us. We will extract your genetic material from your blood to perform genetic testing.

Unaffected Testing
If you are not affected with ALS or a related disease, the results of genetic testing will not be provided to you.

Affected Testing
If you are affected with ALS or a related disease, and we are able to identify the genetic cause of your illness, then you will be informed of the results of genetic testing. If we are unable to identify a genetic mutation in the genes known to cause the disease that you have, then we will use your blood sample to identify new genes that cause this disease.

The results of genetic testing will come out in one of four ways:

1. Positive for a genetic change that is known to cause your disease. If this is the case then the specific name of the genetic change will be provided to you.
2. Negative for any genetic change known to cause your disease. If this is the case then the genetic change that causes your disease is one that has not yet been discovered. This information will be communicated to you.
3. Positive for a genetic change that has not previously been known to cause your disease. If this is the case then you will be informed of this new discovery.
4. Inconclusive test results. This can result from a variety of reasons, including the finding of a genetic change that is not known to cause your disease or from technical problems in performing the test.

The results of genetic testing do not impact your current disease status. If you already have a neurological disease, then finding out whether or not we can identify a genetic mutation does not change anything.

A Federal law called the Genetic Information Nondiscrimination Act (GINA) generally prohibits health insurance companies, group health plans, and most employers to discriminate against you based on your genetic information. GINA does not protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance. GINA also does not protect you against discrimination based on an already-diagnosed genetic condition or disease.

Genetic Counselling
If you have ALS or a related disease, the results of genetic testing will be provided to you by the study doctor or genetic counselor. Genetic counselling is often useful and appropriate when people are learning about their genes. During the session, you will be able to ask any questions that you may have regarding your test results. We will follow up by mailing you a letter explaining your genetic test results.

Risks Associated with the Procedure

- Collecting blood from a vein in someone's arm is a standard medical procedure. Sometimes there may be some discomfort, bruising or a feeling of faintness. Rarely this procedure may cause infection.
- It is important to understand that the results of the test do not change anything related to your condition.

- Undergoing genetic testing and/or learning the results of genetic testing for mutations known to cause ALS or a related disease may cause psychosocial stress. You should also think about the implications for your children and other family members. Once you know that you definitely carry a gene mutation, each of your children is at a 50% risk of also carrying the genetic change. The study doctor or genetic counselor will discuss these issues with you in detail.
- Aside from the risks listed above there may be other risks to genetic research that we do not yet know about.

Symptom Questionnaire
A member of the research team will ask you questions to collect information about the earliest and current signs of disease. You will be asked about the nature and location of your initial symptoms, the date that symptoms first appeared, and how the symptoms progressed in your body. We may look in your medical records to collect information about your symptoms. This procedure may take up to 20 minutes of your time.

Risks Associated with the Procedure
- There are no risks associated with this procedure.

Daily Activities and Quality of Life Questionnaires
You will complete paper-and-pencil questionnaires asking about what you are able to do on a daily basis and how you feel physically, mentally and socially.

Risks Associated with the Procedure
- There are no risks associated with this procedure.

Neurological Examination
A neurologist will perform the neurological examination. It is a physical examination that tests your muscle strength, coordination and reflexes. This procedure will take about 10 minutes of your time.

Risks Associated with the Procedure
- There are no risks associated with this procedure.

Respiratory Muscle Function
A member of the research team will measure the strength of your breathing using one or more techniques or devices. These breathing tests typically require breathing in and/or breathing out into a machine that measures things such as your vital capacity (the amount of air you can exhale from your lungs), the force of your breathing (either while breathing in or while breathing out), and the pressure generated in your airways when you breath in/out or sniff. We will ask you to perform each of these tests while you are sitting up. Each test will be repeated between three to five times. This procedure may take up to 30 minutes of your time.

For visits completed remotely (at your home), you will be provided with a device that you can use at home. A member of the research team will teach you how to perform the test before your first remote visit.

Risks Associated with the Procedure
- The measurement of your respiratory function is an entirely non-invasive procedure. The risks associated with this procedure are minimal.

Edinburgh Cognitive and Behavioral ALS Screen (ECAS)
ECAS is a neuropsychological screening tool that is used to detect cognitive and behavioral impairments that are associated with ALS and related disorders. During testing, a member of the

research team will ask you to answer questions and perform small tasks. You will be able to provide answers vocally or in writing. This procedure may take up to 30 minutes of your time.

When you complete this procedure, we will ask you to identify someone who knows you well and has close contact with you (e.g., spouse, caregiver, family member or friend) and who may be willing to answer 13 questions about your symptoms and functioning. These questions can be administered in person or over the phone. If the person of your choice agrees to be contacted and wants to serve as your informant, he/she will receive a separate consent before he/she can complete the questionnaires.

Risks Associated with the Procedure

- There is a risk of boredom or embarrassment that may result from not being able to do certain tasks during the evaluation.
- You may occasionally become more aware of any cognitive and behavioral impairment that you may have.

Extended Neuropsychological Testing

You will complete this procedure only if your ECAS evaluation (see above) shows that you may have cognitive and behavioral impairments. The procedure is performed to test a wide range of cognitive functions, including language, attention/executive functions, memory, visuospatial skills, motor skills, as well as mood and behavior. A neuropsychologist will perform the evaluation, which will include standard, paper-and-pencil measures of memory and thinking, as well as standard self-report questionnaires of personality, mood and quality of life. This procedure may take up to 3 hours of your time.

If you complete this procedure, we will ask you to identify someone who knows you well and has close contact with you (e.g., spouse, caregiver, family member or friend) and who may be willing to fill out brief questionnaires about your symptoms and functioning. If this person agrees to be contacted and wants to serve as your informant, he/she will receive a separate consent before he/she can complete the questionnaires.

Risks Associated with the Procedure

- There is a risk of boredom or embarrassment that may result from not being able to do certain tasks during the evaluation.
- You may occasionally become more aware of any cognitive and behavioral impairment that you may have.

Spastic Paraplegia Rating Scale (SPRS)

This procedure is a physical examination that will be performed by a neurologist to determine the severity and progression of disease. It will only be performed in patients with spastic paraplegia and primary lateral sclerosis. During the evaluation, the neurologist will evaluate various physical aspects, including any spasticity, weakness, contractures and pain, as well as your ability to walk, climb stairs and arise from a chair. Your bladder function will also be assessed. This procedure will take about 15 minutes of your time.

Risks Associated with the Procedure

- There are no risks associated with this procedure.

Collection of Biological Specimens

Biological samples are collected at each in-person visit and may be also be collected remotely.

Blood Sample Collection

A trained professional will collect a blood sample from a vein in your forearm when you come to the research site. A standard sterile technique will be used during sample collection. We may collect up to 72 ml (about 14 teaspoons) of your blood at each collection. This procedure will take about 10 minutes of your time. If you cannot come to the site or if you are a family member and you are completing your visit remotely (at your home), we may send you a kit in the mail for the remote collection of a sample of your blood. Your name, address, and phone number will be provided to the University of Miami in order to ship the blood collection kit. The kit will include the necessary supplies for a health-care professional to collect your blood sample. We will also send you a special kit for shipping your blood sample back to us.

Urine Sample Collection
You will be provided with a sterile cup and you will be asked to provide a sample of your urine. If you cannot come to the site or if you are a family member and you are completing your visit remotely (at your home), we will send you a kit in the mail for the remote collection of a sample of your urine. Your name, address, and phone number will be provided to the University of Miami in order to ship the urine collection kit. We will also send you a special kit for shipping your urine sample back to us.

Cerebrospinal Fluid (CSF) Sample Collection
A neurologist may perform a lumbar puncture (also known as a spinal tap) to collect a sample of your CSF during an in-person visit. You will be asked to lie on your side and curl up in the fetal position or to sit upright and round your back. The skin of your lower back will be cleansed and a numbing medicine will be injected in the skin and underlying tissues. Enough time will be given for the numbing medicine to take effect. A needle will then be inserted into your lower back into the fluid-containing sac that surrounds the spinal cord and nerve roots. The needle is inserted below the level of the spinal cord to avoid the risk of damage to the spinal cord. A standard sterile technique will be used during sample collection, and a sterile bandage will be applied after collection. We want to collect up to 16 ml (about 3 teaspoons) of your CSF at each collection. This procedure will take about 30 minutes of your time.

You may decide not to undergo a lumbar puncture to provide a CSF sample. Participating in this procedure is your choice.

Risks Associated with the Procedures
- Venipuncture (blood draw) carries a small, but standard risk of mild pain and possible bruising and bleeding at the blood draw site, infection and/or fainting from the needle sticks required to obtain blood samples.
- There are no risks associated with the urine sample collection.
- A lumbar puncture (CSF sample collection) may be an uncomfortable procedure. Efforts to minimize discomfort will include the use of local anesthesia before the procedure to numb the collection site as well as the expertise of the skilled neurologist.
- The most common side effect of a lumbar puncture is a headache. This is typically treated with bed rest, fluids and caffeine. If the headache persists, an epidural blood patch may be required. This involves drawing about 10-20 ml (2-4 teaspoons) of your own blood and injecting it into your lower back to 'plug the hole' in the fluid-filled sac that was caused by the lumbar puncture needle. There are also small risks of bleeding, infection and nerve root injury, but these are extra-ordinarily uncommon.

Survival Assessment
A member of the research team will contact you to ask about how you are doing and if you had a tracheostomy or required permanent assisted ventilation.

Risks Associated with the Procedure

- There are no risks associated with this procedure.

RESEARCH STUDY RECORDS

All of the information collected from you for this study will be entered into a secure computer database. We will ask for your permission to keep your contact information in a database so that we may contact you in the future to update our research records.

Information collected in the study is used for research purposes. Clinical reports are not generated and we operate under a principle of strict separation between clinical and research records. As such you will not be provided with reports from the study procedures in which you participate.

TRANSFER OF DATA TO A DATA CENTER AND FEDERAL DATA REPOSITORY

The clinical information collected for this study will be stored in a computer database at the Data Management and Coordinating Center (DMCC) at the University of South Florida in Tampa, FL. The data management center uses several layers of protection for the clinical data stored in its computer database. It meets all of the local and federal security requirements for research data centers.

This study involves the complete analysis of the genetic material that will be extracted from your biological sample. Information obtained from this analysis (genotypic data), as well as information about disease signs and symptoms (phenotypic data), will be entered into one or more scientific repositories maintained by organizations such as the New York Genome Center (NYGC) and the Federal Government. One such repository is the Database of Genotypes and Phenotypes (dbGaP), a data repository at the NIH. All data and information will be submitted via a secure transmission process to a high security network within NIH.

A data repository provides a way for researchers to store the information collected during the research study for future research studies; and to share data from different research studies in order to advance scientific discovery. Only researchers with an approved study may be able to see and use your information, along with information from many other people.

Only de-identified data, which does not include anything that might directly identify you, will be shared with study investigators and approved investigators from the general scientific community for research purposes. However, because your genetic information is unique to you, there is a small chance that someone could trace it back to you. The risk of this happening is very small, but may grow in the future. Researchers will always have a duty to protect your privacy and to keep your information confidential.

FUTURE USE OF SAMPLES FOR RESEARCH

All biological samples collected from you will be stored at the University of Miami Hussman Institute for Human Genomics (HIHG) Biorepository (1501 NW 10th Ave, Miami, FL 33136). The principal investigator will own the samples that you donate and he will not release them back to you, your legally authorized representatives or your family members. Your samples will be stored for as long as they are useful, unless you ask us to destroy them sooner. You may request that your sample be destroyed at any time simply by contacting the principal investigator at the phone number listed on the front of this form.

The principal investigator plans to use your samples for future research. We will ask for your permission for the future scientific use of your samples. If you give your permission, you will not be contacted again for consent or authorization to use these if the requirements for informed consent and HIPAA authorization are waived by the Institutional Review Board (IRB). If these requirements are not waived,

and your samples are sought for future, new research, investigators will request your consent and ask you to complete a new HIPAA form.

Information important for research may be stored with your samples, including your race, ethnicity, sex and medical history, but your confidentiality will be protected to the extent permitted by law. The principal investigator may share your samples with other scientists for research purposes. All samples will be given a code. This code will allow the samples to be used without the researchers knowing your name.

You will not have rights to information obtained from the analysis of your samples. This information may contribute to the creation of new diagnostic tests, new medicines or other uses that may have commercial value. The University of Miami will own the rights to any tests, drugs or treatments that are developed from research on your samples. You will not share in any proceeds the University of Miami might receive from such commercial products. There are no plans to provide financial compensation to you if this occurs.

ALTERNATIVES

You have the alternative to choose not to participate in this research study. This choice will not affect your current or future care or that of any of your family members.

Genetic Testing and Counseling

An alternative is for you to personally seek genetic counseling and obtain genetic testing through a commercial laboratory. This is currently done, for example, by Athena Diagnostic Laboratories in Boston, MA and you can get more information about their costs and services at 800-394-4493. Genetic counselors have special training in helping people understand what genetic testing results mean. You can find a genetic counselor by contacting the National Society of Genetics Counselors at http://www.nsgc.org or with the help of your personal physician.

Commercial laboratories have strict quality control guidelines, which research laboratories do not. However, we have confidence that the research laboratories we are using will produce a reliable and valid result. You do have the option of having testing performed through a commercial laboratory at your own expense. One additional difference between these laboratories is that commercial laboratories only test for mutations in genes that are known to cause a specific disease. Testing in a research laboratory enables us to search for new genes when we don't find a mutation in a gene already known to cause your neurological disease.

BENEFITS

Your participation in the study will provide doctors, researchers and scientists with a better understanding of the biology of ALS and related neurodegenerative diseases.

You may also receive direct benefits by taking part in the study: 1) you may learn more about ALS and related disorders, 2) you may have the opportunity to receive genetic counseling and learn the results of your genetic testing, 3) you will have access to the research team for questions about symptoms, and 4) with your permission, we may contact you in the future regarding opportunities to participate in other related research projects.

RISKS

There is always a risk to your privacy when you share information about yourself. However, the information that we collect from you in this study will be kept confidential, and there are practices in place to protect your personal information.

Overall, the risks associated with your participation in this study are minimal and are less significant than the potential benefits to you and society from gaining further knowledge about the causes, disease progression, and prevention of ALS and related neurodegenerative diseases.

CONFIDENTIALITY

We will keep your research records confidential. However, people other than those who are part of the local research team [Principal Investigator (PI), study coordinator, research nurses, and all other research staff] may need access to your research records:

1) People from various government agencies and this institution's departments that make rules and policies about how research is done.

2) People from various government agencies and departments at both the University of Miami and the institution where this study is taking place that are responsible for making sure that research studies are conducted correctly. This includes people working for the Department of Health and Human Services (DHHS), the Office for Human Research Protections (OHRP), the University of South Florida Institutional Review Board (IRB), the IRB at this institution, University of South Florida Research Integrity and Compliance, the Rare Diseases Clinical Research Network Data Management and Coordinating Center, the University of Miami Institutional Review Board (IRB), and the offices of Research Compliance & Quality Assurance (RCQA) at the University of Miami.

3) People from the National Institutes of Health (NIH), which pays for the study.

4) Select researchers at other study centers may be given access to the information you provide about yourself and your family members for your family tree (pedigree).

By law, anyone who looks at your research records must keep them completely confidential.

In order to keep your personal information confidential, we will use a study number rather than your name on research records when possible. We will not create medical records for you based on your participation in this research study. Your name and other facts that might point to you will not appear when we present this study or publish its results.

CERTIFICATE OF CONFIDENTIALITY

To help us protect your privacy, we have obtained a Certificate of Confidentiality from the National Institutes of Health (NIH). With this Certificate, the investigator cannot be forced to disclose information that may identify you, even by a court subpoena, in any federal, state, or local civil, criminal, administrative, legislative, or other proceedings. The investigator will use the Certificate of Confidentiality to resist demands for information that would identify you. However, the Certificate cannot be used to resist demands from personnel of the United States Government for information that is used for auditing or evaluation of federally-funded projects or for information that must be disclosed in order to meet the requirements of the federal NIH.

Even though there is a Certificate of Confidentiality for this study, the investigator continues to have ethical obligations to report child or elder abuse or neglect and to prevent you from carrying out any

threats to do serious harm to yourself or others. If keeping information private would immediately put you or someone else in danger, the investigator will release information to protect you or another person. The investigator may also release personal information about you for medical necessity. Finally, the Department of Health and Human Services (DHHS) personnel may request personal information about you while performing audits, carrying out investigations of DHHS grant recipients, or evaluating DHHS funded research projects. Information disclosed in all above cases will not be protected by the Certificate of Confidentiality.

You should understand that a Certificate of Confidentiality does not prevent you or a member of your family from voluntarily releasing information about yourself or your involvement in this research. If an insurer, employer, or other person learns about your participation in the study and obtains your written consent to receive research information, then the investigator may not use the Certificate to withhold that information. This means that you and your family must also actively protect your own privacy.

REGISTRATION ON CLINICALTRIALS.GOV

A description of this clinical trial will be available on http://www.ClinicalTrials.gov, as required by U.S. Law. This Web site will not include information that can identify you. At most, the Web site will include a summary of the results. You can search this Web site at any time.

COSTS AND COMPENSATION

There will be no costs to you if you take part in this research study. We will not pay you for the time you volunteer while being in this study.

However, if you have ALS or a related disorder, we will give you $40 for each of the five scheduled in-person study visits you complete to cover travel related expenses. If you are asked to come to the clinic only to perform the neuropsychological testing, we will give you $40 for travel expenses for that visit. You may be paid up to $240 in total, since there are 5 in-person visits scheduled and one additional visit may be required. If you stop coming to the research site for study visits, withdraw from the study, or are withdrawn by the principal investigator, you will stop receiving reimbursements. Only study participants with ALS or a related disorder will receive reimbursements.

You will be issued a UT Southwestern Greenphire ClinCard, which can be used as a credit or debit card. You will also receive instructions on how to use the card. In order to receive study payments, your name, address, date of birth and Social Security Number (SSN) will be collected from you by the research staff. All information will be stored in a secure fashion and will be deleted from the UT Southwestern Greenphire ClinCard system once the study has been completed

COMPENSATION FOR STUDY-RELATED INJURY

It is important that you report any illness or injury to the research team listed at the top of this form immediately.

Compensation for an injury resulting from your participation in this research is not available from the University of Texas Southwestern Medical Center at Dallas.

You retain your legal rights during your participation in this research.

QUESTIONS, CONCERNS OR COMPLAINTS

If you have any questions, concerns or complaints about this study, call your study doctor at the phone

number listed on the front of this form.

If you have questions about your rights, general questions, complaints, or issues as a person taking part in this study, call the UT Southwestern Institutional Review Board (IRB) Office at 214-648-3060

VOLUNTARY PARTICIPATION / WITHDRAWAL

You should only take part in this study if you want to volunteer. You should not feel that there is any pressure to take part in the study, to please the study doctor or the research staff. You are free to participate in this research or withdraw at any time. If you withdraw, we will not collect any new research data from you, but will use data and biological samples that have already been collected unless you specifically request otherwise in writing. To make this request, please write to your study doctor at the address listed on the front of this form.

The principal investigator may remove you from the study if he/she believes that you do not qualify for the further participation or if you are unable to complete study procedures.

During the time that you participate in the study, we may learn new things about ALS or a related disease that you need to know or that make you want to stop participating in the study. You will be told of any new findings that are discovered while you are taking part in the study.

DESIGNATION OF AN AUTHORIZED REPRESENTATIVE

In the event that you become incapacitated or unable to meet some of the physical demands associated with data collection, or if we are unable to communicate with you for any other reason, we would like for you to authorize a friend or family member to interact with members of the research team on your behalf. Please indicate your willingness to designate an authorized representative BY INITIALING the appropriate box below:

I give permission for my authorized representative to discuss my condition with my study doctor and his/her staff.	

I do NOT give permission for my authorized representative to discuss my condition with my study doctor and his/her staff.	

If you grant us permission to contact an authorized representative, please provide his/her contact information below:

Name: _____

Address: _____

_____

Phone Number: _____

Email Address: _____

Please answer the additional questions below by saying or checking YES or NO.

	YES	NO
Do you authorize your study doctor and his/her team to communicate with any caregiver I may have in the future, including but not limited to home health nurses, family members and social workers, should your authorized representative become unavailable?		
Do you authorize your study doctor and his/her team to communicate with any referring physician or healthcare provider as part of your overall treatment and care by that health professional?		

AUTHORIZATION TO USE AND DISCLOSE PROTECTED HEALTH INFORMATION (HIPAA LANGUAGE)

What is the purpose of this form?
This authorization describes how information about you and your health will be used and shared by the researcher(s) when you participate in this research study ("Research Project"). Health information is considered "protected health information" when it may directly identify you as an individual. By signing this form you are agreeing to permit the researchers and other others (described in detail below) to have access to and share this information. If you have questions, please ask a member of the research team.

Who will be able to use or share my health information?
UT Southwestern Medical Center may use or share your health information with Jaya Trivedi, MD andher staff at UT Southwestern Medical Center ("Researchers") for the purpose of this research study.

Will my protected health information be shared with someone other than the Researchers?
The Researchers may share your health information with others who may be working with the Researchers on the Research Project ("Recipients") for purposes directly related to the conduct of this research study or as required by law. These other people or entities include:

- The University of Miami- The sponsor includes any people, entities, groups or companies working for or with the sponsor or owned by the sponsor. The sponsor will receive written reports about your participation in the research. The sponsor may look at your health information to assure the quality of the information used in the research.

- Rare Diseases Clinical Research Network (RDCRN): These organizations need access to your health information to assist the Researchers in the Research Project.

- The UT Southwestern Institutional Review Board (IRB). This is a group of people who are responsible for assuring that the rights of participants in research are respected. Members and staff of the IRB at UT Southwestern may review the records of your participation in this research. A representative of the IRB may contact you for information about your experience with this research. If you do not want to answer their questions, you may refuse to do so.

- Some additional people, agencies and businesses that may receive and use your health information are investigators at other sites who are assisting with the research; central laboratories, reading centers or analysis centers; the Rare Diseases Clinical Research Network Data Management and Coordinating Center, the University of South Florida Institutional Review

Board (IRB), the University of South Florida Research Integrity and Compliance Office, the University of Miami Institutional Review Board (IRB) and the offices of Research Compliance & Quality Assurance (RCQA) at the University of Miami.

- Representatives of domestic and foreign governmental and regulatory agencies may be granted direct access to your health information for oversight, compliance activities, and determination of approval for new medicines, devices, or procedures.

Medical information collected during this study and the results of any test or procedure that may affect your medical care may be included in your medical record. The information included in your medical record will be available to health care providers and authorized persons including your insurance company.

How will my health information be protected?
Whenever possible your health information will be kept confidential as required by law. Federal privacy laws may not apply to other institutions, companies or agencies collaborating with UT Southwestern on this research project. There is a risk that the Recipients could share your information with others without your permission. UT Southwestern cannot guarantee the confidentiality of your health information after it has been shared with the Recipients.

Why is my personal contact being used?
Your personal contact information is important for the UT Southwestern Medical Center research team to contact you during the study. However, your personal contact information will not be released without your permission.

What health information will be collected, used and shared (disclosed)?
The Researchers will collect your past medical history from the EPIC electronic medical record system. Past medical history will include all outpatient visits to UT Southwestern including physical examinations, medications, laboratory tests and procedures performed.

Will my health information be used in a research report?
Yes, the research team may fill out a research report. (This is sometimes called “a case report”.) The research report will not include your name, address, or telephone or social security number. The research report may include your date of birth, initials, dates you received medical care and a tracking code. The research report will also include information the research team collects for the study.

Will my health information be used for other purposes?
Yes, the Researchers and Recipients may use your health information to create research data that does not identify you. Research data that does not identify you may be used and shared by the Researchers and Recipients in a publication about the results of the Research Project or for other research purposes not related to the Research Project.

Do I have to sign this authorization?
No, this authorization is voluntary. Your health care providers will continue to provide you with health care services even if you choose not to sign this authorization. However, if you choose not to sign this authorization, you cannot take part in this Research Project.

How long will my permission last?
This authorization has no expiration date. You may cancel this authorization at any time. If you decide to cancel this authorization, you will no longer be able to take part in the Research Project. The Researchers may still use and share the health information that they have already collected before you

canceled the authorization. To cancel this authorization, you must make this request in writing to: Jaya Trivedi, MD, UT Southwestern Medical Center, 5323 Harry Hines Blvd, Dallas TX 75390-9036.

Will I receive a copy of this authorization?
Yes, a copy of this authorization will be provided to you.

Signatures:

By signing this document you are permitting UT Southwestern Medical Center to use and disclose health information about you for research purposes as described above.

_____ _____
Signature of Research Participant Date Time: AM/PM

CONSENT TO TAKE PART IN THIS RESEARCH STUDY

Please answer the questions below by saying or checking YES or NO.	**YES**	**NO**
Are you willing to undergo a lumbar puncture for CSF sample collection?		
May we use your biological samples for genetic testing?		
If applicable, may we share the results of your genetic testing with members of your family?		
May we use your biological samples for future scientific research?		
May we contact you in the future to update our research records?		
May we contact you in the future to let you know of other related research opportunities?		

Statement of Person Taking Part in the Study
If the statements below are true and you want to take part in the study, please sign the form.

Please indicate if the statements below are TRUE or FALSE.	**True**	**False**
You freely give your consent to take part in this study.		
You understand that by signing this form you are agreeing to take part in research.		
You have received a copy of this form to take with you.		

_____ _____
Signature of Person Taking Part in Study Date and Time

_____
Printed Name of Person Taking Part in Study

Statement of Person Obtaining Informed Consent
I have carefully explained to the person taking part in the study what he or she can expect. I hereby

certify that when this person signs this form, to the best of my knowledge, he or she understands: 1) what the study is about, 2) what procedures will be performed, 3) what the potential benefits might be, and 4) what the known risks might be.

Signature of Person Obtaining Informed Consent	Date and Time
Printed Name of Person Obtaining Informed Consent	