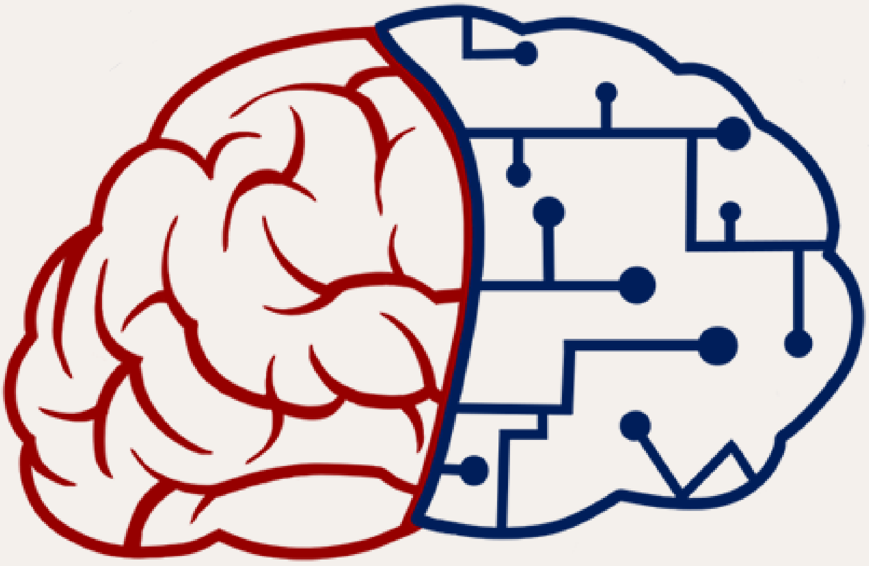
PENN FRONTOTEMPORAL DEGENERATION CENTER



OBSERVATIONAL RESEARCH PROGRAM

CONTENTS

Penn Frontotemporal Degeneration Center

CLINICAL RESEARCH AT THE PENN FTDC

Research studies are a way to help improve diagnosis and treatment of Frontotemporal Degeneration (FTD) and all related neurodegenerative conditions.

This type of research is called observational research since it involves collecting data and samples without an intervention. Our interdisciplinary team learns a great deal about FTD by collecting data and biological samples from patients to study the biology of FTD and related disorders over time. This work contributes to improving diagnosis, developing new markers of prognosis, and discovering therapeutic targets that can enhance and further the development of treatment trials aimed at treating the underlying biology of FTD and related disorders.

These National Institutes of Health (NIH) funded research programs include patients, as well as healthy volunteers, and the study of DNA, brain imaging, neurocognitive testing, and the collection of blood and cerebrospinal fluid to analyze for biological markers of FTD. We collaborate on these projects with many other neurodegenerative disease centers, within Penn, nationally, and internationally. The knowledge we gain helps collaborators worldwide move the scientific understanding of disease forward and improve diagnosis and treatment.

Participation in research has enormous impact on advancing the clinical care for FTD and related disorders.

University of Pennsylvania Centralized Observational Research Repository on Neurodegenerative Disease (UNICORN)

The purpose of this research program is to develop a better understanding of the causes and natural course of FTD and related neurodegenerative diseases by creating a large collection of different types of data. These diseases cause damage to brain cells (neurons), and they slowly worsen over time. They may cause problems with thinking, memory, movement, language, and behavior. The data and samples collected in this study are analyzed for several ongoing NIH funded studies and will be stored securely for future use.

AMILIAL FRONTOTEMPORAL DEGENERATION

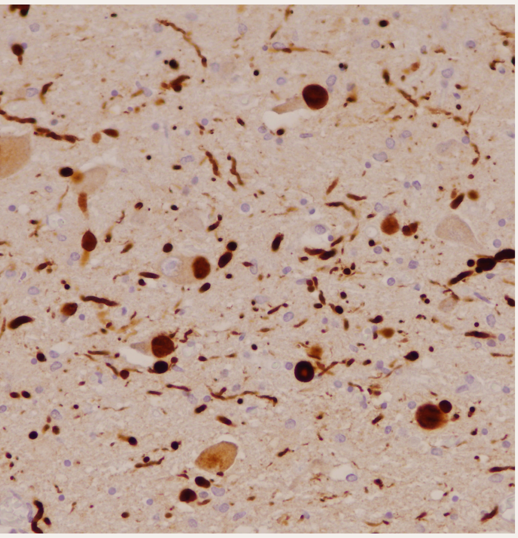
- Genetic causes of FTLD
- About 25.1% of FTLD patients have a strong family history
- Basis of inherited disease defined in about 75% of these patients
  - Chromosome 17 MAPT→ tau
  - Chromosome 17 GRN→ TDP-43
  - Chromosome 9 C9orf72→ TDP-43
  - Chromosome 9 VCP
  - Chromosome 3 CHMP2B
  - Chromosome 1 TARDBP

What types of activities are involved?

Core activities for this study include lumbar puncture, MRI (3T), cognitive testing, neurological examination, speech tasks, blood draw, family and medical history collection, and questionnaires. By studying and integrating these data, our goal is to learn more about these diseases so we can make better tests and treatments for them in the future. Activities can be scheduled in the same day or spread out over different days. We follow up on a yearly basis to repeat these activities. The repeat visits help us learn how these diseases change over time. The study is designed to include annual visits for at least 3 years, if possible, but it is a voluntary study, which means that you can participate for any amount of time as you would like or are able to.

There are other activities that you may be eligible for depending on your diagnosis and ability to participate. These activities include conversation tasks, which help us develop new objective measures of language, computerized tasks and brief questionnaires that help us develop new measures to reflect social behavior in FTD conditions, wearable technologies to measure heart rate and other physiologic outcomes, and 7T MRI, which is a higher strength FDA approved MRI scan that allows us to take ultrahigh resolution images of the brain to help identify protein patterns that are usually only seen under the microscope.

ClinicalTrials.Gov ID NCT04715399



Dementia With Lewy Bodies Consortium

The goal of the Dementia with Lewy Bodies Consortium (DLBC), established by the Cleveland Clinic, is to develop a collection of data from patients with Lewy body dementia, Parkinson's Disease Dementia, or mild cognitive impairment (MCI) suggestive of an early stage of Lewy body dementia. Patients will be followed over a period of five years with detailed clinical information (e.g. medical history, cognitive testing), collection of biospecimens (e.g. blood, cerebrospinal fluid), and imaging data. The establishment of this data will also promote additional future research, including therapeutic trials, by having a readily available subject sample. Subjects across the United States are being enrolled at 10 study sites.

ALLFTD Study

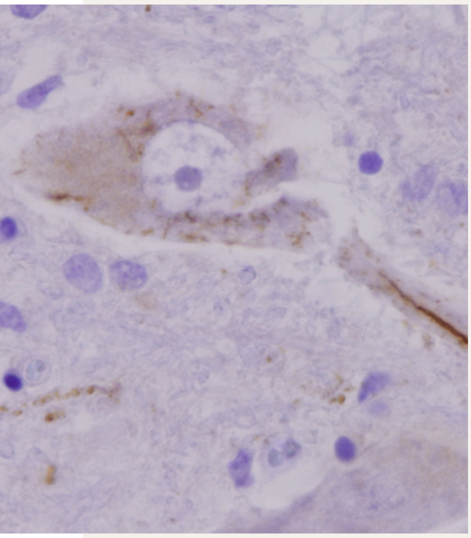
The ALLFTD Study is an NIH funded research study for Frontotemporal Lobar Degeneration. The knowledge gained from ALLFTD and similar studies are driving the development of therapies for this rare disease. The goal of this study is to enroll individuals with FTLD and those at risk of developing FTLD to observe changes in the brain over time. You do not need to know if you carry an FTLD associated mutation to participate in ALLFTD. Activities include cognitive testing, biofluids collection, MRI, optional genetic testing and counseling, and neurological assessment. 23 academic centers in the US and Canada participate as enrolling sites. Many people participate because they want the research to benefit their children and others in the family.

www.allftd.org

Positron Emission Tomography Studies

Some of the specialized studies in our center involve Positron Emission Tomography Imaging (PET/CT), which is a specialized type of imaging. While the MRI provides a structural picture of the brain, the PET scan allows us to visualize unique proteins in the brain we hope will improve our ability to diagnose FTD and related conditions. In our studies, we are looking at a protein called tau. Tau has been shown to build up in the brains of patients with injury to brain cells, including those with conditions like Alzheimer's disease, Parkinson's disease, Lewy body disease, and Frontotemporal degeneration. PET imaging uses a specialized radioactive PET tracer that highlights the tau protein in the brain and allows us to form images of tau binding in the brain. We combine this specialized imaging with data we gather during patient's participation in our core research program like brain MRI, biofluids, cognitive test results, etc. and hope that this will provide information that could be used to refine and confirm diagnosis for patients in the future.

ClinicalTrials.Gov ID NCT05456503



CReATe - ALS Studies

CReATe, also known as the Phenotype, Genotype, Biomarker 2 (PGB2) research study, is a prospective, longitudinal, observational study aiming to learn more about amyotrophic lateral sclerosis (ALS) and other related neurodegenerative diseases by identifying new biomarkers and linking the observable signs and symptoms of disease with genetic factors that may contribute to it. The long-term goal is to use this information to advance the development of therapies for this group of neurodegenerative disorders. Participation in this research study will include semi-structured interview, questionnaires, neuromuscular assessment, collection of biological samples, respiratory function testing, and neuropsychological testing.

ClinicalTrials.Gov ID NCT04875416

BRAIN DONATION

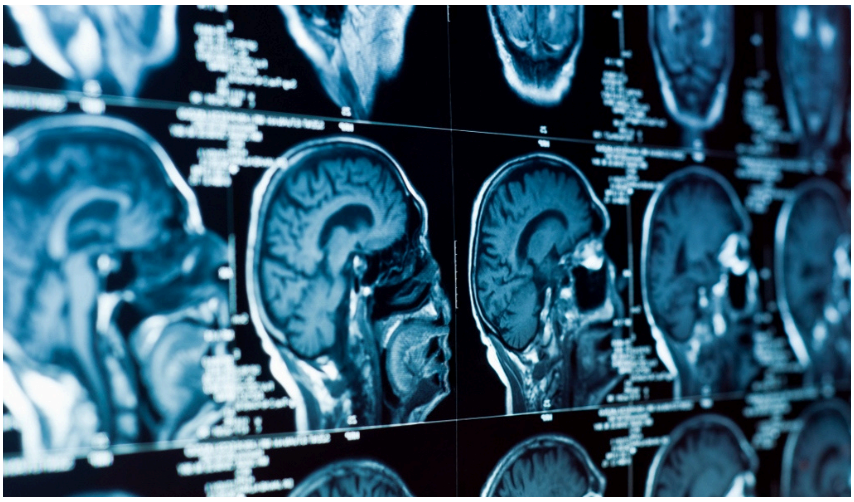
Brain donation is a deeply meaningful contribution.

The gift of brain donation is critical to advancing science of FTD research and accelerating discovery of new diagnostics and mechanisms of disease needed to develop treatments to slow or stop FTD and related disorders.

At Penn, we perform NIH funded research and share de-identified tissue and data broadly with the scientific community, so that as a team the FTD research community can advance discoveries.

As part of the brain donation program, patient families receive a final neuropathological diagnosis in an autopsy report which is reviewed with them by their treating physician.

www.pennftdcenter.org/brain-donation



OUR TEAM

David Irwin MD

Co-Director, Associate Professor

Dr. David Irwin has dual training in cognitive neurology and neuropathology and his lab focuses on integrating human brain histopathology and molecular techniques with imaging methods to discover therapeutic targets and develop tissue-sensitive biomarkers to facilitate clinical trials for emerging therapies for FTD, LBD, AD and related disorders.

Lauren Massimo PhD, CRNP, FAAN

Co-Director, Associate Professor

Lauren Massimo is a graduate of the PhD program at the University of Pennsylvania. She is an Assistant Professor in the School of Nursing at the University of Pennsylvania. Dr. Massimo's research focuses on identifying the cognitive and neural basis for decline in neurodegenerative disease.

Corey McMillan PhD

Co-Director, Associate Professor

Dr. Corey McMillan completed his graduate training at the University of Edinburgh. The overall mission of Dr. Corey McMillan's Bioinformatics in Neurodegenerative Disease (BiND) Lab, housed within the Penn FTD Center, is to use multimodal and bioinformatic approaches to improve our understanding of the biological basis of neurodegenerative conditions.

EVENTS

Penn
Frontotemporal
Degeneration Center
Caregiver
Conference

Penn FTD Center Annual Caregiver Conference

The Penn FTD Center's Annual Caregiver Conference is for those diagnosed with FTD, and their caregivers, family, and friends to access information and support. With presentations from leading experts in neuropsychology, clinical care, genetics, cognitive neuroscience, and more, attendees will hear the latest on innovative treatments, diagnostics, and practical caregiver strategies. As always, our goal is to empower caregivers and deliver real-time updates.

Penn
Frontotemporal
Degeneration Center
Familial Conference:
Education and Support
for Familial FTD/ALS

Penn FTD Center Annual Familial Conference

Our team understands ALS, FTD, and related disorders can impact an entire family even when just one person is diagnosed. However, we also recognize the unique challenges and situations that arise from familial or genetic diseases. We have designed this exclusive event to recognize and address these topics for people living with or at risk for a familial and/or genetic form of FTD/ALS. You will have the chance to hear from genetic counselors, neurologists, and social workers on the genetics of FTD and ALS—and have the chance to ask your questions in real-time.

FTD Clinical Research
Learning Institute

FTD Clinical Research Learning Institute

The FTD Clinical Research Learning Institute is a one-day virtual program that empowers individuals diagnosed with FTD, their caregivers and loved ones to be strong advocates in the field of FTD research by educating them on the clinical research process. Certification as an FTD Research Ambassador provides opportunities to influence and improve the FTD research process. Research Ambassadors will also be able to share information about the research process, and address common misconceptions about research, to patients and caregivers through support groups, conferences, social media, and other platforms. There is no cost for this program.

Penn Frontotemporal Degeneration Center

Contact Us

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For General Questions or Research Interest: 215-349-5863

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www.pennftdcenter.org

Newsletter

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