GLOSSARY

Augmentative and alternative communication (AAC) - Strategies used to assist or replace communication for individuals with communication disorders. Some examples include flash cards, gestures or applications for electronic devices.

Agrammatism - A symptom of aphasia characterized by grammatical errors, such as using the incorrect verb tense, number or gender, or incorrect word order.

Agnosia - Literally, a “loss of knowing.” For example, visual agnosia is the inability to recognize an object by sight.

Akinesia - Very slow or rigid mobility. The loss or absence of voluntary movement.

Alzheimer’s disease - A brain disease that primarily affects memory, however the disease progressively affects other cognitive abilities including language, behavior/personality, visual/spatial perception, motor abilities, and ability to carry out daily activities. This is the most common form of dementia in persons over age 65. Alzheimer’s is associated with the proteins tau and beta-amyloid.

Allele - Refers to different versions of the same gene.

Amyloid - A starch-like protein that in one configuration accumulates and is related to disease. Abnormal accumulations of amyloid in the brain are involved in Alzheimer’s disease, but not FTD. Identifying amyloid in brain imaging (amyloid PET imaging) can differentiate FTD from Alzheimer’s disease.

Amyotrophic lateral sclerosis (ALS) - Also known as Lou Gehrig’s disease, is a disease of the nerve cells in the brain and spinal cord that control voluntary muscle movement. ALS is associated with abnormal accumulations of the protein TDP-43 and can be associated with symptoms of FTD. 10-15% of patients with FTD may also develop symptoms of ALS. C9orf72 gene mutations can cause FTD and ALS.

Anomia - Difficulty with naming objects or actions, some refer to this as word-finding difficulty, a common symptom of dementia.

Apathy - Lack of interest or concern for activities, others, or self-care.

Aphasia - An acquired disorder of speech, reading, writing and/or comprehension of others. While the most common cause of aphasia is stroke, it can also result from head injury, brain tumor or other neurological causes, such as primary progressive aphasia.

Apraxia - An inability to perform purposeful tasks or movements when asked, even though a person understands the request or command and has the physical ability to perform the task. Apraxia is a symptom associated with corticobasal syndrome.
Atrophy - Degeneration of an organ or body part; shrinkage or loss of tissue or cells. AD, PPA, bvFTD, CBS and PSP are each associated with atrophy in specific areas of the brain. ALS can show atrophy in the spinal cord as well as in the brain.

Basal Ganglia - Several large clusters of brain cells deep in the cerebral hemispheres that contribute to cognition, voluntary movement and coordination. Neurodegeneration in parts of the basal ganglia is involved in Huntington disease, Parkinson’s disease, and corticobasal degeneration.

Behavioral neurology - A subspecialty of neurology that studies the neurological basis of behavior and cognition. Behavioral neurologists and specialists are trained in the diagnosis and treatment of dementia syndromes.

Behavioral variant FTD (bvFTD) - A subtype of FTD in which patients experience personality changes and loss of executive function, such as disorganization, loss of judgment, apathy, inappropriate behavior and lack of insight. Also sometimes called frontotemporal dementia.

Biomarker - A physiological characteristic that can be measured as a way to detect illness and diagnose a disease. It can be a protein measured in blood or cerebrospinal fluid (CSF), or a compound used in conjunction with an imaging technique to track specific pathology. Biomarkers for FTD are needed so we can identify during the patient’s life which brain changes are responsible for his or her symptoms.

Bradykinesia - Slowness of movement. Can be a symptom of nervous system disorders, especially Parkinson's disease, or a side effect of medications.

C9orf72 - A gene of unknown function with a specific type of mutation, called a hexanucleotide repeat (a large repeat of a section of genetic material) on chromosome 9. The mutation can cause hereditary FTD and/or ALS.

CHMP2B - A gene that makes the charged multivesicular body protein 2b. Variations in the CHMP2B gene are associated with a very rare form of hereditary FTD.

Corticobasal degeneration (CBD) - A progressive neurological disorder characterized by loss of cells in the cerebral cortex and the basal ganglia. It usually begins as a movement disorder, with affected individuals showing a unilateral (one sided) movement disorder, often involving muscle rigidity or spasm. People with CBD often develop the cognitive features associated with FTD. CBD pathology is caused by accumulation of the tau protein.

Corticobasal syndrome (CBS) - The clinical diagnosis given to describe the characteristic movement and motor symptoms and cognitive features above, before definite confirmation of pathology. CBS symptoms are sometimes found on autopsy to be caused by Alzheimer’s pathology.
Cerebellum - A large structure at the lower back part of the brain responsible for the coordination of movement and balance.

Cerebral cortex - The largest portion of the brain, which is responsible for the processes of thought, perception and memory, and plays a key role in advanced motor function, social abilities, language, and problem solving.

Cerebrospinal fluid (CSF) - A clear fluid that circulates nutrients within the brain and spinal cord. A lumbar puncture procedure produces a CSF sample which may be examined for abnormal protein levels, infection or cancer cells.

Chromosomes - Typically, there are 46 chromosomes (23 pairs) in every human cell. Each chromosome is one very long, folded strand of DNA in combination with proteins that contains hundreds or thousands of genes.

Clinical diagnosis - A diagnosis based on observable signs, symptoms and medical history of a disease in conjunction with laboratory and other diagnostic testing, but without confirmation of disease pathology.

Cognition - A term given to mental processes including perception, memory, awareness, reasoning, judgment, intellect, imagination and social skills.

CT Scan - A series of X-rays of the head (or other body part) are compiled to visualize bleeding, tumors or other injury.

Contractures - Permanent shortening of a muscle or tendons caused by remaining immobile for a long period of time. Once they occur, contractures resist stretching or relaxation through exercise. They affect range of motion and can cause pain, and may limit a caregiver’s ability to bathe and dress the person.

Delirium - A temporary state of mental confusion that develops quickly and usually fluctuates in intensity and may be caused by pain, dehydration, medications, or acute illness.

Delusions - Unshakable beliefs in something untrue. These beliefs defy normal reasoning and remain firm even when overwhelming proof is presented to dispute them.

Dementia - A generic term that is used to indicate significant impairment in two or more areas of cognitive function (memory, language, visuo-spatial, executive function). There are many causes of dementia, some progressive neurodegenerative disease like AD or FTD, others that are non-progressive, like stroke.

Differential diagnosis - The treating physician’s short list of likely causes for a patient’s symptoms. The list can be honed with further observation, laboratory tests and imaging.
Disinhibition - A lack of restraint or ability to manage or edit an immediate, impulsive response to a situation. It can be manifested in behavior in several ways, including disregard for social conventions and poor risk assessment.

DNA (Deoxyribonucleic acid) - DNA is a long, double-stranded compound that codes for genetic information.

Double-blind trial - A double blind trial is a clinical experimental condition in which neither participants nor researchers know who is receiving an active treatment and who is receiving a placebo.

Dysarthria - Difficulty controlling or coordinating the muscles used in speech or weakness of those muscles. Dysarthria is characterized by slurred or slow speech, unusually soft speech or other changes in speech quality that make understanding difficult.

Dysphagia - Difficulty in swallowing which may be associated with esophageal obstruction as well as certain neurodegenerative or motor disorders involving the esophagus.

Dyspraxia - Difficulty in the coordination and execution of learned motor movements. See apraxia.

Dystonia - Recurrent involuntary muscle contractions that may manifest as twisting, writhing or unusual postures or positions.

Electroencephalography (EEG) - A non-invasive, diagnostic technique that indicates cortical brain activity, similar to an EKG for the heart. EEG may assist in the diagnosis of seizure disorders and coma.

Executive function - A set of cognitive skills that control and regulate other abilities and behaviors such as planning and strategizing, changing tasks appropriately, self-monitoring, attention, and abstract thinking.

Familial - A broad term used to indicate that more than one person in a family has a trait or disease. The apparent clustering of affected individuals may occur by chance or due to a genetic mutation.

FTD with motor neuron disease (FTD/MND) - This form of FTD may have the same behavioral/language changes as other forms of the disease, but it also involves the nerve cells that control voluntary movement, called motor neurons. Motor symptoms may include: muscle weakness, muscle atrophy, twitches, cramps, tight and stiff muscles.

Frontal lobe - The part of the brain that controls executive function, executing movements, and processing emotion.

FUS (Fused in sarcoma protein) - Approximately 10% of FTD cases are associated with accumulation of the FUS protein in the brain.

Gait apraxia - Loss of the natural sequencing of the movements and balance required to walk properly.
Gene - A sequence of DNA that carries the instructions to make a specific protein or other substance needed for proper functioning.

Gene sequencing - A test that deciphers the genetic code of a gene or genes by reading each nucleotide. Gene sequencing is used to detect genetic differences that may related to disease.

Genetic testing - analysis of DNA to look for gene sequences related to disease or other unusual traits.

Hereditary - The genetic transmission of a trait, condition, or disorder from parent to offspring.

Inclusion - A small collection of proteins found within a nerve or brain cell which would ordinarily have been dispersed.

Induced pluripotent stem cells (iPSC cells) - A powerful research tool developed in 2007 that allows a researcher to take a skin cell (or other adult cell) from a person and generate stem cells, which can be grown and differentiated into various types of tissue containing that person’s genome. iPS cells from people with FTD will facilitate study of the disease.

Logopenic Primary Progressive Aphasia (PPA) - A type of PPA characterized by problems with repetition of sentences, but preserved single word comprehension and grammar production. Often found to be associated with Alzheimer’s pathology upon autopsy.

MAPT - The microtubule associated protein tau gene. Mutations in MAPT are associated with some hereditary FTD cases.

Motor neuron disease (MND) - A progressive loss of the neurons for voluntary movement associated with amyotrophic lateral sclerosis (ALS) or Lou Gehrig’s disease and several other progressive neurological disorders.

Microtubules - Tiny hollow fibrous rods that provide structure and facilitate movement within cells. The tau protein is involved in stabilizing microtubules.

MRI (Magnetic resonance imaging) - A diagnostic test that uses radio waves and magnets to image the brain (or other body parts) with greater detail than the CT scan.

Mutation - A change in DNA that affects a gene’s ability to function properly, often resulting in an abnormality or disease.

National Alzheimer’s Coordinating Center (NACC) - A database owned and operated by the National Institute on Aging (NIA) where information on all patients seen at nationally funded Alzheimer Disease Centers (ADCs) is stored. An additional data module for FTD patients has been developed to permit collection of standardized data on FTD patients seen at different clinical sites across the US.
Neurofibrillary tangle - Pathological clusters of the protein tau in brain cells of people with Alzheimer’s disease.

Neuroimaging - Techniques, such as CT, MRI, PET, that produce images of the brain and spinal cord that may assist in the diagnosis of neurological diseases. Advances in neuroimaging technology are fueling important advances in FTD diagnosis and treatment.

Neurotransmitter - Naturally produced chemicals, such as serotonin or acetylcholine, by which nerve cells communicate.

Parkinsonism - The motor symptoms that are commonly seen in Parkinson’s disease and some people with FTD, such as tremors, rigidity, slowing and walking/balance difficulties.

Pathology - The study of the organs and tissues to determine precise changes that lead to disease.

Pathological diagnosis - A diagnosis made by examining tissues under a microscope to determine the neuro-chemistry associated with the clinical symptoms. A pathological diagnosis of FTD will include identification of any abnormal proteins, such as tau, TDP-43 or FUS.

Pedigree - A family tree that is specifically used to record health history and chart diagnoses and causes of death.

Penetrance - The likelihood that a genetic mutation will result in disease.

Phonemic paraphasias - Errors involving use of the incorrect sound ("ped" instead of "bed") or transposition of the sound ("efelant" for "elephant").

PET scan - A type a neuroimaging technique called functional imaging that creates an image of brain activity. PET scans can detect changes earlier than CT or MRI, and are often used to complement CT or MRI data.

Pick bodies - A specific type of cellular inclusion made up of the protein tau and found in the brain cells of some people with FTD.

Pick’s disease - A former name for FTD, named for the physician Arnold Pick who first described the condition. It is now reserved as a pathological diagnosis for cases of FTD in which Pick bodies are present.

Placebo - An intervention administered despite knowing it will not have an effect on the target symptom, usually used in the context of clinical trials to prove a new drug’s effectiveness.
Primary progressive aphasia (PPA) - The umbrella term for the language variants of FTD. PPA encompasses agrammatic/non-fluent, semantic and logopenic variants.

Progressive nonfluent aphasia (PNFA) - One of the language variants of FTD, characterized by changes in the ability to speak, write and understand speech. Now called non-fluent/agrammatic PPA.

Progressive supranuclear palsy (PSP) - A rare movement disorder included among FTD related disorders and Parkinson’s-plus disorders. Patients experience paralysis of vertical eye movement often resulting in falling and difficulty reading, which make clinical diagnosis relatively accurate. PSP is always caused by abnormal accumulation of the tau protein.

Progranulin - Mutations in the progranulin gene (PGRN), cause a deficiency in production of the progranulin protein. This deficiency is associated with development of aggregates of the TDP-43 protein in brain cells. Mutations in PGRN are responsible for some cases of hereditary FTD.

Prosopagnosia - The inability to identify a person by the appearance of his face.

Protein - Chains of amino acids. Most proteins serve a specific structural or enzymatic function in the body.

Semantic variant PPA - One of the language variants of FTD. Patients lose the meaning of words but can still produce speech that is fluent (if empty of meaning). Semantic PPA is often caused by accumulation of the protein TDP-43. Also known as semantic dementia.

Semantic memory - The memory of meaning, understanding and other concept-based knowledge. Remembering that a robin is a bird with a red breast is one example of semantic knowledge.

Semantic paraphasia - The substitution of a word that is closely related to the target word, as in "cat" for "dog."

Sporadic - A disease is said to be sporadic when no other family members have a related disease and there is no known genetic cause.

TAR DNA - binding protein (TARDBP) – A protein in the nervous system that is made by the TARDBP gene. Mutations in this gene are associated with hereditary ALS and rare cases of hereditary FTD.

Tauopathy - Refers to neurodegenerative diseases which are associated with abnormal accumulation of tau protein.

TDP-43 - The abnormal version of the TAR DNA binding protein. Buildup of TDP-43 is associated with approximately 50% of FTD cases and 100% of ALS.
Tau - A protein used in the brain to support the microtubules in the axons of neurons. Abnormal accumulation of tau is seen in about 40% of FTD cases.

Temporal lobes - The brain lobes located on the sides of the brain near the ears which are responsible for hearing, language comprehension, memory, and visual and emotional processing.

Tremors - Involuntary shaking movements most easily noticed in the hands and head when present. Tremor is a major symptom in Parkinson’s disease but is also found in other conditions.

Valosin-containing protein (VCP) - A protein made by the VCP gene. Mutations in this gene cause a very small portion of hereditary FTD and a specific condition called inclusion body myopathy with Paget disease of the bone.

We welcome input on terms that should be added to the Glossary.