FTD DISORDERS REGISTRY®

Research Survey & Clinical Trials Glossary

Summer 2020 Newsletter

Autosomal Dominant — A pattern of inheritance in which an affected individual has one copy of a mutant gene and one normal gene on a pair of autosomal chromosomes.

Autosomal Recessive — A pattern of inheritance in which an affected individual has two copies of the mutant gene.

Biased — Systemic errors that encourage one outcome over another.

Citizen Scientist — The practice of public participation and collaboration in research to increase scientific knowledge. Through citizen science, people share and contribute to data monitoring and collection programs.

Clinical Study — A research study involving human volunteers (also called participants) to answer a specific health question.

Clinical Trial (also known as Interventional Study) — A type of clinical study in which participants are assigned to groups and receive a treatment to help researchers learn the effects of the interventions on a health condition.

Contact Registry — A type of registry that includes contact information and basic demographic data. A contact registry needs to collect enough information to determine who qualifies for a research study.

Control/Control Group — The group of individuals in a clinical trial assigned to a comparison intervention.

C9orf72 — A mutation in the chromosome 9 gene open reading frame 72 allows a series of nucleotides to repeat too many times, which leads to abnormal function of proteins.

De-identify — The process used to prevent someone's personal identity from being revealed in clinical studies.

Efficacy — The ability of the intervention to produce the desired therapeutic effect on the disease or condition being studied.

Eligibility Criteria — The requirements that people who want to participate in a clinical study must meet.

Emerging — Newly created or noticed and growing in strength or popularity.

Facilitate — To help cause a process or action.

Food and Drug Administration (FDA) — The branch of the federal government that approves new drugs.

GLOSSARY continues on Page 2

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GLOSSARY continues from Page 1

Gene Mutation — A permanent change in the DNA sequence that makes up a gene. FTD disease-causing mutations are most often seen in: *C9orf72*, Microtubule-associated protein tau (*MAPT*), Progranulin (*GRN* or *PGRN*), and Valosin-Containing Protein (*VCP*).

Genetic or Gene Testing — A type of medical test that discovers changes in chromosomes, genes, or proteins; usually performed by collecting blood or saliva.

Healthy Volunteer — A person who does not have the disorder or disease being studied.

Heterozygous Mutations — A permanent change of only one allele.

Homozygous Mutations — An identical permanent change in both the paternal and maternal alleles.

Infographic — A visual representation of information or data.

Informed Consent — A process used by researchers to tell enrolled participants about the risks and benefits of the clinical study.

Intervention — A treatment or action taken to prevent or treat disease, or improve health in other ways.

Interventional Study (also known as Clinical Trial) — A type of clinical study in which participants are assigned to groups and receive a treatment to help researchers learn the effects of the interventions on a health condition.

Microtubule-associated protein tau (*MAPT*) — A mutation in this gene causes a permanent change to genes responsible for making tau protein. This mutation reduces the gene's ability to bind, which interrupts many cell functions.

Natural History Studies — A study that follows a group of people over time who have, or are at risk of developing, a specific medical condition or disease. Two of the largest FTD examples of these studies have been the ARTFL (Advancing Research and Treatment for Frontotemporal Lobar Degeneration) and the LEFFTDS (Longitudinal Evaluation of Familial Frontotemporal Dementia Subjects) studies which have been combined into the ALLFTD (ARTFL LEFFTDS Longitudinal Frontotemporal Lobar Degeneration) Study.

Observational Study — A clinical study in which participants are identified as belonging to study groups and are watched over time for biomedical or health outcomes.

Orphan Drug — A medical agent developed to treat a rare disease due to its limited market.

GLOSSARY continues on Page 3

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GLOSSARY continues from Page 2

Outcome Measure — A specific measurement to evaluate the effect, both positive and negative, of an intervention on participants. Example outcomes are changes in health, function, or quality of life.

Patient-centric — The process of putting the patient at the forefront to improve the overall experience.

Pharmacokinetics — The study of how the body absorbs, distributes, and gets rid of the drug.

Pharmacodynamics — The study of the effects of drugs on the body.

Phase/Trial Phases — The stage of a clinical trial studying a drug or other treatment, based on definitions developed by the Food and Drug Administration (FDA). These are broken out as Early Phase 1 (formerly listed as Phase 0), Phase 1, Phase 2, Phase 3, and Phase 4.

Placebo — A placebo is an inactive pill or liquid that contains no medicine.

Progranulin (*GRN* or *PGRN*) — A mutation in this gene causes a permanent change in the instructions for making the progranulin protein. This mutation reduces progranulin levels or results in loss of function.

Protocol — The written description of a clinical study.

Randomization — The process in which two or more treatments are assigned to volunteers by chance rather than by choice.

Rare Disease — A disease that affects fewer than 200,000 people in the United States. Scientists estimate that FTD affects about 60,000 people.

Registry — An electronic database of information about individuals with a specific disorder or medical condition.

Research Registry — A collection of information about individuals, usually focused around a specific diagnosis or condition to assess health outcomes. In some registries, patients and their families report the information. In others, the information is collected and reported by medical professionals.

Sortilin (*SORT1*) — A protein that in humans is encoded by the *SORT1* gene on chromosome 1. It is involved in the sorting, transport, recycling, and degradation of other proteins.

Valosin-Containing Protein (*VCP*) — A mutation in this gene causes a permanent change in the single protein building block in VCP genes. This mutation keeps the gene from breaking down protein, which causes buildup and affects normal cell function.

Join the Registry. Tell Your Story. Advance the Science.

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