

## Amyloid Plaques

Amyloid plaques are accumulations of abnormally-configured proteins deposited in the liver, kidneys, spleen, or other tissues. In the brain of someone affected by Alzheimer's disease, abnormal levels of an amyloid protein called beta-amyloid 42 clump together to form plaques that collect between neurons and disrupt cell function.

## Biomarker

A biomarker is a biological marker to measure change. It's a reliable predictor and indicator of disease and disease progression. Examples of biomarkers include glucose for diabetes and cholesterol for heart disease. The biomarkers currently used for Alzheimer's disease are brain imaging known as PET scans. There are two FDA approved types: one that can detect amyloid plaques and one that can detect tau tangles. Biomarkers found in cerebrospinal fluid (CSF) or blood are used in research studies but are not yet ready for clinical practice.



## Biotech (Biotechnology)

Biotechnology (often shortened to "biotech") companies conduct research on the use of live organisms, such as bacteria or enzymes, to develop medicines. Conversely, pharmaceutical companies research and create medicines using chemicals and synthetic processes. Both biotechnology and pharmaceutical companies are involved in Down syndrome research to tackle challenges facing the community, including the high risk of Alzheimer's disease.

## Clinical Endpoint

This is an event that can be measured to prove the efficacy of a treatment being tested in a clinical trial. Endpoints can be objective (e.g., increased survival rates of patients) or subjective (e.g., patient offering a "symptom score" to measure their experience). Sometimes, trials will use a "surrogate" endpoint to show that although there is no provable causal relationship between their tested treatment and a traditional clinical endpoint, there is a demonstrable effect on a biomarker related to the condition/disease being treated.





# RESEARCH GLOSSARY

[LuMindIDSC.org/research](http://LuMindIDSC.org/research)

## Clinical Trial

The U.S. National Institutes of Health (NIH) define a clinical trial as a research study in which one or more human research subjects are prospectively assigned to one or more interventions (which may include placebo or other control) to evaluate the effects of those interventions on health-related biomedical or behavioral outcomes. Interventions may be medical products, such as drugs or devices; procedures; or changes to participants' behavior, such as diet.



## DSC2U

DSC2U, or "Down Syndrome Community to You," is digital tool created by the Massachusetts General Hospital Down Syndrome Program that helps a caregiver prepare for his/her loved one's doctor appointment via a questionnaire/survey tool developed by clinical experts to facilitate an optimal doctor's visit.



## DS-CTN

The Down Syndrome Clinical Trial Network (DS-CTN) is a collaboration of Down syndrome-related medical and academic research clinics located across the U.S. DS-CTN sites serve as drivers of research studies and clinical trials, focused primarily on the diagnosis, treatment, and prevention of Alzheimer's disease onset in Down syndrome.



## FDA (Food and Drug Administration)

The U.S. Food and Drug Administration is responsible for protecting the public health by ensuring the safety, efficacy, and security of human and veterinary drugs and the U.S. food supply. After conducting research on the efficacy of a drug, biotech and pharmaceutical companies need to send their results to the FDA showing that it is safe, effective, and meets regulatory standards.





## Hypoglossal Nerve Stimulation

Hypoglossal nerve stimulation is a type of treatment for obstructive sleep apnea, a common condition in people with Down syndrome. The treatment involves using an implantable pacemaker-sized pulse generator to stimulate the hypoglossal nerve, which starts at the base of an individual's brain and travels down the neck to the tongue. By electrically stimulating the nerve, the tongue is moved out of the way to ensure breathing is possible.

## Hypothesis

A hypothesis (plural "hypotheses") is a proposed explanation for a scientific phenomenon, which is then tested within the structure of a scientific study.



## Industry

Industry generally means a group of companies that conduct similar business, e.g. "the cosmetics industry," "the film industry." In the context of LuMind IDSC and Down syndrome research, "industry" most often refers to the pharmaceutical and biotech industries, who fund research with the goal of producing medicines and/or therapeutic solutions to challenges facing the Down syndrome community, such as Alzheimer's disease and obstructive sleep apnea.



## IRB (Institutional Review Board)

An IRB, or "Institutional Review Board," is defined by the FDA as a group that has been formally designated to review and monitor biomedical research involving human subjects. An IRB has the authority to approve, require modifications in (to secure approval), or disapprove research and any corresponding materials (such as marketing/advertising content). FDA regulations require that IRBs have at least five members and take into consideration race, gender, and cultural background to build a diverse membership.





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## LIFE-DSR

The “Longitudinal Study for the Enhancement of Down Syndrome Research” is a landmark study founded by LuMind IDSC is collecting key data from 270 adults with Down syndrome to learn how they change with age. Insights from the study are critical for developing Alzheimer’s therapies for this population. LIFE-DSR is operated through the DS-CTN, with support from industry companies via the LuMind IDSC Research Consortium.



## LuMind IDSC Research Consortium

The LuMind IDSC Research Consortium accelerates Down syndrome research in pursuit of significant medical and therapeutic outcomes. The Consortium (currently Merck, AbbVie, and Lilly, along with NDSS) will expand LIFE DSR research to include key sub-studies.

## myDSC

myDSC, which is short for “my Down Syndrome Community,” is LuMind IDSC’s digital platform for reliable Down syndrome resources and tools. It is a free membership portal with hundreds of curated resources, customized libraries of information, and special member discounts at external vendors.



## Natural History Study

A natural history study collects information about the natural history of a condition in the absence of an intervention, from the condition’s onset until either its resolution or the individual’s death. Researchers observe participants as they are by recording medical, physical, and behavioral data points, like: height, weight, blood/ plasma samples, key behaviors, sleeping patterns, and blood pressure. Researchers use the information collected from all study participants to better understand the clinical profile of a given condition, such as DS.





## Obstructive Sleep Apnea

In terms of the Down syndrome community, “regression” is broadly defined as the loss of skills over time. Examples include mood instability, loss of speech ability, and social withdrawal. One major Down syndrome regression-related condition being researched today is Down syndrome disintegrative disorder (DSDD), which may occur in children and young adults.

## PI (Private Investigator)

The principal investigator is the researcher leading a clinical trial or scientific research project. The principal investigator prepares and carries out the plan for the research. Once the initial research has been conducted, the PI analyzes the data and reports the results.



## Placebo Effect

A placebo is a fake treatment with no known clinical benefit given to one group of a clinical trial, while the other group (the “control group”) receives the actual treatment being tested. The placebo effect is a beneficial health outcome resulting from a member of the placebo group’s anticipation that the tested treatment will help.

## Preclinical Research

Before clinical trials for potential new treatments can begin on humans, they must go through preclinical research that proves they are safe enough to be tested that way. Preclinical studies take place in two ways: *in vitro* (Latin for “within the glass”), in which the treatment is tested with cell cultures in a Petri dish or test tube; and *in vivo* (Latin for “within the living”), in which the treatment is tested on animal subjects.



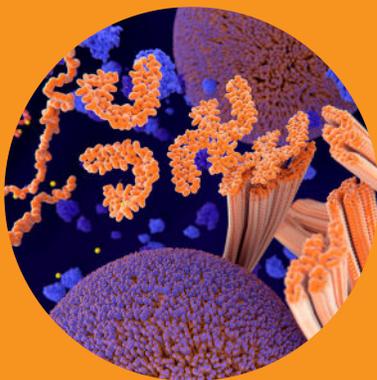


## Regression

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## Sponsor

In the context of a clinical trial, a sponsor is a person, company, institution, group, or organization that oversees or pays for the trial. The sponsor is responsible for oversight of the trial and communication with the FDA. The longer term “trial sponsor” also refers to this same idea.



## Tau Tangles

Tau tangles (also known as neurofibrillary tangles) are abnormal accumulations of a protein called tau that collect inside neurons in specific regions of the brain involved in memory, harming the synaptic communication between them. Scientists believe there is a relationship between the accumulation of amyloid plaque and tau tangles. As the amount of beta-amyloid in the brain increases, a tipping point is reached that causes abnormal tau to spread throughout the brain, leading to memory decline and loss of skills.

## Translational Research

Translational research is conducted with the goal of solving a particular problem; for example, reducing the risk of Alzheimer’s disease in adults with Down syndrome. It’s the process of transforming research discoveries from the laboratory into practical medical or therapeutic protocols, policies, approaches, and treatments.

