Glossary of Terms

List is not complete nor comprehensive

Term	Meaning/Definition
2FA	Two Factor Authorization
ANDA	Abbreviated New Drug Application is an application for a U.S. generic drug approval for an existing licensed medication or approved drug.
ASO	Antisense Oligonucleotide - are small, synthetically created pieces of DNA or RNA that can bind to specific molecules of RNA and block them from making a protein. ASOs can also be used to alter the process of splicing which is a cut-and-paste process for DNA. This can be useful to either include or exclude an important part of the gene that is impacted due to a mutation. In either case, the final goal is to have a mature RNA that will be made into a properly functioning protein
Basket trials	Patients all receive the same treatment that targets the specific mutation or biomarker. Basket trials may allow new drugs to be tested and approved more quickly than traditional clinical trials.
BGTC	Bespoke Gene Therapy Consortium
Biobank	A biorepository, a collection of biological samples (such as blood) and health information
Biomarker	A biological molecule found in blood, other body fluids, or tissues or a measureament of some function that is a sign of a normal or abnormal condition or response to treatment.
BLA	A biologics license application is defined by the U.S. Food and Drug Administration as “a request for permission to introduce, or deliver for introduction, a biologic product into interstate commerce”.
CDER	Center for Drug Evaluation and Research - As part of the U.S. Food and Drug Administration (FDA), CDER regulates over-the-counter and prescription drugs, including biological therapeutics and generic drugs.
CDISC	Clinical Data Interchanges Standards Consortium - An organization that creates and maintains standards for clinical research. http://www.cdisc.org
CDMO	A contract development and manufacturing organization provides end-to-end drug development and manufacturing services to biotechnology and pharmaceutical companies.

Cell lines	A specific population of cells that can maintain in vitro culture for an extended period of time, and is usually clonal meaning that the entire population originated from a single, common, ancestor cell. Used in vaccine production, testing drug metabolism and cytotoxicity, antibody production, and the study of gene function
CIRM	California Institute for Regenerative Medicine -The California Institute for Regenerative Medicine was created in 2004 after 59% of California voters approved California Proposition 71: the Research and Cures Initiative, which allocated $3 billion to fund stem cell research in California.
COA	Clinical Outcome Assessment - A measure that describes or reflects how a patient feels, functions, or survives.
Compassionate use	Also call Expanded access - is the use of an unapproved drug or medical device under special forms of investigational new drug applications (IND) or IDE application for devices, outside of a clinical trial, by people with serious or life-threatening conditions who do not meet the enrollment criteria for the clinical trial in progress
CRO	A contract research organization provides support to the pharmaceutical and biotechnology industries in the form of research services outsourced on a contract basis. These services may include assay development, clinical development, clinical-trial management, pharmacovigilance, and other.
CZI	Chan Zuckerburg Initiative -The Chan Zuckerberg Initiative was founded in 2015 to help solve some of society's toughest challenges — from eradicating disease and improving education to addressing the needs of our local communities. Our mission is to build a more inclusive, just, and healthy future for everyone.
PRO	Patient-reported outcomes/data-Information about a patient's health that comes directly from the patient. Examples of patient-reported outcomes include a patient's description of their symptoms, their satisfaction with care, and how a disease or treatment affects their physical, mental, emotional, spiritual, and social well-being.
Data standard	A formally governed set of formats, principles, or terms that specify how data should be collected, stored, or exchanged.
DCP	Data Collection Platform / Data Collection Program
DNA	deoxyribonucleic acid: an extremely long macromolecule that is the main component of chromosomes and is the material that transfers genetic characteristics in all life forms, constructed of two nucleotide strands coiled around each other in a ladderlike arrangement with the sidepieces composed of alternating phosphate and deoxyribose units and the rungs composed of the purine and pyrimidine bases adenine, guanine, cytosine, and thymine: the genetic information of DNA is encoded in the sequence of the bases and is transcribed as the strands unwind and replicate.
DPA	Data Protection Addendum - A data processing agreement — also called a data processing addendum or DPA — is a legal contract in which you determine the rights and obligations of the parties involved in data processing. Most of the time that includes your business and any third-party services you use.
DTA	A Data Transfer Agreement - is a legal contract governing the transfer of non-human subject data or completely de-identified human subject data. It sets out the related protections, rights, and obligations of both parties and delineates the specific purpose(s) for which the data may be used.
DUA	Data Use Agreement-Definition - is a legal contract between the entity that owns access to a data source, typically a dataset or database, and a secondary entity that will receive the data, or a subset of it, for reuse.

EDC	Electronic Data Collection; refers to the database system used to enter and capture data from a clinical trial
FAIR data principles	A set of guidelines for making data Findable, Accessible, Interoperable, and Reusable. https://www.go-fair.org/fair-principles/
FDA	Food and Drug Administration, the federal agency responsible for protecting the public health by assuring the safety, efficacy, and security of drugs, biological products, medical devices, etc.
GARD	Genetic and Rare Diseases Information Center https://rarediseases.info.nih.gov/
H2L	Hit to lead (H2L) also known as lead generation is a stage in early drug discovery where small molecule hits from a high throughput screen (HTS) are evaluated and undergo limited optimization to identify promising lead compounds.
Human Phenotype Ontology (HPO)	A standardized, computable vocabulary of phenotypic abnormalities found in human disease.
IND	An investigational new drug is a substance that has been tested in the laboratory and has been approved by the U.S. Food and Drug Administration for testing in people.
IND-Enabling Dataset	An Investigational New Drug (IND) application is the first regulatory step that drug developers must take when preparing an investigational drug for human clinical studies. During a new drug's early preclinical development, it has to be demonstrated that a compound is reasonably safe for use in humans and is efficacious enough to justify commercial development. IND-enabling studies are conducted and datasets are generated to evaluate potential toxicity risks prior to human studies and to estimate starting doses for clinical trials. According to the Food and Drug Administration (FDA), IND applications must include data on: 1. Animal pharmacology and toxicology studies 2. Manufacturing information 3. Clinical protocols and investigator information The specific IND-enabling studies needed are unique for each drug candidate.
IPS cells	Induced Pluripotent Stem Cells can differentiate into any type of cell in the body. These can be used as disease models.
IRB	Institutional Review Board - an administrative body established to protect the rights and welfare of human research subjects
Longitudinal Study	Tracking the same variable over time. As in the same patient data points over many years or a lifespan.
MOU	Memorandum of Understanding - A memorandum of understanding (MOU) is a document describing the broad outlines of an agreement that two or more parties have reached.
MSA	Master Service Agreement - A master service agreement, sometimes known as a framework agreement, is a contract reached between parties, in which the parties agree to most of the terms that will govern future transactions or future agreements.

MTA	Material Transfer Agreement - A Material Transfer Agreement (MTA) is a contract governing the transfer of materials between two parties. It defines the rights of the provider and the recipient with respect to the materials and any derivatives. MTAs can help to ensure a common understanding as to what is being shared, for what purpose, and how it can be used. MTAs regularly govern the transfer of biological materials, such as samples but can also cover associated data, such as meta data or the clinical state of the donor.
Mutation Agnostic	particular disease-causing variation (aka mutation) in a gene. Because of the great diversity of mutations within a particular rare disease, therapy that only targets a specific mutation can be challenging and slow to develop. This highlights the need for mutation-agnostic treatments where molecules are able to mimic the overall function of a gene instead of correcting the effects of one particular mutation or type of mutation. Mutation-agnostic therapies have the potential to benefit patients that have different mutations but have the same underlying disease.
N of 1 trials	Studies of treatments developed for one or a few patients.
Natural History Study	An observational study intended to track the course of a disease over time. Its purpose is to identify demographic, genetic, environmental, and other variables (e.g., treatment modalities, concomitant medications) that correlate with the disease's development and outcomes. Natural history studies are likely to include patients receiving the current standard of care and/or emergent care, which may alter some manifestations of the disease.
NCATS	National Center for Advancing Translational Sciences
NDA/CDA	Non-Dislosure Agreement/ Confidential Disclosure Agreement
NIH	National Institutes of Health
NINDS	National Institute of Neurological Disorders and Stroke
NORD	National Organization for Rare Disorders
Ontology	A set of terms and relationships that describes the knowledge in a domain. Modern ontologies are stored in a computable format that allows special programs called reasoners to make additional inferences about the data or knowledge associated with the ontology.
Organoid	Organoids are 3-dimensional mini-organ-like structures that are engineered from stem cells to grow in the laboratory. Organoids mimic many aspects of the complex structure and function of human tissues and organs and are used to study organ structure and function outside of the body. To date, researchers have been able to produce organoids that resemble the brain, kidney, lung, intestine, stomach, and liver.
PAG	Patient Advocacy Group
Phase 1 Clinical Trial	Phase I studies of a new drug are usually the first that involve humans recieved the therapy.
Phase 2 Clinical Trial	Determine the effectiveness of an experimental drug on a particular disease or condition in a sample volunteer group of approximately 100 - 300
Phase 3 Clinical Trial	Demonstrates whether or not a product offers a treatment benefit for a certain disease or condition.
Preclinical	A stage of research that begins before clinical trials and during which important feasibility, and drug safety data are collected, typically in laboratory animals.

PRO	Patient Reported Outcome
RaDaR	Rare Diseases Registry Program - online resource website that provides patient groups with guidance on how to develop registries for rare diseases. Good-quality registries help support research programs and stimulate treatment development. The RaDaR website is an educational platform based on the NCATS Toolkit for Patient-Focused Therapy Development.
Registry	A database that collects and stores specified types of information that are usually related in some way. In the context of the therapy development pathway, a registry usually collects information about patients who have a specific disease or condition and may be referred to as a patient registry.
RMAT	Regenerative Medicine Advanced Therapy - is a designation given by the Food and Drug Administration to drug candidates intended to treat serious or life-threatening conditions under the 21st Century Cures Act. A RMAT designation allows for accelerated approval based surrogate or intermediate endpoints.
RNA	ribonucleic acid: any of a class of single-stranded molecules transcribed from DNA in the cell nucleus or in the mitochondrion or chloroplast, containing along the strand a linear sequence of nucleotide bases that is complementary to the DNA strand from which it is transcribed: the composition of the RNA molecule is identical with that of DNA except for the substitution of the sugar ribose for deoxyribose and the substitution of the nucleotide base uracil for thymine
RNA-based therapies	RNA-based therapeutics are a new class of medications that are based on modulating RNA molecules. RNA are go-between molecules between DNA and protein and facilitate the decoding of genetic instructions from DNA to the protein-making machinery. The main types of RNA therapeutics are those based on: * messenger RNA (mRNA) where mRNAs drive the transient production of certain proteins in the cytoplasm of the cell (for instance, to replace defective proteins or present antigens for vaccination) *antisense RNA (asRNA) where antisense oligonucleotides can alter RNA and reduce, restore, or modify protein expression. *RNA interference (RNAi) is a cellular process that induces the degradation of particular RNA targets. siRNAs are small interfering double-stranded RNA molecules that can be used to silence the expression of defective genes. RNA aptamers are short single-stranded RNAs that can bind to a variety of targets, such as proteins, peptides, and carbohydrates. RNA aptamers can be
SAAS	Software As A Service -a method of software delivery and licensing in which software is accessed online via a subscription, rather than bought and installed on individual computers.
SoC	Standard of Care - refers to the type of care (e.g., procedures, assessments) and frequency of care that a patient receives as part of usual treatment from a physician, i.e., not as part of a clinical trial. These can be unique to a certain diagnosis or disease state. Similar in concept to Best practices.

SOE/SOA	Schedule of Events / Schedule of Assessments - refers to the list of procedures/assessments required of each patient in a clinical trial. Usually expressed in text and as a table, the SOA also indicates the frequency and/or volume of assessment (eg., blood collected) and if there is a specific order in which the procedures/assessments must be performed.
SOP	Standard Operating Procedure - A standard operating procedure is a set of written instructions that describes the step-by-step process that must be taken to properly perform a routine activity. SOPs should be followed the exact same way every time to guarantee that the organization remains consistent and in compliance with industry regulations and business standards.
SOW	Statement of Work - A statement of work is a document routinely employed in the field of project management. It is the narrative description of a project's work requirement. It defines project-specific activities, deliverables and timelines for a vendor providing services to the client
TPSP	Third Party Service Provider - A third-party service provider is any unaffiliated person, company, or entity that performs services for a company. Third-party service providers are paid for their services, but do not have a stake, share, or equity in the company.
UDN	Undiagnosed Disease Network - The Undiagnosed Diseases Network (UDN) is a research study backed by the National Institutes of Health that seeks to provide answers for patients and families affected by these mysterious conditions.
UDP	Undiagnosed Disease Program - The National Institutes of Health (NIH) Undiagnosed Diseases Program is part of the Undiagnosed Disease Network (UDN), an NIH Common Fund initiative that focuses on the most puzzling medical cases referred to the NIH Clinical Center in Bethesda, Maryland. It was organized by the National Human Genome Research Institute (NHGRI), the NIH Office of Rare Diseases Research (ORDR) and the NIH Clinical Center.
Umbrella Trail	A research study that tests and compares two or more study treatments (potential therapies) for one disease or condition.
Vector	Vectors are vehicles by which to deliver genetic material into a cell for gene therapy. There are two types of vectors: viral and non-viral. Viral vectors are built using parts of a virus that help with delivering genetic material to the right part of cells. Non-viral vectors depend on physical or chemical methods to deliver genetic material into a cell, including chemical disruption and use of electrical currents. As of now, viral vectors are the most common vehicle used in gene therapies.