
GLOSSARY OF TECHNICAL TERMS

This glossary contains explanations of certain technical terms used in this document. As such, these terms and their meanings may not correspond to standard industry meanings or usage of these terms.

“ADOD”	Alzheimer’s disease and other dementias, refer to a group of neurodegenerative disorders
“amniocentesis”	a medical procedure that removes amniotic fluid from uterus for prenatal testing
“bioinformatics”	a subdiscipline of biology and computer science concerned with the acquisition, storage, and analysis of biological data, most often DNA and amino acid sequences
“BoBs assays”	BACs-on-Beads, a modification of comparative genomic hybridization used to detect DNA copy number variations and chromosomal aneuploidies
“CAGR”	compound annual growth rate
“cfDNA”	cell-free DNA, refers to all non-encapsulated DNA in the blood stream, cerebrospinal fluid and other body fluids
“chorionic villus sampling (CVS)”	a prenatal testing procedure using a small sample of placental tissue to detect chromosomal anomalies and genetic disorders in the fetus
“chromosomal aneuploidy”	a genetic disorder caused by the presence of an abnormal number of chromosomes in a cell
“Class I medical device”	medical devices with the lowest level of risk, which require compliance with general controls but typically do not need pre-market approval from the regulatory authorities
“Class II medical device”	medical devices with a moderate level of risk, which requires the regulatory approval from the regional MPA
“Class III medical device”	the medical device with highest risk level, which requires the regulatory approval from the NMPA
“CMA”	Chromosome microarray analysis, a method that uses microarray chips to conduct high-resolution genome-wide analysis for detecting copy number variations (CNVs)

GLOSSARY OF TECHNICAL TERMS

“CNV”	copy number variations, a phenomenon in which sections of the genome are repeated and the number of repeats in the genome varies between individuals
“CRO”	Contract Research Organization, handling tasks such as clinical trial management, data analysis, and regulatory compliance to support the development and approval of new products
“ <i>de novo</i> sequencing”	sequencing a novel genome where there is no reference sequence available for alignment
“ELISA”	referring to enzyme-linked immunosorbent assay, which is used to measure antibodies, antigens, proteins and glycoproteins in biological samples
“fluorescent immunoassay”	a sensitive technique that can be used in the measurement of many compounds, including drugs, hormones and proteins
“full-length transcriptome sequencing”	a sequencing method that captures and sequences entire RNA molecules from end to end
“gene expression”	the regulated process by which the genetic information encoded in DNA is transcribed into RNA and often translated into functional proteins
“genome assembly”	the reconstruction of the whole genome sequence by aligning and merging sequence reads generated from the genetic sequencing
“GMP”	good manufacturing practices, the aspect of quality assurance that ensures that medicinal products are consistently produced and controlled to the quality standards
“Hi-C technology”	a high-throughput genomic and epigenomic technique to capture chromatin conformation, detecting genome-wide chromatin interactions in the cell nucleus
“impact factors (IF)”	a journal-level metric used to measure the importance of a journal
“InDels”	Insertions/deletions, genetic mutations where small segments of DNA are inserted or deleted from the genome
“Independent Clinical Laboratory (ICL)”	a laboratory certified to perform diagnostic and/or clinical tests independent of an institution or a physician's office
“IVD product”	in vitro diagnostic products, the reagents, instruments and systems intended for use in diagnosis of disease or other conditions

GLOSSARY OF TECHNICAL TERMS

“karyotyping”	the process of pairing and ordering all the chromosomes of an organism, thus providing a genome-wide snapshot of an individual’s chromosomes
“KOLs”	acronym for Key Opinion Leaders; refers to renowned physicians that influence their peers’ medical practice
“laboratory developed test (LDT)”	a type of in vitro diagnostic test that is designed, manufactured and used within a single laboratory
“long-read HiFi sequencing”	a single-molecule, real-time sequencing technology (SMRT) that provides incredible single-molecule read accuracy across long reads of tens of kilobases in length or more
“methylation”	the addition of methyl groups to DNA nucleotides, which modulates gene expression without altering the underlying sequence and is frequently analyzed in genetic testing
“microdeletion”	disorders caused by the deletion of small segments of DNA that are too small to be detected
“microduplication”	disorders caused by a chromosomal change in which a DNA fragment on chromosome is abnormally copied
“molecular diagnostics”	a field of medical testing that involves analysing biological markers in the genome and proteome
“mosaicism”	the presence of two or more cell lines with different genetic makeup or chromosomal constitution derived from a single zygote
“MPA”	Medical Products Administration, which is a regional regulatory authority responsible for overseeing the approval and regulation of medical products
“MRI”	a medical imaging technique that uses a magnetic field and computer-generated radio waves to create detailed images of the organs and tissues inside the body
“mRNA”	messenger RNA, an RNA produced by transcription that carries the code for a particular protein from the nuclear DNA to a ribosome in the cytoplasm
“multi-omics”	a biological analysis approach in which the data sets are multiple “omes”, such as the genome, proteome, transcriptome, epigenome, metabolome and microbiome

GLOSSARY OF TECHNICAL TERMS

“neurodegenerative disease”	a range of conditions characterized by the progressive loss of specific groups of neurons
“NGS”	Next-Generation Sequencing, a massively parallel sequencing technology for determining the sequence of nucleotides in a targeted region of an entire genome or DNA or RNA
“NIPT”	non-invasive prenatal testing, a screening test during which the peripheral blood of pregnant women is collected to screen chromosomal anomalies of fetuses
“NMPA”	National Medical Products Administration, the regulatory authority in China responsible for the supervision and management of pharmaceuticals, medical devices and cosmetics
“organogenesis”	the developmental process during embryogenesis in which the three germ layers differentiate and give rise to the internal organs of a living organism
“PCR”	polymerase chain reaction, a method widely used to rapidly make millions to billions of copies of a specific DNA sample
“phosphorylated tau-181 protein (p-Tau-181)”	a modified form of the tau protein, serving as a biomarker for Alzheimer’s disease and other neurodegenerative disorders
“rRNA”	ribosomal RNA, a key structural and catalytic component of ribosomes, which facilitate mRNA translation and polypeptide assembly
“sensitivity”	the ability of a test to correctly identify those with the disease (true positive rate)
“single-cell sequencing”	an advanced sequencing examines the nucleic acid sequence information from individual cells with optimized genetic sequencing technologies
“SMA”	Spinal muscular atrophy, a genetic disorder characterized by the progressive degeneration of motor neurons in the spinal cord
“SMO”	Site Management Organization, an entity that provides services to manage clinical trial sites, including the recruitment and training of clinical research coordinators
“SNV”	Single nucleotide variants, genetic variations involving a change of a single nucleotide in the DNA sequence, which can impact gene function and contribute to genetic diversity or disease

GLOSSARY OF TECHNICAL TERMS

“SV”	Structural variants, large-scale genetic alterations involving rearrangements, duplications, deletions, or inversions of DNA segments
“spatial sequencing”	an advanced genomic technique that captures nucleic acid sequence information while preserving the spatial context of cells within tissues
“specificity”	the ability of the test to correctly identify those without the disease (true negative rate)
“stereo sequencing”	a high-throughput spatial transcriptomics technology
“target capture sequencing”	a new sequencing technology that allows researchers to extract genome information from exons or regions of interest in the human genome with a wide selection of panels
“transcriptome sequencing”	also known as RNA sequencing or RNA-seq, uses high-throughput sequencing technology to analyze RNA
“trisomy”	the presence of three copies of a chromosome rather than the normal two copies.
“tRNA”	transfer RNA, a small RNA molecule that serves as a link between mRNA molecule and the growing chain of amino acids
“UPD”	uniparental disomy, a genetic anomaly where both copies of a chromosome are inherited from one parent instead of one copy from each parent
“ β -amyloid 1–42 (A β 1–42)”	a peptide consisting of 42 amino acids derived from the amyloid precursor protein, being a major component of found in the brains of individuals with Alzheimer’s disease