## **Bibliography**

- CEN/TS 17688-1:2021, Molecular in vitro diagnostic examinations Specifications for pre-examination processes for Fine Needle Aspirates (FNAs) Part 1: Isolated cellular RNA
- EN ISO 14155, Clinical investigation of medical devices for human subjects Good clinical practice (ISO 14155
- EN ISO/IEC 17043:2010, Conformity assessment General requirements for proficiency testing (ISO/IEC 17043:2010)
- EN ISO 17511:2021, In vitro diagnostic medical devices Requirements for establishing metrological traceability of values assigned to calibrators, trueness control materials and human samples (ISO 17511:2020)
- EN ISO 20166-1:2018, Molecular in vitro diagnostic examinations Specifications for pre-examination processes for formalin-fixed and paraffin-embedded (FFPE) tissue Part 1: Isolated RNA (ISO 20166-1:2018)
- EN ISO 20166-2:2018, Molecular in vitro diagnostic examinations Specifications for pre-examinations processes for formalin-fixed and paraffin-embedded (FFPE) tissue Part 2: Isolated proteins (ISO 20166-2:2018)
- EN ISO 20166-4:2021, Molecular in vitro diagnostic examinations Specifications for pre-examination processes for formalin-fixed and paraffin-embedded (FFPE) tissue Part 4: In situ detection techniques (ISO 20166-4:2021)
- EN ISO 22174:2005, Microbiology of food and animal feeding stuffs Polymerase chain reaction (PCR) for the detection of food-borne pathogens General requirements and definitions (ISO 22174:2005)
- ISO 5725-1:2023, Accuracy (trueness and precision) of measurement methods and results Part 1: General principles and definitions
- ISO 5725-2:2019, Accuracy (trueness and precision) of measurement methods and results Part 2: Basic method for the determination of repeatability and reproducibility of a standard measurement method
- ISO 9000:2015, Quality management systems Fundamentals and vocabulary
- ISO 13495:2013, Foodstuffs Principles of selection and criteria of validation for varietal identification methods using specific nucleic acid
- ISO 14621-1:2019, Space systems Electrical, electronic and electromechanical (EEE) parts Part 1: Parts management
- ISO 14971, Medical devices Application of risk management to medical devices
- ISO 15190, Medical laboratories Requirements for safety
- ISO 15193:2009, In vitro diagnostic medical devices Measurement of quantities in samples of biological origin Requirements for content and presentation of reference measurement procedures

- ISO 15194:2009, In vitro diagnostic medical devices Measurement of quantities in samples of biological origin Requirements for certified reference materials and the content of supporting documentation
- ISO 17123-1:2014, Optics and optical instruments Field procedures for testing geodetic and surveying instruments Part 1: Theory
- ISO 17822:2020, In vitro diagnostic test systems Nucleic acid amplification-based examination procedures for detection and identification of microbial pathogens Laboratory quality practice guide
- ISO 17966:2016, Assistive products for personal hygiene that support users Requirements and test methods
- EN ISO 18113-1:2009, In vitro diagnostic medical devices Information supplied by the manufacturer (labelling) Part 1: Terms, definitions and general requirements (ISO 18113-1:2009)
- ISO 20387, Biotechnology Biobanking General requirements for biobanking
- ISO 20395:2019, Biotechnology Requirements for evaluating the performance of quantification methods for nucleic acid target sequences. qPCR and dPCR
- ISO/TS 20658, Medical laboratories Requirements for collection, transport, receipt, and handling of samples
- ISO/TS 22692, Genomics Informatics Quality control metrics for DNA sequencing
- ISO 22949-1:2021, Molecular biomarker analysis Methods of analysis for the detection and identification of animal species in food and feed products (nucleotide sequencing-based methods) Part 1: General requirements
- ISO 35001, Biorisk management for laboratories and other related organisations
- ISO/IEC Guide 99:2007, International vocabulary of metrology Basic and general concepts and associated terms (VIM)
- [1] Compendium of Chemical Terminology. Gold Book. International Union of Pure and Applied Chemistry. Version 2.3.3., 2014
- [2] Horwitz W. (1990). Nomenclature for sampling in analytical chemistry (Recommendations 1990). International Union of Pure and Applied Chemistry. 1990, 62, 1193 p. 1206
- [3] Calvert J.G. (1990). Glossary of atmospheric chemistry terms (Recommendations 1990). International Union of Pure and Applied Chemistry. 1990, 62, 2167 p. 2173
- [4] Khoury M.J. Genetics and Genomics in practice: The continuum from genetic disease to genetic information in health and disease. Genet. Med. 2003 Jul-Aug, 5 (4) pp. 261–268. DOI:10.1097/01.GIM.0000076977.90682.A5
- [5] Global Harmonization Task Force: Clinical Evidence for IVD medical devices Key Definitions and Concepts, Authoring Group: Study Group 5 of the Global Harmonization Task Force, Date: November 2nd, 2012

- [6] Wu Ct., Morris J.R. Genes, genetics, and epigenetics: A correspondence. Science. 2001 Aug 10, 293 (5532) pp. 1103–1105. DOI:10.1126/science.293.5532.1103
- [7] Théry C. et al. Minimal information for studies of extracellular vesicles 2018 (MISEV2018): a position statement of the International Society for Extracellular Vesicles and update of the MISEV2014 guidelines. J. Extracell. Vesicles. 2018, 7 (1) p. 1535750
- [8] Jennings L.J. et al. Guidelines for Validation of Next-Generation Sequencing-Based Oncology Panels: A Joint Consensus Recommendation of the Association for Molecular Pathology and College of American Pathologists. J. Mol. Diagn. 2017 May, 19 (3) pp. 341–365 Epub 2017 Mar 21. DOI:10.1016/j.jmoldx.2017.01.011
- [9] Statistical guidance on reporting results from studies evaluating diagnostic tests (2007) Guidance for Industry and FDA staff
- [10] Considerations for Design, Development, and Analytical Validation of Next Generation Sequencing (NGS) Based In Vitro Diagnostics (IVDs) Intended to Aid in the Diagnosis of Suspected Germline Diseases Guidance for Stakeholders and Food and Drug Administration Staff. 2018
- [11] MedTech Europe, 2020, Clinical evidence requirements for CE certification under the in-vitro diagnostic regulation in the European Union
- [12] WHO document https://www.who.int/observatories/global-observatory-on-health-research-and-development/analyses-and-syntheses/target-product-profile/who-target-product-profiles
- [13] Schlattmann P. Statistics in diagnostic medicine. Clin. Chem. Lab. Med. 2022 Mar 31, 60 (6) pp. 801–807. DOI:10.1515/cclm-2022-0225
- [14] Witwer K.W. et al. Standardization of sample collection, isolation and analysis methods in extracellular vesicle research. J Extracell Vesicles Co-Action Publishing. 2013, 2 p. 20360
- [15] Mateescu B. et al. Obstacles and opportunities in the functional analysis of extracellular vesicle RNA An ISEV position paper. J Extracell Vesicles Taylor and Francis Ltd. 2017, 2017 p. 6
- [16] Christina U. et al. Noninvasive diagnosis of urothelial cancer in urine using DNA hypermethylation signatures—Gender matters. Int. J. Cancer. 2019, 145 pp. 2861–2872
- [17] Wang Y., Navin N.E. Advances and Applications of Single-Cell Sequencing Technologies. Mol. Cell. 2015, 58 pp. 598–609
- [18] Sho S. et al (2017), Precision oncology using a limited number of cells: optimization of whole genome amplification products for sequencing applications. BMC Cancer volume 17, Article number: p. 457 (2017)
- [19] https://clinicalgenome.org/affiliation/gcep/#ep\_table\_heading
- [20] https://www.pharmgkb.org/
- [21] Whirl-Carrillo M. et al. An Evidence-Based Framework for Evaluating Pharmacogenomics Knowledge for Personalized Medicine. Clin. Pharmacol. Ther. 2021 Sep, 110 (3) pp. 563–572 Epub 2021 Jul 22. DOI:10.1002/cpt.2350

- [22] Matthijs G. et al. Guidelines for diagnostic next-generation sequencing. Eur. J. Hum. Genet. 2016 Jan, 24 (1) pp. 2–5 Epub 2015 Oct 28. DOI:10.1038/ejhg.2015.226
- [23] Pengelly R.J. et al. (2013), A SNP profiling panel for sample tracking in whole-exome sequencing studies. Genome Medicine. 2013 Sep 27; 5(9): 89. doi: 10.1186/gm492. eCollection 2013
- [24] Pengelly R.J. et al. Erratum to: a SNP profiling panel for sample tracking in whole-exome sequencing studies. Genome Med. 2015 May 7, 7 (1) p. 44. DOI:10.1186/s13073-015-0163-1
- [25] Souche E. et al. Recommendations for whole genome sequencing in diagnostics for rare diseases. Eur. J. Hum. Genet. 2022 Sep, 30 (9) pp. 1017–1021 Epub 2022 May 16. DOI:10.1038/s41431-022-01113-x
- [26] Deun J Van et al. EV-TRACK: Transparent reporting and centralizing knowledge in extracellular vesicle research. Nat. Methods. 2017 Feb 28, 14 (3) pp. 228–232. DOI:10.1038/nmeth.4185
- [27] Amorim M.G. et al. A total transcriptome profiling method for plasma-derived extracellular vesicles: Applications for liquid biopsies. Sci. Rep. 2017 Oct 31, 7 (1) p. 14395. DOI:10.1038/s41598-017-14264-5
- [28] http://varnomen.hgvs.org
- [29] ACMG guidelines: https://www.acmg.net/docs/standards\_guidelines\_for\_the\_interpretation\_of\_sequence\_variants .pdf
- [30] Navin N.E. Cancer genomics: one cell at a time. Genome Biol. 2014, 2014 (15) p. 452
- [31] Zeb Q. et al. An Overview of Single-Cell Isolation Techniques. Single-Cell Omics. Chapter 6. Academic Press, 2019, pp. 101–35., https://doi.org/10.1016/B978-0-12-814919-5.00006-3
- [32] Shackleton M. et al. Heterogeneity in cancer: cancer stem cells versus clonal evolution. Cell. 2009 Sep 4, 138 (5) pp. 822–829. DOI:10.1016/j.cell.2009.08.017
- [33] Cristofanilli M. et al. (2004). Circulating tumor cells, disease progression, and survival in metastatic breast cancer. The New England Journal of Medicine. 2004, 2004 Aug 19;351(8):781-91. doi: 10.1056/NEJMoa040766
- [34] Reichard A. et al. Best Practices for Preparing a Single Cell Suspension from Solid Tissues for Flow Cytometry. Cytometry A. 2019 Feb, 95 (2) pp. 219–226 Epub 2018 Dec 6. DOI:10.1002/cyto.a.23690
- [35] Guillaumet-Adkins A. et al. Single-cell transcriptome conservation in cryopreserved cells and tissues. Genome Biol. 2017 Mar 1, 18 (1) p. 45. DOI:10.1186/s13059-017-1171-9
- [36] Lafzi A. (2018) Tutorial: guidelines for the experimental design of single-cell RNA sequencing studies, Nature protocol 2018 Dec;13(12):2742-2757. doi: 10.1038/s41596-018-0073-y
- [37] De Luca F. et al. Mutational analysis of single circulating tumor cells by next generation sequencing in metastatic breast cancer. Oncotarget. 2016 May 3, 7 (18) pp. 26107–26119. DOI:10.18632/oncotarget.8431
- [38] Luecken M.D., Theis F.J. Current best practices in single-cell RNA-seq analysis: a tutorial. Mol. Syst. Biol. 2019 Jun 19, 15 (6) p. e8746. DOI:10.15252/msb.20188746

## CEN/TS 17981-1:2023 (E)

- [39] Macaulay I.C. et al. Single-Cell Multiomics: Multiple Measurements from Single Cells. Trends Genet. 2017 Feb, 33 (2) pp. 155–168. DOI:10.1016/j.tig.2016.12.003
- [40] Navin N.E. et al. Tumour evolution inferred by single-cell sequencing. Nature. 2011, 472 (7341) pp. 90–94 https://doi.org/10.1038/nature09807
- [41] Wang J. et al. Genome-wide single-cell analysis of recombination activity and de novo mutation rates in human sperm. Cell. 2012 Jul 20, 150 (2) pp. 402–412. DOI:10.1016/j.cell.2012.06.030
- [42] Zong C. et al. Genome-wide detection of single-nucleotide and copy-number variations of a single human cell. Science. 2012 Dec 21, 338 (6114) pp. 1622–1626. DOI:10.1126/science.1229164
- [43] Chen C. et al. Single-cell whole-genome analyses by linear amplification via transposon insertion (LIANTI). Science. 2017 Apr 14, 356 (6334) pp. 189–194. DOI:10.1126/science.aak9787
- [44] Zahn H. et al. Scalable whole-genome single-cell library preparation without preamplification. Nat. Methods. 2017 Feb, 14 (2) pp. 167–173 Epub 2017 Jan 9. DOI:10.1038/nmeth.4140
- [45] Zhu Z. et al. (2018). Progress and challenges of sequencing and analyzing circulating tumor cells. Cell biology and toxicology vol.34,5 (2018): 405-415. doi:10.1007/s10565-017-9418-5