



MEDICINA DE PRECIZIE

Topics

1. Human Genome. Genomics
2. Epigenetics and epigenomics. Pharmacogenetic and pharmacogenomic.
3. Genetic variability (gene mutation, SNP, CNV, indels, rearrangements)
4. Molecular diagnostic techniques and NGS technologies (WGS, WES, RNA-seq)
5. HGVS nomenclature, guidelines, recommendations for description of sequence variants
6. Genetic testing: current approaches
7. Advanced Technologies in Personalised Medicine: Single-Cell RNA-seq, Transcriptomics, optical gene mapping (OGM)
8. Genetic Variant Analysis (NGS): Pathogenicity assessment according to ACMG/AMP guidelines

References

1. Peter Russell. iGenetics: Pearson New International Edition. A Molecular Approach, 3 ed
2. Mark Sanders, John Bowman Genetic Analysis: An Integrated Approach, Global Edition, Pearson Education Limited
3. HGVS nomenclature.
<https://hgvs-nomenclature.org/stable/recommendations/general/>
4. HGNC Guidelines. <https://www.genenames.org/about/guidelines/>
5. Richards S, Aziz N, Bale S, Bick D, Das S, Gastier-Foster J, Grody WW, Hegde M, Lyon E, Spector E, Voelkerding K, Rehm HL; ACMG Laboratory Quality Assurance Committee. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med. 2015 May;17(5):405-24. doi: 10.1038/gim.2015.30.
6. Ronald Cohn, Stephen Scherer and Ada Hamosh. Thompson and Thompson Genetics and Genomics in Medicine. Elsevier, 2024
7. Peter D Turnpenny Sian Ellard, Ruth Cleaver Emery's Elements of Medical Genetics and Genomics, Sixteenth Edition Ed Elsevier, 2021
8. Medical Genetics and Genomics, 4th Edition – Fully Revised and Updated. Mircea Covic, Ionel Sandovici, Eusebiu Vlad Gorduza, Dragoș Ștefănescu. Ed. Polirom, 2024
9. https://www.illumina.com/content/dam/illumina-marketing/documents/products/illumina_sequencing_introduction.pdf
10. <https://documents.thermofisher.com/TFS-Assets/CSD/Reference-Materials/next-generation-sequencing-ebook.pdf>
11. <https://www.cap.org/member-resources/articles/optical-genome-mapping-a-tool-with-significant-potential-from-discovery-to-diagnostics>





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UNIVERSITATEA DE MEDICINĂ,
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ȘCOALA DOCTORALĂ DE
MEDICINĂ ȘI FARMACIE



12. Wallace SE, Bean LJH. Educational Materials — Genetic Testing: Current Approaches. 2017 Mar 14 [Updated 2020 Jun 18]. In: Adam MP, Feldman J, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2025. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK279899/>

