

# Chronic disease markers analysis using high-throughput sequencing approaches

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**Motivation and Aim:** With the recent development of sequencing technology and the rapid reduction of sequencing costs, high-throughput sequencing is revolutionizing basic life science research and clinical research from various aspects. High-throughput sequencing produces millions of sequencing reads at a time, and the alignment or assembly of these reads allows determination of various mutations at the genomic level, accurate gene expression quantification at the transcriptomic level, and identification of histone or DNA modification at the epigenomic level. The resulting accumulation of multi-omics information has opened up a new era of finding effective disease markers and studying their roles in disease occurrence and development [1, 5]. We aimed collect papers on biomarkers in a thematic journal issue to highlight recent findings in this field based on sequencing.

**Methods and Algorithms:** We have organized special journal issue (Research Topic) “High-throughput sequencing-based investigation of chronic disease markers and mechanisms” at *Frontiers in genetics* journal focuses on the genomics studies on cancer and chronic diseases (<https://www.frontiersin.org/research-topics/21036/high-throughput-sequencing-based-investigation-of-chronic-disease-markers-and-mechanism>). Using high-throughput sequencing, various markers of chronic diseases (such as cancer, heart disease, diabetes, and arthritis) have been developed at all omics levels, which have been used for diagnosis and classification of diseases, prediction of treatment effects, and prevention of diseases [6]. This research topic aimed at developing new chronic disease markers at four levels (i.e., genome, epigenome, transcriptome, and translome) with the help of high-throughput sequencing, and delineating potential marker-related mechanisms for chronic disease development. More specifically, this research topic contains contributions including:

1. Identification of novel biomarkers and prediction signatures for chronic disease detection (especially in early-stage) or prognosis prediction using high-throughput sequencing;
2. Analysis the possible pathological causes of markers as well as the potential roles they play in disease initiation and development;
3. Applications of new high-throughput sequencing techniques facilitating the development of more effective biomarkers of chronic disease;

4. New algorithms or tools for *in silico* identification of effective chronic disease markers based on high-throughput sequencing data.

**Results:** In this Research Topic a total of 14 papers could be arranged by two main areas – the cancer studies, and the works on the other chronic diseases such as allergy. Lung cancer is one of the leading causes of cancer-associated death in the world. This Topic complements recent Research Topics “Bioinformatics of Genome Regulation” and “Association between Individuals’ Genomic Ancestry and Variation in Disease Susceptibility” in *Frontiers in Genetics* considered more genetic background rather than molecular mechanisms of the human diseases [3, 4]. Non-small cell lung cancer comprises about 85 % of all lung cancers challenging treatment problems. We collected at the Research Topic the group of papers on lung cancer [2]. Traditional Chinese medicine formula Feiyaning has been clinically administered in China for more than a decade and raised attention due to its anticancer effect in lung cancer. Wang et al. (2021) revealed the migration-associated genes involved in antitumor effects of herbal medicine Feiyaning on lung cancer cells using RNA-Seq and ATAC-Seq analysis. Recent works presented novel biomarkers in gastric cancer [8].

**Conclusion:** Overall, we have compiled several special journal issued on biomedical problems [3, 4]. We hope that the readers will find these papers collections a stimulating reading and consider participation in the next conferences and journal issues in this area ("Medical Genetics, Genomics and Bioinformatics–2022" – [https://www.mdpi.com/journal/ijms/special\\_issues/Medical\\_Genetics\\_2022](https://www.mdpi.com/journal/ijms/special_issues/Medical_Genetics_2022); [https://www.mdpi.com/journal/genes/special\\_issues/Transcriptional\\_Regulation\\_Tumor](https://www.mdpi.com/journal/genes/special_issues/Transcriptional_Regulation_Tumor)).

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## References

1. Anashkina A.A., Leberfarb E.Y., Orlov Y.L. Recent Trends in Cancer Genomics and Bioinformatics Tools Development. *Int J Mol Sci.* 2021;22:12146. doi: 10.3390/ijms222212146.
2. Chang Y., Wang Y., Li B. et al. Whole-Exome Sequencing on Circulating Tumor Cells Explores Platinum-Drug Resistance Mutations in Advanced Non-small Cell Lung Cancer. *Front Genet.* 2021;12:722078. doi: 10.3389/fgene.2021.722078.
3. Das R., Tatarinova T.V., Galieva E.R., Orlov Y.L. Editorial: Association between individuals' genomic ancestry and variation in disease susceptibility. *Front Genet.* 2022;13:831320. doi: 10.3389/fgene.2022.831320.
4. Orlov Y.L., Anashkina A.A., Klimontov V.V., Baranova A.V. Medical genetics, genomics and bioinformatics aid in understanding molecular mechanisms of human diseases. *Int J Mol Sci.* 2021;22(18):9962. doi: 10.3390/ijms22189962.
5. Orlov Y.L., Baranova A.V., Hofstaedt R., Kolchanov N.A. Computational genomics at BGRS\SB-2016: introductory note. *BMC genomics.* 2016;17(Suppl. 14):996. doi: 10.1186/s12864-016-3350-6.
6. Voronova V., Glybochko P., Svistunov A. et al. Diagnostic value of combinatorial markers in colorectal carcinoma. *Front Oncol.* 2020;10:832. doi: 10.3389/fonc.2020.00832.
7. Wang C., Li P., Peng Y. et al. Integrative RNA-Seq and ATAC-Seq analysis reveals the migration-associated genes involved in antitumor effects of herbal medicine feiyaning on lung cancer cells. *Front Genet.* 2021;12:799099. doi: 10.3389/fgene.2021.799099.
8. Wang Y., Fang Y., Zhao F. et al. Identification of GGT5 as a novel prognostic biomarker for gastric cancer and its correlation with immune cell infiltration. *Front Genet.* 2022;13:810292. doi: 10.3389/fgene.2022.810292.