УДК 575.1:616+004

THE FUTURE OF GENETIC DIAGNOSTISC TESTS

N. KARAGEN, MD PhD
Professor at Pamukkale University, medical Faculty, department of medical biology, Turkey

Pamukkale University, Dept. of Geophysical Engineering, Denizli, Turkey

Abstract. Molecular genetic tests become increasingly sophisticated. Until recently, genetic tests were carried out on one or a few loci. The advent of large-insert clone arrays and, later, next generation sequencing method changed this by allowing a patient's entire genome to be searched at improved resolution, thereby allowing the detection of medium to large genomic areas.

Detecting large genome areas and many mutations by genetic test allows us to detect numerous mutations related by particular disease or particular condition. Development of detection methods of genes have brought many new aspects in gene related health conditions and also the treatment of certain diseases in this new era of clinical diagnostics tests.

Detecting large genom areas produces huge data to be analyzed which also leads the development of genome screening-analizing softwares. Having this big data and developed softwares also open a demand for work force in Information Technologies to do bioinformatic analysis. Therefore, a new area has been developed combining clinical medicine and in research and medicine.

Data privacy will be another key component of the future of genetic tests. We must address concerns about the privacy of patient data to take full advantage of large-scale analysis of aggregated patient datasets, and to allow the healthcare industry to fully realize the economic benefits of cloud computing. Protecting personel data also requires also legal legislations.

Whole genom analysis or detecting certain mutations also leads the pharmacetical companies to discover new mutation specific drugs and alsopatient or individual lead gene terapy.

This work is supprted by the Project PauBap, Project number: 2018KRM002-550

Key words: diagnostisc tests, molecular genetic, information technologies.

References

[1]. Mattick JS, Dinger M, Schonrock N, Cowley M. Whole genome sequencing provides better diagnostic yield and future value than whole exome sequencing. Med J Aust. 2018 Apr 9

[2]. Hayashi M, Watanabe A, Muramatsu M, Yamashita N. Effectiveness of personal genomic testing for disease-prevention behavior when combined with careful consultation with a physician: a preliminary study. BMC Res Notes. 2018 Apr 3;11(1):223.

[3]. Goodenberger ML, Thomas BC, Kruisselbrink T. Response to Commercial Genetic Testing and the Future of the Genetic Counseling Profession. J Genet Couns. 2018 Mar 23.

[4]. Middleton A. Society and personal genome data. Hum Mol Genet. 2018 Mar 7.