https://doi.org/10.53040/cga11.2021.011 UDC:575.1+612.6.05

INFORMATION TOOL FOR NEW GENERATION SEQUENCE DATA INTERPRETATION

Viorel MUNTEANU, Rodica MARTEA, Maria DUCA
Moldova State University, Center of Functional Genetics, Republic of Moldova

E-mail: *mduca2000@yahoo.com*

The completion of the Human Genome project, which described the sequence of nucleotides in the DNA molecule and identified about 3 billion pairs of nucleotides for a total of about 23,000 to 25,000 genes, personalized medicine became a major challenge for biomedicine. Every year, researchers identify new genes associated with certain pathologies, then create and test new drugs that target the activity of these genes. Moreover, approximately 150-200 genetic tests are used in clinical practice and tests have been developed that include gene panels for many of the most common polygenic diseases.

An important role in the development of genomics-based therapies and testing of predisposition to certain diseases lies in molecular analysis techniques, including the development and implementation of state-of-the-art sequencing (SNG) technologies, which have changed the perspectives of analysis and understanding of living beings generally, including allows the assessment of genome variability in populations.

The challenge in reporting SNG results appears at the intersection between the necessary and useful information for personalized medicine, regarding the appropriate level of detail, so that the use and limitations of the analysis are clearly understood by the user. The time required to evaluate an SNG test can vary between hours and weeks. The final stage of the analysis is represented by the elaboration of the resulting clinical report. The clinical report of SNG results must contain sufficient information to communicate the results of the test and its limitations.

Based on these desiderata, the purpose of the research was to develop information tool for new generation sequence data interpretation, which would allow stakeholders, especially doctors, to understand and interpret properly, and then make clinical decisions in accordance with the results. The classification of variants forms the basis of clinical decisions, where the quality of the classification of variants is critical for the well-being and treatment results. Without careful interpretation and evaluation of the evidence, the results of the sequencing are data without biomedical interest.

The analysis of these data was performed by using bioinformatics and biostatistics software (R, Cytoscape) and multiple databases (COSMIC, TCGA), having as reference criteria the set of standards and instructions for interpreting sequence variants, developed by the American College of Genetics and Medical Genomics.

The study was carried out with the support of the State Program "Precision medicine in the prevention, diagnosis and treatment of pathologies" with the number 18.80.07.16A/PS, 2018-2019.