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EDITORIAL COMMENTS – OPEN ACCESS

A Potential Method to Help Predict Genetic Diseases and Arrange Healthcare: Copy Number Variation Analysis

Munis Dundar, Saide Betül Arslan

Genetic testing is continually gaining more importance in medical practice. Development of new genetic tests and increasing the availability to these tests enable the conduction of more genetic studies in scientific communities. Increasing information about the genetic effect on disease etiology, pathology, and treatment response helps to achieve better public health planning.

Gene sequencing is currently the gold standard to detect mutations when a specific predefined target is established. However, single-gene point mutations cannot be detected in many cases, not even using more extensive tests such as clinical exome sequencing or whole-exome sequencing. In such cases, copy number variation (CNV), rather than point mutations, may be the cause of diseases.

CNV is the variation in DNA segments that should normally be two copies due to gain or loss. CNV can be detected by microarray or multiplex ligation-dependent probe amplification. It is not yet known which CNV is pathogenic or is only polymorphism. However, a comparison of genetic data with clinical histories will help in understanding the disease-causing potential of CNV and its association with diseases.

Neurodevelopmental diseases and intellectual disability are thought to be diseases with the highest CNV association. In a retrospective study, Ceylan et al. detected 43 types of CNV in 300 people with an intellectual disability or global developmental delay (1). Crawford et al. found that common diseases such as diabetes, hypertension, obesity, and renal failure may be associated with CNV. The association of CNV with intellectual disability or global neurodevelopmental delay was not investigated because only middle-advanced-aged patients were included (2).

Prevention and follow-up of common diseases, such as hypertension, obesity, and diabetes, are significant in terms of public health planning to reduce mortality. Ozturk et al. found that the prevalence of hypertension in individuals aged >30 years is 34.6% in Kayseri (3). Sahin et al. estimated the prevalence of obese and overweight students to be 28.6% (4). Genetic risks of these individuals who are already at a high risk related to environmental factors can also be determined by analyzing CNV. Consequently, their lifestyles could be regulated, and they may be referred to personal treatments. The concept of personalized medicine includes genomic risk assessment along with other clinical trials, and personalized medicine and personal genomic language are commonly used (5).

In a study by Crawford et al., 54 CNV regions for approximately 380,000 participants were scanned. The participants' health records and relationships were investigated. Their results show that the diseases with the highest relation with CNV included hypertension, diabetes, neuropathies, renal failure, and obesity. Moreover, 330 genotypic phenotype correlations were found. One aim of their study, as an additional gain, was to allow the determination of the pathophysiology of diseases during follow-up after identifying the possible diseases (2).

Craddock et al. concluded that CNV is not associated with common diseases (6). However, their study included a small group and few diseases. On the other hand, the study by Crawford et al. is more comprehensive, extensive, and up-to-date. Therefore, more studies including more groups and diseases are required to increase conspicuity and reliability.

Craddock et al. also demonstrate the benefits of establishing a national database. Their study highlights the importance of creating a database on common CNV and polymorphism or related diseases in our society. Accordingly, secondary health protection can be regulated by modifying lifestyle according to the risk of a disease encountered in one's future life. An important outcome of the Human Genome Project was that electronically compiled genomic data could be used to identify the desired genes in certain genome parts.

Genomics (the study of genes, their functions, and related techniques) has become a crucial science for developing

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Department of Medical Genetics, Erciyes University Faculty of Medicine, Kayseri, Turkey

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Correspondence
Munis Dundar,
Department of Medical Genetics, Erciyes University Faculty of Medicine, Kayseri, Turkey
Phone: +90 352 437 06 00
e-mail: dundar@erciyes.edu.tr

an understanding of life processes and their evolution. Since the advent of the Human Genome Project, significant advances have been made in developing an understanding of DNA and RNA sequence information and how it can be implemented in biotechnology. Newly derived sequencing and bioinformatics tools have added to the torrent of new insights gained, because of which DNA apps of the "sequence once and query often" type are becoming a reality (7).

CNV analysis provides important contributions to the evaluation, pathogenesis, etiology, and treatment of common diseases. It is important to implement the necessary steps to design the existing tests, create a database in this direction, and share results with clinicians.

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