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Opinion Article

Application of Genomics Approach in Development of Diagnostic Medicine

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DESCRIPTION

Gene interactions include all physiological processes in both health and disease. Diagnostic medicine, which helps assess human health, will greatly benefit from genomic research. Genomic databases and bioinformatics have emerged as valuable tools for discovering genotypic and phenotypic diagnostic markers. Genomic medicine is an emerging medical field involving the use of genomic information about individuals as part of clinical care (such as diagnosis and treatment decisions) and the health and policy implications of this clinical use. These tools have already led to the discovery of genes associated with colon cancer, atherosclerosis, osteoporosis, and many other diseases. Gene sequence information provides insight into the identity and function of unknown genes, and comparisons of gene expression patterns between normal and diseased tissues which help in defining the role of specific genes in the disease. These techniques enable the simultaneous evaluation of multiple diagnostic leads, achieving economies of scale and enabling the analysis of complex gene interactions. There are several challenges in diagnostic genomics, such as the discovery and analysis of low-expressed genes and the development of versatile genetic testing tools. Genomic medicine is already impacting oncology, pharmacology, rare diseases, undiagnosed diseases, and infectious diseases. Early diagnosis of disease greatly increases the chances of successful treatment, and genomics can detect disease long before symptoms appear. The Diagnostic Genomics program is designed to provide scientists and clinicians with genomics knowledge and skills and to overcome existing barriers to the integration of genomics into healthcare. Unprecedented advances in genomics and related technologies are creating tremendous potential for change in both medicine and the life sciences. Innovative genomics applications include:

- Incorporation of Next-Generation Sequencing (NGS) into clinical diagnostics (applying both whole-genome and exome sequencing).
- Single-cell sequencing studies
- Quantification of gene expression changes (whole-transcript sequencing) (including Tome Sequencing)
- Pharmacogenomics
- Blood-based cell-free DNA testing.

Technological breakthroughs in genomics are driving the healthcare landscape towards personalized healthcare with the potential to deliver better health outcomes. Healthcare is being transformed by clinical genomics day by day. Genomic techniques are mainly used in medical genetics and cancer to improve diagnosis and patient care. Clinical genomics uses sequencing techniques to support patient diagnosis and treatment. Medical professionals use diagnosis in approximately 70% of their decision-making to select the appropriate treatment for their patients. Recent technological advances have led to the proliferation of molecular genetics and genomic testing as part of clinical diagnostic services. The association of genotype and phenotype has been a central part of biology for a century. Advances in nucleic acid sequencing technology have provided translational medicine with extraordinary momentum and a fundamental understanding of chronic, metabolic, and genetic diseases. Genomic technology and molecular diagnostics are enabling the vision of precision medicine and driving the advancement of healthcare. The impact of these technologies will have a positive impact on healthcare, from prevention to diagnosis to treatment. There is a need to integrate these technologies into clinics and hospitals to guide medical decisions towards the most effective disease prevention and personalized targeted therapies based on genetic makeup. Fast, low-cost, large-scale DNA sequencing has propelled genomics into mainstream medicine, facilitating a revolutionary shift to precision medicine. Health care providers can use genomic data to identify patients at increased risk of developing certain diseases and to plan better treatment. This approach may be particularly useful for proactively treating cognitive or behavioral disorders before people show signs of illness. The simultaneous development of genomic technologies such as DNA arrays for rapid analysis of multiple genes will improve the discovery and evaluation of potential diagnostic medicine.

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