

Impact of Genomics on the Future of Healthcare

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Abstract. Genomics has become an increasingly mature discipline, but there are still many problems to be solved. One of them is how to use genomics to personalize precision therapy for the population. This paper introduces the potential application of genomics in the prediction, prevention, diagnosis, and treatment of diseases. Discusses the frontier problems of genomics research, difficulties in the development of genomics, and puts forward new thinking for the application of genomics in public health.

Keywords: Genomics, NHGRI, genome sequencing.

1. Introduction

In 1986, Thomas H. Roderick proposed the concept of genomics. Genomics mainly studies the structure, function, evolution, localization and editing of genes, as well as their effects on organisms. Since its development, genomics has been divided into sub-disciplines, including functional genomics, structural genomics, epigenomics and metagenomics. With the development of new science and technology, many new technologies have been applied in the research of genomics. Such as artificial intelligence, third-generation sequencing, three-dimensional genomics and so on.

Genomics and its related technologies are also increasingly being used in medical fields. In the early 21st century, scientists from the United States, Britain, France, Germany, Japan and China completed the human Genome Project. (Wei Dong, 2021) The completion of this project gradually brought the life science in the field of genes into people's vision. People are starting to think about how genomic technology can be applied to more fields, such as agriculture, botany, zoology, and of course, medicine and public health.

Genomic technology has been used in the field of public health. Genomics and related technologies have been used to predict, prevent, diagnose and treat diseases. These four application directions are also popular in the frontier of genomics research.

Prenatal genetic examination and neonatal genetic examination are the most extensive applications of genomic technology in disease prevention. Down syndrome is a focus area of prenatal genetic testing. Glenn E. Palomaki, PhD et al. screened the fetus for Down syndrome by detecting DNA in maternal plasma, with a detection rate of 98.6%, a false positive rate of 0.20%, and a detection failure rate of 0.8% (Palomaki et al., 2011). (Palomaki et al., 2011) The NBS program, launched in the last century, tested newborns for diseases such as phenylketonuria, hemoglobinopathy and biotinase deficiency, using a variety of testing methods. Genetic testing in the NBS program is less accurate but more expensive as testing programs increase. As a result, NGS technology has been gradually applied in genetic testing, including whole exome sequencing (WES) and whole genome sequencing (WGS) (Alfares et al., 2018). (Alfares et al., 2018) If NGS technology is gradually applied to newborn genetic examination in the future, it will provide more accurate prediction of disease, and reference for disease prevention and personalized treatment.

In addition, genomics can also be applied to the risk assessment of complex diseases, that is, to carry out more precise examination of people who have participated in genetic testing and have abnormal results (Khoury, Janssens, & Ransohoff, 2013). (Khoury, Janssens, & Ransohoff, 2013) Through further examination, the incidence rate of the subjects can be estimated, combined with the surrounding environmental factors of the subjects, the specific disease development process can be simulated, and prevention can be realized on the basis of prediction. (Chatterjee, Shi, & García-Closas, 2016).

There are a growing number of commercial agencies that perform genetic testing by collecting saliva from a subject and then performing polymerase chain reaction (PCR) on the DNA contained in the oral epithelial cells. After a period of time, the subjects received a report. The report will present the test results, along with an analysis of the results, and inform the subjects of their risk of disease. Such programs are still seen as a commercial endeavor, but perhaps in the future, genetic testing programs could become government-led health checks for everyone. With the guidance and support of the government, genetic testing data can be collected to detect and avoid increased risk factors in susceptible populations, and to detect and control familial epidemics, such as atrial fibrillation (Kany et al., 2021). (Kany et al., 2021) This will also help health authorities formulate policies, coordinate medical resources, provide medical security, reduce the incidence of high-risk groups and prevent diseases.

In the aspect of infectious disease prevention, changing the pathogen genotypes of common infectious diseases can reduce the risk of infection and incidence. For example, mycoplasma genitalium is the main pathogen causing urinary tract infections (Peric, Weiss, Vulliemoz, Baud, & Stojanov, 2019). It is the smallest organism that can live independently discovered so far. There are about 482 genes (Colman, Hu, Litaker, & Bott, 1990). (Peric, Weiss, Vulliemoz, Baud, & Stojanov, 2019) Fookes et al. 's work has assembled complete synthetic genomes by performing transformation-related recombinant clones in *S. cerevisiae*, which are then isolated and sequenced (Fookes et al., 2017). (Fookes et al., 2017) It may be possible in the future to edit infectious pathogens at the genetic level to induce them to mutate in a way that makes them less virulent and less infectious.

2. Diagnosis

AReduce the cost of genetic testing, expand the scale of testing, and use genetic testing results as one of the diagnostic means. At present, researchers have established automated platform by combining genome sequencing technology with electronic medical records to accelerate the diagnosis of severe diseases in newborn infants (Clark et al., 2019) In the diagnosis of some diseases with large genetic and phenotypic heterogeneity, genetic testing has become one of the important detection indicators, providing diagnostic basis for doctors. Jiang et al. conducted WES detection and KGGSeq informatics analysis on a sibling with severe muscular atrophy and distal skeletal abnormality, and found GDAP1 (induction-induced ganglioside differentiation related protein 1) mutation. (Jiang et al.)

The level of disease is determined by the degree of genetic variation in the cells. Gleason patten (GP) is a pathological grading system that evaluates the degree of malignancy of tumors according to the degree of differentiation of glands and the way of tumor growth in the stroma, and is a key indicator for predicting the prognosis of prostate cancer patients.(Mayer, Simone Ii, Turkbey, & Choyke, 2022) In 1974, Donald F. Gleason first proposed GP scoring system based on tumor tissue and cell morphology.(DF & GT, 1974) This scoring system has been modified many times, but its grading system still has limitations. Wang Tao et al. used TCGA to analyze gene expression differences between GP4 and GP3 prostate tissue cells (Wang Tao et al., 2018). (Wang et al., 2018) The study found that cyclin in GP4 prostate cancer tissue is upregulated, leading to more active cell division, which explains why GP4 cells are more malignant at the genetic level (Wang Tao et al., 2018). Perhaps, in the future cancer diagnosis and evaluation system, the degree of genetic variation of cancerous cells can be used as one of the indicators.

3. Treatment

In the course of disease treatment, it is very important to understand the pathogenesis. According to the patient's gene to determine the treatment plan, research and development of molecular level targeted drugs for gene, targeted treatment development, avoid side effects (Rossing, Sørensen, Ejlersen, & Nielsen, 2019) found that RNA interferes with oncogenes and tumor suppressor genes

to improve drug effectiveness (Fatemian & Chowdhury, 2014). (Fatemian & Chowdhury, 2014) Genomics can also be applied to basic medical research to explain the pathogenesis of complex diseases, such as sporadic adrenal tumors (Igaz et al., 2006).

4. Frontiers of genomics research

In 2020, the National Human Genome Research Institute (NHGRI) of the United States proposed a strategic vision to improve human health in the frontier of genomics. This is the third vision proposed after the completion of the human Genome sequencing project in 2003. It is mainly divided into four areas: 1. Guiding principles and values of human genomics 2. Sustaining and improving solid research in genomics 3. Breaking down barriers that impede progress in genomics 4. Compelling Genomics Medicine Project (Green et al. 2020).

Today, genomics is increasingly complex. In the process of developing and applying genomics, researchers must be more careful to implement the guiding principles and values of human genomics, and to minimize the impact of genomics on the current sound and mature legal system, society in the process of scientific research ethical impact. (McEwen et al., 2014)

Previously, the basic research of genomics was mainly divided into the exploration of the structure and function of the genome (Lander et al., 2001), the innovation of genome statistics and computational methods (Schatz, 2015), the interaction between genomics and social and environmental factors (Morris, Davies, Hemani, & Smith, 2020), and the cultivation of the genomics workforce (Stephens et al., 2015). In the future, the development of genomics needs to consolidate basic research, aim at breakthroughs and innovations, and combine other disciplines, such as the integration of information science, statistics, and artificial intelligence science when developing genomic data science.

Despite the rapid development of genomics, the various parts of genomics are still facing uneven development, and their productivity and interconnection are not fully developed. The small scale of DNA synthesis and editing, the limitations of genomic data and experimental technology (Z et al., 2019), the lack of a clear understanding of genomic variation and its impact on human health from a biological point of view (Rehm et al., 2015), the lack of clinical application of genomic technology is still a matter of genomics developmental disorder (Khera et al., 2018). Therefore, focusing on breaking down barriers is also an important part of the vision. (Green et al., 2020)

At the same time, during the development of genomics, there have been many novel topics in the field of biomedicine. For example, using experimental data and computational models, we can more fully understand the relationship and role between genes and regulatory elements, and predict the phenotype of cells and organisms. (Green et al., 2020).

5. Obstacles to the application of genomics

Supporting technology is not perfect, massive data analysis is difficult. Rapid advances in biotechnology over the past two decades have made sequencing easier and cheaper. But the combination of biotechnology and computing remains an obstacle. Mendel statistics and analysis of pea characters is the application of computational technology and statistical knowledge in the field of biology. However, in addition to genetic factors, there are also environmental factors affecting biological traits. How to distinguish environmental factors from genetic factors affecting biological traits, how to correctly analyze data and avoid false correlation are still obstacles to progress. In addition, as sequencing technology evolves, the storage of measured genetic data also needs to be considered.

Genomic data is difficult to monitor and may be used for other studies. The use of medical records is a very common situation in the medical process, doctors can more accurately diagnose the patient's condition through the patient's basic information, past medical history, and allergy history. At present, medical and health organizations and health departments in many countries have established

electronic health records by uploading patients' personal health information to databases Wu et al. (2017) However, it is difficult to guarantee the security of genomic data. Ayozy et al.'s study shows that the widely used genomic data sharing beacon protocol still has vulnerabilities, and attackers can use the correlation in genomic and clustering technologies to reconstruct the genomes of victims (Ayozy, Ayday, & Cicek, 2021).

At present, genome sequencing technology is not perfect, is a factor of social instability, and may cause potential danger. If the security of the genetic information of the subjects is not guaranteed, it may affect their schooling, work, insurance, and other aspects. Genome sequencing technology is not yet popularized. If gene sequencing technology is difficult to develop in developing countries, it will be difficult to ensure the diversity of genetic samples. New medical methods designed according to populations in developed areas may not be universal due to race, gender, etc. This may lead to a significant backwardness in the level of medical care in underdeveloped areas, and at the same time exacerbate racial, ethnic and gender discrimination.

6. Epilogue

In the decades of rapid development of genomics, genomics has provided a new platform for human cognition of biology. The combination of biotechnology and multifaceted scientific fields has moved public health from groups to precision. At a time when the new coronavirus is still raging around the world and has an impact on human life, the development of genomics has given us confidence in fighting the epidemic. Although no one has been able to accurately predict when the epidemic will end, at least the struggle between humans and the virus has taught our existing medical system and medical methods some experience and lessons, giving researchers and medical staff new insights into public health. Thinking has also brought new challenges and opportunities for the development of genomics.

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