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Genomic Research Unlocking New Avenues for Chronic Disease Treatment

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ABSTRACT

As the world's population ages, mortality increasingly reflects the ravages of complex chronic diseases, particularly cancer and heart disease. A person's risk of succumbing to a chronic disease is linked to his or her genetics. This genome-centric view of causation is motivated by the technologic ability to detect and manipulate genes, and fosters the notion that genetic factors are necessary determinants of disease that operate in a causal background of diverse exposures. Chronic venous disease is highly prevalent in the Western side. The recent developments in sequencing technology, clinicians and geneticists alike are embarking on a journey to identify and unravel the genetic candidates of chronic venous disease. Currently substantial evidence to suggest the presence of genetic influences in the aetiology and pathology of venous disease. The researchers should be urged to foster collaborative links and design a genome-wide case-control association study as an international consortium to provide a statistically robust paradigm in the field of chronic venous disease genetics. Family history is a well-known risk factor for developing many common chronic diseases, such as diabetes, asthma, and coronary heart disease, and reflects inherited genetic and shared environmental contribution in disease. The future directions include incorporating genotype information to partition DNA-transmitted genetic versus environmental variance in phenotype in family history to decompose the various components of risk influenced by familial disease. An accurate diagnosis is also crucial for genetic counseling and family planning and allows reproductive options, such as prenatal or pre-implantation genetic testing. It allows screening of at-risk family members, which may also be important in determining their eligibility as kidney transplant donors. At present, phenotype-associated multigene panels and ES are the preferred diagnostic MPS-based testing modalities, but it is expected that when GS becomes more feasible, both in terms of cost-effectiveness and complex data interpretation, GS-based diagnostic testing will replace most current testing modalities.

Keywords: Complex chronic diseases, Genetics, Genetic testing, Genotype information, Familial disease.

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1. Introduction

The Human Genome Project began in 1990 with the goal to map the entire human genome. Although genomic technology is used for various purposes in healthcare, such as pharmacogenomics, this paper will focus on genomic

screening of some specific sub forms of chronic diseases due to the prevalence and cost associated with these types of diseases. Traditional approaches of medicine and surgery have helped to treat many patients; however, there is a wide

scope of indications for which conventional treatment is either inefficient or burdensome to the patient. Such indications, often associated with an underlying genetic cause, require multiple rounds of intervention over an extended period of time, making them chronic by nature. Chronic disease prevention in primary care aims to identify individuals at increased risk who can then be given tailored disease prevention advice including lifestyle modification, disease screening, or preventive treatment. Current approaches to chronic disease risk assessment use a combination of factors including demographics, family history, lifestyle, physiological measurements, and biomarkers. The family medical history currently makes an important contribution to risk assessment for many diseases as the risk of a number of cancers, type 2 diabetes, and ischemic heart disease are all increased in the presence of a family history; this risk increases with more affected relatives. The genomics could then support tailored cancer screening, the integration of genomics data with family history, sociodemographic, behavioral, and environmental risk factors into risk assessment tools, and even pharmacogenetically-informed prescribing such as for nicotine replacement treatment. However, there are still a number of issues that remain important to address before the incorporation of any of these approaches into routine general practice¹⁻⁴.

As genomics-based approaches begin to be implemented both within health services and, increasingly, through direct-to-consumer testing, there is still much research to be undertaken. In a recent issue *Middle mass* and colleagues reported a qualitative study investigating the introduction of genetic testing for cardiovascular disease in primary care. They emphasized that it will be important to build on the patient's motivation to undertake genetic testing both for themselves and their families; to understand how patients deal with conflicting findings between genetic and conventional cardiovascular risk assessment; to learn how to promote responses that reinforce healthy behavior change and reduce false reassurance; and to look at the potential benefit of genetic testing for younger people who may have more to gain from risk reducing behavior.

Chronic illnesses are mostly characterized by

- Complex causes
- Many risk factors
- Long latency periods
- A long illness
- Functional impairment or disability
- Common Chronic Diseases

List of chronic disease conditions

- Cardiovascular Disease
- Stroke
- Lung Cancer
- Colorectal Cancer
- Depression
- Type 2 Diabetes
- Arthritis
- Osteoporosis
- Asthma
- Obesity

- Chronic Obstructive Pulmonary Disease (COPD)
- Chronic Kidney Disease
- Oral Disease

Chronic Diseases in Genomic Research

- Cardiovascular Diseases: Including coronary artery disease, hypertension, and stroke.
- Diabetes: Affecting blood sugar regulation and a major area of genetic study.
- Cancer: Research investigates genetic markers and mutations linked to various cancer types.
- Neurological Disorders: Such as Alzheimer's and Parkinson's disease.
- Autoimmune Conditions: Including rheumatoid arthritis, lupus, and Crohn's disease.
- Respiratory Diseases: Like asthma and chronic obstructive pulmonary disease.
- Chronic Kidney Disease: With significant research focusing on genetic risk factors, particularly in populations of African ancestry.

Working pattern of Gene Analysis⁵⁻⁹

Identifying Genetic Variants

Advances in high-throughput sequencing technologies allow for the creation of large genetic datasets that can be analyzed using bioinformatics tools.

Next-Generation Sequencing

This technology helps identify specific genetic changes, such as mutations and single nucleotide polymorphisms, linked to a disease.

Genome-Wide Association Studies

GWAS are used to identify genetic markers that are more common in people with certain chronic diseases.

Bioinformatics

Specialized software and algorithms process and analyze large genomic datasets to find patterns and insights into disease development and treatment responses.

Applications in Chronic Disease Treatment

Personalized Medicine

By understanding a patient's unique genetic makeup, treatments can be tailored to their specific genetic blueprint, improving outcomes and reducing side effects.

Drug Discovery

Genomics helps identify new therapeutic targets by highlighting genes and pathways involved in disease pathogenesis, leading to the development of innovative treatments.

Risk Assessment and Prevention

Genetic analysis can identify individuals at higher risk of developing certain chronic diseases, allowing for early diagnosis, preventative measures, and proactive management.

Monitoring Treatment Efficacy

Biomarkers derived from genetic analysis can be used to monitor disease progression and the effectiveness of treatments, allowing for adjustments to the treatment plan.

Genetic Screening

Genetic screening utilizes molecular biology techniques to detect specific genetic variants in an individual's DNA that may be associated with genetic diseases or disease risk. Specifically, genetic screening typically includes the following steps: First, DNA is extracted from the

individual's blood, saliva, or other tissue samples. Next, various molecular biology techniques, such as polymerase chain reaction and DNA sequencing, are used to analyze the target gene regions and detect specific genetic variants. Finally, professionals assess the individual's health risks or diagnosis based on the test results and other clinical information. Genetic screening has a wide range of applications and purposes, including newborn screening, prenatal screening, adult disease risk assessment, pharmacogenetic testing. These screenings help with early detection of genetic diseases, prediction of disease risks, guidance for treatment plan formulation, and assistance with reproductive decisions.

Principles of Genetic Screening

The principles of genetic screening are based on the diversity of the human genome. Our genome contains many variants, including single nucleotide polymorphisms, copy number variations, insertions and deletions, etc. Certain variants may affect gene function and thus be associated with specific diseases or disease risks. Genetic screening assesses an individual's risk of developing certain genetic diseases or makes a diagnosis by detecting these key variants. The specific principles of genetic screening involve a variety of molecular biology techniques. Currently, the most commonly used methods are polymerase chain reaction and DNA sequencing. It is a rapid and sensitive DNA amplification technique that selectively amplifies the target gene region by using specific primers and detects variants within it. DNA sequencing is a more comprehensive and high-throughput method that determines the exact sequence of DNA fragments and identifies various variants within them.

Mechanisms of Genetic Diseases

Genetic diseases are primarily caused by abnormalities in genes or chromosomes. Chromosomal abnormalities, such as the absence of some or all chromosomes, are referred to as aneuploidy. Changes or mutations in genes are the root cause of many hereditary diseases.

Technical Methods of Genetic Screening

Polymerase Chain Reaction

Principle: Exponential amplification of target DNA fragments using specific primers and DNA polymerase.

Application: Commonly used for detecting specific gene mutations, such as CFTR gene mutations in cystic fibrosis.

DNA Sequencing

a. Sanger Sequencing

Application: Suitable for mutation detection and verification of known genes.

Next-Generation Sequencing

Application: Whole exome sequencing, whole genome sequencing, targeted gene panel sequencing, etc.

Microarray Technology

Application: Single Nucleotide Polymorphism (SNP) detection, Copy Number Variation (CNV) analysis.

Digital PCR

Application: Rare mutation detection, copy number variation analysis.

Long-Read Sequencing

Application: Structural variation detection, sequencing of complex regions.

2. Types of Genetic Screening

Prenatal Genetic Screening: Prenatal genetic screening is an examination performed before the birth of a fetus to identify whether the fetus has a risk of certain genetic diseases or chromosomal abnormalities. This screening is usually performed at specific times during pregnancy and can help prospective parents understand the health status of the fetus and provide important information for subsequent medical decisions¹⁴.

Currently, common prenatal genetic screening methods include maternal blood Down syndrome screening, non-invasive prenatal genetic testing, and invasive prenatal diagnosis. Maternal blood Down syndrome screening assesses the risk of the fetus having Down syndrome by measuring specific markers in maternal blood; non-invasive prenatal genetic testing screens for common chromosomal abnormalities in the fetus by analyzing free fetal DNA fragments in maternal blood; invasive prenatal diagnosis requires obtaining fetal tissue samples and can more accurately diagnose genetic diseases and chromosomal abnormalities.

Newborn Genetic Screening

Newborn genetic screening is an important public health measure aimed at early detection and treatment of certain genetic diseases that may endanger infant health. This screening is usually performed shortly after the infant's birth by testing the infant's blood or other tissue samples to screen for a range of genetic diseases.

Adult Disease Risk Genetic Screening

Adult disease risk genetic screening aims to identify an individual's genetic risk of developing certain common diseases (such as cardiovascular disease, type 2 diabetes, etc.). By analyzing specific genetic variants, these screenings can assess an individual's disease susceptibility and help them take preventive measures.

Cancer Genetic Screening

Cancer genetic screening mainly targets certain hereditary tumor syndromes, such as hereditary breast and ovarian cancer syndrome, Lynch syndrome, etc. These diseases are associated with specific gene mutations, and family members carrying the mutations have a higher risk. Currently, multiple cancer genetic screenings have been applied clinically, such as BRCA1/2 gene testing (breast and ovarian cancer), MLH1/MSH2 gene testing (colorectal cancer), etc. For high-risk populations, cancer genetic screening can help them detect tumors early or take preventive measures (such as prophylactic surgery), thereby improving prognosis. However, cancer genetic screening also has some limitations. In addition, positive results may bring a heavy psychological burden to those being screened, and some preventive measures (such as prophylactic mastectomy) may also have a negative impact on quality of life.

Pharmacogenetic Screening

Pharmacogenetic screening utilizes an individual's genetic information for drug therapy, with the aim of achieving personalized medication and reducing adverse reactions. This screening mainly targets genes related to drug metabolism enzymes, drug targets, and drug transport proteins. Some common pharmacogenetic screening items

include CYP2C19 genotype (clopidogrel metabolism), VKORC1 and CYP2C9 genotypes (warfarin dosage), TPMT genotype (azathioprine toxicity), etc. With the help of these screenings, clinicians can adjust drug dosages or choose alternative drugs based on the patient's genotype, thereby improving efficacy, reducing the risk of adverse reactions.

Benefits of Genetic Screening

Early Detection of Potential Health Risks

One of the most significant benefits of genetic screening is that it can identify potential health risks early, even before clinical symptoms appear. This provides valuable time for taking preventive measures and early treatment, which helps improve disease prognosis and quality of life.

Personalized Medicine and Prevention Strategies

Another major advantage of genetic screening is that it can provide personalized medical and prevention strategies. The traditional "one-size-fits-all" treatment model ignores individual differences among patients, while genetic screening can "tailor" medical plans according to each person's unique genetic background and disease risk.

3. Application of Genetic Screening

Lifestyle Adjustments

An important application of genetic screening is to guide individuals in adjusting their lifestyles based on their own genetic characteristics to prevent diseases and promote health. For some common chronic diseases such as cardiovascular disease and diabetes, early identification of genetic risk factors can help high-risk populations take preventive measures in a timely manner, such as controlling diet, increasing exercise, quitting smoking, and limiting alcohol consumption.

Regular Check-ups and Monitoring

For certain genetic diseases or individuals carrying high-risk genetic variants, regular check-ups and monitoring are key measures for prevention and early intervention. For example, for BRCA1/2 gene mutation carriers, regular breast X-ray examinations, breast magnetic resonance imaging (MRI), and transvaginal ultrasound examinations can detect breast and ovarian cancers early and improve treatment outcomes¹⁵⁻²².

Preventive Treatments

For certain high-risk populations, preventive treatments or surgeries can greatly reduce the incidence and mortality of specific diseases. For example, for Lynch syndrome patients, prophylactic colorectal resection can significantly reduce the risk of colorectal cancer¹³⁷. For BRCA1/2 gene mutation carriers, prophylactic bilateral mastectomy and salpingo-oophorectomy can reduce the risk of breast and ovarian cancers.

Current status of genomic screening for chronic diseases

Currently, genomic screening is utilized for monogenic subforms of some common chronic disorders with a complex etiology typically including gene-gene and gene-environment interaction. For instance, in cancer and cardiovascular disorders, genomic screening can be used for the detection of hereditary breast and ovarian cancer, hereditary forms of colorectal cancer, and familial hypercholesterolemia. In such cases, genomic information is mainly oriented to disease diagnosis, prognosis and

individualized treatment/management and allows for prevention in family members. Rather than opting for population screening for these conditions, cascade screening of first degree family members of index cases has proven to be effective for the aforementioned conditions and is, therefore, recommended by prominent international bodies.

Importance of Genetic Screening

Genetic screening occupies an increasingly important position in modern medicine and personal health management.

The following points illustrate the significant implications of genetic screening

1. Early Identification of Health Risks:

Genetic screening can detect an individual's risk of developing certain diseases before clinical symptoms appear. This provides valuable time for early intervention and prevention, which helps improve disease prognosis and reduce the disease burden on individuals and society.

2. Personalized Medicine and Prevention Strategies:

The results of genetic screening provide key information for medical professionals, helping them formulate medical and prevention plans tailored to individual characteristics. This precision medicine approach can improve treatment efficacy, reduce adverse reactions, and optimize the implementation of preventive measures.

3. Family Risk Assessment:

Many diseases are hereditary, and the results of genetic screening are of great significance for the health management of both the person being screened and their family members. After identifying the risk of familial diseases, family members can take appropriate preventive and monitoring measures to detect and treat potential health problems early.

4. Reference for Reproductive Decisions:

Genetic screening can help prospective parents understand their risk of carrying genetic diseases, providing an important reference for reproductive decisions. This helps reduce the incidence of genetic diseases in offspring and improve the health level of newborns.

5. Promoting Scientific Research and Medical Progress:

The large amount of data generated by genetic screening provides valuable resources for scientific research. By analyzing these data, researchers can gain in-depth understanding of the genetic basis of diseases, discover new disease markers and therapeutic targets, and promote medical progress and innovation²³⁻²⁹.

Genetics and genomic principles

DNA, genes, and genomes constitute the fundamental structural components of an organism's biological framework. DNA double helix with structural base pairing is the most widely recognized DNA structure. It is evident from this structure that DNA is structurally dynamic and capable of adopting alternative secondary structures. A genome is an organism's complete set of DNA sequences. Although people in this world may look different, all human genomes are highly similar^[10]. It includes all of an organism's genes and non-coding sequences. Most genomes consist of a linear polymer of DNA wrapped around octameric histone protein complexes to generate a chromatin structure resembling beads on a string.

Integration of genomic data into clinical practice

It is a slow process yet advancement in knowledge is increasing the use of genomic data and genomic medicine in clinical care. Advancement in genetics brings genetic medicine and genetic data into clinical practice improving the diagnosis of rare diseases, illness related risk improvement, and treatment efficiency through advanced measurement and methods. Next-generation sequencing has changed the genomics and not only improve the method but also lowering the costs, can perform rapid genome sequencing and has several medical uses.

Genetic testing is important for the detection of inherited and acquired disorders, and also for treatment responses. Multiple genetic tests are used including targeted single-gene assays, gene panels, whole-exome sequencing, and whole-genome sequencing. Chromosomal testing use for detecting changes in chromosomes like additional or missing copies and any large segment modifications. Exome sequencing improves genetic diagnosis and aid in the prenatal identification of structural abnormalities or genetic disorder. Combining copy number variant and single nucleotide variant analyses increases accuracy, whereas low-pass genome sequencing provides higher resolution. Combining copy number variant sequencing and karyotyping improves the identification of prenatal pathogenic chromosomal abnormalities, enhancing the accuracy of prenatal diagnosis.

Gene therapy in cardiovascular diseases

Genetics play an important role in cardiomyopathies. Pathogenic variants in MYH7 gene, MYBPC3 gene are the most common in encoding abnormal sarcomeric proteins causing Hypertrophic cardiomyopathy. TTN gene, LMNA gene are the most commonly implicated genes in dilated cardiomyopathy. DSC2, DSG2 genes are implicated in arrhythmogenic right ventricular cardiomyopathy. Pathological variant genes testing is implicated to improve prognosis via early screening. Screening of first-degree relatives is also implicated via serial ECGs and echocardiography.

Applications of genomic medicine in oncology

Over the past decade recent advances in genomic medicine has enhanced diagnosis and management of neoplastic diseases by knowing underlying molecular process. Cancer genomic profiling has shown to detect gene amplification, gene deletion mutation, gene fusion of the target genes.

Rare genetic disorders

There are a wide variety of rare unexplained genetic disorders and developmental anomalies. Their apt diagnosis can help the patients understand their condition better. Recent advancements with NGS, which includes whole genome sequencing, whole exam sequencing, whole mtDNA sequencing, targeted exam sequencing and RNA sequencing, has countered the limitations of more traditional methods like Sanger method, karyotyping, and chromosomal arrays for rare genetic diseases.

Pharmacogenomics

Pharmacogenomics leverages genomic biomarkers to predict individual responses to drug efficacy and toxicity. While factors like disease severity, diet, and other medications also influence drug responses, genetic

differences significantly impact drug metabolism and action. Despite the growing body of research, replicating findings remains a challenge. Genome-wide association studies have identified genetic variations associated with psychiatric disorders and drug responses, but most findings lack consistent replication. The FDA includes pharmacogenomics information in drug labels, highlighting its growing recognition³⁰⁻³⁹.

Genomic medicine and personalized treatment

One of the main advantages is increased diagnostic accuracy. Comprehending genetics differences allows clinicians to properly diagnose disorders that might otherwise be missed using conventional approaches, resulting in more accurate and earlier disease identification. Enhanced therapeutic efficacy and safety are also significant advantages. Personalized plans based on genomic data can assist in selecting the best medications and dosages, lowering adverse drug reactions and enhancing therapeutic success rates.

Clinical trials on genomics and personalized treatment

Personalized medicine in oncology is adapting treatment to individual patient features, especially genomic and molecular markers. This strategy seeks to give the “right treatment for the right person at the right time” by using genetic information to guide therapeutic decisions. The potential of genomics-based tailored treatment, demonstrating that magnesium supplementation can regulate DNA methylation in the TMRSS2 gene, which is critical for SARS-CoV-2 viral entry.

Applications in Chronic Disease Treatment Personalized Medicine:

By understanding a patient's unique genetic makeup, treatments can be tailored to their specific genetic blueprint, improving outcomes and reducing side effects.

Drug Discovery

Genomics helps identify new therapeutic targets by highlighting genes and pathways involved in disease pathogenesis, leading to the development of innovative treatments.

Risk Assessment and Prevention

Genetic analysis can identify individuals at higher risk of developing certain chronic diseases, allowing for early diagnosis, preventative measures, and proactive management.

Monitoring Treatment Efficacy

Biomarkers derived from genetic analysis can be used to monitor disease progression and the effectiveness of treatments, allowing for adjustments to the treatment plan.

4. Conclusion

Our review states that genetic research is a scientific field focused on understanding how genes, which are made of DNA influence traits, health, and disease. It involves studying DNA, RNA, chromosomes, and other genetic material to identify how variations lead to diseases or affect responses to treatments. The goal is to improve disease detection, treatment, and prevention by finding the genetic and environmental factors that contribute to health conditions. The genetic testing involves examining a person's blood or other tissues to determine whether he or she has a change in his or her genetic material⁴⁰. The

genetic testing may be useful in determining whether an individual has a genetic condition or may develop one in the future. The information gained from genetic testing may be helpful in a number of ways such as diagnosing a genetic disease, starting treatment, or initiating prevention strategies, as well as making life decisions such as career choice and family planning.

The genomic medicine has increased our knowledge of genetic variations, resulting more accurate diagnosis and personalized treatment. DNA sequencing technology, genetic data integration into clinical care, and the use of multi-omics techniques are among the most significant developments. The future research should focus on increasing access to genetic technology, tackling ethical challenges, and enhancing bioinformatics facilities. Clinical practice needs to change to include these developments, providing fair and efficient individual treatment. Addressing the existing health challenges important for achieving its full assurance, leading to more customized, precise, and efficient treatments that improves outcomes for patients.

5. Bibliography

- [1] Kumar V, Cotran RS, Robbins SL. *Robbins Basic Pathology*. 6th ed. Philadelphia: W.B. Saunders; 2000.
- [2] Musunuru K, Hickey KT, Al-Khatib SM, et al. Basic concepts and potential applications of genetics and genomics for cardiovascular and stroke clinicians. *Circulation*. 2015;8:216–42.
- [3] Watson J, Crick F. Molecular structure of nucleic acids: a structure for deoxyribose nucleic acid. *Nature*.1953;171:737–38.
- [4] Sanger F, Thompson EOP. The amino-acid sequence in the glycol chain of insulin. 2. The investigation of peptides from enzymic hydrolysates. *Biochem J* 1953;53:366–74.
- [5] Maria Giani A, Roberto Gallo G, Gianfranceschi L, et al. Long walk to genomics: history and current approaches to genome sequencing and assembly. *Comput Struct Biotechnol J*. 2020;18:9–19.
- [6] Staden R. A strategy of DNA sequencing employing computer programs. *Nucleic Acids Res* 1979;6:2601–10.
- [7] Baer R, Bankier A, Biggin M, et al. DNA sequence and expression of the B95-8 Epstein-Barr virus genome. *Nature*.1984;310:207–11.
- [8] Heather JM, Chain B. The sequence of sequencers: the history of sequencing DNA. *Genomics* 2016;107:1–8.
- [9] Goetz LH, Schork NJ. Personalized medicine: motivation, challenges, and progress. *Fertil Steril* 2018;109:952–63.
- [10] Alzu'bi AA, Zhou L, Watzlaf VJM. Genetic variations and precision medicine. *Perspect Health Inf Manag* 2019;16:1a.
- [11] Juengst E, McGowan M. Why does the shift from 'personalized medicine' to 'precision health' and 'wellness genomics' matter? *AMA J Ethics* 2018;20:e881–e890.
- [12] Shastry BS. SNPs: impact on gene function and phenotype. *Methods Mol Biol*. 2009;578:3–22.
- [13] Say YH. The association of insertions/deletions (INDELs) and variable number tandem repeats (VNTRs) with obesity and its related traits and complications. *J Physiol Anthropol*. 2017;36:25.
- [14] Shaikh TH, Gai X, Perin JC, et al. High-resolution mapping and analysis of copy number variations in the human genome: a data resource for clinical and research applications. *Genome Res* 2009;19:1682–90.
- [15] Lerman C, Schnoll RA, Lw H, Jr, et al. Use of the nicotine metabolite ratio as a genetically informed biomarker of response to nicotine patch or varenicline for smoking cessation: a randomised, double-blind placebo-controlled trial. *Lancet Respir Med*.2015;3:131–38.
- [16] Manolio TA, Chisholm RL, Ozenberger B, et al. Implementing genomic medicine in the clinic: the future is here. *Genet Med* 2013;15:258–67.
- [17] Manolio TA, Rowley R, Williams MS, et al. Opportunities, resources, and techniques for implementing genomics in clinical care. *Lancet* 2019, 394: 511–20.
- [18] McCombie WR, McPherson JD, Mardis ER. Next-generation sequencing technologies. *Cold Spring Harb Perspect Med* 2019;9:a036798.
- [19] Chau MHK, Choy KW. The role of chromosomal microarray and exome sequencing in prenatal diagnosis. *Curr Opin Obstet Gynecol*. 2021;33: 148–55.
- [20] Chrzanowska NM, Kowalewski J, Lewandowska MA. Use of Fluorescence in Situ Hybridization (FISH) in diagnosis and tailored therapies in solid tumors. *Molecules* 2020;25:1864.
- [21] Ferrara K, Pollard R, Borden M. Ultrasound microbubble contrast agents: fundamentals and application to gene and drug delivery. *Annu Rev Biomed Eng* 2007;9:415–47.
- [22] Gill SL, O'Neill H, McCoy RJ, et al. Enhanced delivery of microRNA mimics to cardiomyocytes using ultrasound responsive microbubbles reverses hypertrophy in an in-vitro model. *Technol Healthc* 2014, 22:37–51.
- [23] Naito Y, Aburatani H, Amano T, et al. Clinical practice guidance for next-generation sequencing in cancer diagnosis and treatment (edition 2.1). *Int J Clin Oncol* 2021;26:233–83.
- [24] Swain SM, Shastry M, Hamilton E. Targeting HER2-positive breast cancer: advances and future directions. *Nat Rev Drug Discovery* 2023;22:101–26.
- [25] Fu K, Xie F, Wang F, et al. Therapeutic strategies for EGFR-mutated non-small cell lung cancer patients with osimertinib resistance. *J Hematol Oncol* 2022;15:173.
- [26] Hughes T, Klairmont M, Sharfman WH, et al. Interleukin-2, Ipilimumab, and Anti-PD-1: clinical management and the evolving role of

immunotherapy for the treatment of patients with metastatic melanoma. *Cancer Biol Ther* 2021; 22: 513–26.

- [27] Maggiore U, Palmisano A, Buti S, et al. Chemotherapy, targeted therapy and immunotherapy: which drugs can be safely used in the solid organ transplant recipients? *Transplant Int* 2021;34:2442–58.
- [28] Onozawa H, Saito H, Sunami K, et al. Lung adenocarcinoma in a patient with a cis-based profiling test: negative diagnosis on initial companion test and successful treatment with osimertinib. *Thorac Cancer* 2020;11:3599–604.
- [29] Martinez-Fernandez De La Camara C, Nanda A, Salvetti AP, et al. Gene therapy for the treatment of X-linked retinitis pigmentosa. *Expert Opin Orphan Drugs* 2018;6:167–77.
- [30] Hanenberg H, Roellecke K, Wiek C. Stem cell genetic therapy for fanconi anemia - a new hope. *Curr Gene Ther* 2017;16:309–20.
- [31] Koromina M, Koutsilieri S, Patrinos GP. Delineating significant genome-wide associations of variants with antipsychotic and antidepressant treatment response: implications for clinical pharmacogenomics. *Hum Genomics* 2020;14:4.
- [32] Zhao W, Meng H. Effects of genetic polymorphism of drug-metabolizing enzymes on the plasma concentrations of antiepileptic drugs in Chinese population. *Bioengineered* 2022;13:7709–45. [DOI] [PMC free article] [PubMed] [Google Scholar]
- [33] Luzum JA, Petry N, Taylor AK, et al. Moving pharmacogenetics into practice: it's all about the evidence! *Clin Pharmacol Ther* 2021;110:649–61.
- [34] Wolf SM, Annas GJ, Elias S. Respecting patient autonomy in clinical genomics: new recommendations on incidental findings go astray. *Science*. 2013; 340(6136):1049–1050.