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# Review article

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# Biobanks in chronic disease management: A comprehensive review of strategies, challenges, and future directions

Wanna Xu<sup>a</sup>, Xiongshun Liang<sup>a</sup>, Lin Chen<sup>a</sup>, Wenxu Hong<sup>a,\*\*</sup>, Xuqiao Hu<sup>a,b,\*</sup>

<sup>a</sup> Shenzhen Center for Chronic Disease Control, Shenzhen Institute of Dermatology, Shenzhen, 518020, China
 <sup>b</sup> Second Clinical Medical College of Jinan University, First Affiliated Hospital of Southern University of Science and Technology (Shenzhen People's

Hospital), Shenzhen, China

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# ABSTRACT

Biobanks, through the collection and storage of patient blood, tissue, genomic, and other biological samples, provide unique and rich resources for the research and management of chronic diseases such as cardiovascular diseases, diabetes, and cancer. These samples contain valuable cellular and molecular level information that can be utilized to decipher the pathogenesis of diseases, guide the development of novel diagnostic technologies, treatment methods, and personalized medical strategies. This article first outlines the historical evolution of biobanks, their classification, and the impact of technological advancements. Subsequently, it elaborates on the significant role of biobanks in revealing molecular biomarkers of chronic diseases, promoting the translation of basic research to clinical applications, and achieving individualized treatment and management. Additionally, challenges such as standardization of sample processing, information privacy, and security are discussed. Finally, from the perspectives of policy support, regulatory improvement, and public participation, this article provides a forecast on the future development directions of biobanks and strategies to address challenges, aiming to safeguard and enhance their unique advantages in supporting chronic disease prevention and treatment.

# 1. Introduction

In the realm of global health, the management of chronic diseases has emerged as an increasingly prominent challenge. Chronic illnesses such as cardiovascular diseases, diabetes, and cancer pose not only a severe threat to individual health but also impose a significant burden on public health systems. In this context, the importance of biobanks has become increasingly evident. By collecting and storing biological samples from patients, such as blood, urine, and tissue samples, biobanks provide invaluable resources for the research and management of chronic diseases.

The application of biobanks in chronic disease research is extensive. They offer researchers unique opportunities to explore the molecular and genetic mechanisms of diseases, thereby advancing the development of new diagnostic methods and treatment strategies. For instance, by analyzing genetic data from biobanks, researchers can identify gene mutations associated with specific chronic diseases, which is crucial for understanding the pathogenesis of diseases and developing personalized treatment plans [1–3]. Additionally, biobanks play a significant role in the prevention and early diagnosis of chronic diseases. Monitoring changes in biomarkers

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<sup>\*</sup> Corresponding author. Shenzhen Center for Chronic Disease Control, Shenzhen Institute of Dermatology, Shenzhen, 518020, China. \*\* Corresponding author.

E-mail addresses: szbloodcenter@hotmail.com (W. Hong), haniahu@hotmail.com (X. Hu).

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can lead to early identification of disease risks, enabling effective preventive measures and intervention strategies [4,5]. This is vital for reducing the incidence of chronic diseases and improving the quality of life for patients. However, the application of biobanks in chronic disease management also faces numerous challenges. These challenges include, but are not limited to, standardization issues in the collection, storage, and processing of samples, as well as related ethical, legal, and data privacy concerns [6]. Despite these challenges, the potential of biobanks in advancing chronic disease research and management cannot be overlooked.

In summary, as chronic diseases continue to pose a significant challenge globally, the role of biobanks in managing these conditions has become increasingly crucial. Through the collection, preservation, and analysis of biological samples, biobanks enable significant advancements in personalized medicine, biomarker identification, and the development of targeted therapies. This manuscript aims to provide a narrative review that explores the current status, inherent challenges, and prospective future of biobanks within the context of chronic disease management, thereby offering insights into their evolving role in enhancing biomedical research and healthcare outcomes.

# 2. Biobanks: overview and development

# 2.1. Definition and types

A biobank, also known as a biological bank, is a term that first appeared in the literature in the 1996 article "Cancer risk and oxidative DNA damage in Man" [7]. Biobanks are typically defined as facilities or collections used for storing and managing human biomedical samples, such as blood, urine, tissue sections, cells, and molecular genetic materials. These repositories can be categorized based on the type of samples, the purpose of collection, or the field of research, for example, cancer biobanks, genetic disease biobanks, or population-based biobanks.

#### 2.2. Historical development and current trends

The concept of biobanks gradually took shape in the mid-20th century, initially focusing primarily on the storage of blood samples. Over time, biobanks have experienced significant growth in both quantity and complexity. Today, these repositories have expanded not only in terms of sample diversity but also in advancements in data management and application domains [8]. For instance, modern biobanks may include everything from simple biological specimens to complex genomic data. In recent years, research on complex diseases such as cancer and neurodegenerative disorders has increasingly relied on these diverse and rich biobanks. With globalization and shifts in disease patterns, the demand for and scope of application of biological samples continue to grow, especially in emerging markets and developing countries.

# 2.3. Technological advancements and their impact

In recent years, the value and application scope of biobanks have greatly expanded, largely due to the rapid development of genomic sequencing technologies and bioinformatics. For example, high-throughput sequencing technologies, such as next-generation sequencing, have made it possible to obtain a vast amount of genetic information from a single sample, which is crucial for understanding the genetic basis of complex diseases [9]. Additionally, advancements in bioinformatics tools have not only improved the efficiency of data management but also facilitated in-depth analysis of these complex datasets. Significant progress in automation and digital methods in sample processing and data storage has enhanced efficiency and reduced errors. For instance, the application of machine learning and artificial intelligence is transforming the interpretation of sample data, making it more feasible to discover new biomarkers and disease-related patterns from large datasets [10-12]. With the rise of personalized medicine, biobanks are increasingly important in the development of new treatment methods and drugs. They provide key information for early diagnosis, prediction of treatment response, and therapeutic evaluation. In summary, biobanks play an indispensable role in modern biomedical research, and with technological advancements, their application and impact are expected to further expand.

#### 2.4. Imaging biobanks: a critical subset in precision medicine

Amidst the diverse categories of biobanks, imaging biobanks have emerged as a critical subset, revolutionizing the landscape of medical research and patient care in the era of precision medicine. These repositories, which store a vast array of medical imaging data, including X-rays, MRI scans, and CT scans, have become pivotal in harnessing the power of imaging biomarkers for disease diagnosis, prognosis, and therapeutic strategies [13].

The significance of imaging biobanks is particularly pronounced in the realm of evidence-based medicine, where they contribute immensely to the identification and validation of quantitative imaging biomarkers. These biomarkers offer a non-invasive means to understand the pathological mechanisms of diseases, monitor disease progression, and evaluate therapeutic responses with unprecedented precision. As underscored by a comprehensive review, imaging biobanks are instrumental in bridging the gap between basic research and clinical applications, facilitating the transition towards more targeted and personalized healthcare solutions [14].

Furthermore, the differentiation between disease-oriented and population-based biobanks, as discussed in the literature, highlights the broad utility of imaging biobanks. Disease-oriented biobanks focus on specific patient groups and are invaluable in studying the onset and progression of diseases, enabling researchers to pinpoint potential therapeutic targets. In contrast, population-based biobanks gather data from a broader demographic, offering insights into disease prevalence, risk factors, and health outcomes on a

population level. Both types of biobanks, powered by the inclusion of imaging data, play a pivotal role in the modern era of AI-driven precision medicine, providing a rich dataset for the development of algorithms that can predict disease risk, detect early disease markers, and personalize treatment plans [15].

In conclusion, the integration of imaging biobanks into the biobanking landscape represents a significant advancement in medical research and patient care. By facilitating the detailed analysis of disease markers and contributing to the development of precision medicine, imaging biobanks underscore the importance of comprehensive data collection and utilization in improving health outcomes.

### 2.5. A bibliometric overview of biobank research in chronic disease management

To provide a comprehensive perspective on the most influential research related to the use of biobanks for chronic disease management, a bibliometric analysis was conducted. This analysis examined literature from 2014 to the present, retrieved from the Web of Science database using the keywords "Biobank" and "Chronic Disease". A total of 1996 research articles were identified through this search.

Fig. 1 presents the core sources contributing to this field, ranked according to Bradford's Law of Scattering. The top 10 journals account for 327 articles, constituting the nucleus of the literature. Scientific Reports, PLOS ONE, and Medicine emerge as the most prolific sources within this core group.

The citation analysis showcases leading research within this field (Fig. 2). The study by Wuttke M. in Nature Genetics (2019) has emerged as a pivotal resource, with 69 local and 398 global citations, evidencing a significant academic footprint [16]. Alongside, Pattaro C.'s 2016 publication in Nature Communications reflects its enduring impact with 29 local citations and a global citation tally of 348 [17]. These works, among others listed, have played a central role in advancing chronic disease discourse and underscore the interconnectedness of current research efforts.

Furthermore, an analysis of the countries contributing to this field based on the corresponding authors' affiliations is depicted in Fig. 3. China emerges as the most prolific nation with 605 articles, although the United States exhibits the highest ratio of corresponding authors at 0.453. The United Kingdom, Germany, and Spain follow as major contributors to biobank research related to chronic disease management.

In summary, this bibliometric overview elucidates the core literature, highly cited works, and geographical distributions underlying research efforts aimed at leveraging biobanks to advance chronic disease management strategies. These findings underscore the global significance of this field and its potential to drive groundbreaking discoveries.

# 3. The global burden of chronic diseases

# 3.1. Definition and classification of chronic diseases

Chronic diseases refer to those conditions that are long-lasting and generally progress slowly. The primary characteristic of these diseases is that they usually do not resolve spontaneously and require long-term medical management and treatment. The scope of



Fig. 1. Core Literature Sources by Bradford's Law. A graph depicting the core sources by Bradford's Law, illustrating a steep decline in the number of articles as the rank of sources increases, with a long tail of sources contributing a relatively small number of articles.



Fig. 2. Most Local Cited Documents in the Field. A bar chart displaying the most globally cited documents in the field.



# Corresponding Author's Countries

Fig. 3. Countries of Corresponding Authors. A horizontal bar chart showing the corresponding author's countries for the publications analyzed, with China and the USA having the highest number of single-country publications (MCP), while the USA and the United Kingdom lead in terms of multiple-country collaborations (SCP).

chronic diseases is quite broad, encompassing but not limited to cardiovascular diseases (such as hypertension, heart disease, and stroke), cancer, chronic respiratory diseases (such as asthma and chronic obstructive pulmonary disease), and diabetes. These diseases are often closely linked to lifestyle factors, such as diet, physical activity, tobacco use, and alcohol consumption, while also being influenced by genetic factors [18].

# 3.2. Global epidemiology of chronic diseases

According to the World Health Organization reports, chronic diseases are the leading cause of death globally, accounting for 71 % of all deaths worldwide. Cardiovascular diseases are the most common cause of death among chronic diseases, followed by cancer,

respiratory diseases, and diabetes. Globally, the prevalence of these diseases is on the rise, especially in low- and middle-income countries. For instance, cardiovascular diseases cause approximately 17.9 million deaths annually, while cancer accounts for about 9.6 million deaths each year. These figures highlight the significance of chronic diseases in the global health landscape [19–21].

#### 3.3. Impact of chronic diseases on public health

Chronic diseases have a profound impact on global public health systems. These illnesses not only lead to a high number of deaths and disabilities but also have a significant economic impact, including increased healthcare costs and decreased labor productivity. For instance, the management and treatment costs of chronic diseases impose a substantial burden on families and societies, especially in resource-limited settings. Moreover, the burden of chronic diseases varies significantly between different countries and socio-economic groups, highlighting issues of global health inequality. The widespread prevalence of chronic diseases also exacerbates the strain on public health systems, particularly in the face of global health crises such as the COVID-19 pandemic [22].

#### 4. Application of biobanks in chronic disease research

#### 4.1. Basic research and molecular mechanisms

Biobanks play a crucial role in understanding the molecular mechanisms of chronic diseases. Through in-depth analysis of these samples, researchers can reveal the molecular and genetic basis of specific diseases, thereby advancing the development of disease treatments and prevention strategies.

In the field of cancer research, the application of biobanks is particularly significant. Genomic sequencing of tumor samples has enabled scientists to identify key gene mutations that contribute to tumor growth and spread. For instance, mutations in the BRCA1 and BRCA2 genes have been confirmed to be associated with a high risk of breast and ovarian cancers [23,24]. Additionally, research on tumor biomarkers has led to the development of more precise treatment methods, such as targeted therapy drugs for specific gene mutations [25]. In the area of cardiovascular diseases, biobanks also play an important role. By analyzing specific proteins and genetic markers in patient blood samples, researchers can better understand the pathogenesis of cardiovascular diseases. For example, studies on inflammatory markers like C-reactive protein (CRP) help assess the risk of heart disease [26]. Moreover, genetic analysis of cardiovascular disease patients has revealed multiple gene loci associated with disease risk, offering possibilities for personalized treatment [27]. Research on diabetes and other metabolic diseases also benefits from biobanks. By analyzing blood and tissue samples from patients, scientists can identify molecular mechanisms affecting insulin resistance and beta-cell function. For instance, research on the insulin signaling pathway aids in understanding the pathogenesis of type 2 diabetes and provides clues for developing new treatment strategies [28,29]. For neurodegenerative diseases like Alzheimer's disease, biobanks offer opportunities to study brain degeneration and functional loss. By analyzing patients' cerebrospinal fluid and blood samples, researchers can explore biomarkers related to the disease, such as beta-amyloid and tau proteins. These studies aid in early diagnosis and monitoring disease progression, as well as in developing new treatment methods [30–32].

With the rise of personalized medicine, biobanks are increasingly important in developing new treatment methods and drugs. They provide key information for early diagnosis, predicting treatment response, and evaluating therapeutic efficacy. For example, analyzing gene expression patterns in tumor samples can tailor personalized treatment plans for cancer patients. In summary, biobanks play an indispensable role in modern biomedical research, and with technological advancements, their application and impact are expected to further expand [33–35].

#### 4.2. Clinical trials and therapeutic evaluation

Biobanks play a crucial role in clinical trials and therapeutic evaluation. They provide researchers with key tools for assessing the effects of new treatment methods, particularly in monitoring the biological effects of drugs or treatment methods and patients' responses to therapy.

During the clinical trial phase of drug development, data from biobanks are essential for evaluating the efficacy and safety of drugs. By analyzing biomarkers in blood, urine, or tissue samples, researchers can monitor the drug's metabolism in the body, its efficacy, and potential side effects. For example, in clinical trials for cancer treatment, measuring tumor marker levels can assess the efficacy of chemotherapy or targeted therapy drugs [36–38]. Biobanks can also be used to evaluate patients' responses to specific treatments. This is particularly important in personalized medicine, as different patients may respond significantly differently to the same treatment. For instance, in diabetes treatment, monitoring blood glucose levels and insulin response can help adjust treatment plans for optimal control [39–41]. The application of biomarkers in clinical trials is increasingly widespread; they can be used for early assessment of treatment effects, monitoring disease progression or recurrence, and predicting treatment outcomes. For example, in cardiovascular disease research, blood cholesterol levels and inflammatory markers are used to evaluate the effectiveness of drug treatments [42–44]. Biobanks also support the evaluation of the long-term efficacy and safety of drugs and treatment methods. By longitudinally tracking patient samples, researchers can monitor the long-term effects of chronic disease treatment and potential delayed side effects. With the advancement of precision medicine, biobanks are playing an increasingly important role in developing personalized treatment plans. By analyzing patients' genetic information and biomarkers, tailored treatment plans can be devised for each patient, thereby improving treatment effectiveness and reducing unnecessary side effects [45–47].

#### 4.3. Discovery and application of biomarkers

Biobanks play a key role in the discovery and validation of new biomarkers. Biomarkers are objectively measurable indicators that can be used to assess biological processes, pathological states, or responses to drug treatments. These markers are of significant value in early disease diagnosis, disease progression monitoring, prognostic assessment, and the development of personalized treatments.

Biobanks provide a wealth of biological samples that can be used to screen and validate potential biomarkers. For example, through genomic and proteomic analysis of tumor samples from cancer patients, researchers can discover specific gene mutations or changes in protein expression patterns associated with tumor development. These findings help identify early indicators of cancer, enabling early diagnosis and treatment [48,49]. Biomarkers play an important role in the early diagnosis of diseases. For instance, tumor markers in the blood, such as prostate-specific antigen (PSA), are widely used for cancer screening [50]. Similarly, cardiac biomarkers, such as cardiac troponin, can be used for the rapid diagnosis of myocardial infarction [51]. Biomarkers can also be used to monitor patients' responses to treatment and disease progression. For example, changes in certain cancer biomarkers during chemotherapy can reflect the tumor's response to treatment [52]. Additionally, biomarker levels can be used to predict disease prognosis, such as the level of BNP (brain natriuretic peptide) in the blood of cardiovascular disease patients being related to the risk of heart failure [53,54]. With the development of personalized medicine, the role of biomarkers in formulating personalized treatment plans is becoming increasingly prominent. For example, patients with HER2-positive breast cancer can receive targeted therapy against HER2. Moreover, by analyzing patients' genetic markers, doctors can predict patients' responses to specific drugs, thereby optimizing treatment plans [55].

Despite significant progress in the discovery and application of biomarkers, challenges remain, including the specificity, sensitivity, and applicability of biomarkers in different populations. Future research needs to address these issues and further explore how to translate biomarker discoveries into clinical practice to improve the diagnosis and treatment of diseases.

### 5. The role of biobanks in chronic disease management

#### 5.1. Prevention strategies and early diagnosis

Biobanks play a crucial role in the prevention and early diagnosis of chronic diseases. Through in-depth analysis of biological samples, researchers can identify early biomarkers of specific diseases, enabling effective intervention before symptoms appear.

Samples in biobanks, such as blood, urine, or tissue samples, contain a wealth of biological information, including DNA, RNA, proteins, and metabolites. By analyzing these biomolecules, researchers can identify early biomarkers associated with specific chronic diseases. For instance, certain patterns of gene expression or changes in protein levels may indicate the development of cancer or cardiovascular diseases. In the early diagnosis of heart disease, biomarkers such as cardiac troponin and brain natriuretic peptide (BNP) are used to assess the risk of heart damage and heart failure [56]. In diabetes, measurements of blood glucose levels, insulin resistance markers, and glycated hemoglobin (HbA1c) are crucial for identifying the early stages of diabetes [57]. Analysis of samples in biobanks allows researchers to better understand the pathogenesis of chronic diseases, thereby formulating effective prevention strategies. For example, identifying biomarkers associated with obesity, hypertension, and high cholesterol can provide personalized lifestyle and dietary recommendations to prevent the development of cardiovascular diseases [58]. The application of biobanks also promotes the development of personalized medicine. By analyzing an individual's genetic and molecular characteristics, doctors can provide more precise prevention and treatment plans. For example, analyzing an individual's genetic background for drug metabolism capabilities can optimize drug selection and dosage, reducing side effects and enhancing treatment effectiveness [59,60].

Despite the broad prospects of biobanks in chronic disease management, challenges remain, including the collection and storage of samples, data privacy protection, and the validation of biomarker effectiveness. Future research needs to address these issues and further explore how to translate discoveries from biobanks into clinical practice to improve the prevention and early diagnosis of chronic diseases.

#### 5.2. Personalized treatment and management

Biobanks play a key role in the personalized treatment and management of chronic diseases. By analyzing an individual's genetic and biomarker information, doctors can design more precise and effective treatment plans for each patient.

In the field of cancer treatment, personalized therapy has become an important trend. By performing genomic sequencing on tumor samples, doctors can identify specific gene mutations and variations, which are crucial for selecting the most appropriate treatment methods. For example, targeted therapies for HER2-positive breast cancer patients [61,62] and treatments for non-small cell lung cancer patients with EGFR mutations [63,64], are based on the individual tumor's genotype.

Personalized treatment is also important in the management of cardiovascular diseases. By analyzing patients' genetic information and cardiovascular biomarkers, doctors can assess patients' responses to specific drugs, thereby optimizing treatment plans. For instance, for patients with hypertension and heart disease, drug selection and dosage adjustments based on genetic background can significantly improve treatment effectiveness and reduce side effects [65–67].

The treatment of chronic kidney disease is also moving towards personalization. By analyzing patients' genetic markers and renal function indicators, doctors can more accurately assess disease progression and treatment response. This helps to formulate more effective treatment plans, including medication, lifestyle adjustments, and renal replacement therapy when necessary [68,69].

Diabetes management is increasingly reliant on personalized strategies. By monitoring blood glucose levels, insulin responses, and other metabolic indicators, doctors can tailor diet, exercise, and medication plans for each diabetic patient. Additionally, drug

selection based on genetic information is playing an increasingly important role in diabetes treatment [70].

Despite the great potential of personalized treatment in chronic disease management, challenges remain, including the interpretation of large amounts of genetic and biomarker data, managing treatment costs, and ensuring all patients have access to high-quality personalized care. Future research needs to address these issues and further explore how to translate the advantages of personalized medicine into widespread clinical practice, improving treatment outcomes and quality of life for patients with chronic diseases.

#### 5.3. Patient education and self-management

Utilizing data from biobanks can significantly enhance patients' understanding of their own diseases and promote more effective self-management. This approach not only helps patients better understand their health status and treatment options but also assists them in adjusting their lifestyles to better control their condition.

By analyzing data from biobanks, patients can gain detailed information about their diseases, including genetic risks and biomarker levels. This information helps patients gain a deeper understanding of their conditions, such as how specific genetic variations may affect the risk and progression of their diseases. This enhanced disease awareness can enable patients to play a more active role in the treatment decision-making process.

Understanding one's genetic and biomarker information can also help patients manage their conditions more effectively. For example, knowing one's sensitivity or risk of side effects to certain medications can help patients work with their doctors to develop more suitable treatment plans. Additionally, monitoring biomarkers can help patients track treatment effectiveness and disease progression.

Information from biobanks can also guide patients in adjusting their lifestyles, such as diet and exercise habits, to better control their diseases. For instance, for diabetic patients, understanding their blood sugar levels and insulin responses can help them adjust their diet and exercise plans to maintain stable blood sugar levels. For cardiovascular disease patients, knowing their cholesterol levels and blood pressure can guide them in adjusting their diet and lifestyle habits to reduce cardiovascular risks.

Effective self-management requires crucial patient education. Healthcare providers need to ensure that patients can understand and interpret data from biobanks and know how to apply this information to their daily lives. This may include providing educational materials, conducting workshops, or individual counseling to help patients better understand their diseases and management strategies.

While utilizing biobank data for patient education and self-management holds great potential, challenges remain, including ensuring the accuracy and interpretability of data, as well as protecting patient privacy and data security. Future research needs to explore how to overcome these challenges while enhancing patients' awareness and ability to manage their diseases.

#### 6. Technological, ethical, and legal challenges

#### 6.1. Data security and privacy protection

Ensuring data security and privacy protection is crucial in the management of biobanks. With the increasing number of samples and advancements in technology, protecting individuals' genetic and health information from unauthorized use or disclosure has become a significant challenge.

To protect data in biobanks, the use of advanced encryption technologies is necessary. This includes encrypting stored and transmitted data to prevent unauthorized access and data breaches. Additionally, strict access control mechanisms are key to safeguarding data security. This means that only authorized researchers and medical professionals should have access to relevant data, and their access should be strictly limited based on their work needs. To further protect patient privacy, data in biobanks often undergo anonymization or de-identification. This involves removing all information that can directly identify an individual, such as names, addresses, and social security numbers, allowing the data to be used for important medical research while protecting individual privacy.

The operation of biobanks also needs to comply with relevant legal and ethical guidelines. This includes ensuring the legality of data collection and use, as well as adhering to legal provisions regarding the protection of personal data. Additionally, from an ethical standpoint, ensuring that patients have full informed consent for the use of their data is very important.

Despite various measures taken to protect data in biobanks, data security and privacy protection continue to face many challenges due to the evolution of technology and the advancement of cyber-attack methods. Future research needs to explore more efficient and secure data protection methods and continuously update legal and ethical guidelines to respond to the changing technological and social environment [71–73].

#### 6.2. Ethical issues and sample collection

The establishment and operation of biobanks involve a range of complex ethical issues, particularly in the process of sample collection. These issues not only relate to the rights and welfare of participants but also involve ethical principles in the use and management of samples.

Ensuring informed consent from participants is a core ethical principle in the sample collection process. This means that participants must voluntarily decide whether to participate after fully understanding the research purpose, methods, potential risks, and benefits. Obtaining informed consent is particularly important and complex for vulnerable populations, such as children, individuals

with cognitive impairments, or economically disadvantaged groups. In these cases, researchers need to take additional measures to ensure that participants fully understand and voluntarily participate. Ownership and usage rights of samples are also significant ethical issues. Participants typically have some form of rights over their biological samples, which requires researchers to respect the wishes and privacy of participants when using these samples. Additionally, issues regarding whether samples can be used for future unknown research projects and how sample data should be handled need to be clarified in the informed consent process. When biobanks are used for research that may bring economic benefits, how to fairly share these benefits is an important ethical consideration. This includes ensuring that participants or their communities receive appropriate returns from the use of their samples, such as through improved medical services or economic compensation.

As the scale and scope of biobanks continue to expand, ethical issues will become more complex. Future research needs to explore how to find a balance between respecting individual rights and promoting scientific research. Additionally, with the increase in international collaborative research, cross-cultural and transnational ethical standards need further development and harmonization [71–73].

# 6.3. Legal framework and compliance

With the rapid development of biobanks, existing legal frameworks face new challenges and demands. These challenges involve not only data protection and privacy rights but also span multiple areas including bioethics and intellectual property rights.

In the operation of biobanks, the protection of data and privacy rights is a core component of the legal framework. As technology evolves, protecting individuals' genetic and health information from unauthorized use or disclosure becomes a significant legal challenge. For example, the European Union's General Data Protection Regulation (GDPR) imposes strict requirements on the processing of personal data, including data from biobanks. Bioethics is another important legal area, involving ethical issues in sample collection, use, and storage. This includes ensuring informed consent from participants, usage permissions for samples, and issues of benefit-sharing. Different countries and regions may have various laws and guidelines to regulate these ethical issues.

As biobanks increase their application in drug development and medical research, intellectual property law becomes an important consideration. This involves how to protect the intellectual property of research findings while ensuring that these findings benefit society fairly. For instance, patent law plays a crucial role in protecting new discoveries and innovations, but it also needs to balance public interest and incentives for innovation.

With the growing role of biobanks in international research collaboration, international legal coordination and cooperation become particularly important. This includes establishing common standards and principles in data sharing, ethical review, and intellectual property protection.

Facing the new challenges posed by biobanks, legal frameworks need to be continuously updated and adapted. This includes regulating emerging technologies, managing cross-border data flows, and adapting to novel biomedical research methods. Future legal developments need to find a balance between promoting scientific research and protecting individual rights, while also adapting to the changing technological and social environment [71–73].

#### 6.4. Technical standards and data interoperability

Biobanks play a pivotal role in medical research by providing valuable biospecimens crucial for advancing our understanding of disease mechanisms and developing personalized therapies. To optimize their value, biobanks must navigate a complex landscape of technical standards, ensure strict ethical and legal compliance, and enable transparent access to high-quality samples. This section delves into the significance of these elements, highlighting foundational standards and models like OMOP and MIABIS, instrumental in refining biobank processes.

The integrity of samples and data under biobank stewardship is crucial for the progress of biomedical research. Initiatives like the Minimum Information About BIobank data Sharing (MIABIS) strive to standardize biobank nomenclature and foster data sharing protocols. MIABIS delineates a structured approach to describing biobanks, sample collections, and associated studies, promoting data interoperability and streamlining the exchange of samples among research entities. The establishment of MIABIS and its core terminology has been pivotal in creating a common language for biobank data sharing, facilitating the efficient sharing of biobank samples and data across research institutions [74].

In response to the demand for detailed biobank queries, MIABIS has expanded its framework to include specific components for describing samples and donors at an individual level. This expansion introduces an "event" component for detailing attributes tangentially related to samples or donors, crucial for contextualizing biospecimen collections. These enhancements in biobanking data schemas underscore the shift towards more nuanced, interoperable datasets, enabling precise and wide-ranging inquiries into biobank holdings [74].

Biobanks operate within a rigorously defined ethical and legal perimeter, ensuring the respectful and lawful handling of human biospecimens. Ethical adherence mandates informed consent from donors, the safeguarding of donor privacy, and the protection of data confidentiality. Legal obligations, conversely, involve compliance with both national and international statutes that regulate biobanking practices, encompassing data protection legislation and human research guidelines. Ensuring the proper quality of samples and data, along with ethical and legal compliance, is essential for maintaining the trust of sample donors and facilitating research that can lead to the discovery of new treatments [75].

The efficacy of biobanks is notably enhanced through the establishment of transparent and streamlined access protocols. Providing researchers with explicit instructions for accessing biobank samples and data is vital for encouraging collaborative endeavors and

amplifying the impact of biospecimens on medical breakthroughs. The standards for biobanking, as outlined, aim to supply researchers with high-quality samples fit for intended use, highlighting the need for transparent and efficient access procedures [75].

In summation, the incorporation of technical standards such as MIABIS, coupled with strict adherence to ethical and legal frameworks, is indispensable for biobank operations. These practices not only safeguard the quality and integrity of biobank resources but also cultivate a spirit of transparency and cooperation within the scientific community. As biobanking methodologies evolve, sustained engagement among stakeholders and the embracement of adaptable, interoperable data models will be essential for surmounting existing challenges and seizing the opportunities inherent in this dynamic domain.

#### 7. Success cases and practical experiences

# 7.1. International biobank case studies

Biobanks around the world have demonstrated their immense value in disease research and patient management. Here are some representative international biobank cases:

In Europe, cancer research biobanks have focused on analyzing thousands of samples to reveal the genetic basis of various cancers. These large-scale biobank projects, using high-throughput genomic sequencing technologies, have identified gene mutations and biomarkers associated with specific types of cancer. These discoveries have not only deepened our understanding of the mechanisms of cancer development but also facilitated the development of targeted treatment methods for specific genetic mutations, significantly improving treatment outcomes [76–84].

In the United States, biobanks focusing on cardiovascular diseases have provided new insights into the prevention and treatment of heart diseases. These biobanks contain a large number of biological samples from patients with cardiovascular diseases, including blood and tissue samples. Through in-depth analysis of these samples, researchers have been able to identify genetic markers associated with the risk of cardiovascular diseases and evaluate the effectiveness of different treatment methods. These studies contribute to the development of personalized prevention and treatment strategies for cardiovascular diseases [85,86].

Beyond cancer and cardiovascular diseases, biobanks have also shown their value in other areas. For example, in research on diabetes, neurodegenerative diseases, and rare diseases, biobanks provide valuable resources that help scientists understand the complex mechanisms of these diseases and develop new treatment methods.

Despite the significant achievements of biobanks in disease research, future research still faces many challenges, including the diversity of samples, data integration and analysis, and addressing ethical and legal issues. Future biobanks need to expand the scale and types of samples while ensuring data quality and reliability, as well as the privacy and rights of participants.

#### 7.2. Innovative models in chronic disease management

The application of biobanks in chronic disease management has demonstrated innovative management models, particularly in personalized medicine and early diagnosis.

In diabetes management, the application of biobanks has significantly changed treatment approaches. By analyzing the genetic data of diabetic patients, researchers have identified genetic factors that affect disease progression and treatment response. This information is used to provide patients with personalized lifestyle and medication recommendations. For example, based on an individual's genetic background, doctors can recommend specific diet plans, exercise regimes, and medication treatments to optimize blood sugar control and reduce the risk of complications. In the management of chronic kidney disease, early diagnosis methods based on biobank sample analysis are changing disease prognosis. By detecting specific biomarkers in blood and urine, doctors can identify signs of kidney function impairment before symptoms appear. This enables early intervention with medication and lifestyle adjustments to slow disease progression and prevent kidney failure [81,87].

#### 7.3. Lessons and insights

Analyzing the success stories of biobanks, we can learn key lessons and insights crucial for the future establishment and operation of biobanks.

The establishment and maintenance of biobanks require comprehensive consideration at the technological, ethical, and legal levels. Technologically, it is necessary to ensure the quality, security, and accessibility of data. Ethically, protecting participants' privacy and rights and ensuring that the informed consent process is transparent and fair is vital. Legally, complying with relevant data protection regulations and bioethical standards is essential to ensure that the operation of biobanks meets international and national legal requirements.

Interdisciplinary collaboration and data sharing are crucial for maximizing the value of biobanks. By collaborating across interdisciplinary teams, expertise from biology, medicine, statistics, and computational science can be combined to more comprehensively understand and utilize the data in biobanks. Additionally, data sharing can promote scientific discoveries and accelerate the development of new treatment methods.

With the rapid development of technology and the constantly changing research environment, it is necessary to continuously update data management and analysis strategies. This includes adopting the latest data analysis tools and algorithms and updating the infrastructure for data storage and processing. Moreover, as new technologies like machine learning and artificial intelligence emerge, the methods of data analysis in biobanks also need to evolve.

Public participation and education are also key to the successful operation of biobanks. Through education and communication, public awareness of the importance of biobanks can be increased, enhancing trust and participation among contributors. Public involvement can also promote transparency and a sense of social responsibility, ensuring that research activities align with societal expectations and values.

Looking to the future, biobanks need to adapt to new scientific discoveries and technological changes while addressing ethical and legal issues. This includes dealing with challenges brought by big data, protecting personal privacy, promoting international cooperation, and addressing emerging ethical and social issues.

# 8. Future outlook and challenges

# 8.1. Potential impact of emerging technologies

With the rapid development of Artificial Intelligence (AI), machine learning, and big data technologies, these technologies are expected to have a significant impact on the operation and data analysis of biobanks.

Improving Data Analysis Efficiency and Accuracy: AI and machine learning technologies can process and analyze large volumes of complex biomedical data, enhancing the efficiency and accuracy of data analysis. This is crucial for understanding complex disease mechanisms and accelerating the development of new treatment methods. For example, AI systems have already shown significant impact in the medical field, assisting doctors in making accurate diagnoses and treatment decisions [88].

Application in Cancer Research: In cancer research, AI and big data technologies are making important contributions to biomedical and disease diagnostics. By analyzing high-dimensional multi-omics data, AI frameworks can extract meaningful information that is difficult to obtain manually, advancing our understanding of cancer biology and improving patient care and clinical outcomes [89,90].

Application in Disease Diagnosis and Treatment: The application of AI technology in disease diagnosis, treatment, and pharmaceutical formulation development is rapidly evolving. Core digital technologies of AI, such as machine learning and deep learning, are being widely applied to support diagnostics and treatment. AI technology plays a key role in multiple medical fields, including the development of new medical systems, improvement of patient information and records, and treatment of various diseases [88].

Enhancement in Parkinson's Disease Management: The application of AI technology in the management of Parkinson's Disease (PD) demonstrates its potential in understanding protein degradation mechanisms [91,92]. AI technologies, such as machine learning and data mining, have been used to identify and describe protein degradation pathways involved in PD, providing potential targets for developing new therapeutic interventions.

# 8.2. 8.2 policy making and funding support

Effective policy support and stable financial investment are crucial for the construction and maintenance of biobanks. Here are some key aspects extracted from the latest literature:

Importance of Policy Framework, Goals of Policy Making: Policy making should aim to support the sustainable development and ethical use of biobanks. This includes ensuring that the operation of biobanks complies with ethical standards, data protection regulations, and intellectual property laws. Cross-Sector Collaboration: Cooperation between government and private sectors is essential for ensuring adequate resource allocation. This collaboration can help balance the protection of scientific freedom and individual privacy rights.

Importance of Funding, Stable Financial Support: Stable and sufficient financial support is key to the long-term maintenance and updating of biobanks. This includes government funding, private sector investment, and potential international financial support. Strategies for Fund Allocation: Funding allocation strategies should consider the diversity and representativeness of biobanks, ensuring that research covers a wide range of populations and disease types.

Challenges Faced, Challenges in Fund Allocation: Fund allocation needs to consider the long-term nature and complexity of research, ensuring that funds can continuously support the operation and research activities of biobanks. Challenges in Policy Making: Policy making needs to continuously adapt to technological developments and societal changes, ensuring that the operation of biobanks complies with the latest ethical and legal requirements.

Future Directions, Coordination of Policy and Funding: Future research needs to explore how to more effectively coordinate policy and funding support to promote the sustainable development and ethical use of biobanks. Strengthening International Cooperation: Enhancing international cooperation, especially in terms of funding support and policy making, can promote the development and data sharing of global biobanks.

#### 8.3. Conclusions: major challenges and research directions

As biobanks grapple with technological advancements and the surging volume of data, they face numerous challenges that define significant research directions for the future. Among the foremost challenges are ensuring data security and privacy, enhancing the representativeness and accessibility of biobanks, and adhering to the FAIR data principles. Addressing these issues calls for innovative solutions in legal compliance, data sharing, and management standards, with blockchain technology emerging as a promising tool.

One of the paramount challenges in the era of big data is safeguarding data security and personal privacy within biobanks. Blockchain technology, characterized by its immutable, encrypted, and distributed ledger system, offers potential solutions to these challenges without compromising security. The technology's application extends beyond finance, proving increasingly relevant to healthcare, particularly in medical imaging and health data management. Blockchain could revolutionize the management of electronic health records, enhance data sharing across biobanks, and improve transparency and traceability in clinical trials. However, challenges such as scalability, regulatory compliance, and integration with existing healthcare data ecosystems persist [93,94].

Blockchain technology also addresses the need to enhance the representativeness and accessibility of biobanks. By facilitating secure, decentralized platforms for data exchange, blockchain supports international collaboration, thus expanding the diversity and utility of biobank resources for global research initiatives. This technology's potential extends to various healthcare applications, including chronic disease diagnosis and management, clinical trial management through smart contracts, and participant-controlled data access, showcasing blockchain as a key catalyst for healthcare innovations [94].

Implementing the FAIR (Findable, Accessible, Interoperable, Reusable) data principles is crucial for biobanks. Blockchain's inherent properties align with the FAIR principles by promoting standardized, transparent, and efficient data management practices. This not only ensures that biobank data are accessible and reusable across different research contexts but also addresses interoperability challenges, making it an integral part of advancing the FAIR data agenda in biobanking [95].

While the integration of blockchain technology into biobanking is still in its infancy, its potential to transform the field is immense. By addressing key challenges such as data security, privacy, and interoperability, blockchain stands as a beacon of innovation in biobanking. The convergence of blockchain with AI and IoT holds the promise of further enhancing patient outcomes, healthcare experiences, and the overall efficiency of biobanks. As the field evolves, ongoing research and collaboration among technologists, legal experts, ethicists, and biobankers will be crucial in leveraging blockchain technology to its full potential.

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# Data availability statement

Data will be made available on request.

# CRediT authorship contribution statement

Wanna Xu: Writing – original draft. Xiongshun Liang: Writing – review & editing. Lin Chen: Writing – review & editing. Wenxu Hong: Writing – review & editing, Project administration, Conceptualization. Xuqiao Hu: Writing – review & editing, Visualization, Project administration, Funding acquisition, Conceptualization.

## Declaration of competing interest

The authors declare that they have no completing interests.

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