Biobanking: technology basics

An introduction for ethics appraisal experts

irecs

Funded by the European Union

Contents

The aims and learning outcomes 3

Aims 3

Learning outcomes 3

What is a biobank? 4

Donating samples to a biobank 5

Types of biobanks and their uses 6

Types of biological samples and associated health-related data 8

Storage in biobanks 11

Sharing of biological samples 13

Bio sample and data sharing quiz 15

Summary 16

End of module quiz 17

Answers to the quizzes 19

Bio sample and data sharing quiz 19

End of module quiz 19

References and further resources 20

The aims and learning outcomes

Aims

The aim of this module is to support research ethics reviewers in learning about biobanking for the review of projects and proposals that include the use of biobanks. The content focuses on key technology basics, in a succinct manner, and signposts further learning opportunities for those who require more in-depth knowledge.

Learning outcomes

At the end of this module, learners will be able to:

1. Explain what is meant by 'biobank', the different types and uses.
2. Describe different types of biological sample and related data and their uses.
3. Discuss matters related to the sources, storage and sharing of biological samples and health-related data.

What is a biobank?

Biobanks are specialised repositories that collect and store biological samples and information from various sources (animals, plants, microorganisms, humans, etc.).The focus of this module is on human biobanks which collect and store biological samples from human donors (e.g., saliva, urine, blood) and health-related data (e.g., health records, family history, lifestyle, genetic, occupational, residential information, etc.) for research purposes and the development of new diagnostic procedures, preventive measures, and treatments. Some biobanks collect health-related data from donors throughout their life, and researchers may continue to access and make use of this data after donors' demise.

These repositories play a crucial role in advancing biomedical research by providing scientists with access to a diverse array of high-quality biological materials and continuously updated health-related data. Biobanks ensure the preservation of sample integrity, allowing for researchers to conduct longitudinal studies and other investigations into various health conditions at multiple scales.

For instance, by analysing samples and data, researchers can search for biological markers, investigate the relationship between biological markers and the sensitivity of diseases to treatment, the aggressiveness of diseases, progression, risk of death, as well as study the genetic and environmental factors that influence the development of certain diseases.

Legal instruments and guidance govern biobank operations to protect the autonomy and dignity of donors, along with their fundamental rights (e.g., private life and data protection) while also advancing the societal benefit of conducting research to address the key public health challenges.

Overall, biobanks contribute significantly to the progress of medical science and personalised medicine.

Donating samples to a biobank

Alison, a 42-year-old woman in robust health was invited to contribute to scientific advancement by donating biological material along with health-related information (data) to a biobank. Alison explained to her friends and family that she wanted to contribute to medical research 'to give something back' after seeing her father's successful treatment for cancer.

The biobank provided Alison with detailed information about the purpose of the biobank, the types of samples and data needed, risks and potential benefits related to the donation to biobanking and the right to withdraw consent. Alison then gave her consent to the donation process, acknowledging her understanding and agreement to participate.

Before donating the samples, Alison underwent a thorough screening process which included a review of her medical history and a physical examination. Staff at the biobank collected samples of Alison's blood, urine, and saliva.

The samples were carefully labelled, processed, and stored in secure, controlled environments. Preservation methods, like freezing or using specialised storage solutions, will maintain the long-term stability and integrity of Alison's samples for many years. Storage conditions also comply with stringent ethical and privacy protocols ensuring the preservation of Alison's privacy and confidentiality.

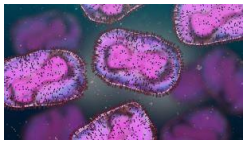
Alison was told that her personal information would be de-identified or pseudonymised to protect her identity. As part of the consent process, Alison also gave permission for the biobank to contact her in the future to follow up on her health as she ages and potentially invite her to participate in further studies beyond the scope of her original donation. Maintaining contact with donors is crucial for biobanks when samples are used for longitudinal studies or when follow-up health information is needed.

Alison's donation is now part of a valuable resource for researchers studying various health conditions, including genetic predispositions and disease markers. Her contribution will allow scientists to conduct longitudinal studies, advancing the understanding of health and potential breakthroughs in personalised medicine. Alison's decision exemplifies the vital role individuals can play in shaping the future of medical research through biobank donations.

Types of biobanks and their uses

Biobanks can be classified according to different criteria. However, they are most often classified as population-based or disease-based biobanks.

Population-based biobanks collect samples and data from a broad cross-section of the population, often with the aim of investigating the complex interplay of genetic, environmental, and lifestyle factors in the development of common diseases. These repositories provide a broad spectrum of genetic and health data, facilitating studies on population-level health trends, genetic variations, and the identification of risk factors for diseases. Population-based biobanks have been pivotal in many significant discoveries about the genetic risk factors for diseases like cancer, diabetes, and heart disease.



Disease-specific biobanks concentrate on samples from individuals affected by a particular health condition. This targeted approach aids researchers in understanding the genetic basis of diseases, developing targeted therapies, and conducting clinical trials. These biobanks provide crucial insights into the pathogenesis of diseases and are essential for the development and testing of new therapeutic interventions.

Overall, these biobank types play pivotal roles in advancing scientific understanding, improving diagnostics, and driving innovations in personalised healthcare, ultimately contributing to the evolution of precision medicine and improved patient outcomes.

In addition, virtual biobanks or virtual repositories are a relatively new management model employed in biobanking, formed of digital rather than physical repositories of biological samples and associated data.

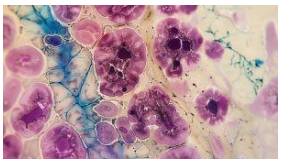
Virtual biobanks or virtual repositories are searchable central databases that leverage existing healthcare databases, linking clinical information with genetic data. This approach streamlines the collection process and fosters collaboration among researchers across different institutions. They store genetic information, clinical records, and specimens, enabling researchers worldwide to access valuable resources remotely. This virtual approach facilitates collaboration, accelerates discoveries, and enhances the efficiency of biomedical studies, ultimately advancing personalized medicine and understanding complex diseases.

Types of biological samples and associated health-related data

Promising research into the identification of proteins that can predict the onset of dementia in older age was made possible through researchers accessing data from blood samples donated by more than 50,000 healthy volunteers to the UK Biobank. Scientists see this work as a major step towards the development of a blood test that could predict the likelihood of someone developing dementia before symptoms start to show, enabling doctors to work with patients to consider treatment options.

Biobanks collect a variety of **biological samples**, each serving specific research purposes.

Blood samples: These samples are a rich source of information, containing cells, proteins, and genetic material. Researchers use these samples to study diseases, identify biomarkers, and analyse genetic variations.



Tissue samples: These samples may be obtained from biopsies from tumours or organs. They can provide insights into disease pathology, progression, and response to treatment. These samples are crucial for cancer research and understanding various medical conditions.

DNA samples: The extraction of this fundamental genetic material can be used for studying an individual's genetic makeup. Genome-wide association studies (GWAS) use these types of samples to identify links between genetic variations and diseases.

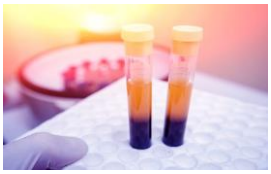
RNA samples: These samples help researchers understand gene expression patterns, providing insights into how genes are activated or deactivated in response to various conditions. This information is crucial for understanding disease mechanisms.

Urine samples: These non-invasive samples are valuable for certain types of studies. For example, they can be used to study kidney function.

Saliva samples: These non-invasive samples which are sometimes collected via a mouth swab contain genetic information for analysis.

Cerebrospinal fluid (CSF): These samples which are obtained through lumbar punctures, are crucial for studying neurological disorders and diseases affecting the central nervous system.

Cell lines: Biobanks may develop these biological cultures from tissues or blood samples. These immortalised cells can be used for long-term research, drug testing, and understanding cellular behaviour.



Plasma and serum: These components of blood are often used to study proteins and other biomolecules. These samples are essential for biomarker discovery and monitoring disease progression.

Faecal samples: Analysing the microbiome through this type of samples helps researchers to understand the role of gut bacteria in health and disease.

Researchers access biobanks' collections of samples to conduct a wide range of studies, including genetic research, biomarker discovery, disease modelling, and the development of

personalised medicine approaches. The diversity of biological samples allows for comprehensive investigations into the complexities of human health and diseases.

In addition to collecting biospecimens from donors, biobanks also collect a wide variety of health-related data. The types of data collected and stored may be categorised into 4 main categories.

Clinical and demographic Information:

- Demographics: Age, gender, ethnicity, and other demographic details.
- Medical history: General health, previous illnesses, surgeries, medications, and family medical history.
- Lifestyle information: Dietary habits, physical activity, and environmental exposures.
- Biometric data: Measurements such as height, weight, and body mass index (BMI).
- Laboratory results: Results from previous medical tests and screenings.

Follow-up data collected throughout the donor's life:

- Disease progression: Information on the development and progression of diseases.
- Treatment history: Details about medical treatments and interventions.
- Outcomes: Patient outcomes, including responses to therapies or interventions.

Genomic Data:

- Genetic sequencing data: Information derived from analysing the donor's genome.
- Genetic variants: Identification of specific genetic variations and mutations.

Imaging data:

- Radiological images: Such as MRI, CT scans, or X-rays for research purposes.

It's important to note that the specific data collected can vary depending on the focus and objectives of the biobank. The contextual information provided by clinical, demographic, and lifestyle data enhances the scientific utility of biospecimens. Researchers can correlate genetic details with health histories, enabling insights into disease mechanisms, personalised treatments, and biomarker discovery. Longitudinal data aids in tracking disease progression, contributing to better prognostics. Collecting comprehensive data alongside biospecimens ensures the quality, relevance, and ethical use of stored samples, fostering advancements in precision medicine and therapeutic development.

Storage in biobanks

Biological samples in biobanks are stored with meticulous care to ensure their preservation, integrity, and long-term stability. The storage process involves several key steps:

1. Sample processing: After collection, biological samples undergo processing to prepare them for storage. This may include centrifugation, separation, and aliquoting to obtain specific components like DNA, RNA, or serum.

2. Barcoding and labelling: Each sample is assigned a unique barcode or identifier, linking it to relevant donor and sample information. This labelling system ensures accurate tracking and minimises the risk of errors during storage and retrieval.

3. Temperature control: Samples are stored at controlled temperatures to prevent degradation. Common storage conditions include ultra-low temperatures (-80°C or -196°C in liquid nitrogen) for DNA, RNA, and tissues, or -20°C for certain proteins and blood components.

4. Cryopreservation: For long-term storage, particularly for cells and tissues, cryopreservation is employed. This involves freezing the samples rapidly to extremely low temperatures, often in liquid nitrogen, to halt biological activity and prevent cellular damage.

5. Automated storage systems: Many modern biobanks utilise automated storage systems with robotic retrieval mechanisms. These systems enhance efficiency, accuracy, and minimise the need for human intervention, reducing the risk of contamination or mishandling.

6. Quality control checks: Regular quality control checks are conducted to ensure sample integrity. This includes monitoring temperature stability, confirming sample identity through barcodes, and assessing sample condition over time.

7. Security measures: Biobanks implement stringent security measures to safeguard samples. Access to storage facilities is restricted, and environmental monitoring systems detect any deviations from optimal storage conditions.

8. Backup systems: Biobanks often have backup systems in place, such as back-up freezers and power sources, to mitigate the risk of sample loss in case of equipment failure or unforeseen events.

9. Data management: Accurate and secure data management systems track the entire lifecycle of each sample, including collection, processing, storage, and retrieval. This comprehensive documentation ensures traceability and transparency.

By adhering to these procedures, biobanks ensure that biological samples remain well-preserved, maintaining their scientific utility for years or even decades. This meticulous

approach is crucial for supporting diverse research initiatives and contributing to advancements in medicine and healthcare.

Sharing of biological samples

International cooperation among biobanks is essential for advancing scientific research, promoting data sharing, and accelerating global progress in healthcare. Biobanks employ several methods to facilitate this collaboration such as adhering to standardised protocols for sample collection, processing, and storage. Common practices ensure compatibility and comparability of data across different institutions and countries. Efforts to ensure data harmonisation ensure that information collected follows standardised formats. This harmonisation allows seamless integration and analysis of datasets from diverse sources.

Biobanks often participate in collaborative networks or consortia that span multiple countries. These networks foster communication, resource sharing, and joint research initiatives. Examples include the International Cancer Genome Consortium (ICGC) and the Global Alliance for Genomics and Health (GA4GH). Data sharing platforms that comply with established standards for security, privacy and interoperability[1] may be used to facilitate the

[1] The term interoperability refers to the ability of different systems, devices, or software to seamlessly exchange and interpret information, allowing them to work together effectively and efficiently without hindrance. It ensures that diverse components can communicate, share data, and collaborate in a cohesive manner, fostering compatibility and synergy across various platforms or technologies.

exchange of data while addressing ethical and legal considerations. Additionally, institutions engaging in international collaboration may establish formal agreements through memorandums of understanding. These agreements outline the terms of collaboration, data sharing, and the responsible use of biological samples.

Collaborative research projects involving multiple biobanks encourage the exchange of data and samples for specific research objectives. These projects leverage the collective expertise and resources of participating institutions. International collaborations may also involve capacity-building initiatives to enhance the capabilities of biobanks in developing countries as exemplified by the establishment of a biobank in Western Visayas in the Philippines. Such inclusivity fosters a more equitable and globally impactful approach to research.

Biobanks navigate complex ethical and legal considerations when sharing data internationally. They establish clear frameworks for obtaining informed consent, protecting donor privacy, and complying with diverse national and international regulations. To regulate access to shared data, international biobanks often establish data access committees that evaluate research proposals and ensure adherence to ethical, legal, and privacy standards.

Bio sample and data sharing quiz

Please answer the following questions to check your understanding of the issues relating to the sharing of biological samples. When you have finished, you can check your answers on page 19.

1. What is a primary consideration when sharing biological samples between biobanks?

a) Regulatory compliance

b) Sample quantity

c) Biobank location

d) Sample age

2. Why is data harmonisation important when sharing data between biobanks? Select all that apply.

a) To ensure that information collected follows standardised formats

b) To allow staff from different biobanks to develop good relations

c) To enable the seamless integration and analysis of datasets from diverse sources

d) To economise on the costs of data sharing

3. What is a key ethical and legal consideration when sharing data internationally between biobanks?

a) Data encryption methods

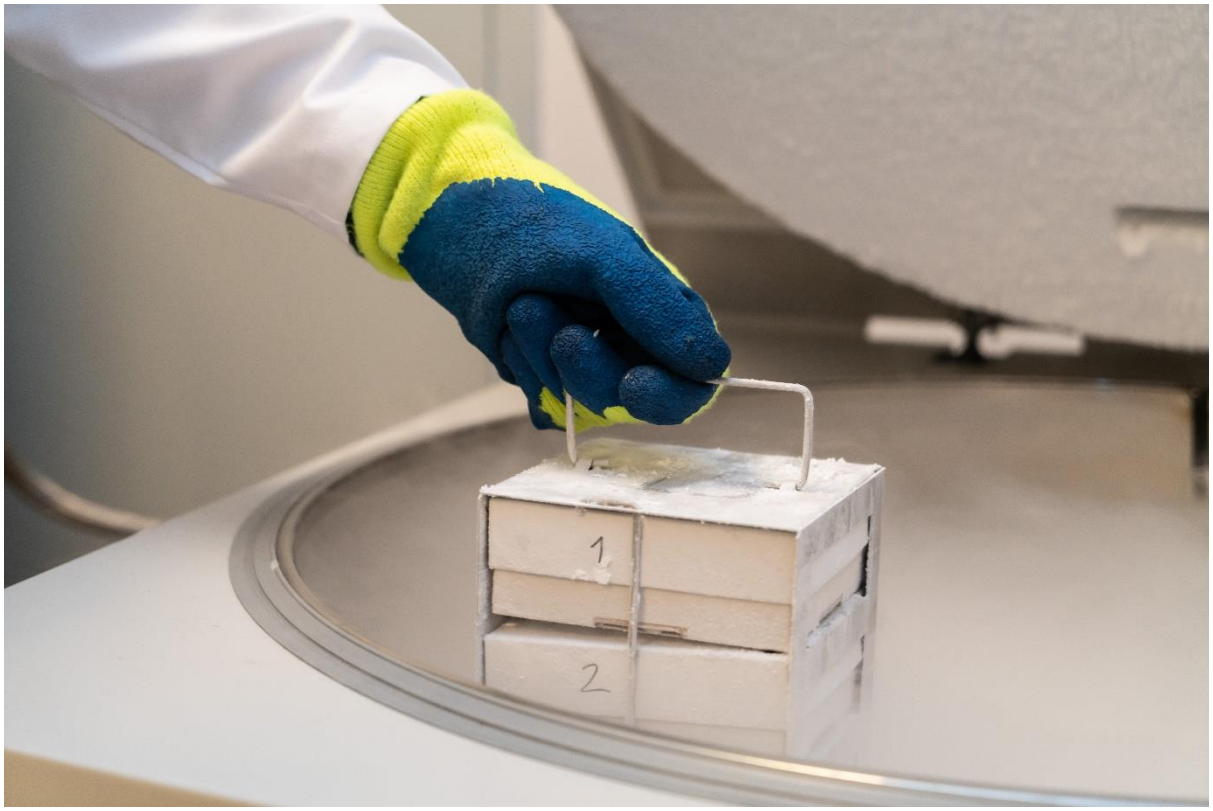
b) Harmonisation of consent processes

c) Speed of data transfer

d) Exclusive ownership rights

By employing effective strategies for the sharing of biological samples, biobanks contribute to a global network of knowledge sharing, accelerating scientific discovery, and promoting advancements in personalised medicine and public health on an international scale.

Summary



In this module we have considered key concepts associated with the use of biobanking technologies, including the different types of biobanks; the different types of biological samples and their uses; and matters related to the sources, storage and sharing of biological samples and health-related data. Now you can try the end of module quiz to see whether your learning from this module addresses the intended learning outcomes.

End of module quiz

End of module quiz

You can try these questions to see whether your learning from this module addresses the intended learning outcomes. When you have finished, you can check the answers on page 19.

1. Which of the following best describes the purpose of biobanks?

A) To conduct genetic engineering experiments on various biological samples.

B) To serve as commercial laboratories for the production of genetically modified organisms.

C) To store and manage biological samples and associated data for research purposes.

D) To facilitate the sale of biological materials to interested parties for scientific research.

2. What distinguishes population-based biobanks from disease-based biobanks?

A) Population-based biobanks aim to represent the genetic diversity of a particular population, while disease-based biobanks focus on samples from individuals affected by specific diseases.

B) Population-based biobanks store samples from individuals with a particular genetic predisposition to diseases, while disease-based biobanks store samples from healthy individuals within a population.

C) Population-based biobanks focus on collecting samples from individuals with specific diseases, while disease-based biobanks collect samples from diverse populations.

D) Population-based biobanks primarily collect samples from deceased individuals, while disease-based biobanks primarily collect samples from living patients undergoing treatment.

3. What types of biological samples are commonly collected by human biobanks?

A) Hair, nail clippings, teeth.

B) Urine, saliva, blood, tissues, DNA, RNA, cerebrospinal fluid, faecal samples.

C) Soil, water, polluted air form areas with high incidences of illness.

D) Samples obtained from food establishments with poor hygiene ratings.

4. **Why are regular quality control checks on stored biological samples important in biobanking?**

A) To ensure compliance with government regulations on biobank operations.

B) To maintain the confidentiality of donor information stored in the biobank database.

C) To monitor the effectiveness of fundraising efforts to support biobank infrastructure.

D) To verify the integrity and stability of stored biological samples over time.

5. **Why is international cooperation among biobanks deemed crucial for advancing scientific research and healthcare globally?**

A) To minimise competition among biobanks and ensure equal distribution of funding.

B) To establish trade agreements for the exchange of biological samples between countries.

C) To facilitate data sharing, promote collaboration, and accelerate progress in healthcare.

D) To prioritise domestic research efforts over international collaborations to protect national interests.

Answers to the quizzes

Bio sample and data sharing quiz

1. A
2. A and B
3. B

End of module quiz

1. C
2. A
3. B
4. D
5. C

References and further resources

Annaratone L, De Palma G, Bonizzi G, Sapino A, Botti G, Berrino E, Mannelli C, … &, Marchiò C; Alleanza Contro il Cancro (ACC) Pathology and Biobanking Working Group (2021) Basic principles of biobanking: from biological samples to precision medicine for patients. Virchows Arch. 2021 Aug;479(2):233-246. doi: 10.1007/s00428-021-03151-0. Epub 2021 Jul 13. PMID: 34255145; PMCID: PMC8275637. Available at: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8275637/

BBMRI-ERIC Common Service ELSI
https://www.bbmri-eric.eu/services/common-service-elsi/

CIOMS (2016) International ethical guidelines for health-related research involving humans
https://cioms.ch/wp-content/uploads/2017/01/WEB-CIOMS-EthicalGuidelines.pdf

Council of Europe Biobanks https://www.coe.int/en/web/bioethics/biobanks/-/asset_publisher/lcb5Z6eMEwYb/content/research-on-biological-materials-of-human-origin-new-recommendations-to-member-stat-1

Council of Europe (2016) Recommendation CM/Rec (2016)6 of the Committee of Ministers to member States on research on biological materials of human origin
https://search.coe.int/cm/Pages/result_details.aspx?ObjectId=090000168064e8ff

European Commission (2018) Data protection in the EU
https://commission.europa.eu/law/law-topic/data-protection/data-protection-eu_en

Harati, M.D., Williams, R.R., Movassaghi, M., Hojat, A., Lucey, G.M., Yong, W.H. (2019). An Introduction to Starting a Biobank. In: Yong, W. (eds) Biobanking. Methods in Molecular Biology, vol 1897. Humana Press, New York, NY. https://doi.org/10.1007/978-1-4939-8935-5_2

Healthtalk.org What is biobanking and why is it important?
https://healthtalk.org/experiences/biobanking/what-is-biobanking-and-why-is-it-important/

ISBER (International Society for Biological and Environmental Repositories) (2024) Best practices for repositories https://www.isber.org/page/BPR

NIH (2024) Genomic Data Sharing (GDS) Policy https://sharing.nih.gov/genomic-data-sharing-policy

OECD Guidelines for Human Biobanks and Genetic Research Databases (HBGRDs)
https://www.oecd.org/health/biotech/guidelines-for-human-biobanks-and-genetic-research-databases.htm

World Medical Association (2016) Declaration of Tapei on ethical considerations regarding health databases and biobanks
https://www.wma.net/policies-post/wma-declaration-of-taipei-on-ethical-considerations-regarding-health-databases-and-biobanks/