# Biobanks: A Definition

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The terms biorepository, Biological resource centre (BRC), biobank refer to structured collections of biological samples and associated data, stored for the purposes of present and future research. Both biorepositories (ISBER 2001) and BRCs (OECD 2007) can include tissues from humans, animals, cell and bacterial cultures, and even environmental samples (see below the OECD definition of BRCs), while a biobank typically handles human biospecimens—such as tissue, blood, urine—and information pertaining to the donors: demography and lifestyle, history of present illness, treatment and clinical outcomes.

## 1 Types of Biobanks

Biobanks can be established within academic medical or research institutions, pharmaceutical/biotechnology companies or as stand-alone organizations. A clear distinction among research, diagnostic and therapeutic biobanks is not always easy (i.e. cord blood stem cells, typically collected for therapeutic purposes, can be used for research, and tumor tissue samples can become the basis of tumor vaccines). Laboratories involved in cell therapy and tissue engineering clinical trials (cell factories) also handle biobanks for clinical use. However, legislation, ethical and social issues, handling of the biological specimens are remarkably different for these different types of biobanks.

Gottweis and Zatloukal (2007) differentiate between four types of research biobanks: clinical case/control based on biological specimens from patients

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with specific diseases and from non-diseased controls (e.g. pathology archives); longitudinal population based biobanks that follow a portion of the population over a large period of time (e.g. Estonian and UK Biobank); population isolate biobanks with a homogenous genetic and environmental setup of the population represented (e.g. the Icelandic Biobank); twin registries with samples from monozygotic and dizygotic twins (e.g. the GenomEUtwin and the Swedish Twin registry).

Rebulla et al. (2007) take the classification even further and differentiate between six types of biobanks: leftover tissue biobanks collected during clinical pathology diagnostic procedures; population biobanks; twin biobanks; disease biobanks form patients suffering from a specific condition; organ biobanks; nonhuman biobanks (e.g. Primate Brain Bank).

A more general distinction within the research biobank domain can be made between population based prospective biobanks (focused on the study of the development of common, complex diseases over time, and mainly based on blood/nucleic acids collection) and biobanks of tissue samples and clinical data (also referred to as disease oriented or clinical biobanks, mainly based on tissue sample collection). This classification has been used by the pan-European Biobanking and Biomolecular Resources Research Infrastructure (BBMRI), which has set two work packages on biobanks, WP2—population-based biobanks and WP3—disease-oriented biobanks.

## 2 Population Based Biobanks for Genetic Research

Genetic studies based on biobanking are becoming increasingly common as researchers recognize the need for large amounts of samples to identify the genetic basis of susceptibility to common complex diseases. Large scale population biobanking projects link genetic data with information on health status, lifestyle and environmental factors. Population biobanks have recently been defined by OECD as "collections of biological material and the associated data and information stored in an organized system for a population or a large subset of a population" (OECD 2006).

Another definition of population biobank has been given by the Council of Europe:

> a collection of biological materials that has the following characteristics: the collection has a population basis; it is established, or has been converted, to supply biological materials or data derived therefrom for multiple future research projects; it contains biological materials and associated personal data, which may include or be linked to genealogical, medical and lifestyle data and which may be regularly updated; iv. it receives and supplies materials in an organised manner (Council Of Europe 2006).

In the international context, the Public Population Project in Genomics ($P^3G$), a not-for-profitinternational consortium (Knoppers et al. 2008), plays an important role for population based genetic research, through open-access research tools

for effective collaboration between biobanks, enabling the international research community to share expertise and resources and facilitate knowledge transfer for the health of populations. The 28 charter members of $P^3G$, international, national or regional not-for-profit organizations, are conducting, or will be conducting large population genomics projects such as biobanks or large-scale cohort studies (N > 10,000 samples).

In Europe, the diversity of populations is a beneficial feature for genetic research, and centralized population-based biobanks have been established in a number of European countries, as Europe's national health care systems have facilitated collection of clinical samples and produced highly reliable health care records.

Biobanks collecting samples from twins are also part in the domain of population biobanks. Twin cohorts and Twin Registries provide a unique competitive advantage for investigations of the role of genetics and environment or life style in the etiology of common diseases. The international GenomEUtwin project[1] (Genome-wide analyses of European twin and population cohorts to identify genes predisposing to common diseases), a collaboration between Twin Registries in the Netherlands, Denmark, Norway, Sweden, Finland, Italy, UK and Australia, aims at identifying genetic variants associated with common diseases by pooling epidemiological and phenotype information from over 600,000 twin pairs, and genotype data from an ascertained fraction of those.

To overcome fragmentation, the most prominent European population biobanks are collaborating within the work package 2 of BBMRI, aimed at providing a strategy to solve the legal, governance and financial challenges involved in the Europe-wide cataloguing and storage of the vast amount of information collected in large epidemiological sample collections and population cohorts. The effort involves epidemiologists, biobankers, clinicians, experts in different fields of laboratory medicine, molecular geneticists and experts in high-throughput 'omics' technologies, with the goal of establishing an European infrastructure for collection, storage, annotation, validation and dissemination of the diverse data collected.

## 3 Disease-Oriented Biobanks

Disease-oriented biobanks (which may also be referred to as clinical biobanks) are placed at the interface between clinical practice and research. They collect biological samples from patients, aiming at discovery and validation of genetic and non-genetic risk factors of diseases. They are usually established in hospitals and research institutes, and multi-centre collections can raise from clinical trials and genetic studies. Two domains of clinical biobanks can be distinguished: tissue banks and rare disease biobanks.

[1] http://www.genomeutwin.org.

## 3.1 Tissue Banks

Tissue banking is a strategic activity for research and innovation in biomedicine in a clinical context, essential for the procurement of high quality samples for translational research. Well annotated and pathologically reviewed case-series (either based on specimens collected and processed in the course of clinical diagnostic activities or in specific tissue collection protocols) are required for identifying biomarkers and molecular targets for therapy, establishing their prevalence and formulating hypotheses on their biological and medical significance in ex vivo analyses. Validation of a potential biomarker requires applying ex vivo analyses within study designs with adequate epidemiological and statistical power; these studies may be constructed using retrospective or prospective collections. Finally, translating biomarkers into clinical practice requires applying them to large series of specimens collected using standard operating clinical protocols.

Tissue banking implies informing patients and obtaining the proper consent, data acquisition, tissue procurement, annotation, preservation, storage, cataloguing, managing of access, processing and distribution. It requires expertise in pathology, cryobiology, quality management, legal/ethical aspects, project management, administration and networking.

Specialized pathology expertise is required to identify and define the nature and origin of the tissues to be kept in the biobank. Pathologists make decisions on what should be biobanked, making sure that the requirements of clinical diagnosis and the optimal preservation of biological products are both respected. Pathology archives represent a special type of tissue repository, that may support tissue banking. The primary role of these archives is to document diagnosis and to support later diagnostic analyses, but they can play a role in research as well.

## 3.2 Rare Disease Biobanks

Rare (orphan) diseases are defined as low prevalence diseases, affecting less than one citizen in 2,000. Rare disease biobanks (also referred to as genetic biobanks) have been recognized as important tools for research and treatment. Biological samples from rare diseases (blood, tissues, cell lines, DNA) are precious, because of their rarity and diversity. This emphasises the need of transnational collaboration, quality control of the samples, training and education of scientists using the biomaterials. Quality of biomaterials and associated information rather than their quantity is critical in rare disease biobanking: small collections or even individual samples may be extremely precious for research, and have direct relevance for patients' health. Most rare disease biobanks work through the active participation of patients and patient organizations, and share benefits with them.

Orphanet, the European portal for rare diseases and orphan drugs, has developed a comprehensive disease coding system and a database listing more than 100 rare disease biobanks in Europe. EuroBioBank is a European network of

rare disease biobanks with a focus on neuromuscular disorders. In Italy, Telethon has in place a national plan for rare disease biobanking that assures high quality standards through regular assessments.

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