

BIORESOURCE PAPER

Cell Line and DNA Biobank From Patients Affected by Genetic Diseases

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The Bioresource, presently storing 10,279 biospecimens, was initially established in 1976 as a private laboratory-collection to maintain rare mutant cell lines from genetic-metabolic diseases. Shortly afterwards, however, data from the sample collection was organised in a database and the sample collection was released to the scientific community. The Biobank has received Telethon grants since 1993, as individual facility, and from 2008 as part of the Telethon Network of Genetic Biobanks (www.biobanknetwork.org).

In 2010, the Biobank has obtained official recognition from Regione Liguria. The Biobank has always provided essential services by establishing, analysing, maintaining, and distributing biospecimens from patients affected by rare genetic diseases. Up to now, the contribution of the Biobank to the scientific community has been expressed in acknowledgement notes in 145 scientific manuscripts.

Keywords: biobanking; biological resources centre; biospecimens; rare diseases; cryopreservation

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(1) Overview

Project description

The Bioresource was established in 1976 at a laboratory situated at the G. Gaslini Institute which has a longstanding tradition and internationally recognized expertise in the diagnosis of, and research into, rare genetic diseases. Therefore, interrelated laboratory activities, supported by skilled clinicians, pathologists, biochemists and molecular biologists, have allowed the establishment, over the course of years, of the Biobank which preserves well-documented biospecimens and associated data.

In fact, the Biobank was initially created as private collection to maintain rare mutant cell lines from genetic-metabolic diseases with the two essential needs to (i) have the availability of the index cases for genetic counselling and prospective prenatal diagnosis in the related families, as well as, (ii) cryopreserve precious biological material from undiagnosed patients for retrospective diagnosis. Shortly afterwards, in 1980, data from the sample collection was organised in a database and the hard-copy catalogue was promoted into the scientific community [1].

However, the Biobank made a genuine step forward, in the mid-90s, when one of the biggest biomedical Italian charity, Telethon Foundation, started to support genetic biobanks through peer review-based annual projects. Indeed thanks to these grants the Biobank has begun an

efficient service of high quality standards, with the first online catalogue in 1997 and its own home page since 2001. Another substantial change and step forward has been taking place from 2008 when the Biobank, under another Telethon project, has been one of the founding partner and coordinator centre of the first Network of Genetic Biobanks (www.biobanknetwork.org) [2].

Classification (1)

Human

Species

Human

Classification (2)

Biological samples and associated data.

Context

Spatial coverage

Via Gerolamo Gaslini, Genova, Liguria, Italy:

- Latitude: 44 degrees, 23 minutes, 29.8428 seconds
- Longitude: 8 degrees, 59 minutes, 17.6526 seconds

Temporal coverage

From 01 January 1976 to present.

Temporal coverage for accessibility

From its start to present.

(2) Methods**Steps**

To access to the biobanking service, users are requested to fill in the relevant pre-printed form to provide data of the patient/donor (e.g. personal, clinical, paraclinical, genetic data, etc.). The form is supplied by the Biobank staff or available online on the Biobank website. Signing the submission form, the user also accepts the policies and rules of the Biobank, that are: (i) providing donor's clinical and laboratory documentation; (ii) sending donor's (or legal guardian's) appropriate written informed consent; (iii) not using the sample for commercial purposes; (iv) not using the sample for another project beyond what was approved; (v) citing the Bioresource in the acknowledgements of any resulting scientific production.

Once the sample is accepted, it undergoes codification and processing. In parallel, data of the coded samples are recorded in the Bioresource database which is protected by individual password assigned by the Director to the biobank staff. Generally, data set includes personal data (name, date of birth, address, ethnic origin), sex, phenotype (affected/not affected), essential anamnestic data, presence of consanguinity, presence of familiarity, clinical anomalies, laboratory anomalies, diagnosis, type of diagnosis and centre performing diagnosis. Traceability of the samples is kept by separate clinical and paraclinical patient's data recordings.

Stabilization/preservation

Glycerol or DMSO (cryopreservation).

Type of long-term preservation

Frozen and Glycerol or DMSO (cryopreservation).

Storage temperature

- From -20°C to -180°C (liquid nitrogen)
- Storage duration: until the sample is undegraded (DNA/RNA) or viable (cell line)

Shipping temperature from patient/source to preservation or research use

Room temperature (18-25°C) and/or dry ice (-80°C) depending on the biospecimen type.

Shipping temperature from storage to research use

Room temperature (flask of cell cultures, DNA samples); dry ice (vials of frozen cell lines, RNA samples).

Quality assurance measures

The Biobank host-laboratory operates in compliance with International Standard for Quality Management System (UNI EN ISO 9000) and International Standard for Quality Management System for clinical laboratory (UNI EN ISO 15189) and Joint Commission on Accreditation of Healthcare Organization. Main measures:

- Procedures to guarantee that the cell lines are free of mycoplasma, bacteria, and fungi.
- Procedures for the sample authentication, based on genetic profile (STR).
- Systematic detection/exclusion of mycoplasma before distribution service.
- Check for DNA integrity by agarose gel electrophoresis.
- Controlled access to the cryocontainer room.
- Regular replenishment of liquid nitrogen by the assigned staff of the host "G. Gaslini" Institute.
- Freezers equipped with an alarm system to ensure an immediate intervention in case of an electrical blackout.
- CO2 incubators equipped with an alarm system to check temperature and % of CO2 variations.
- Both computer files and hard copies for data collecting undergo encoding procedure.
- Clinical revision of the patients' data chart by clinicians experts in the field.

Source of associated data

Medical records, laboratory reports, instrumental findings, results from previous studies on those samples.

Ethics statement

Consent is sought using a form approved by the local Ethics Committee. In addition, the Biobank operates in compliance with:

- Recommendations issued by the Oviedo Agreement, 1997 (Oviedo - Convention on Human Rights and Biomedicine 04 April 1997) and the Additional Protocol concerning Genetic Testing for Health Purposes (CETS n. 203), 2008 (Strasbourg, 27 November 2008).
- Telethon-SIGU guidelines, 2003.
- International Declaration on Human Genetic Data, 2003 (16 October 2003, 32nd session, the General Conference of UNESCO).
- Italian Data Protection Authority (Garante Privacy), Personal Data Protection Code, Legislative Decree no. 196, 30 June 2003, published in Official Gazette No. 174 of the Italian Republic, 29 July 2003.
- Italian Data Protection Authority (Garante Privacy), General Authorisation for the processing of genetic data, 12 December 2013, published in Official Gazette No. 302 of the Italian Republic, 27 December 2013.
- Italian Data Protection Authority (Garante Privacy), General Authorisation to process personal data for scientific research purposes, 01 March 2012, published in Official Gazette No. 72 of the Italian Republic, 26 March 2012.

Constraints

N/A

(3) Bioresource description**Object name**

Disease oriented biobank

Bioresource name

- Cell line and DNA Biobank from patients affected by Genetic Diseases
- Acronym: IGG-GB

Bioresource location

Centro di Diagnostica Genetica e Biochimica delle Malattie Metaboliche, Istituto Giannina Gaslini, Via Gerolamo Gaslini 5, 16147 Genova, Italy.

Bioresource contact

- geneticbiobank@ospedale-gaslini.ge.it
- Ph. +39 010 5636 (2792) (2609)

Bioresource URL

<http://dppm.gaslini.org/biobank>

Identifier used

N/A

Bioresource type

Genetic Biobank

Type of sampling

Disease based, sampled in clinical care.

Anatomical site

N/A

Disease status of patients/source

Patients affected and healthy/carrier relatives.

Clinical characteristics of patients/source

General data (age, gender, disease,...) and all those characteristics of interest for that given pathology (on request).

Size of the bioresource

- To date: 8,220 individuals/10,279 samples
- Average of the incoming samples per year: approx. 350 samples
- Average of the outgoing samples per year: approx. 300 samples

Vital state of patients/source

Alive at the sampling.

Clinical diagnosis of patients/source

Rare genetic-metabolic disorders: amino acid metabolism disorders, carbohydrate metabolism disorders, chromosome aberrations, connective tissue disorders, dysmorphic syndromes, hormone disorders, lipid metabolism disorders, lysosomal disorders, membrane transport disorders, metal disorders, mitochondrial function disorders, neurologic disorders, organic acid metabolism disorders, osteochondrodysplasias, peroxisome disorders, purine and pyrimidine disorders, repair defective and chromosome instability syndromes, miscellany.

Pathology diagnosis

N/A

Control samples

Healthy relatives of affected patients.

Biospecimen type

Cell lines (EBV-lymphoblasts, fibroblasts, amniocytes, trophoblast cells), DNAs, RNAs, blood, sera, plasma, other.

Release date

N/A

Access criteria

Basic rules to access to Biobank service are applied to all researchers, including Biobank staff, according to the following criteria:

- The Biobank must guarantee the storage of an adequate aliquot of sample derived from affected individuals in order to offer the possibility of retrospective analysis on the basis of more recent scientific acquisitions;
- Sample distribution only to qualified professionals working at research or medical institutions engaged in health-related research or health care;
- Appropriate justification for use (description of the research project, grant sponsor, expected results);
- Project employing the samples in agreement with the Biobank mission;
- Payment for shipping cost and a partial coverage of the cost of some basic procedures (cost recovery) related to the Biobank distribution service;
- Guarantee of proper acknowledgments in the resulting scientific publication.

(4) Reuse potential

The Biobank, being a member of a Network, manages all sample request through a shared IT infrastructure on the proper website upon users' registration. All the submitted requests are gathered in a unique Request Control Panel, a tool where each Network Partner can manage their own requests after the Committee approval of the submitted project. No samples are sent before receiving the "Material Transfer Agreement" form signed by the Principal Investigator. Published and/or non-published analytical results obtained are gathered through the relevant "Confirmation of sample use" form that the Biobank periodically sends to the Users.

Up to now, the contribution of the Biobank has been expressed in acknowledgement notes in 145 scientific manuscripts.

Author roles

- Filocamo M., Biobank Director
- Mazzotti R., Annotation of data and DNA/RNA sample processing
- Corsolini F., Cell line processing
- Stroppiano M., Quality control

- Stroppiana G., Cell line processing
- Grossi S., Collector of samples and annotation of data
- Lualdi S., Collector of samples and annotation of data
- Tappino B., Collector of samples and annotation of data
- Lanza F., Collector of samples and annotation of data
- Galotto S., Sample request and Shipping management
- Biancheri R., Clinical record revision

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References

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