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 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

**Funding Statement:** This work was supported by Telethon Foundation (grant GTB12001) and by grants from "Cinque per mille e Ricerca Corrente, Ministero della Salute".
Temporal coverage for accessibility
From its start to present.

(2) Methods
Steps
To access 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 acknowledgement 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 Gianna 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.

To date, 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
Acknowledgements

We would like to thank the host G. Gaslini Institute (Genoa, Italy). We thank Lorena Casareto, data manager of the Network coordination office, for her continuous assistance and Giovanni Premuda (SoftWerk, Genova, Italy) for the development of IT platform. Finally, the authors would also like to give special thanks to Lucia Monaco, Chief Scientific Officer and Marco Crimi, Research Program Manager of Telethon Foundation (Italy) for their support.

References

1. Filocamo, M, Superti-Furga, A, Uras, A, Borrone, C, Gatti, R and Durand, P 1982 Banca di cellule umane mutanti: interesse per la diagnosi, la prevenzione e lo studio delle malattie metaboliche. Italian Journal of Pediatrics, 8:313.

2. Filocamo, M, Baldo, C, Goldwurm, S, Renieri, A, Angelini, C, Moggio, M, Mora, M, Merla, G, Politano, L, Garavaglia, B, Casareto, L and Bricarelli, F D 2013 Telethon Network of Genetic Biobanks: a key service for diagnosis and research on rare diseases. Orphanet Journal of Rare Diseases, 8(1):129. DOI: http://dx.doi.org/10.1186/1750-1172-8-129