Patient organisation direct access to Biobanks

Marco Crimi, Ph.D.
Research Program Manager

Telethon Foundation, Italy
**MISSION, VISION AND VALUES**

**Mission**

To advance biomedical research towards cures for Muscular Dystrophy and other genetic diseases

**Vision 2015**

To convert the results of excellent, selected and sustained research into available therapies

Telethon’s “Core” activities

1. Fundraising
2. Fund allocation and management
3. Development of scientific research
ABOUT TELETHON

The Italian **Telethon** Foundation was funded in 1990 and is the 1\textsuperscript{st} biomedical charity in Italy with a focus on genetic disorders.

**Funding initiatives to date**

- Almost 2400 projects funded, >450 genetic disorders & € 354 Mi, INCLUDING:
  - Research projects
  - Intramural research (TIGEM and HSR-TIGET institutes)
  - A Career program
  - Research services and facilities, including **genetic biobanks**

**Achievements**

- 8500+ scientific publications of high average impact
- First successful and safe gene therapy carried out on 14 children affected by ADA-deficiency
  - Several new projects currently in the clinical pipeline – ref. “**Italian research on genetic diseases: worthy ideas deserve care**", F. Sofia (Poster 364 & Theme 4, May 25\textsuperscript{th} 15:00)
- Creation of the Italian platform of Neuromuscular clinical centers through a dedicated program
- Creation of the first **Italian network of genetic biobanks**
Biobanks are service units for reliable storage and professional management of biological material and corresponding clinical data.

Telethon Genetic Biobanks:
- Focused on diseases of Genetic origin
- Samples are available for research purposes only
- A portion of samples is preserved for the patients’ advantage to allow retrospective analyses

The protection of individual privacy and personal integrity must be a focus of any discussion on the collection of individual samples in Biobanks.

Some of the most relevant adopted principles:
- Recommendations issued by the Oviedo Agreement, 1997
- International Declaration on Human Genetic Data, 2003

March, 2009
THE TELETHON NETWORK OF GENETIC BIOBANKS (TNGB)

- Telethon has been funding Biobanks since **1993**
- In 2008, Telethon merged its Biobanks into a Network (**10 Biobanks**)
THE TELETHON NETWORK OF GENETIC BIOBANKS (TNGB)

- Telethon has been funding Biobanks since **1993**
- In 2008, Telethon merged its Biobanks into a Network (**10 Biobanks**)

- High quality component biobanks undergoing yearly assessments by an independent body (Telethon Scientific Committee)
- Management model according to Telethon standards
- Searchable, dynamic catalogue
- Uniform operative procedures and rules
- Integrated workflow for sample deposition and requests
- Strong interaction with patient organizations
• Telethon has been funding Biobanks since 1993
• In 2008, Telethon merged its Biobanks into a Network (10 Biobanks)
• >600 genetic diseases

More than 50,000 collected samples

MAJOR DISEASE CATEGORIES
• Neuromuscular diseases:
  - muscular dystrophies
  - congenital myopathies
  - mitochondrial disorders
  - spinal muscular atrophies
• Metabolic disorders
• Parkinson disease & parkinsonism
• Mental retardation
• Chromosome aberrations
• Skeletal dysplasia
• White matter disorders
Telethon has been funding Biobanks since 1993.
In 2008, Telethon merged its Biobanks into a Network (10 Biobanks).
>600 genetic diseases
Approved funds (2008 – 2012): 2.2 M€
Telethon has been funding Biobanks since 1993.
In 2008, Telethon merged its Biobanks into a Network (10 Biobanks).
>600 genetic diseases
Approved funds (2008 – 2012): 2.2 M€
Partial cost recovery system (users are asked to contribute for sample processing and shipping costs) has recently been implemented to enhance sustainability.

THE TELETHON NETWORK OF GENETIC BIOBANKS (TNGB)
TNGB: ORGANIZATION AND GOVERNANCE

The funding body

TELETHON FOUNDATION
Telethon Scientific Committee

Assessment with yearly reports

The decision-making body

NETWORK BOARD
Coordinator + Biobank Directors

Supported by a Coordinator emeritus
(F. Dagna Bricarelli)

The consulting body

ADVISORY BOARD
Legal, ethical and technical experts
and stakeholders

Agreed principles (ethical guidelines, governance and policies, benefits and duties) are stated in the:

TNGB CHARTER
The TNGB Biobanks professionally manage the storing of RD samples

The Biobanks collect samples from clinicians or researchers after obtaining the Informed consent forms and clinical data (a). The main benefits for the donors are:

- Samples are stored in repositories according to shared high quality Standards (SOPs)

www.biobanknetwork.org
The workflow of TNGB activities – SAMPLE DISTRIBUTION

The TNGB acts as a single virtual biobank thanks to its IT infrastructure.

The IT infrastructure manages all TNGB activities for the public website which hosts the on-line catalogue. The IT system also manages the complete in/out samples workflow.
THE TNGB COLLECTION AND RESULTS

**Cumulative samples stored**

- Start: ~30,000
- 2008: ~40,000
- 2009: ~45,000
- 2010: ~50,000
- 2011: ~55,000

**185 articles acknowledged the TNGB**

- 2008: 50 articles
- 2009: 60 articles
- 2010: 70 articles
- 2011: 105 articles

**Impact Factor:** 5.7 (averaged)

- ~5,000 samples distributed/year

**Cumulative samples stored**

- Other types: 5,000
- DNAs: 2,500
- RNAs: 2,500
- Tissues: 2,000
- Cell lines: 2,000

**Requested:**

- 2008: 3,000
- 2009: 4,000
- 2010: 5,000
- 2011: 6,000

Source: http://www.biobanknetwork.org
Preamble: the Ring14 association and the Telethon Biobank in Genoa (Galliera Hospital) signed an operative agreement in 2010

- Samples of patients affected by Ring14 started to be rapidly collected and centralized in a single biorepository

- Telethon covers the costs for the biobanking of these samples

- To date, a total of 110 DNAs, 30 peripheral blood lymphocytes, and 80 lymphoblast cell lines derived from subjects affected by Ring chromosome 14 are listed on the TNGB online catalogue

- The Ring14 samples have already been worldwide requested and are being studied in relevant scientific research projects
In February 2012, Telethon & UNIAMO signed a core agreement regarding biobanks

**Main tasks of the parties**

**Promoting the TNGB activities** within the Associations of patients, which will:
1. collect and ship the samples to TNGB
2. share clinical data with TNGB

**Facilitating the access** of patients affected by genetic disorders to the TNGB Biobanks, which will:
1. provide services of sample storage & distribution
2. inform the Association about scientific results

**Major “common” Benefit**
To improve the knowledge and solicit research on rare genetic diseases, i.e. those that have not been so far substantially addressed by the international scientific community
In February 2012, Telethon & UNIAMO signed a core agreement regarding biobanks.

Very recently, other Italian Associations of Patients have expressed their interest in collaborating with the TNGB project within the UNIAMO-Telethon framework agreement:

- Mowat-Wilson Syndrome
- invdup15 Syndrome
- Wilson’s disease
- …
EuroBioBank

EuroBioBank is the 1st operating network dedicated to rare disease research in Europe.

The EBB Network was established in 2001 by two patient organizations: AFM and EURORDIS, who has since coordinated the network up to 2011. EBB received EC support (2007-2011 within the European Network of Excellence TREAT-NMD in the FP6).

As of January 2012, Telethon has undertaken the EBB coordination. Prof. Hanns Lochmuller (Newcastle University) is the Scientific Coordinator.

The EBB Network is currently composed of 23 members (including all TNGB biobanks) from 8 European countries. Over 440,000 samples are available and can be requested via the online catalogue.

RD-CONNECT is a recently approved FP7 project (now in the negotiation phase) aimed at the development of an integrated platform in which clinical profiles will be combined with -omics data and sample availability for RD research.

- Telethon is coordinating the Biobanking WorkPackage

http://www.rd-connect.eu
THE TNGB STAKEHOLDERS

- Physicians
- Patients (beneficiaries)
- Scientists
- Regulatory / ethics
- EU partners: EURORDIS, EBB, BBMRI...
- Biobank managers
- Family associations
THANK YOU!

Name

Marco Crimi, Ph.D.
Research Program Manager

Address

Fondazione Telethon (Scientific office)
P.za Cavour, 1 - 20121 Milan (Italy)
mcrimi@telethon.it
+39.02.202217.208