Patient organisation direct access to Biobanks

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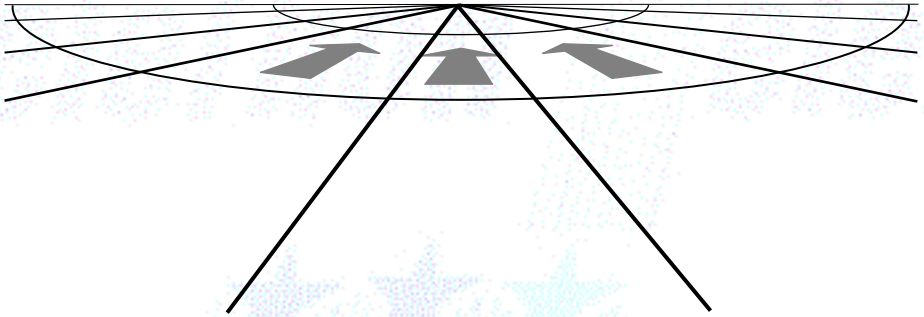
MISSION, VISION AND VALUES

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 managment***
3. ***Development of scientific research***

ABOUT TELETHON

The Italian **Telethon** Foundation was funded in 1990 and is the 1st 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 25th 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

March, 2009

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
- Italian Data Protection Authority, General Authorization for the processing of genetic data, 2007. Revised in 2011

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**)

BIOBANK	DIRECTOR	INSTITUTE
Cell line and DNA Biobank from patients affected by Genetic Diseases	**Mirella Filocamo**	Istituto Giannina Gaslini - Genova
Galliera Genetic Bank	Chiara Baldo	E.O. Ospedali Galliera - Genova
Human genetic Bank of patients affected by Parkinson Disease and parkinsonisms	Stefano Goldwurm	Centro Parkinson, Istituti Clinici di Perfezionamento - Milano
Cells lines and DNA bank of Rett syndrome and other X-linked mental retardation	Alessandra Renieri	Policlinico Le Scotte - Siena
Genomic Disorder Biobank from patients with Williams-Beuren syndrome and other genomic disorders	Giuseppe Merla	IRCCS Casa Sollievo della Sofferenza - San Giovanni Rotondo (FG)
Neuromuscular bank of tissues and DNA samples	Corrado Angelini	Venetian Institute of Molecular Medicine - Padova
Bank of DNA, cell lines and nerve-muscle-cardiac tissues	Maurizio Moggio	Centro D. Ferrari, Fond. IRCCS Cà Grande Osp. Maggiore Pol. - Milano
Cells, tissues and DNA from patients with neuromuscular diseases	Marina Mora	Istituto Neurologico Carlo Besta Fondazione IRCCS - Milano
Naples Human Mutation Gene Biobank	Luisa Politano	Seconda Università di Napoli - Napoli
Cell Line and DNA Bank of Paediatric Movement Disorders	Barbara Garavaglia	Istituto Neurologico Carlo Besta Fondazione IRCCS - Milano

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  - 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

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- **>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**

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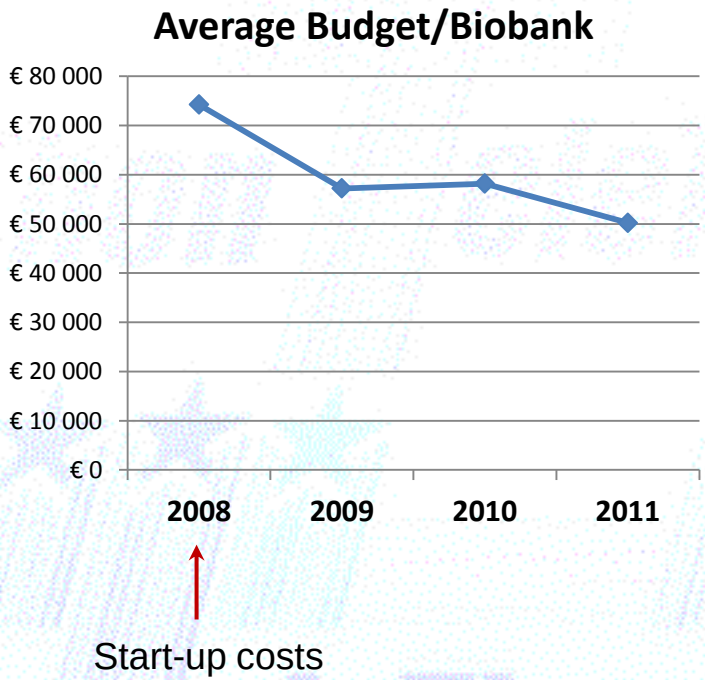
- Approved funds (**2008 – 2012**): **2.2 M€**



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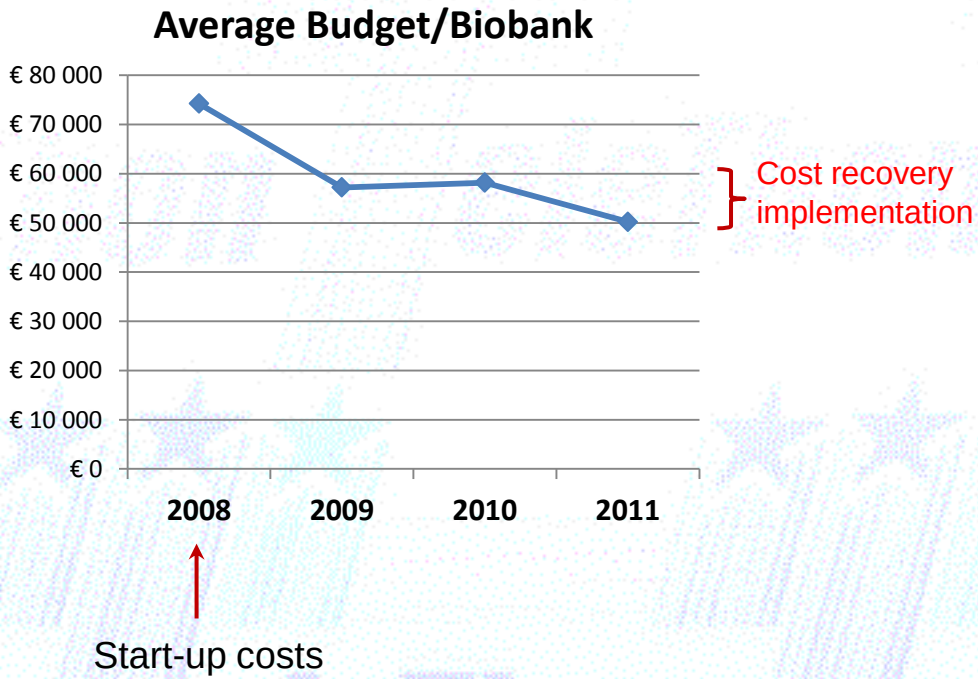
- **Partial cost recovery system** (users are asked to contribute for sample processing and shipping costs) has recently been implemented to enhance sustainability



TNGB: ORGANIZATION AND GOVERNANCE

Approval / appeal panel

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
http://www.biobanknetwork.org/page.php?ID=TheCharter

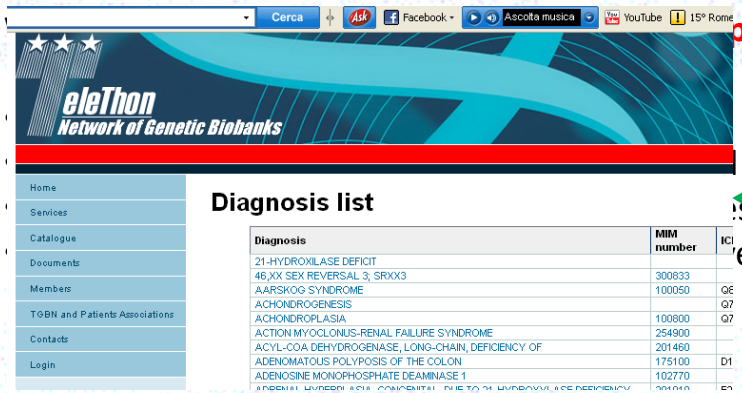
The workflow of TNGB activities – SAMPLE COLLECTION

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)

a



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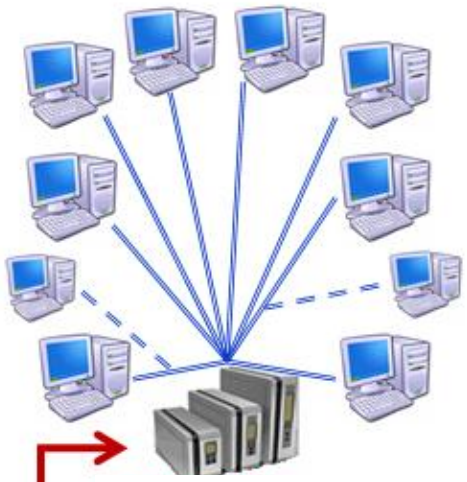
www.biobanknetwork.org

The workflow of TNGB activities – SAMPLE DISTRIBUTION

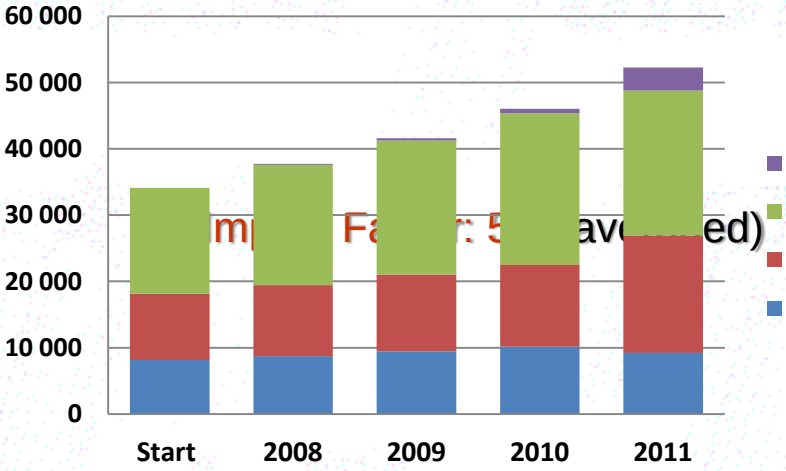
The TNGB acts as a single virtual biobank thanks to its IT infrastructure

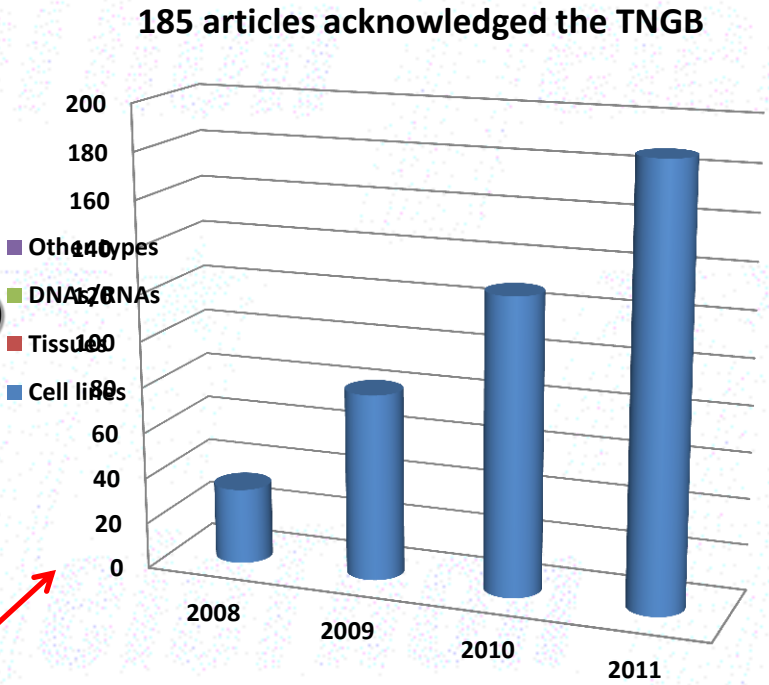
The IT infrastructure manages all TNGB activities for the public website which hosts the **on-line catalogue**. The IT system also manages the complete in/out samples workflow.

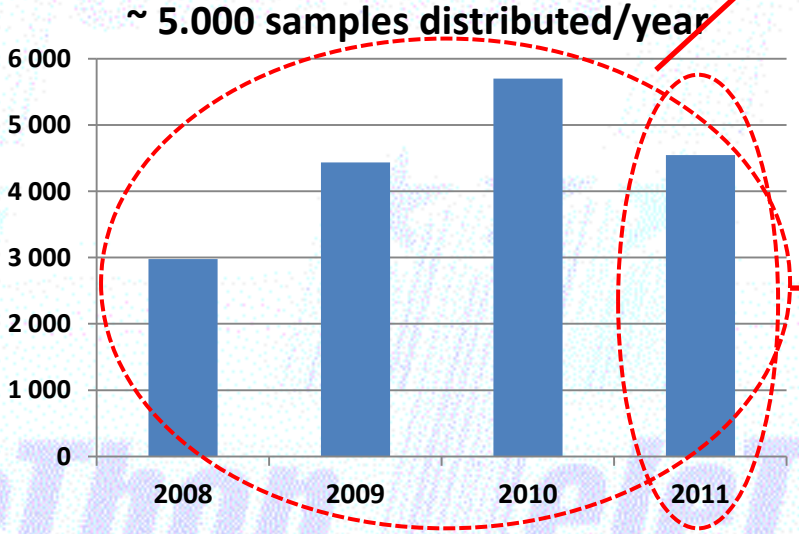


TNGB-hub

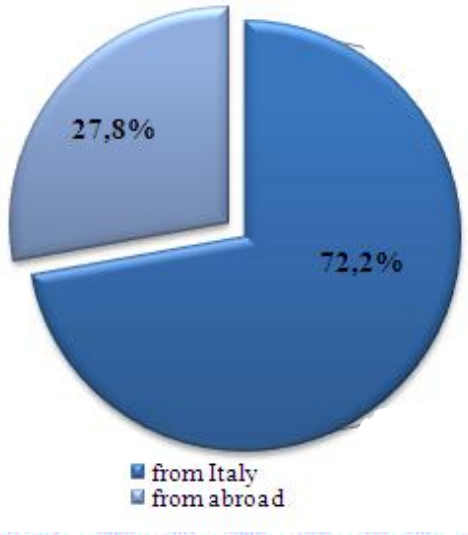
THE TNGB COLLECTION AND RESULTS







requested



Source: http://www.biobanknetwork.org

A STRATEGIC ALLIANCE

UNIAMO
FEDERAZIONE ITALIANA
MALATTIE RARE

UNIAMO is the National Italian *Federation* pooling more than100 Associations of patients affected by rare diseases

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

A STRATEGIC ALLIANCE

In February 2012, Telethon & UNIAMO signed a core agreement regarding biobanks

UNIAMO
FEDERAZIONE ITALIANA MALATTIE RARE

Telethon
Network of Genetic 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

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UNIAMO
FEDERAZIONE ITALIANA MALATTIE RARE

TeleThon
Network of Genetic 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
- …

TELETHON BIOBANK PROJECTS: INTERNATIONAL PERSPECTIVE

EuroBioBank

EURORDIS Rare Diseases Europe

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.

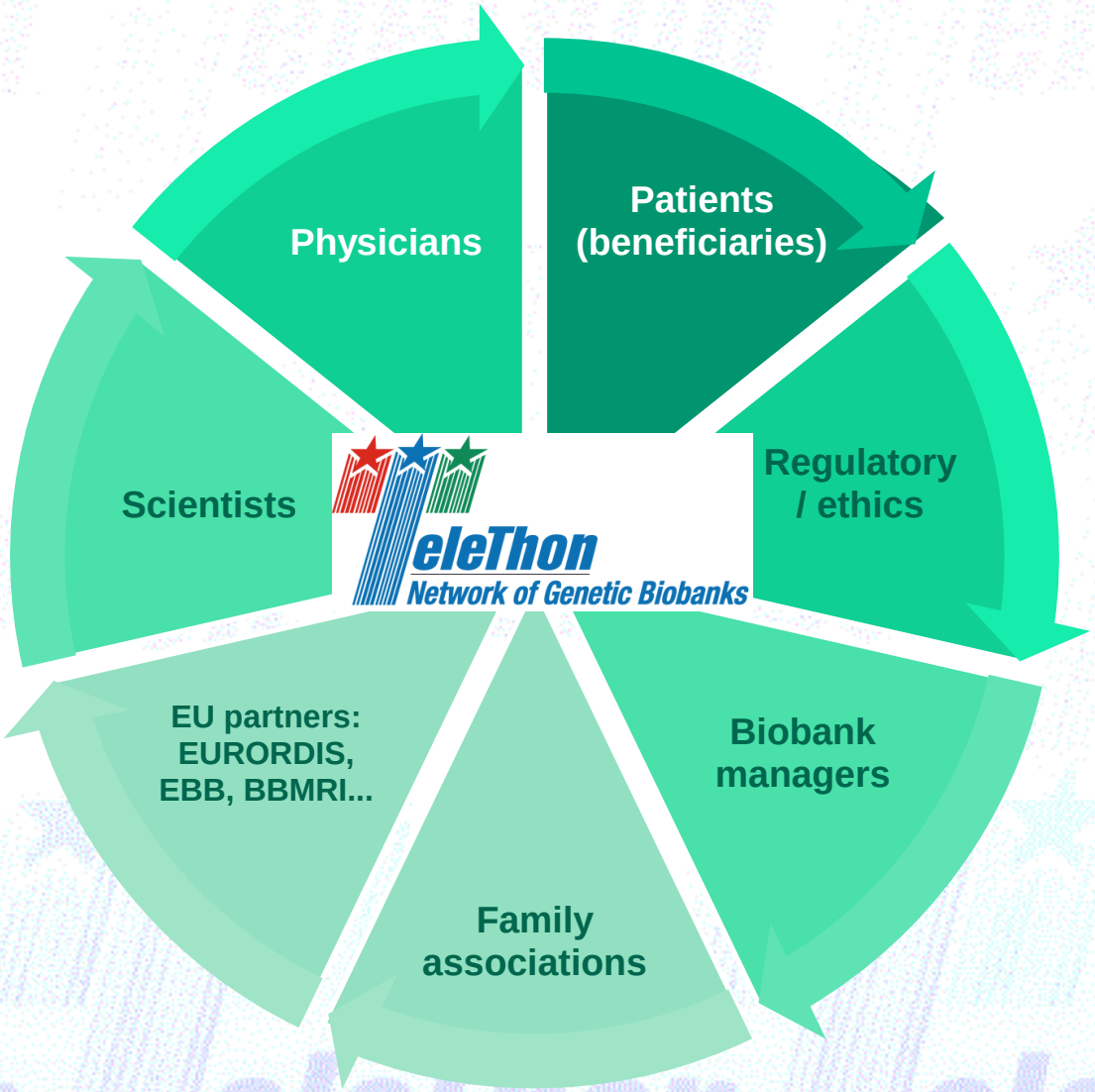
SEVENTH FRAMEWORK PROGRAMME

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



THANK YOU!

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