WECAN SmartStart in Cancer Patient Advocacy

WECAN MASTERCLASS in Cancer Patient Advocacy

Basics of biobanks and registries – types, value, challenges, relevance for patient advocacy

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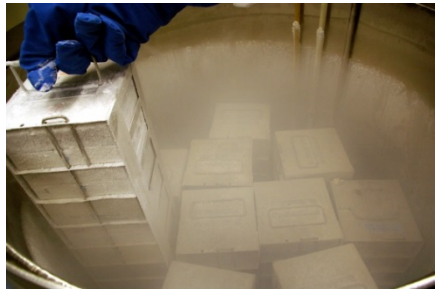
What is a biobank?

Biobank is an organised collection of human biological material and associated information stored **for one or more research purposes**



Biobanks are important research infrastructures for:

- brokering access for researchers who might not otherwise be able to access the needed materials for their research.
- Warranting the quality of the biological materials over time; support research reproducibility and openness to future research technologies.
- managing the related ethical and legal issues.

Types of biobanks

Disease-based

Contain biological samples taken from patients with specific diseases, from carriers and health control individuals. eg. Cancer, cystic fibrosis, etc

Population-based

Contain samples from subsets of a population with or without a certain disease. eg. regions, ethnics. Contain homogenous genetic material of the population.

Biobanks can vary in:

- Size
- Health status of participating individuals
- Type of biological samples stored

NOT biobanks

- Repositories of biological material having specific regulations:
  - Organs for transplant
  - Samples for therapeutic purposes
  - Blood for transfusion
  - Embryos, sperms, oocytes for IVF

- Repositories of human tissue created for diagnostic or clinical purposes
  - Guthrie cards/dried blood cards, cells for skin burns

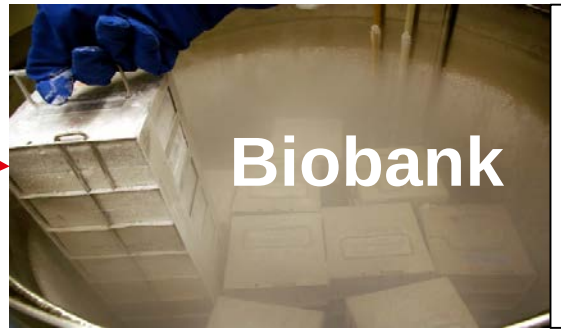
- Collections of samples and data made for obtaining regulatory approval
  - (eg. Clinical trials for new drugs)

How does a biobank operate?

Patients, Participants

Informed Consent

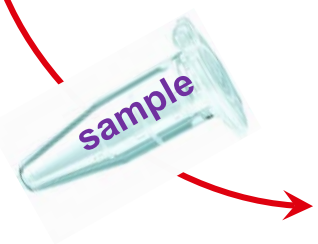
Blood, cells, DNA, urine, etc

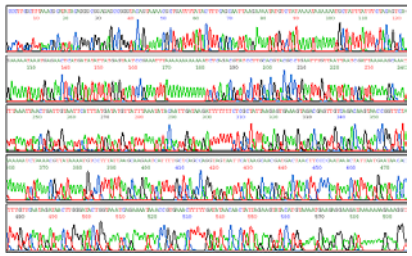


Sample Catalogue Request Form

Users

Material Transfer Agreement





Biobanks support research advancement

1999

5 male babies died in early infancy in Italian family due to a rare immunodeficiency disorder **IPEX**
(immunodysregulation polyendocrinopathy enteropathy X-linked)

Galleria Hospital, Genoa, Italy

Genetic counselling
Deposit of biological samples in a Biobank

Samples banked

2001

Scientists in USA requested these samples and identified FOXP3 as causative gene for IPEX.

Wildin et al., 2001 Nat Genet

X-linked neonatal diabetes mel
enteropathy and endocrinopat
syndrome is the human equiva
mouse scurfy

To determine whether human X-linked neonatal diabetes mellitus, e
endocrinopathy syndrome (IPEX; MIM 304930) is the genetic equivale
(*sf*) mouse, we sequenced the human ortholog (*FOXP3*) of the gene m
mice (*Foxp3*), in IPEX patients. We found four non-polymorphic
mutation affects the forkhead/winged-helix domain of the scurfin protein, indicating
that the mutations may disrupt critical DNA interactions.

Gene discovery

2006

Samples used in the development and validation of the first genetic test for prenatal diagnosis

Perroni et al., 2006 Prenat Diagn

Immune dysregulation, polyendocrinopathy, enteropathy, X-linked
(IPEX): report of the first prenatal mutation testing

Immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome (IPEX, MIM 304930) is a rare X-linked recessive disorder of immune regulation, characterized by enteropathy, eczema, anemia, thrombocy-

diagnosis. As autopsy specimens of t
had been collected, DNA could be obta
analysis using three microsatellite ma
DXS1003 and DXS1208) segregating with the syndrome

Prenatal testing

2011

Samples used in pathophysiology studies

Passerini et al., 2011 Eur J Immunol

European Journal of Immunology

Functional type 1 regulatory T cells develop
of FOXP3 mutations in patients with IPEX sy

Laura Passerini[1], Sara Di Nunzio[1], Silvia Gregori[1], Eleonora Gambineri[2], Massimiliano Cecconi[3], Markus G. Seidel[4], Giantonio Cazzola[5], Lucia Perroni[3], Alberto Tommasini[6], Silvia Vignola[7], Luisa Guidi[8], Maria G. Roncarolo[1,9] and Rosa Bacchetta[1]

Disease mechanism

What is a registry?

- an organised system that uses **observational methods** to collect uniform data on a patient population defined by a particular **disease**, **exposure** or **condition** (e.g. age, pregnancy, specific patient characteristics), and which is followed over **time**.

- Patient disease registries may be established by public organisations such as *academia* or medical *research associations* of *health care* professionals or ***patients***.

- They may have different objectives, such as:
  - describe the natural history of a disorder,
  - to monitor the efficacy or safety of treatments,
  - to describe the impact of a disease on patients' health and quality of life or
  - to identify patients suitable for new treatments.

European Medicine Agency, EMA/763513/2018

Elements to consider for a registry

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- **Patient population**: enrolment of patients and avoid selection bias.
- **Time elements**: accurate knowledge and recording of dates of important events.
- **Core data elements**: a list of core data elements to be collected in all patients is proposed.
- **Terminologies**: common terminologies for diseases, diagnostic tests, symptoms and other relevant data. Local or national terminologies should be mapped to international terminologies.
- **Quality management** – ensure data accuracy and timeliness.
- **Safety analysis**: registries conducted by organisations such as academia or medical research associations should follow the national requirements.
- **Governance**: Most registries have a governance model relying on principles and constraints based on their mandate, operating procedures, legal environment or funding sources. Principles of data ownership, informed consent and data security in accordance with the General Data Protection Regulation (GDPR).

Registry in steps of drug development

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Inform design of clinical trials

Clinicians

Support regulatory, reimbursement decisions

Regulatory Authority

Discovery → Pre-Clinical → Clinical → Market Authorisation → Post-Authorisation

Natural history studies, quality of life measures

Researchers

Identify suitable patients for trials

Industry

Safety commitments, real world

Patients

Patient organisations

Similarities between registries and biobanks

- Extremely valuable for reseach
- Collection of resources (data, biological samples)
- Require good data management and quality
- Require good governace for their operations
- Involve key stakeholders: patients, clinicians, reseachers
- National and international networks and standards
- Need to be sustainable

Patients can be involved in several steps

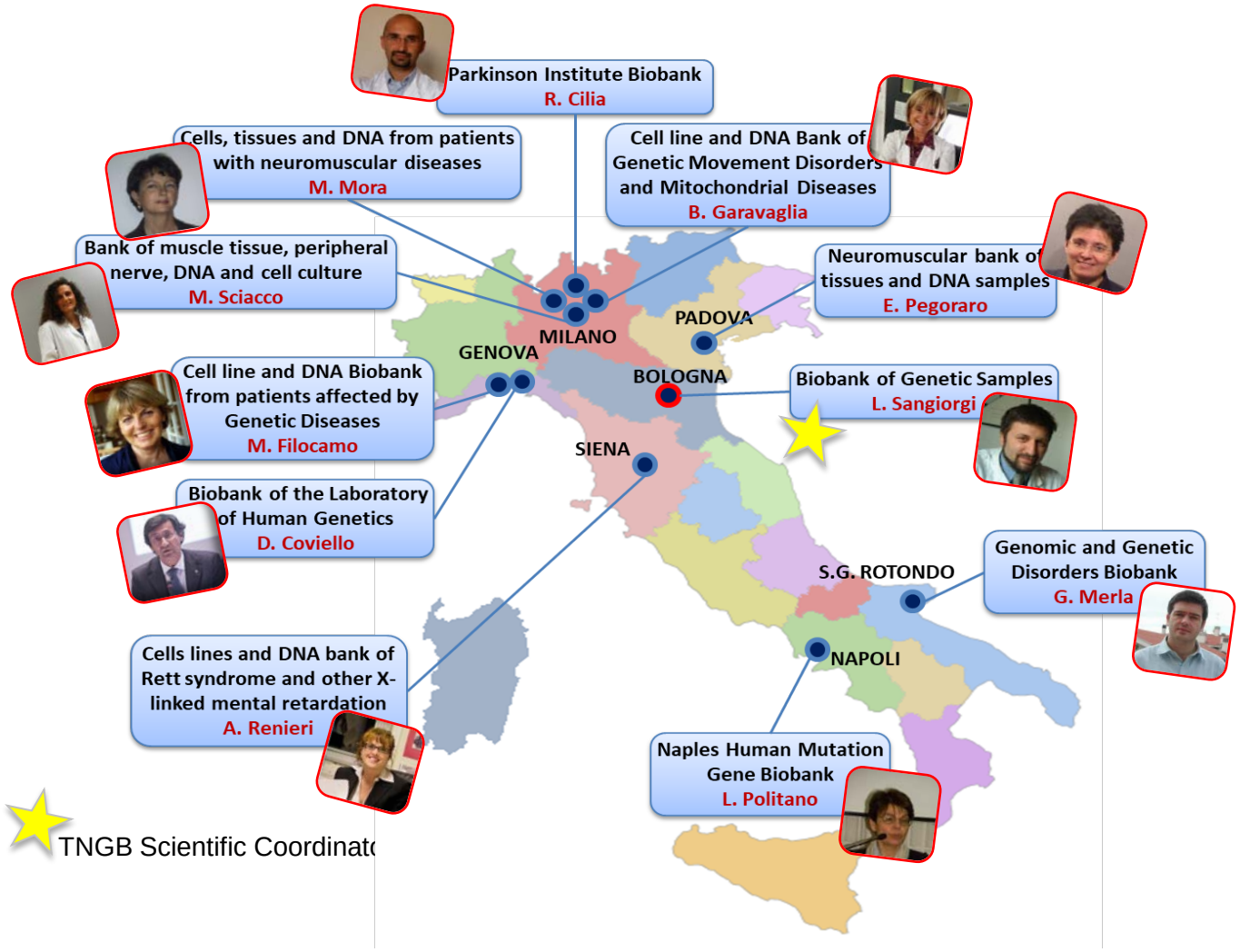
- Drafting of informed consent forms
- Governance of biobank/registry
- Creation of dedicated sample collections, databases
- Drive formation of biobank networks and collaborations

Why get involved?

- Suitable informed consent forms
- Express your needs and wishes
- Be a player in research process
- Advance research!

Case I: Co-creation with biobanks

Telethon Network of Genetic Biobanks



http://biobanknetwork.telethon.it/

123,000 biological samples in the catalogue

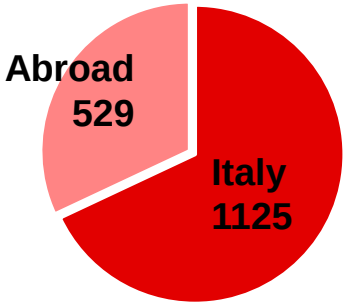
1,500 genetic diseases

24 types of samples stored (DNA, RNA, tissue, blood, plasma, cell lines, etc)

5000+ aliquots distributed per year

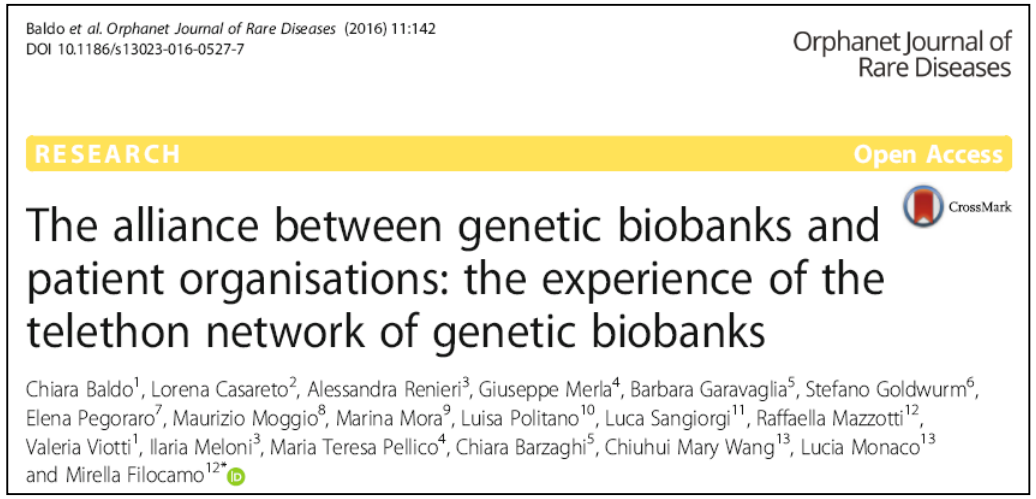
611 scientific articles acknowledged TNGB as source of biological materials (2008-2018)

TNGB registered users (2018)



Collaboration between patient organisations and biobanks

Opportunity for patient organisations to set up sample collections related to their diseases.
Formalisation of collaboration for dissemination and sample collections.


Baldo et al. Orphanet Journal of Rare Diseases (2016) 11:142
DOI 10.1186/s13023-016-0527-7

Orphanet Journal of Rare Diseases

RESEARCH Open Access

The alliance between genetic biobanks and patient organisations: the experience of the telethon network of genetic biobanks

Chiara Baldo[1], Lorena Casareto[2], Alessandra Renieri[3], Giuseppe Merla[4], Barbara Garavaglia[5], Stefano Goldwurm[6], Elena Pegoraro[7], Maurizio Moggio[8], Marina Mora[9], Luisa Politano[10], Luca Sangiorgi[11], Raffaella Mazzotti[12], Valeria Viotti[1], Ilaria Meloni[3], Maria Teresa Pellico[4], Chiara Barzaghi[5], Chiuhui Mary Wang[13], Lucia Monaco[13] and Mirella Filocamo[12*]

Co-creation
Education & Training

Acknowledgement for the co-created collections

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ACKNOWLEDGMENTS

The financial support of Telethon Italy (grant GEP 14131) is gratefully acknowledged. A.R.P. is supported by the National Institute of Neurological Disorders and Stroke under award K08NS078054. The authors thank the Galliera Genetic Bank, member of the Telethon Genetic Biobank Network (project GTB12001), funded by Telethon Italy, and the Associazione Italiana Mowat Wilson ONLUS (AIMW), for assistance in managing patients' samples. We also thank all cooperating family members for contributing the medical and neuroimaging data necessary for this study. In addition, we are grateful to Luca Valcavi for helping with the design and adaptation of the figures and tables.

Official journal of the American College of Medical Genetics and Genomics **ORIGINAL RESEARCH ARTICLE** Genetics inMedicine

Open

Neuroimaging findings in Mowat–Wilson syndrome: a study of 54 patients

Garavelli L et al., 2016

Research-ready resources for patient-led Calls

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GRUPPO FAMIGLIE Dravet ASSOCIAZIONE ONLUS

CALL OF PROPOSALS 2019

The Associazione Gruppo Famiglie Dravet (GFD), Italy is dedicated to support the highest quality basic or clinical research on Dravet syndrome (DS). In pursuing its objective, this year, GFD will select and support **research projects** aimed at understanding the pathogenesis of Dravet syndrome and at paving the way to new treatments that will ultimately lead to better quality of life for people with DS. **Funds were made available thanks to contributions from the GFD and the Swiss Dravet Syndrome Association (SDSA).**

If necessary, access to relevant, well-characterized patient populations or suitable biomaterial collections must be demonstrated. In this regard, please know that the GFD has an agreement with the Telethon Network of Genetic Biobanks (**TNGB**), by which biological samples and related clinical data from individuals with Dravet syndrome are collected, preserved and offered to the scientific community. Interested applicants can refer to the online catalogue of the **TNGB**, to identify biosamples to employ in their proposed research.

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Case II – Be a part of governance

Ambrosini *et al. Orphanet Journal of Rare Diseases* (2018) 13:176
https://doi.org/10.1186/s13023-018-0918-z

Orphanet Journal of Rare Diseases

RESEARCH Open Access

The Italian neuromuscular registry: a coordinated platform where patient organizations and clinicians collaborate for data collection and multiple usage

Anna Ambrosini[1*], Daniela Calabrese[2], Francesco Maria Avato[3], Felice Catania[4], Guido Cavaletti[5], Maria Carmela Pera[6], Antonio Toscano[7], Giuseppe Vita[7], Lucia Monaco[1] and Davide Pareyson[2]

Slides curtesy of Dr Anna Ambrosini, Fondazione Telethon

https://doi.org/10.1186/s13023-018-0918-z

The Italian Neuromuscular Registry platform

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- "Associazione del Registro" (**ADR**), a legal entity founded in 2009 by Fondazione Telethon and several neuromuscular patient organizations, with the goal to involve patient organisations in the governance.

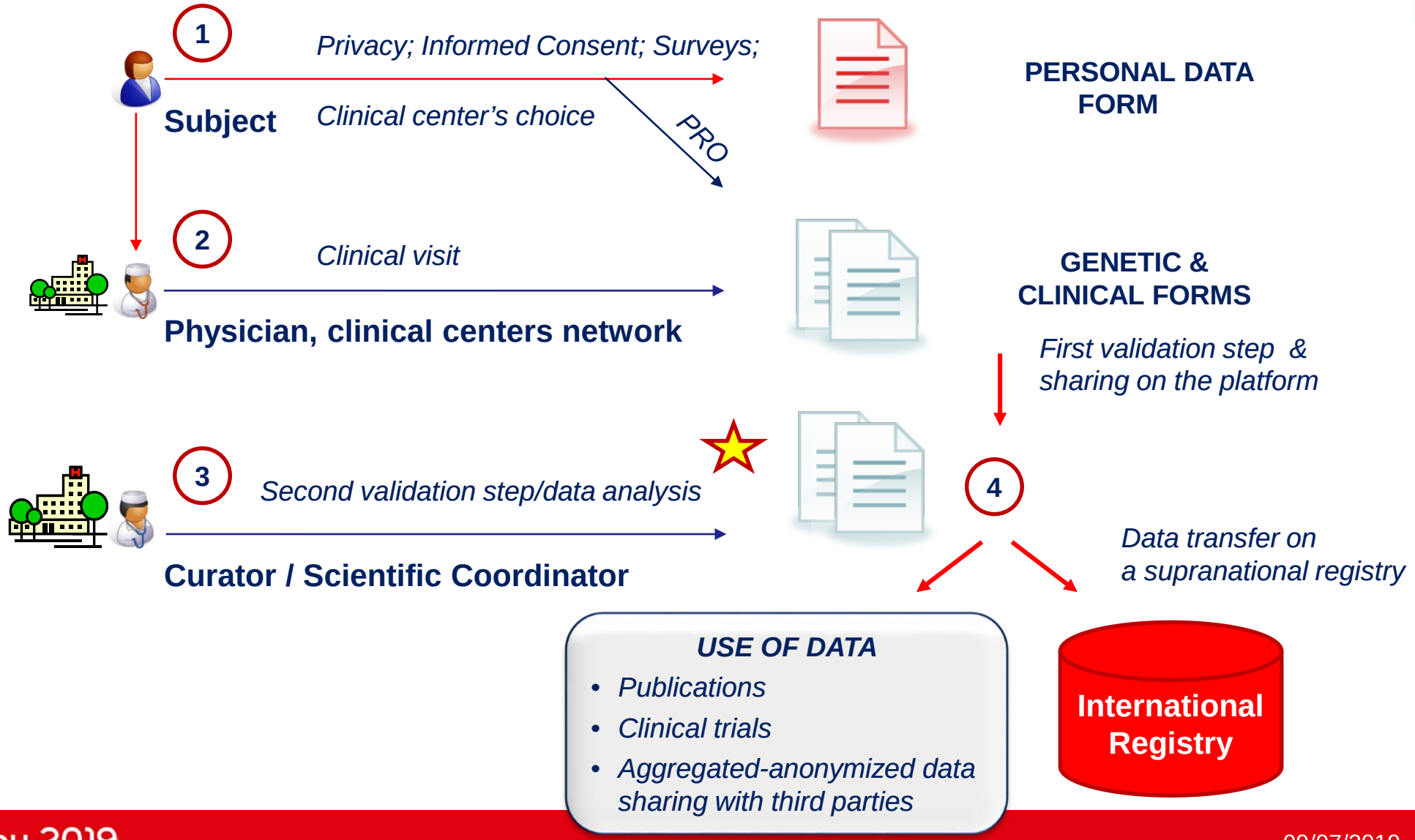
- Disease registries, data on 2000+ individuals living with a neuromuscular disease condition

- Current members
  - ACMT-Rete (Charcot Marie Tooth Disease)
  - AISLA (Motor Neuron Disease)
  - ASAMSI (Spinal muscular atrophy)
  - Famiglie SMA (Spinal muscular atrophy)
  - UILDM (Muscular Dystrophy)
  - Fondazione Telethon

- Stewardship and data sharing according to legal & ethical principles
- Equal rights and responsibilities among members
- Inclusion warranty to patients, patient organisations, clinicians

www.registronmd.it

Standard Operating Procedures



ADR Governance

Executive Board

(a representative from each participating member)

Access Board

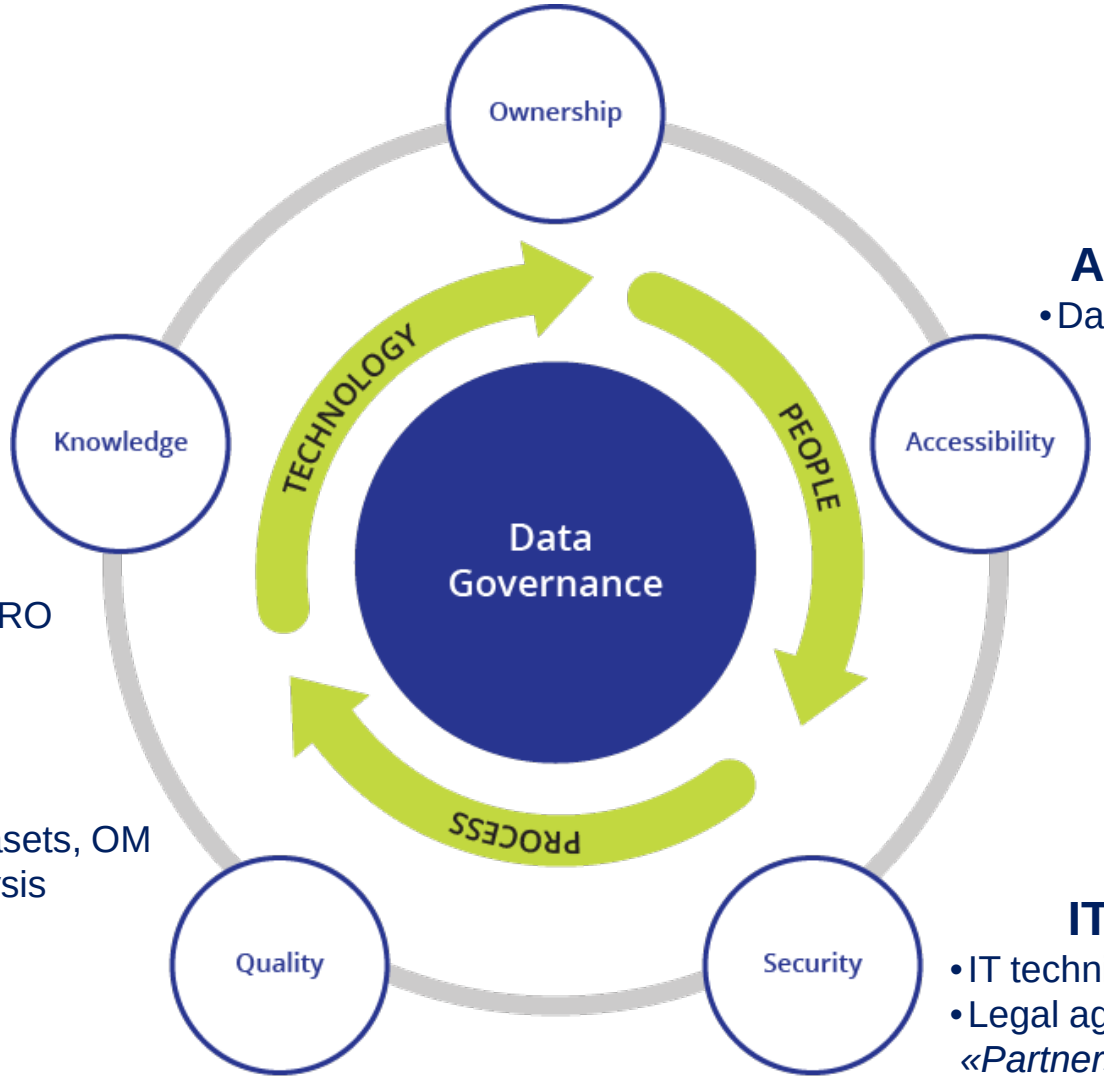
(independent; ethics, legal and scientific competence)

Scientific coordinators and registry boards

(Coordinator and PIs of the clinical centres)

ADR

- Ownership of the process
- Stewardship of the data
- *Pt organisations'empowerment*



Access Board

- Data sharing approval

Patients

- Active participation in PRO

Clinicians

- Up to date minimal datasets, OM
- Data validation & analysis

IT supplier

- IT technical solutions
- Legal agreement with ADR *«Partner»*

Take home messages

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- Biobanks and registries are fundamental resources for research
- Biobanks and registries share many common operational features

- Require collaboration with multiple stakeholders to ensure their success
- Management and sustainability can be challenging

- **Patients can and are being involved in many aspects of rare disease registries and biobanks**