II INTERNATIONAL SUMMER SCHOOL

Rare disease and orphan drug registries

Reporting and Dissemination

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Organised by Istituto Superiore di Sanità

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Scientific Research Excellence

- Necessary to improve health
- Substainability supported by the quality (trustable results, solid base for innovation)
- Coverage of European populations/ global /patients
- Interoperability
- Ethical and legal compliance
- Quality of tools used
- Quality of tools developed (services)
- Reduce duplication and improve research quality
- Policy of dissemination: the “open (access, open data etc) paradigm” help to “feel the gap” in translational medicine

To evaluate excellence we need indicators and tools

Output indicators of excellence

- Publications
- Patents
- Trained highly skilled people
- Healthcare innovation (SMEs, Pharma R&D in Europe)
- Improved healthcare

Indicators of scientific research

Some of the indicators used in the evaluation of scientific research are:

- Impact factor
- Article citations
- Journal citations

➢ **Right reference citation and retrieval of a contribution at the base of evaluation**

HEALTH RESEARCH EXCELLENCE: REPORTING, DISSEMINATION and SCIENCE QUALITY

Plan

To Plan: experimental study

Quality of published available literature is essential in research project design

- Overview **existing knowledge on topic** :

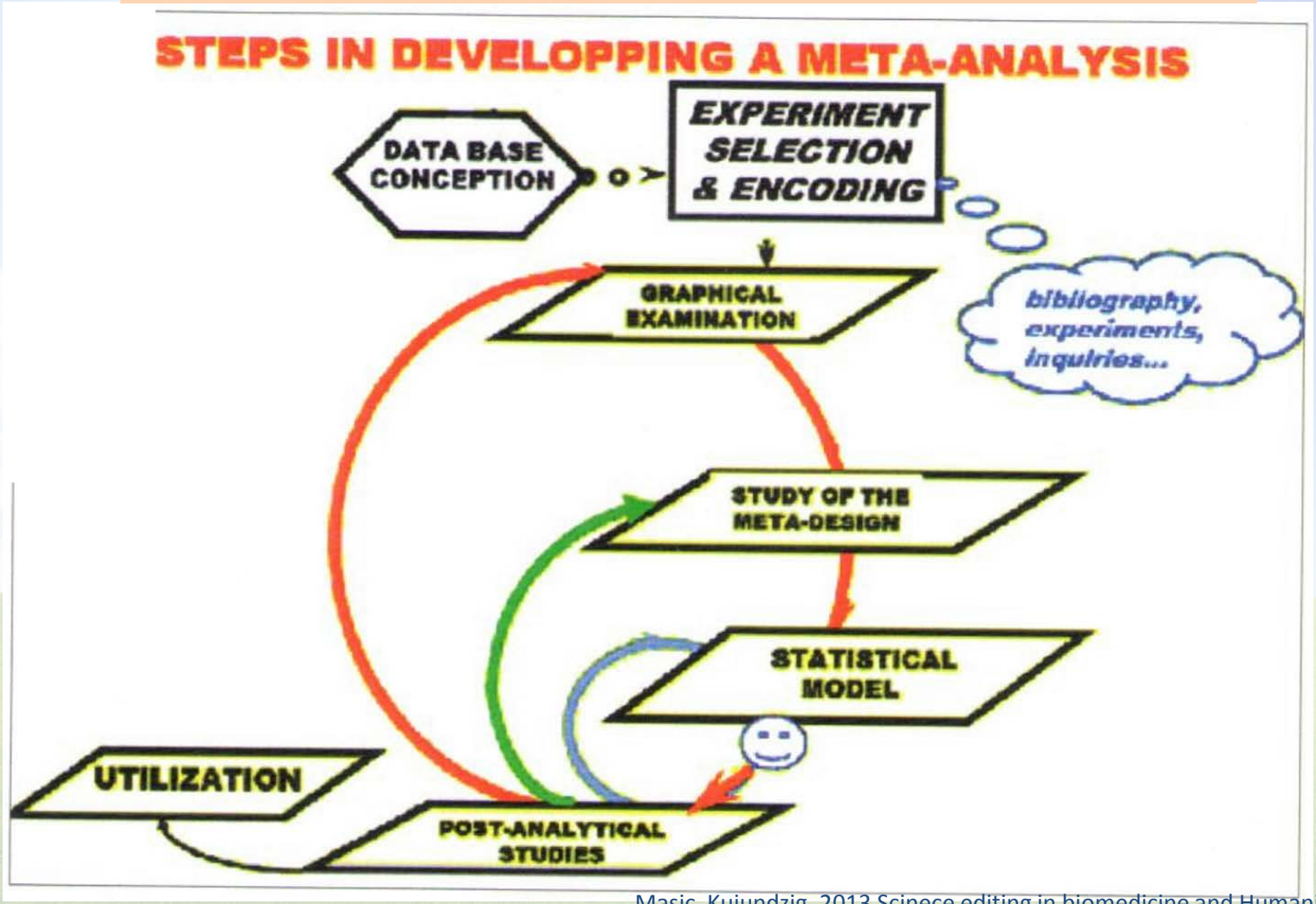
  - Search on suitable website (i.e: health disciplines: WHO, Medline, Embase, ISI Web of Science, CINHAL, ScopeMed, PsycINFO, etc.)

- ***Literature*** :
  - Trustable journal
  - Easily retrieval (key words, consolidated journal)
  - Well written (format, content)
  - Allow reproducibility of experimental work
  - Easy to read the articles (Editorial policy, online access etc)

To Plan: literature-based studies

Plan

Secondary studies: the existing data sets, review, meta-analysis



Masic, Kujundzig. 2013 Scinece editing in biomedicine and Humanities, Avicena,

To Perform

Main factors affecting quality f the performance of the research:

- Infrastructure / organization factors
- Funding
- Instrument
- Human resource
- Management /organization

- Methodologies (depending as well as from literature)
  - adopt the principles of Good Scientific Practice (GSP) and Good Laboratory Practices (GLP)

- Tools / Services for the research (Database, Biosamples, Health data) in other words **Bioresource quality**

Biological Resource

- **1992, United Nations adopted Convention on Biological Diversity (Rio Convention)**

1999, *OECD* (Organisation for Economic Co-operation and Development) Workshop on Scientific and Technological Infrastructure – Support for BRCs (quoted from OECD 2001, p12)

OECD* "...consist of service providers and repositories of the living cells, genomes of organisms, and information relating to heredity and the functions of biological systems. BRCs contain collections of culturable organisms (e.g. micro- organisms, plant, animal and human cells), replicable parts of these (e.g. genomes, plasmids, viruses, cDNAs), viable but not yet culturable organisms, cells and tissues, as well as databases containing molecular, physiological and structural information relevant to these collections and related bioinformatics."

Mesh term

Biological Specimen Banks [N02.278.065]

Facilities that collect, store, and distribute tissues, e.g., cell lines, microorganisms, blood, sperm, milk, breast tissue, for use by others. Other uses may include transplantation and comparison of diseased tissues in the identification of cancer.

- Blood Banks [N02.278.065.200]
- Milk Banks [N02.278.065.600]
- Sperm Banks [N02.278.065.700]
- Tissue Banks [N02.278.065.900]

BIORESOURSES: a tool for research

The **bioresource** is the assets/patrimony for the research which includes the **biological sample**, the **epidemiological and clinical data** associated with the sample, (including **omics**, and investigations based on **imaging** and on **sound** for clinical purposes)

The bioresource, if of quality, is a patrimony with an increasing capital: its value increases along with the quantity and quality of the associated information

Biobanks and physical biosamples repositories are the keepers/custodians of physical bioresources, having the role of warrantors and being responsible for sample management as well.

Portal, dataset , registries platforms are custodians and manager of scientific basic knowledge for excellent development of science.

The International Critical Need for Quality Biospecimens

As part of the planning phase for the National Cancer Institute's (NCI) new national biospecimen resource, called **caHUB, the Office of Biorepositories and Biospecimen Research conducted a survey** of researchers funded by NCI, as well as federal agencies, cancer centers, industry, foundations, and advocacy groups about the need for quality human biospecimens. The survey results indicated that a**cquiring quality biospecimens was a major barrier to progress in many research areas, including discovery and validation of new diagnostic assays**.

NCI Launches National Biobank
Clinical Laboratory News March 2010: 36 (3).

To Perform: Quality of the bioresource

Quality of data / information / reporting /tool (Reliable tools) [*Dataset, Registry, Biobank*]

- Efficiency for updating the data (including patients follow up) [*Dataset, Registry, Biobank*]
- Quality of samples (DNA / RNA /biological fluids, tissue
- Sample quantity (i.e.: available nucleic acids)
- Morphological control
- Professional Management
- Ethical standards
- Certification and accreditation
- Policy access (Fear, Clear, no Time consuming etc)

Reporting: primary publications

Report

A "**Primary contribution**" in a published journal.

- "original" contribution : any research report /original opinion about primary (experimental, epidemiologic) study;
- based on existing /data studies , but express the author work (review, editorial, commentary etc)

Biomedical journals can be divided into four main categories:

1. Narrow specialized journals;
2. General medical journals;
3. Classic journals;
4. Primary scientific journals.

The major **catalogues of existing scientific published journals** are:

- JCR: Journal Citation Reports
- DOAJ: Directory of Open Access Journals

Reporting: secondary publications

Report

Secondary publications basic task was to monitor the rate of appearance and development of primary publications regardless of the forms.

The common characteristics of secondary publications is that these publications **point to the information provided by the primary publication (give the description and content), and facilitate the choice of proper primary publication**.

Nanopublications for single, attributable and machine-readable assertions in scientific literature.

Nanopublication, generally refer to **microattribution** (a form of data citation) defined as "a scholarly contribution smaller than a journal article being ascribed to a particular author" or *a small scholarly contribution being ascribed to a particular author*.

Reporting: writing an article

Type contribution:

research article, review, quote, strategy, review, guidelines, clinical trials, editorial, letter, posters and presentations etc.

- **Choice:** check relevance of your contribution and choose the right type of contribution
- **Pre-defined contribution form**: poster, abstract, special issue

Structure of the article

The most adopted basic structure is:

- I–Introduction,
- M–Methods (Methods and/or Materials),
- R–Results,
- A–and
- D–Discussion and Conclusion.

For didactic reasons is formed the acronym **IMRAD.**

- References or Citation list

Tools for good reporting: Instructions for authors

The International Committee of Medical Journal Editors (ICMJE)

This Committee is a group of medical editors and representatives of selected related organizations who improve the quality of medical science and its reporting .
Their recommendations for the Conduct, Reporting, Editing, and Publication of Scholarly Work in Medical Journals, is named **Uniform Requirements for Manuscripts Submitted to Biomedical Journals (URM) .**

European Association of Science Editors (EASE)

EASE is an internationally community of individuals from diverse backgrounds, linguistic traditions and professional experience.
The **EASE Guidelines for Authors and Translators of Scientific Articles to be Published in English** are a useful, easy tool for Authors at international level, thanks to their translation in 20 languages at international level .

Committee on Publication Ethics (COPE)

COPE is a forum for editors and publishers of peer-reviewed journals to discuss all aspects of publication ethics. It also advises editors on how to handle cases of research and publication misconduct. Resources available from COPE include "Code of conduct" for Editors, publishers and reviewers.

Tools for good reporting: guideline

Reporting guidelines: Can their use make the work of systematic reviewers and guideline developers better?

In recent years, application of rigorous methodology in the development of systematic reviews and clinical guidelines has triggered more intensive scrutiny of published health research. The need to critically assess methodological quality of studies, examine possible biases and compare findings, beneficial or harmful, across different studies has highlighted serious shortcoming in primary research reporting. These deficiencies hamper the development of systematic reviews, which subsequently impacts on the development of clinical guidelines and ultimately on patients' care. Unfortunately, systematic reviews themselves are not immune to reporting shortcomings or indeed shortcomings in their conduct.photo3big

Reporting guidelines are tools developed to aid accurate and complete reporting of key aspects of research studies. In 2008, the **EQUATOR (Enhancing the QUAlity and Transparency of health Research) Network** was launched.

Quality of reporting

EQUATOR supports wider practical implementation of reporting guidelines by all relevant parties to increase the usability and value of health research.

equator network

Enhancing the QUAlity and Transparency Of health Research

Visit the EQUATOR Spanish Website

Home | Library | Toolkits | Courses & events | News | Blog | About us | Contact

The resource centre for good reporting of health research studies

Library for health research reporting

The Library contains a comprehensive searchable database of reporting guidelines and also links to other resources relevant to research reporting.

Search for reporting guidelines

Visit the library for more resources

Key reporting guidelines

CONSORT	Full Record | Checklist | Flow Diagram
STROBE	Full Record | Checklist
PRISMA	Full Record | Checklist | Flow Diagram
STARD	Full Record | Checklist | Flow Diagram
COREQ	Full Record
ENTREQ	Full Record
SQUIRE	Full Record | Checklist
CARE	Full Record | Checklist
SAMPL	Full Record
SPIRIT	Full Record | Checklist

Dissemination: Author consideration submitting a manuscript to a scientific journal

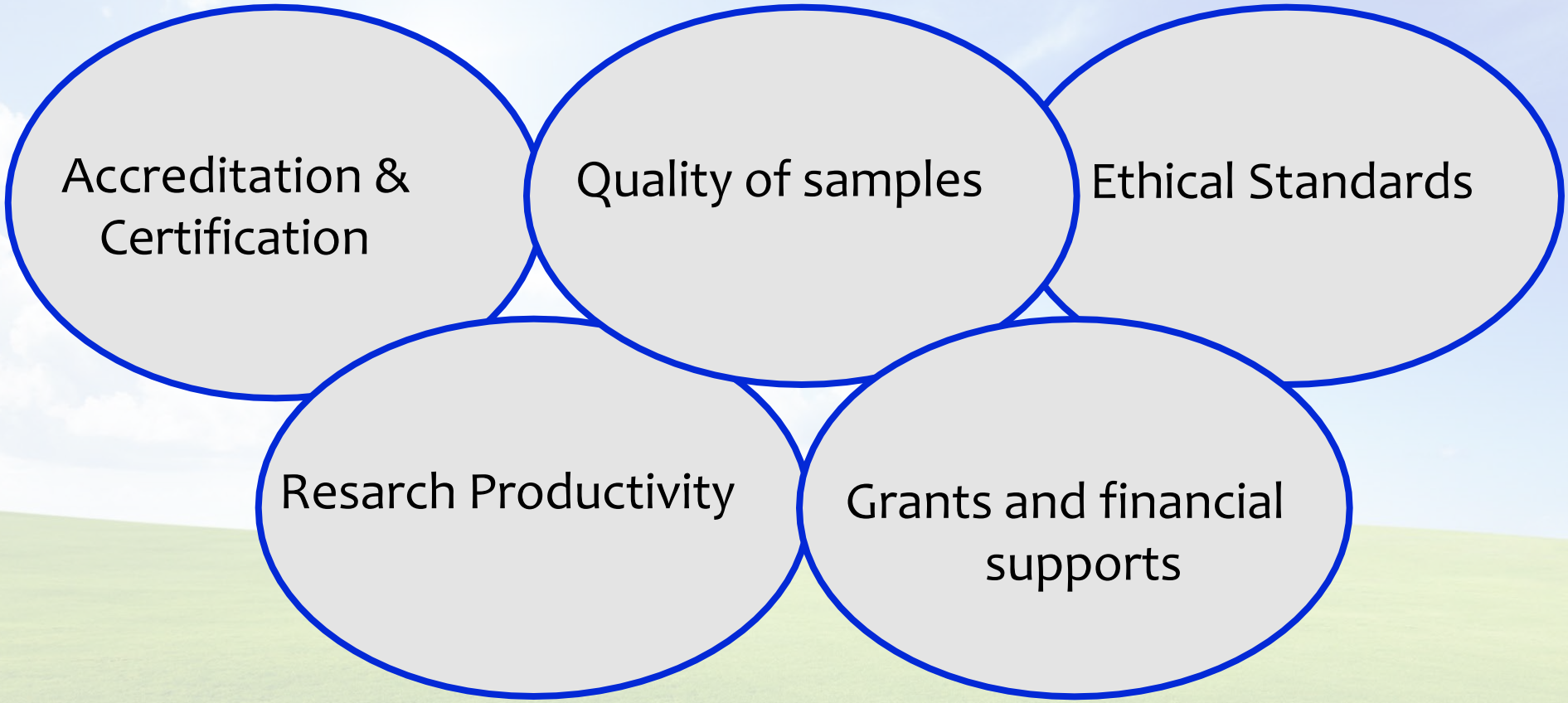
Journal prestige	Impact factor; Inclusion in several databases; Consolidated standards, trustworthy issuing, Confidence in Peer-reviewed process
Target audience	Journal subject category; Journal audience (i.e. base, translational, restricted niche)
Dissemination	Fast and capillary deliver, Traceability by major search engines, Included in major indexing databases, accessibility of articles
Costs related to publishing	Subscription costs. Costing for OA publication (paid OA option). Manuscript submission fee. Additional charges f (tables, color etc). Relation between costs and other factors such as copyright policy,
Copyright issues	Management of intellectual property rights. Self-archiving policy (permission granted to authors to post articles in public repositories, embargo period). Negotiation of copyright conditions (i.e. SPARC addendum).
Publishing practice and process	Familiarity with a certain publisher or specific journal. Acquaintance with the editorial board of a journal. Clarity of instructions for authors. Friendliness of manuscript submission system. Traceability of manuscript submission process. Speed of publication process (time lapse between submission and publication).
Services offered by a journal	Citation service, monitoring access (i.e. web usage statistics, online counts). Technical quality of reprints and/or online display of articles (i.e. layout, design, editing, print), friendly access to the journal ; Language. Facilities provided by publishers to support authors (i.e. translating services). Contents Sharing through social networks Access through mobile applications .

With minor changes from.: Poltronieri, Bravo et *et al. JECCR* 201J3, 32:- 34

HEALTH RESEARCH EXCELLENCE, SCIENCE QUALITY AND BIORESOURCE CITATION

BRIF
Bioresource Research Impact Factor

Bioresource-based research
Criteria to get a high impact on health



Toward a high Bioresource Research Impact factor

Biological Resource

2001, OECD, Biological Resource Centres

- **Conceptualization of biological resources as common public good**
- **Competing discourses of venture capital, scientific progress, data protection, etc**
- **Biological resources – living organisms, cells, genes, and related information – are the essential raw material for the advancement of biotechnology, human health, and research and development in life sciences“**

Bioresources: a tool for research

Biological samples
with associated data

Databases
organized collection of data - created to operate large quantities of information by inputting, storing, retrieving and managing that information

Bioresources

Registries
systematic collections of a clearly defined set of health and demographic data for patients with specific health characteristics, held in a central database for a predefined purpose

Biorepositories
Plants, microorganisms etc.

The BRIF initiative

Human bioresources are key components of biomedical research. Yet, their role is underestimated and the work provided to setting up and maintaining a valid bioresource is not recognized.

Cambon-Thomsen et al. *Nat Genet* 2003, 34:25–26

The BRIF initiative is a work in progress, currently developing a framework for:

- **creating a tool for calculating research impact of bioresources based on a metric (algorithm) and on the use of a unique digital resource identifier**
- **assessing requirements for citation/acknowledgement of bioresources in order to trace their use in research**

initiative: to recognise bioresource

WHY?

- not visible enough;
- not acknowledged adequately;
- difficult to trace;
- difficult to assess

- *lack of indicators describing*

efficient usage and management of BR

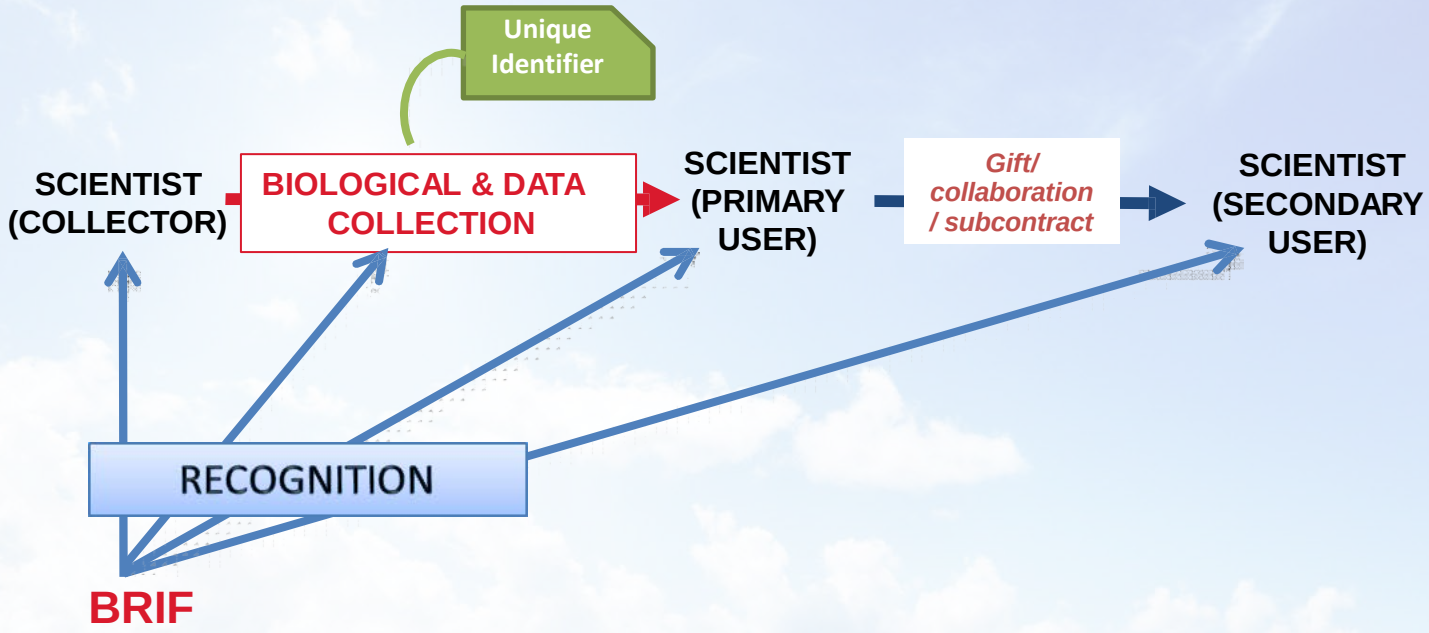
- *lack of a unique BR identification*

system to trace them precisely

- *lack of standards for BR citation in*

the scientific literature

The BRIF initiative : develop an impact



General information on the Bioresource, its use and the results of the research based on it

REVIEWERS, EDITORS

FUNDING BODIES

INDUSTRY

INDIVIDUAL

MEDICAL AND GENERAL PUBLIC

SCIENTIFIC READERS

Competences / parties represented in BRIF

- Biobank partners
- Computational biologists
- Computer scientists
- Genome/genetics scientists
- Epidemiologists
- Jurists, lawyers
- Ethicists
- Bibliometricists
- Journal Editors
- Researchers/users

Organization : Working subgroups

‘BRIF & Digital Identifiers’

co-chaired by G. A. Thorisson, University of Leicester, UK and P.A. Gourraud, University of California SF, USA Pierreantoine.Gourraud@ucsf.edu

‘BRIF Parameters’

chaired by B. Parodi, National Inst. Cancer Res. Genoa, IT barbara.parodi@istge.it

‘BRIF in Access & Sharing Policies’

co-chaired by E. Rial-Sebbag, Inserm UMR1027, Toulouse, FR and J. Harris, Norwegian Institute of Public Health, Oslo, Norway emmanuelle.rial@univ-tlse3.fr, Jennifer.Harris@fhi.no

‘BRIF dissemination’

chaired by L. Mabile, Inserm UMR1027, Tlse, FR laurence.mabile@univ-tlse3.fr

‘BRIF and Journal Editors’

‘BRIF and digital identifiers’ subgroup

‘Digital identifiers - some background

The subgroup address the various issues referred to identification, bioresources need to be assigned actionable digital identifiers or IDs. In order to fulfill the requirements of the scholarly record, bioresource ID should be

- persistent
- globally unique
- citable
- resolvable

3 broad categories of “stuff” to identify:

i) **Digital resources** : Resources that actually “lives” in computers (born-digital or digitized content): datasets and databases

ii) **Physical resources :** Resources corresponding to actual physical things: samples, groups of samples, experimental instruments, etc.

iii) **Project-level and other “meta” resources :** Higher-level aggregates of things, projects, organizations, consortia etc

iv) **Database relevant for health research**

‘BRIF parameters’ subgroup

Points to address: Evaluation of Bioresource Quality

To identify the different parameters to take into account when calculating the BRIF, it has been decided to focus on two types of entity providing a service to the scientific community:

i) biobanks of human biomaterials

ii) databases of information relating to human subjects research.

The aim is to provide a measure of the extent to which bioresources contribute to research.

What are the parameters to take into account when calculating the BRIF?

The parameters need to be objective and easily verifiable, and the calculation of a BRIF needs to be as simple as possible

A wide range of parameters are being considered for inclusion, including some indicators of **bioresource value, efficiency and research productivity**.

BRIF in sharing policies' subgroup

Attempting to measure the impact of a bioresource supposes, as its premise, that the research resource is actually being used.
Use of a biobank (or research database) is contingent upon many factors, but the ***access and sharing policies*** certainly play a major role in facilitating or hindering use. Various components such as the level of constraints imposed on users or the level of simplicity/complexity of the procedures to gain access are pivotal in creating an environment that will stimulate or discourage use of a given bioresource.

Points to address:
to study and develop an appropriate set of tools that could eventually be integrated in the overall access and sharing policies of the bioresources.

The BRIF work for reporting and dissemination

BRIF Journal Editors Subgroup

Elena Bravo ISS, Alessia Calzolari- ISS; Anne Cambon-Thomsen – INSERM; Paola De Castro – ISS ; Laurence Mabile –INSERM; Federica Napolitani – ISS; Anna Maria Rossi - ISS

Objective of the BRIF- Editorial subgroup:

The BRIF Journal Editors subgroup is dedicated to the relation with scientific journal editors. The subgroup agreed to submit proposals to international associations, committees and other organisations of scientific editors, in order to sensitize them on specific issues related to bioresources, BRIF project and, possibly, proceed to amend their editorial guidelines accordingly

Need of standardized citation: economical implication

Bioresources and Biobanks potential

The entire potential of biobanks as incubator for economic development and the related positive effects on health will vanish unless a **large investment on the quality of the work done by biobanks**, and on the **organization and coordination of the network of** activities.

Need of standardized citation : to select a bioresource for my research

- ✓ Quality of both biological samples and linked annotations, based on international standards and guidelines
- ✓ Scientific and medical background
- ✓ Quality of management
- ✓ Well defined access policy
- ✓ Expertise
- ✓ Willingness and reactivity
- ✓ Ethical and regulatory issues

Need of standardized citation: Current citation of bioresources use in scientific literature

- **multiplicity of sections where bioresources can be acknowledged (Material & Methods, Acknowledgements, References…)**

- **Bioresource acknowledgement or citation placed outside the main paper (or in online supplementary materials)**

- **typing errors or approximation of the bioresource name/identification;**
- **multiplicity of names for a given bioresource; different languages;**

- **cascade use of resources (*e.g.* Family samples that are themselves part of several other projects**

- **acknowledgement of persons instead of bioresources itself**

- **absence of acknowledgement for the bioresource used (negligence)**

- **suitable to refer to one type of bioresource but not for any derived, or secondary bioresources**

- **No standardized way to incentivise researchers to acknowledge properly the bioresource used**

- **Websites that no more exist**

Difficulties in the identification of bioresources

Most of the citation can be detected via full-text mining **(not indexed in Pubmed or Web of Science**).

With minor changes from.: Mabile *et al. GigaScience* 2013, **2**:7

Standardize bioresource citation: Editor' subgroup work

- Promote sharing of data and biological samples
- Recognize the work of setting up and maintaining a valid bioresource
- Reward the efforts involved in making the bioresources available to others

What is needed

- Sensitize journal editors to BRIF issues
- Standardize citations in journal articles
- Modify editorial guidelines
- Inform the scientific community on the issue relevance

Awareness on BRIF

Standardizing citations of research biobanks for a possible evaluation of Bioresource Research Impact Factor

Paola De Castro[1], Federica Napolitani[1], Anne Maria Rossi[1], Carlo Pastrello[2], Anne Cambon-Thomsen[2] and Elena Bravo[1]

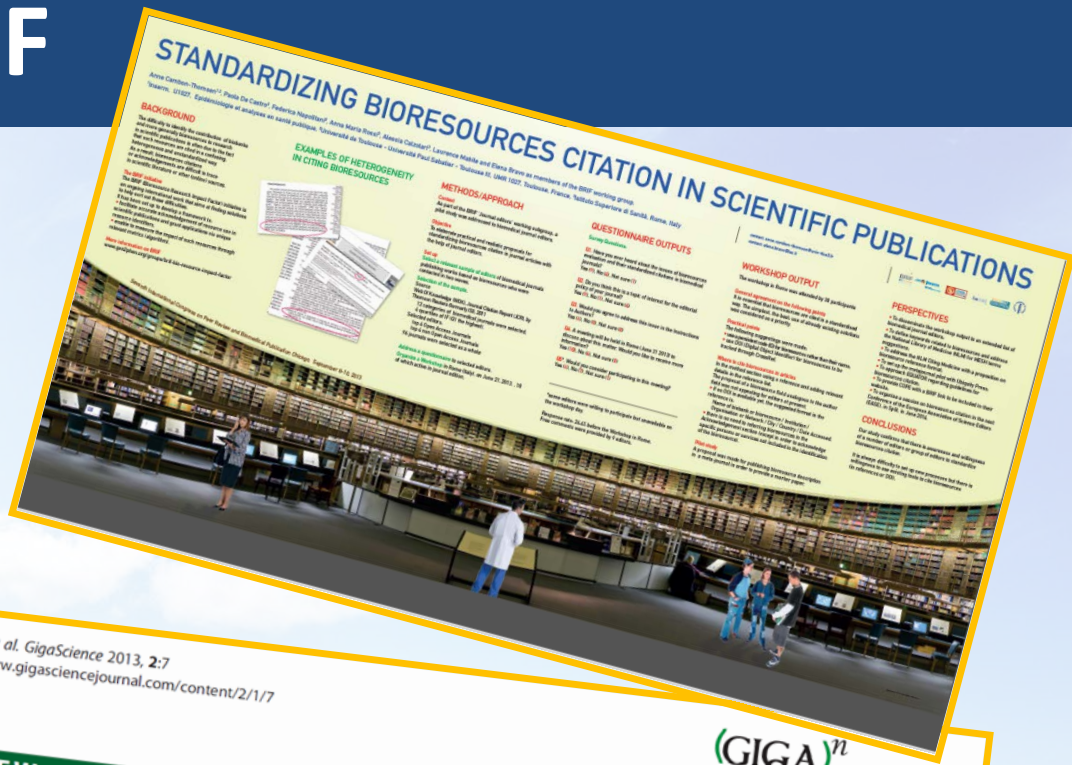
[1] Istituto Superiore di Sanità, Rome, Italy
[2] Inserm, Université de Toulouse, Toulouse, France

Introduction

The Bioresource Research Impact Factor (BRIF)

The subgroup "BRIF and journal editors"

References



STANDARDIZING BIORESOURCES CITATION IN SCIENTIFIC PUBLICATIONS

BACKGROUND

EXAMPLES OF HETEROGENEITY IN CITING BIORESOURCES

METHODS/APPROACH

QUESTIONNAIRE OUTPUTS

WORKSHOP OUTPUT

PERSPECTIVES

CONCLUSIONS

Mabile et al. GigaScience 2013, 2:7
http://www.gigasciencejournal.com/content/2/1/7

REVIEW

(GIGA)n SCIENCE

Open Access

Quantifying the use of bioresources for promoting their sharing in scientific research

Mabile[1,2], Raymond Dalgleish[3], Gudmundur A Thorisson[3,4], Mylène Deschênes[5], Robert Hewitt[6], nter[7], Elena Bravo[8], Mirella Filocamo[9], Pierre Antoine Gourraud[10], Jennifer R Harris[11], Paul Hofman[12], uffmann[13,14], Maria Angeles Muñoz-Fernàndez[15], Markus Pasterk[16], Anne Cambon-Thomsen[1,2*]

rking group

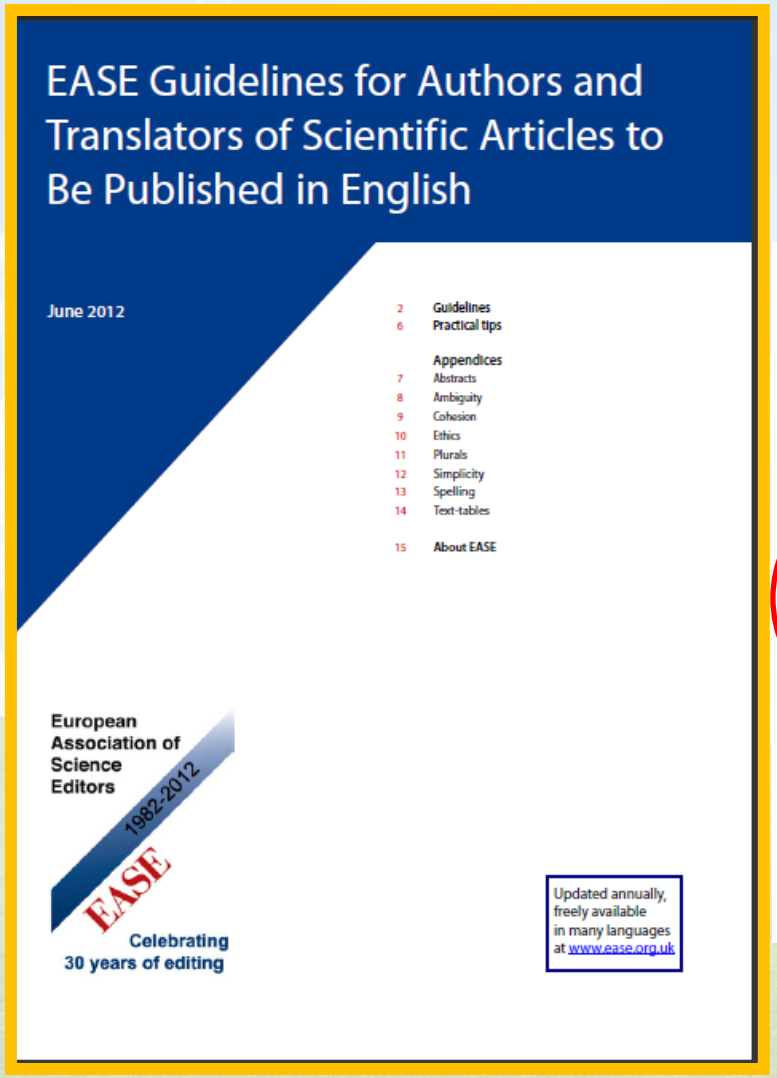
European Science Editing 36 May 2013; 39(2)

Citation of bioresources in journal articles: moving towards standards

Elena Bravo[1], Anne Cambon-Thomsen[2], Paola De Castro[1], Laurence Mabile[2], Federica Napolitani[1], Mariarosaria Napolitano[1], Anna Maria Rossi[1]

talk about diving beneath the surface of the written word (Tom Jefferson) and the relationship between translation and travel literature (Loredana Polezzi). Printing, diving and travel were all represented in the Aldine colophon. As I learned about Manuzio's work, I found further semblances to MET and METM12. Like Aldus & Co., MET attracts a multifaceted group of scholars, educators, linguists, translators and editors. Like Manuzio, MET members attain to high standards and are concerned with trends in publishing. And, in keeping with the motto, the planning of METM12 was characterized by perseverance, aiming for

Inclusion of Bioresources in EASE Guidelines


EASE Guidelines for Authors and Translators of Scientific Articles to Be Published in English

June 2012

European Association of Science Editors 1982-2012 EASE Celebrating 30 years of editing

Updated annually, freely available in many languages at www.ease.org.uk

The insertion has been added in the section '**Methods**'

- **Methods**: describe in detail how the study was carried out (e.g. study area, data collection, criteria, origin of analysed material, sample size, number of measurements, age and sex of participants, equipment, data analysis, statistical tests, and software used). All factors that could have affected the results need to be considered. Sources of experimental materials obtained from biobanks should be mentioned with full names and identifiers, if available (Cambon-Thomsen et al. 2011). If you cite a method described in a non-English or inaccessible publication, explain it in detail in your manuscript. Make sure that you comply with the ethical standards (e.g. WMA 2008) in respect of patient rights, animal testing, environmental protection, etc.

available in 20 languages

(www.ease.org.uk/publications/author-guidelines)

BRIF Meeting – Rome, June 21st 2013

Standardizing Bioresources Citation In Journal Articles: The Editors Point of View

Objective of the workshop

To elaborate practical and realistic proposals for harmonizing bioresources citation in journal articles with the help of journal editors.

Critical point

An ID and/or DOI which identify a human Bioresources is not available

Outcome

General agreement on Standard citation format

STANDARD BIORESOURCE CITATION

Where to cite

Cite bioresource in the "**Methods**" section (not in the acknowledgements) and add relevant details in the **reference list**

How to cite

- DOI or ID / Name of biobank / bioresource / City / Country
- Organisation / Network
- Date accessed (MTA or DTA)

Suggestions

- To identify bioresources using a persistent code (ID) rather than the name is better in order to avoid confusions. Alternatively, or complementary, the DOI (Digital Object Identifier) system, which refer to the publication that describe in detail the bioresource, seems to be the more appropriate so that bioresources could be **tracked through CrossRef**.

OJB initiative: a DOI for standardized citation

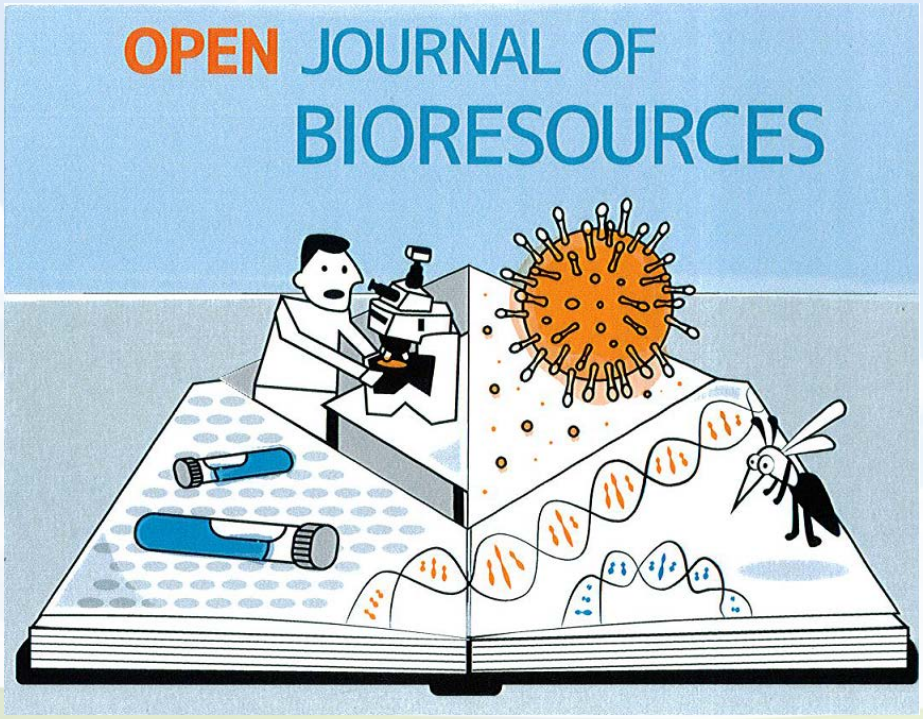
Marker paper (DOI) for a bioresource

A new meta-journal dedicated to bioresources description, the Open Journal of Bioresources (OJB) http://openbioresources.metajnl.com/). OJB has the purpose of giving to bioresources the possibility to describe their own characteristics, providing the biomedical community with easy access methodological “marker papers” for bioresources and biobanks. OJB features peer-reviewed short papers helping researchers to locate and cite bioresources with high reuse potential. Both the resources and the OJB papers are citable and this will be tracked to provide authors with metrics on reuse and impact.

OJB initiative

(1) Bioresource Overview Title Paper Authors Paper Author Affiliations Abstract Project description Classification Keywords Context: Spatial & temporal coverage	(3) Bioresource description Bioresource name , Bioresource location , Bioresource contact Bioresource URL, Identifier used Bioresource type , Type of sampling Disease status of patients/source Clinical characteristics of patients/source: age, gender, treatment information, etc. Vital state of patients/source, Clinical diagnosis of patients/source Pathology diagnosis, Control samples Biospecimen type ,, Anatomical site , Release date Access restrictions
(2) Methods Steps, Stabilization/preservation Type of long-term preservation Storage temperature Shipping temperature from patient/source to preservation or research use Shipping temperature from storage to research use Quality assurance measures, Ethics Statement Constraints	(4) Reuse potential Acknowledgements Funding statement Author Roles References

OJB initiative



OPEN JOURNAL OF
BIORESOURCES

upmetajournals

OJB features peer reviewed short papers helping researchers to locate and cite bioresources with high **reuse** potential.

Making bioresources more openly discoverable has enormous benefits not only for the research community and the **wider public,** but for the producers of the bioresources as well. Both the resources and the OJB papers are **citable** and this is tracked to provide authors with metrics on reuse and **impact.**

We work with major biobanks and other repositories to ensure that the associated bioresources are professionally preserved, and **accessible.**

Accepting submissions now:
http://openbioresources.metajnl.com

Ubiquity Press
http://www.ubiquitypress.com

Benefit from a standardized citation of bioresource?

Stakeholders

- researcher
- staff (any level)
- funding agencies
- initiators
- contributors
- annotators
- the institutions patients
- network organisation
- research participants
- policy decision maker

BIORESOURCES IN RARE DISEASES

Statistical power

Statistical power may depend on a number of factors. Some of these factors may be particular to a specific testing situation, but at a minimum, power nearly always depends on the following three factors:

- the statistical significance criterion used in the test
- the magnitude of the effect of interest in the population
- the sample size used to detect the effect

In rare disease "networking" is mandatory for the advancement of research

Network and make the (few) information available

- **Adequate Number of data / samples**
- **Adequate Quality of samples**
- **Re-use of data**

Dataset as a bioresource:ORPHANET

There is no disease so rare that it does not deserve attention

orphanet

The portal for rare diseases and orphan drugs

Inserm

Orphanet is the reference portal for information on rare diseases and orphan drugs, for all audiences. Orphanet's aim is to help improve the diagnosis, care and treatment of patients with rare diseases.

Orphanet services

- An inventory of rare diseases and a classification of An encyclopaedia of rare diseases in English and French
- An inventory of orphan drugs at all stages of development.
- A directory of expert resources: expert clinics, medical laboratories, ongoing research projects, clinical trials, registries, patient organisations,
- An assistance-to-diagnosis tool
- Guidelines for emergency medical care and anaesthesia.
- A collection of thematic reports, the Orphanet Reports Series, focusing on overarching themes

BIORESOURCES IN RARE DISEASES

ORDR – NIH RD-Hub
Rare Diseases Human Biospecimens /Biorepositories

RD-Hub was developed by the Office of Rare Diseases Research to serve as a central portal where researchers, professional societies, patient advocacy groups and other interested parties can locate and identify biorepositories and specimens needed for their research with a focus on rare diseases.

The RD - HuB website contains a searchable database of human biospecimens collected, stored, and distributed by biorepositories in the United States and around the world for research use. Searchable fields include:

(1) Repository name
(2) Disease
(3) Specimen Type
(4) Anatomic Source
(5) Processing Method
(6) Storage Method
(7) Imaging.

BIORESOURCES IN RARE DISEASES

Australia - OPHG

Office of Population Health Genomics

Established in 2001 to lead in the translation of genomics knowledge into health benefits, the Office has three sections:
Service Evaluation and Monitoring – Monitors genetic service delivery and engages the community to develop evidence-based policies.
Service Planning – Evaluates advances in genetic technologies for implementation into clinical practice.
Screening Policy – Considers emerging evidence and engages with stakeholders to develop policy on screening issues.

These Sections work to:

- Translate information from genetic research to improve the health of people in WA
- Review and evaluate the use of the best available genetic technology within budget
- Promote new partnerships in population health genomics at a local, national and international level.
- Provide policy advice
- Keep people informed
- Collaborate with groups that have an interest in genetics, genomics and gene technology
- Ensure that we do this in a way that is consistent with the ethical, legal and cost-effective community needs

BIORESOURCES IN RARE DISEASES

EBB assets

EuroBioBank is a European network of biological banks established in 2001. The EuroBioBank Network is fully dedicated to supporting research into rare diseases by facilitating access to quality human biological resources (DNA, cells and tissues) and their associated data from patients with rare diseases.

Key EBB assets

- **26 biobanks** from 11 countries (EU, Israel, Turkey and Canada) (including all TNGB biobanks)
- 440,000 samples stored
- Dedicated website, **catalogue** (updated 2x/yr), **charter** and **standard operating procedures** accessible online

BIORESOURCES IN RARE DISEASES

TNGB, Telethon Network of Genetic Biobanks

Fondazione Telethon has been supporting genetic biobanks since 1993. Current investment: 400 k€/yr

In **2008**, Telethon created the **TNGB** including all active Telethon biobanks

Key TNGB assets

- 10 biobanks in 6 Italian cities
- 600+ diseases, 50,000+ samples stored
- Yearly assessments by a dedicated scientific committee
- **Searchable, dynamic catalogue** of biological samples accessible online
- **Integrated workflow** for sample deposition and requests
- Strong interaction with **patient organizations**

BIORESOURCES IN RARE DISEASES

RD-Connect http://rd-connect.eu/

RD-Connect is a unique global infrastructure project that links up databases, registries, biobanks and clinical bioinformatics data used in rare disease research into a central resource for researchers worldwide.

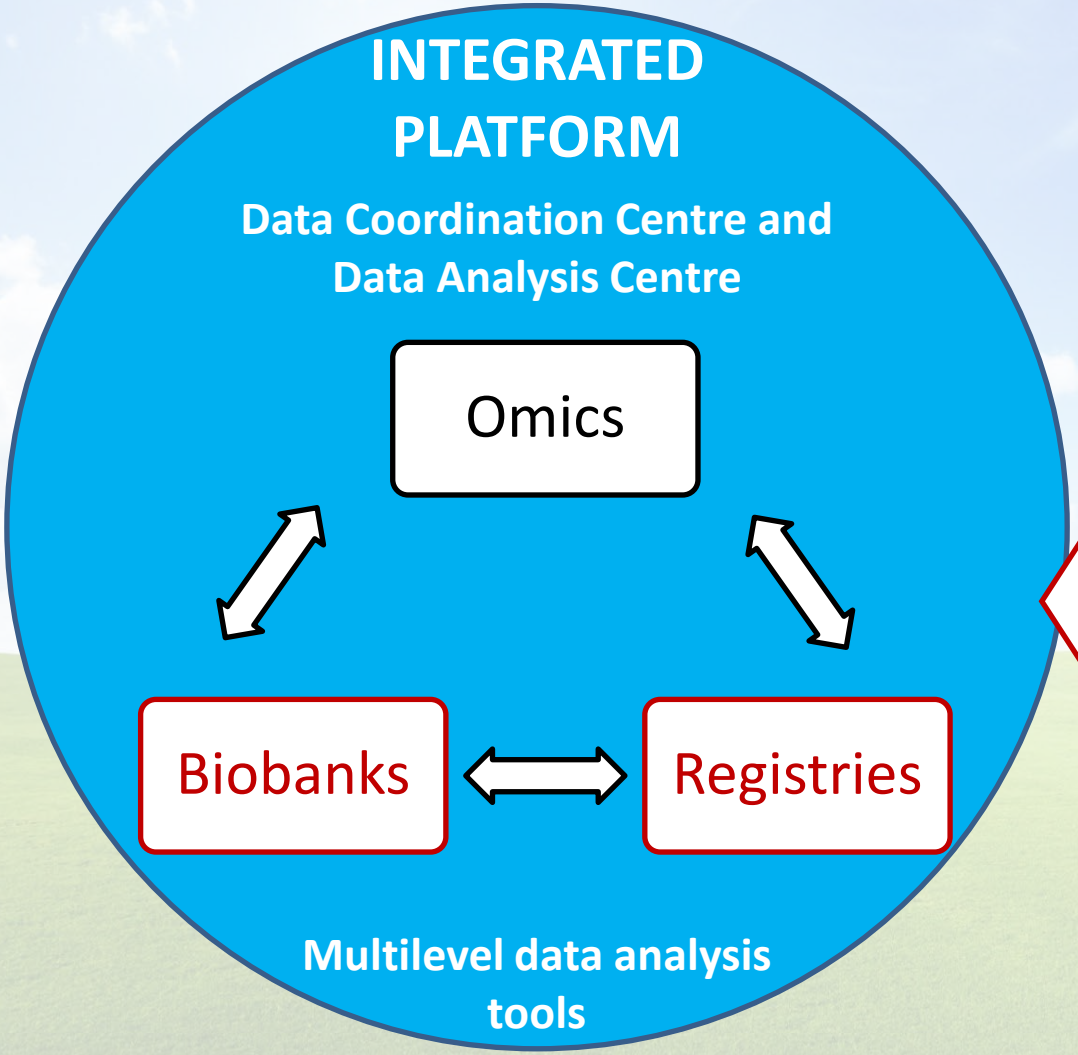
In a six-year project funded by the European Union but uniting researchers across the world, it will develop an integrated research platform in which complete clinical profiles are combined with -omics data and sample availability for rare disease research, in particular research funded under the International Rare Diseases Research Consortium (IRDiRC).

Databases and patient registries

- Map current status of RD registries (building on existing knowledge e.g. EPIRARE mapping but incorporating a research focus)
- Develop procedures and rules to facilitate contacting patients for confirmation of research findings; for example consent processes
- Develop standardised strategies and procedures for registries' interaction with pharmaceutical companies on surveillance registries for follow-up of patients on a particular drug
- Implement standardized coding system
- Develop and implement standard operating procedures and quality control
- Develop mechanisms for registering undiagnosed patients with harmonised consent for use of data for research

BIORESOURCES IN RARE DISEASES

RD-Connect: the Platform, its Users



Users

- NeurOmics/ EURenOmics
- IRDiRC projects

- RD scientists
- Clinicians
- Industry

BBMRI: network and Legal European consortium

[] the pan- European **Biobanking and Biomolecular Resources Research Infrastructure (BBMRI)** will build on existing sample collections, resources, technologies and expertise, which will be specifically complemented with innovative components and properly embedded into European scientific, ethical, legal and societal frameworks.

[ ]to expand and secure competitiveness of European research and industry in a global context and to attract investments in pharmaceutical and biomedical research facilities, by establishing the Biobanking and Biomolecular Research Infrastructure-European Research Infrastructure Consortium - hereinafter referred to as "BBMRI-ERIC".

The ERIC Legal Framework

Internationally recognized legal entity

Establishement of operational sites in different Member States that operate under one legislation

VAT exemption

BBMRI-ERIC and BRIF

- The European Commission policy encourages data sharing and reuse: a policy that is at the basis of the recently constituted European BBMRI-ERIC

- To facilitate the fair access to bioresources, the idea of assigning a unique ID has also been adopted in principle by the BBMRI-ERIC

- BRIF initiative is in workplan of BBMRI-ERIC, therefore playing a major role in BRIF implementation.

Indicators of good research productivity*

- Number of publications
  - Biobank member as co-author
  - Biobank member cited in the Acknowledgements section
  - Biobank cited in Materials and Methods section
  - Biobank cited in Acknowledgements section

- Impact factor of the journal
  - Cumulative impact factors per year and per collection
  - Cumulative impact factors per year for the biobank

* So far very difficult to evaluate

Hofman V et al. BIOPRESERVATION AND BIOBANKING. 2013;11:235-244

Goog Reporting disseminate good science

Major aspect that are often under evaluated by authors:
Good reporting is guarantied that dissemination:

- **Authorship** (different from contributor and Acknowledgement;)

- **Plagiarism** (means literary theft; taking others authorship; edition of another's work under its own name. Plagiarism is defined as "the intentional or unintentional copying the words of others". According to the World Association of Medical Journal plagiarism is to take a series of six words or from 7 to 11 words or overlapping set of 30 letters.

- **Reducing our irreproducibility** Nature **496**, 398, 2013)