orphanet

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Procedural document:
Data collection and registration of biobanks in Orphanet

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I. INTRODUCTION

1. Purpose/objectives

Orphanet offers, amongst a range of expert resources on rare diseases (RD), a directory of biobanks which are important to all domains of biomedical research, and particularly valuable in RD research. The Orphanet directory of biobanks aims to help:

- Experts working in the field of rare diseases to establish collaborations.
- Patients and general public to retrieve information on a particular disease.
- Experts to gain visibility and the ability to analyse the evolution of the field.

This document aims to define the set of criteria to be used when collecting, registering, and updating biobanks on rare diseases for the Orphanet database.

2. Disclaimer

- This procedural document is part of the OrphaNetWork Direct Grant (831390) which has received funding from the European Union's Health Programme (2014-2020).

- The content of this procedural document represents the views of the author only and is his/her sole responsibility; it cannot be considered to reflect the views of the European Commission and/or the Consumers, Health, Agriculture and Food Executive Agency or any other body of the European Union. The European Commission and the Agency do not accept any responsibility for use that may be made of the information it contains.

- Any information provided hereafter is not intended to replace professional health care. Orphanet cannot be held responsible for harmful, truncated or erroneous use of any information found in the Orphanet database.

3. Range of application

The present procedure applies to all biobanks and network of biobanks registered in Orphanet. The registration and update of biobanks is performed by the Orphanet national teams (ONT) having signed a Network Agreement and a Data Transfer Agreement (DTA) with the Orphanet Coordinating Team (OCT).

4. References

• Orphanet Standard Operating Procedures

• Regulation (EC) N°141/2000 of the European Parliament and of their Council of 16 December 1999 on orphan medicinal products

5. Definitions

Contact person of biobank/collection: Person to be contacted to obtain information concerning the biobank.

Coordinator of multicentric biobank: Manager assigned the responsibility for the coordination of several centres collecting patient samples and participating in a multicentric biobank.

Coordinator of network of biobanks: Manager assigned the responsibility for the coordination of managers of biobanks participating in a network of biobanks.

Data transfer agreement (DTA): Contract between the providing and recipient institutions that governs the legal obligations and restrictions as well as compliance with applicable laws and regulations related to the transfer of such data between the parties.

European Reference Networks (ERNs): Virtual networks involving healthcare providers across Europe. They aim to tackle complex or rare diseases and conditions that require highly specialised treatment and a concentration of knowledge and resources.

Funding body: Organisation that provides funds to fulfill a specific and predetermined purpose (to perform a research project, to develop a patient registry, etc.) which achievement is carried out by the beneficiary(ies) of the financial support.

Geographical coverage: Geographical distribution of patients included in the biobank.

Information scientist (IS): Member of the Orphanet team with a scientific and/or medical background in charge of collecting, producing and updating information provided in the Orphanet database.

International quality control (IQC) team: Team of persons in charge of the pre-release quality control of data registered in the database.

International Rare Disease Research Consortium (IRDiRC): Consortium uniting national and international governmental and non-profit funding bodies, companies (including pharmaceutical and biotech enterprises), umbrella patient advocacy organisations, and scientific researchers to promote international collaboration and advance rare diseases research worldwide.

Multicentric biobank: A collaborative biobank collecting patient information in more than one centre in a single country.

Network of biobanks A group of biobanks between which a relationship exists. An Orphanet network of biobanks is always multinational, including a coordination center and nodes in different countries.

Orphadata: A platform developed by Orphanet to provide the scientific community with

comprehensive, high-quality and freely accessible datasets related to rare diseases and orphan drugs, in a reusable format.

Orphanet coordinating team (OCT): French US14 Inserm-based team coordinating the Orphanet Network, producing the English Orphanet nomenclature and its scientific annotations and responsible for coordination of the production of the scientific content and for all Network activities including translation and IT developments.

Orphanet national teams (ONT): An Orphanet team based in one of the member countries of the Orphanet Network as per the Orphanet Network Agreement, and responsible for the collection of data on national expert resources. Some of the national teams are also in charge of the translation of the Orphanet nomenclature and/or the Orphanet database.

Orphanet network: The ONTs contributing to the Orphanet Database in the framework of the signed Network Agreement and DTAs.

Manager of biobank/collection: Individual who assumes full scientific responsibility for a biobank.

Rare disease (RD): A disease that affects less than 5 in 10,000 persons in Europe, as defined by the European Regulation on orphan medicinal products (Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products). In order to be registered in Orphanet, the disease must be described in at least two independent individuals, confirming that it is not an incidental association of clinical signs.

Rare disease biobank: Any collection of human biological samples (e.g. tissues, blood and derivatives, other body fluids, cells, DNA) and associated data such as clinical and research data, whose donors have been diagnosed with a rare disorder, a rare form of a common disorder, or can be defined by a group of rare disorders and that is to be used for research to contribute to the understanding of the physiology and diseases of humans.

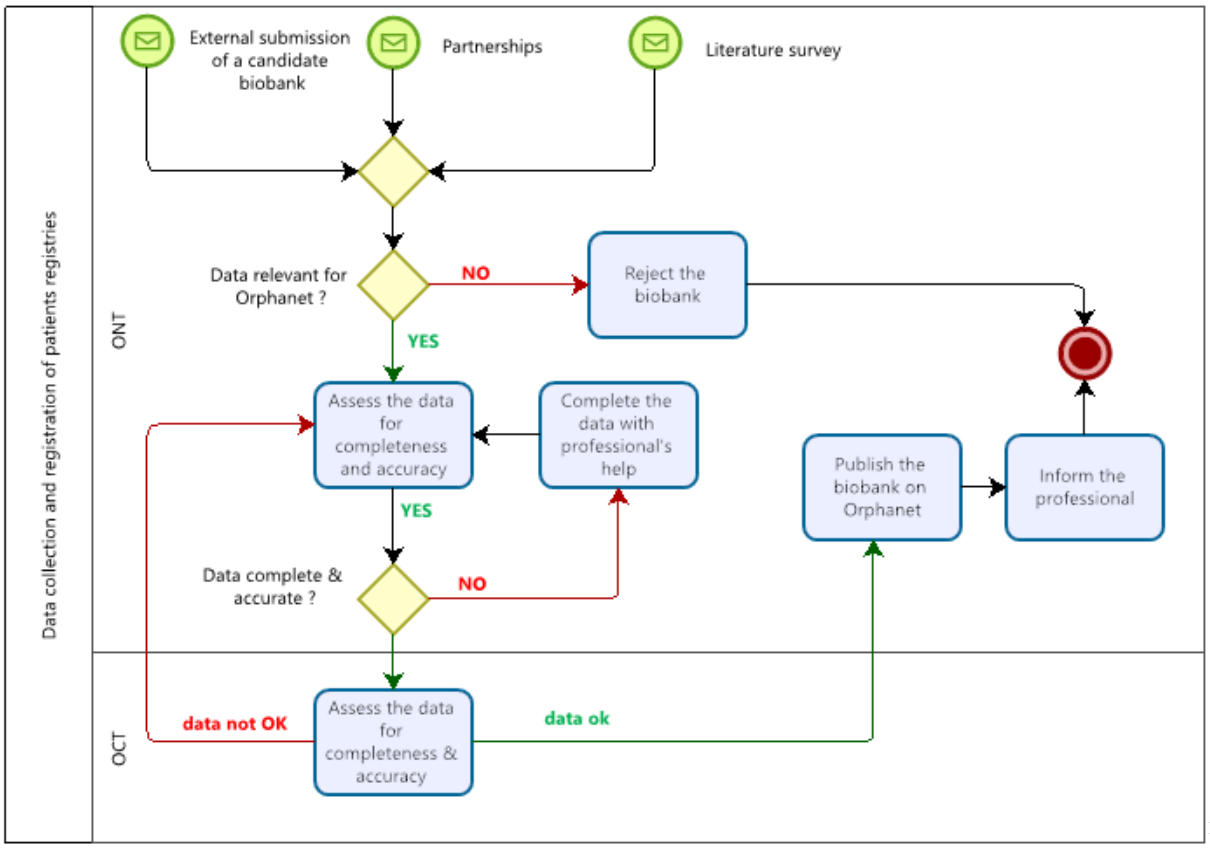
6. Filing and updates

This document is updated by the coordinating team as often as necessary and at least once a year. The most up-to-date version is available on the Orphanet website:
https://www.orpha.net/orphacom/cahiers/docs/GB/Biobanks_in_Orphanet_R2_R&D_BB_EP_08.pdf

II. METHODOLOGY

1. Flowchart

The general process for biobanks data collection, registration, validation, and its quality control is presented below:



[1]

2. Description

The various steps of the flowchart above are explained in more details in the following sections.

The process of registration / update of biobanks or network of biobanks starts with either one or a combination of the cases below:

- A professional asking for the registration of activity through any form (e-mail, phone calls, etc.), or sending an email request for update of its registered biobanks.
- An exchange of data through a partnership with a source of data (i.e. national authorities, etc.).
- Biobanks selected from peer-reviewed publications, the RD-Connect catalogue, the BBMRI catalogue, and IRDiRC.
- A post-release quality control task focused on biobanks or network of biobanks.

[1] Powered by bizagi Modeler

Networks of biobanks are registered by the Orphanet coordinating team, whereas Orphanet national teams are responsible for the registration of national biobanks.

An annual update is organised and launched by the Orphanet coordinating team where all professionals registered in the database are invited to review and update their activities. Orphanet national teams are responsible for following up professionals' feedback.

2.1 Sources of information

National teams are in charge of identifying the sources of information for biobanks in their country and are advised to establish partnerships with their national funding agencies to obtain lists of funded biobanks and be as exhaustive as possible.

If establishing a partnership, national teams must inform the coordinating team as some types of partnerships require the signature of a data transfer agreement (DTA).

The mains sources of information are:

- Professionals declaring a biobank through any form (e-mail, phone calls, etc.), or sending an email request for update of its registered biobanks.
- Biobanks selected from peer-reviewed publications.
- List of biobanks through a partnership with national authorities.

The **coordinating team** collects the biobanks established by ERNs. It also collects biobanks operating in the countries outside of the Orphanet network if they are present in the RD-Connect catalogue, present in the BBMRI catalogue, or funded by members of the International Rare Disease Research Consortium (IRDiRC).

2.2 Data selection

Orphanet national teams (ONTs) are responsible for data selection of biobanks that are established in their own country as well as for the submissions of biobanks they receive via individual requests. Selection of networks of biobanks lies not in the hands of the ONTs but is the responsibility of the Orphanet coordinating team (OCT).
The data selection starts by verifying that the biobank meets the inclusion criteria for Orphanet.

a) Inclusion criteria:

In order to be registered in Orphanet, a candidate biobank should be:

- Any collection of human biological samples (e.g. tissues, blood and derivatives, other body fluids, cells, DNA) and associated data such as clinical and research data, whose donors have been diagnosed with a rare disorder, a rare form of a common disorder, or can be defined by a group of rare disorders and that is to be used for research to contribute to the understanding of the physiology and diseases of humans.

b) Exclusion criteria:

Orphanet does not register a biobank if it is a:

- A collection of biological material with no specificity but that might be useful in the field of rare diseases.
- A private collection, except if it is **open for collaboration.**

Please note that in order to be included in the Orphanet database, a (network of) biobank(s) should be hosted within a country which is part of the Orphanet network (description of the Orphanet network and list of network members available via the links below). Non-Orphanet network biobanks will be registered only if they are funded by a member of IRDiRC.
https://www.orpha.net/consor/cgi-bin/Education_AboutOrphanet.php?lng=EN
https://www.orpha.net/orphacom/special/eproc_SOPs.pdf

2.3 Data assessment

A candidate biobank will go through a mandatory assessment and validation process at the national level. All biobanks, regardless of their origin/source, go through this assessment process. First and foremost, this step aims to ensure the relevance of each new entry in Orphanet as a specific resource on rare diseases. If a biobank is deemed not relevant for the Orphanet database (e.g. not focused on a rare disorders or group of rare disorders), the request to register the biobank is then immediately rejected by the information scientist of the Orphanet country where the request originated. The professional who submitted the suggestion to add the biobank is informed of the decision.

If the biobank complies with the Orphanet inclusion criteria for biobanks, the information scientist (IS) analyses the information to ensure that the mandatory dataset (described below) is provided and is coherent. Eventually, the IS makes the necessary corrections before submitting to pre-release quality control.

In case of inconsistency or missing information, the IS will contact the manager or the contact person of the biobank in order to clarify or obtain the information needed. Once this information obtained and assessed, the IS submits the candidate biobank to the pre-release quality control (see next section).

a) Orphanet dataset for a biobank

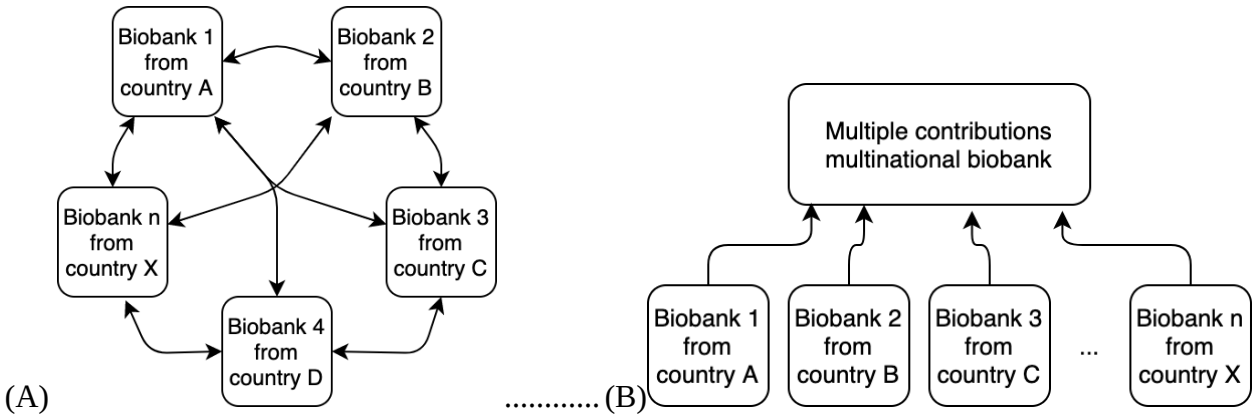
- Official title (in local language and in English).
- Website of the biobank or a description of it.
- Start and end dates of biobank.
- Recruitment status (recruiting or not).
- Disease(s) and/or group(s) of diseases concerned.

- Is it a multicentric national biobank, i.e. does the biobank collect data from several collecting sites within the country?
- Geographical area (regional, national, European, or global), i.e. area of collection of patient samples included in the database.
- Name and contact details (email address +/- phone number) of the manager and/or contact person of the biobank.
- Name of the funding body(ies).
- Is the biobank funded by a member of IRDiRC or is it an IRDiRC-recognized Resource (see https://irdirc.org/research/irdirc-recognized-resources/) at the time the biobank starts?
- Is it a biobank endorsed by a European Reference Network (ERN) member?
- RD-Connect ID if the biobank is present in the RD-Connect catalogue.

b) <u>Orphanet dataset for a network of biobanks</u>

- Official title (in local language and English).
- Website of the network of biobanks or a description of it (study plan: objectives, eligibility criteria, etc.).
- Disease(s) or group of diseases concerned.
- Geographical coverage (European or global) of the network of biobanks.
- Name and contact details (email address +/- phone number) of the coordinator of the biobank network.
- Name of the funding body(ies).
- Is the network of biobanks funded by a member of IRDiRC?

Note that all Orphanet networks of biobanks are multinational. Orphanet recognises two types of biobank networks represented below: (A) A larger physical multinational entity centralising and aggregating data from several smaller national biobanks; (B) A web of interoperable federated biobanks all communicating and forming a virtual single large biobank providing access to each node of the network.



2.4 Pre-release quality control (PrRQC)

Once the candidate biobank passes the national assessment step, the coordinating team (located in France) performs a pre-release quality control to assess the relevance and correctness of the data collected by the national teams. This quality control is mainly focused on the disease(s) linked as well as on the coherence of the whole dataset.

If some information is missing or needs correction, the request is sent back to the national teams who take the necessary steps (incl. contacting the professional if necessary) to complete the dataset originally submitted. After necessary corrections have been implemented by the national team, the biobank is submitted once again to the coordinating team for another round of quality control. Once relevance and correctness of the dataset have been ensured at both the national team level and at the coordinating team level, the candidate biobank is ready to be published.

2.5 Data publication

Once all the quality control steps have been completed, the patient candidate biobank is considered validated and is then published on the Orphanet website. The corresponding ONT is then in charge of informing the professional(s) that the activity has/have been published.

The information on biobanks is accessible on Orphanet via the "Directory of ongoing research projects, clinical trials, registries and biobanks", section "Registries & biobanks". They can be also accessed from the "Additional information" section at the bottom of each disease page.

Information on biobanks can also be retrieved from Orphadata after signing a Data Transfer Agreement (DTA) or a service contract.

2.6 Post-release validation

The post-release quality control for patient registries is performed through the quality control projects, which are organised by the coordinating team on a regular basis in order to check the completeness and consistency of the data.

For any questions or comments, please contact us: contact.orphanet@inserm.fr

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