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Procedural document: Orphanet Rare Diseases Nomenclature Production in National Language

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Instituts thématiques Inserm
Institut national de la santé et de la recherche médicale

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Table of contents

I. Introduction

1. Purpose/objectives

- This document aims to describe how the Orphanet rare disease nomenclature produced in English is produced in other languages of the Orphanet Network.

2. Disclaimer

- This procedure is part of the RD-ACTION joint action which has received funding from the European Union's Health Programme (2014-2020).

- The content of this procedure represents the views of the author only and is his/her sole responsibility; it can not 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.

3. Range of application

Production (Translation and adaptation) of the Orphanet Rare Disease nomenclature in languages other than English can be performed by the relevant Orphanet National Team (ONT) having signed a Data Transfer Agreement (DTA) with the Orphanet Coordinating Team (OCT) or people designated/appointed by the country coordinator. Third parties other than ONT can carry out production of the Orphanet Rare Disease Nomenclature in National language after a DTA has been signed between the ONT and the OCT and a third party if relevant.
Once the production started, the ONT or the third party engages to maintain and update the production in National Language in parallel as the English Production is carried out by the OCT. ONT are entirely responsible for the publication and quality of the Orphanet Rare Disease Nomenclature in their language.
Orphanet RD Nomenclature in national language implementation in the Database and diffusion is carried out by the OCT.

The Orphanet Rare disease nomenclature is accessible through the "Rare diseases" tab on the Orphanet website (if website translated into National Language), through the Orphanet Report Series "List of rare diseases and synonyms in alphabetical order», through the Orphadata website and for the english version also through the Orphanet Rare diseases Ontology (ORDO).

4. References

- Orphanet Inventory of Rare Disease Procedure
- Disease Naming Rules in English
- Linearization Rules for Orphanet Classifications
- Orphanet Standard Operating Procedures
- Data Transfer Agreement between OCT and ONT, template
- Data transfer Agreement between OCT and third party, template

5. Definitions

Orphanet coordinating team (OCT): It is the French US14 Inserm based team who coordinates the Orphanet Network, produces the English Orphanet Nomenclature and its scientific annotations and is also responsible for coordination of the production of the scientific content and of all Network activities including translation.

Orphanet National Team (ONT): teams located in each participating country of the Orphanet Network, and endorsed by national authorities. A ONT is composed, at least, of a country coordinator who is responsible for the national Orphanet activities including translation. It can also include one or several information scientists, translation staff and a project manager.

Orphanet Country Coordinator: He/She is designated by the Institution participating in the Orphanet Network. He/she participates in the Orphanet Management Board, organizes the governance of the project at national level, including liaison with learned societies, national authorities and patient organisations, and the build-up of the Orphanet team if applicable. He/She is responsible for all the data production (either core-data or national data) and data quality management, and translation carried out within the ONT.

The Orphanet Management Board: it is the governing board of the Orphanet Network. It is composed of all the country coordinators of the ONT within the network. It is chaired by the project coordinator at the Inserm.

Third Party: An institution in charge of Nomenclature Production in National Language different form the Institution hosting the Orphanet National team.

Data Transfer agreement (DTA): National teams sign a DTA with the OCT governing legal obligations and restrictions of both parties as well as compliance with national laws related to the Orphanet Database content translation and national data production.
If the RD nomenclature production in National Language is carried out externally, an additional DTA defining the collaboration is to be signed between the OCT and the Third Party in charge of it, the ONT also notifies this agreement.

Translation staff: Staff in charge of nomenclature translation/adaptation into national language either within the Orphanet team or appointed by it or within the Third Party if relevant.

Medical staff: Staff in charge of the medical validation of the Nomenclature in National Language either within the Orphanet team (Country Coordinator, Project Manager...) or

appointed by it (National Advisory Board, physisicians...)

National Advisory Board: ONT can decide to set up a National Advisory Board, its members being nominated by the appropriate legitimate institutions (learned societies, national authorities, etc.), which are defined at country level. National Advisory Board members contribute with their expertise to Orphanet at country level and validate any database content concerning resources listed for the country in question as well as Oprhanet Rare Disease Nomenclature production in national language if relevant.

The Orphanet inventory of rare disorders: it is an inventory organised via a classification system that is based on a spectrum of entities defined by the typology below:
A Group of phenomes is a collection of clinical entities, sharing a given characteristic and are therefore classified together. Groups of phenomes can be clinically homogeneous or, on the contrary, can be used to put together clinically heterogeneous disorders for the needs of organizing the classification. In that case they are called "categories"
Disorders are a set of entities including diseases, syndromes, anomalies and particular clinical situations. Disorders are clinically homogeneous entities described in at least two independent individuals, confirming that the clinical signs are not associated by fortuity please refer to the procedure avialable online for more detailed information.

This typology is available only via Orphadata and ORDO, please refer to the procedure for more information.
A rare disorder is defined according to the European legislation defining a prevalence threshold of not more than five affected persons per 10'000 (Regulation (EC) N°141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products)

Every entity of the nomenclature is defined by:
- An **ORPHA number**: unique identifier attributed randomly by the database and stable through time.
- A **preferred term**: the most generally accepted name in the medical community.
- **Synonyms**: perfect equivalents in the scope of the preferred term they are attached to. As many synonyms as necessary are added to a preferred term.
- **Acronyms**: included only when actually used in literature. Convenience acronyms used in Orphanet summaries that have no use in the scientific community are not included. Several entities can share the same acronym.
- **Keywords**: significant terms for a disease or group of disease that are usefully retained for redirecting users to relevant diseases, but do not fit the defining criteria of a preferred name, a synonym or an acronym.

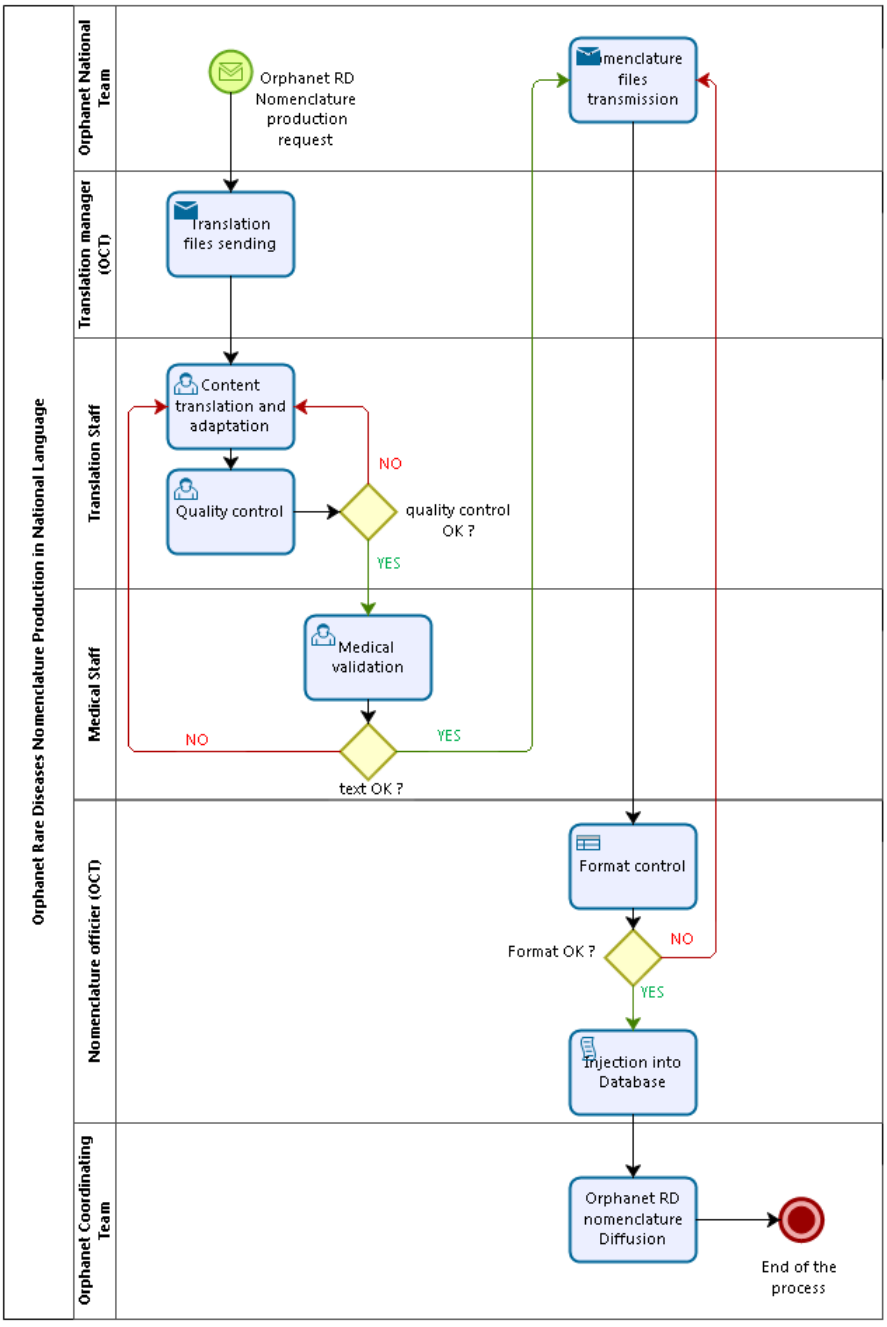
Translation Report: This report is sent every two to three months to the teams involved in the Orphanet international website content translation and/or in the Orphanet Encyclopaedia translation and/or Orphanet inventory of Rare Disease production in National Language. It includes all the necessary information for carrying out monthly translation activities by the ONT.

6. Filing and updates

These procedures are updated annually by the Translation manager in the coordinating team. The most up-to-date version is available on the Orphanet website.

II. METHODOLOGY

1. Flowchart



2. Description

A. National language Orphanet Rare Disease Nomenclature production launch

The country coordinator should send a request to launch production of the Orphanet nomenclature of rare diseases in National Language to the International Coordinator and the Translation Manager specifying the person in team who will be the contact person(s) for this activity.
Nomenclature production in National Language can be performed externally by a third party, after signature of a DTA to define the collaboration between ONT, OCT and the institution in charge of the Nomenclature production in National Language.

For any new demand for production of the Orphanet Nomenclature in National Language, a file containing the whole Orphanet Nomenclature in English is sent (workflow described in section B, Initial Phase). Then, files containing new and modified terms in the English Nomenclature are provided on a regular basis for maintenance of the nomenclature in the National Language (workflow described in section C, Update and maintenance phase).
Please note: a Naming Rules document exists for the English Nomenclature. It is strongly recommended to adapt it to the National Language, in order to have a referential for consistency and quality control.

B. Initial phase

1. Files retrieval

The Translation Manager sends the files containing the Orphanet Nomenclature terms in English to be translated/adapted and a copy of this procedure to the contact person(s) for this activity. Technical instructions on how to handle those files are also sent to the contact person(s).

2. Content translation and inclusion of additional national terms (adaptation)

The content translation is carried out by the translation staff at country level. It can be the ONT, or people designated/appointed by the country coordinator or a third party.

Inclusion of additional terms in National Language: if relevant, perfect synonyms can be added as needed and if relevant (and unavoidable) preferred term (PT) can be adapted to reflect real national use.

Please note: PT are chosen in English according to the Naming Rules (here) to reflect medical practice. Any modification in the preferred term during production in National Language should be discussed with the Nomenclature Team in order to decide whether it is pertinent to apply this change also to the English Nomenclature, or to allow for a different PT in the National Language that would be justified by a different local use.

Steps 3 and 4 are of crucial importance as the medical information should be accurate, medically useful, and coherent.

3. Quality control

This step is set up by the translation staff who should assess the implementation of the formal and editorial rules in the Nomenclature in National Language according to the Naming rules document.

4. Validation of the Nomenclature in National Language

The Nomenclature in National Language must be validated by Medical Staff (the Country Coordinator or physician(s) designated by him/her (*i.e.* a member of the national Scientific Advisory Board)). People having validated the nomenclature can be acknowledged on the national website.

5. Validated Nomenclature files transmitted to the Nomenclature Officer

ONT are entirely responsible for the publication and quality of the Orphanet Rare Disease Nomenclature in their language. Therefore only material sent by ONT through its contact person(s) for National Languages Nomenclature production will be accepted by the Nomenclature Officer even in case of involvement of a Third Party.

6. Format quality control

The nomenclature Officer verifies format conformity of the transmitted files which ensure correct integration in the database.

7. Integration of the Rare Disease Nomenclature in National Language into the Database

Upon reception by the OCT this content is injected into the database by the Nomenclature Officer.

8. Diffusion of the Orphanet Rare Disease Nomenclature in National Language

The Rare Disease Nomenclature in National Language will be made available by the OCT:
- through the “Rare diseases” tab on the Orphanet website, if the website is available into National Language
- through the Orphanet Report Series “List of rare diseases and synonyms in alphabetical order»,
- through the Orphadata website as a separate downloadable file
- through the OrphanetRare diseases Ontology (ORDO) when developed in other languages than English .

C. Update and maintenance phase

1. Files Retrieval

A file containing the new and updated **Orphanet Rare Disease Nomenclature terms** in English is compiled by the nomenclature officer every 2-3 months. The Translation Manager integrates the new and updated terms document in the Translation report, which is regularly sent to the national teams, and also available at: https://network.orpha.net/network/cgi-bin/articles.php?lng=en&pg=97

2 to 7. Steps 2 to 7 are identical as the ones described in B section.

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

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http://www.orpha.net/orphacom/cahiers/docs/GB/eproc_Nomenclature-production-National-language.pdf