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Procedural document: Translation of the Disease Summary Texts of the Orphanet Encyclopedia for Professionnals

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I. Introduction

1. Purpose

For patients affected by a rare disease, obtaining a timely and accurate diagnosis is key to accessing the appropriate medical expertise. The Orphanet website, via the relevant Rare Disease page, provides disease summary texts that are primarily intended for healthcare professionals, with an emphasis on clinically relevant information and with a particular focus on the disease definition, clinical description, diagnosis, and management of a rare disease.

The rare disease page disease summary texts are available in English and are translated into Dutch, French, German, Italian, Polish, Portuguese, Spanish; moreoverthese texts can also be translated into any other language, if the appropriate funding is available. Disease summary texts can also be retrieved from Orphadata after signing a Data Transfer Agreement (DTA) or service contract.

This document aims to outline the Orphanet procedure for the translation of disease summary texts.

2. Disclaimer

- This publication is part of the project OrphaNetWork Direct Grant (831390) which has received funding from the European Union's Health Program (2014-2020).
- The content of this publication 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.
- The information provided by Orphanet in the disease summary texts is based on published scientific articles, but may not apply to specific cases due to the extensive variability of disease expression. Given the rarity of these diseases, the treatments outlined in the abstracts are not always evidence based. The provided information is not intended to replace existing local, regional or country specific recommendations and guidelines. Certain information may be shocking to some readers. It is of the utmost importance to check if the provided information is relevant or not to a specific case.
- Information found on the Orphanet website is updated on a regular basis. It may happen that new discoveries are made in between updates and these do not yet appear in the disease profile. The date of the last update of the disease summary text is always indicated. Professionals are always encouraged to consult the most recent publications before making any decisions based on the information provided.
- Orphanet cannot be held responsible for harmful, truncated or erroneous use of any information found in the Orphanet database.

3. Range of application

This procedure concerns all Orphanet team members or individuals designated by the country coordinator involved in the translation of disease summary texts.

4. References

The Orphanet Standard Operating Procedures available on the Orphanet website here.
The Procedure on creation and update of disease summary texts in English for the Orphanet Encyclopedia for professionals

5. Definitions

Abstract: Texts composed of up to 10 of the following sections: disease definition, epidemiology, clinical description, aetiology, diagnostic methods, differential diagnosis, antenatal diagnosis (if relevant), genetic counselling (if relevant), management and treatment, and prognosis.

Disease definition: A short text stating the group of disorders that the clinical entity belongs to, and listing the major clinical characteristics (e.g. clinical, pathological, radiological, etc.) that define the entity and differentiate it from other entities classified within the same clinical group.

Disease summary text: This term is inclusive of the two text formats (disease definitions and abstracts).

Expert (Expert reviewer): A medical doctor or researcher with prominent clinical experience in a rare disease or a group of rare diseases, and identified by Orphanet based on published articles (particularly reviews and guidelines), involvement in expert centers, expert networks, and/or in dedicated research activities including clinical trials.

Medical validation staff: Staff in charge of the medical validation of the summary information and of the translation of (medical) Orphanet content in national language.

Medical writer: An information scientist, responsible for producing and/or curating the disease summary texts (disease definitions and abstracts).

National coordinator: Designated by the Institution having signed the Network Agreement. He/She has the duty to set up a national team and coordinate it. He/She participates in the Orphanet MB, organises the governance of the project at national level, including liaison with learned societies, National authorities and patient organisations. 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

Orphanet National team (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 Knowledge base: Orphanet is the International Rare Diseases and Orphan Drugs knowledge base, an organised and dynamic collection of information about rare diseases and orphan drugs where data from multiple sources are archived, reviewed, distilled and manually annotated by experts and quality controlled according to published procedures.

National Advisory Board (Scientific advisory board): A consultative group for the Orphanet national team. Its members can be nominated by the appropriate legitimate institutions (learned societies, national authorities, etc.), which are defined at country level. National Advisory Board members contribute their expertise to Orphanet at country level and validate any database content concerning resources listed for the country in question as well as translation of the Rare Disease Nomenclature and of the Encyclopaedia in the national language, if relevant.

Translation report (T-rep): Report containing Orphanet content to be translated in languages other than English by the Orphanet national team**.**

Translation staff: Staff in charge of translation of the relevant material sent through the Translation Report and according to the relevant procedures (either within the ONT or appointed by it or within the Third Party if relevant). He/She is under the supervision of the country National Coordinator to whom he/she should report.

6. Filing and updates

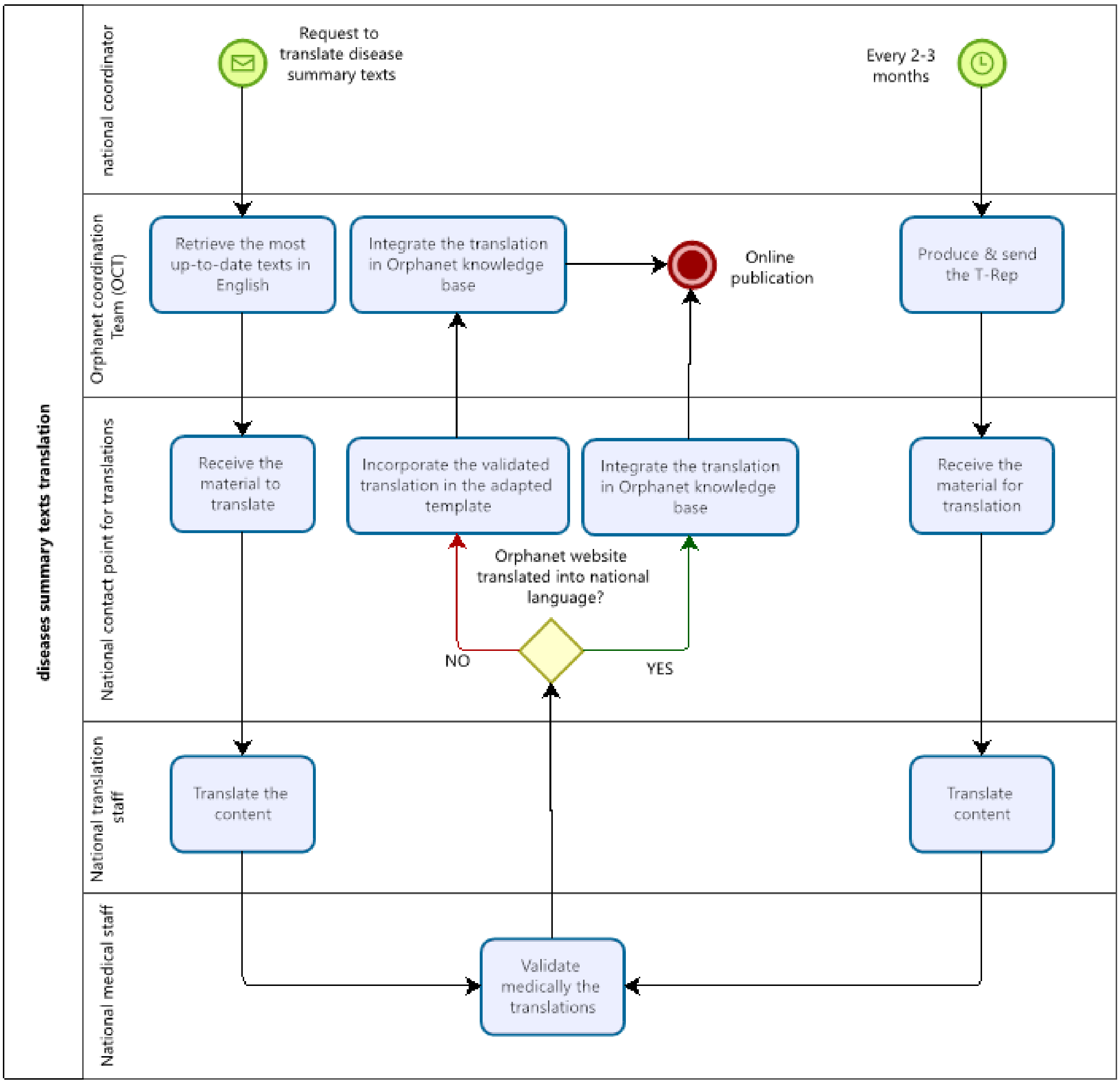
This document is updated by the coordinating team as often as necessary and at least once a year.

II. Methodology

1. Flowchart

There are two phases in the Disease summary text translation process: the initial phase and the monthly updates:

- A first batch with the most up-date abstracts and definitions is translated by the Orphanet national team
- The Orphanet coordinating team sends to the ONT the texts produced or updated during the 2-3 last months



2. Description

a. Initial phase

The national coordinator of an Orphanet national team makes a request to translate disease summary texts to the Orphanet coordinating team, specifying who in the team will be the contact point for translations.

b. Monthly updates

Every 2-3 months the OCT produces and sends to the ONT the T-rep that contains, among other materials, all the summary texts published since the last generation of the T-rep.

c. Content translation

The content translation is carried out by the translation staff at country level. It can be the national team, or individuals designated by the national coordinator.

Translations should be exact, without adding new elements, and correct from a medical point of view.

d. Medical validation of the translation

The translation must be validated by the medical staff, *i.e.* the national coordinator or a physician designated by him/her (*i.e.* a member of the national Scientific Advisory Board).

This step is of crucial importance as the medical information should be accurate, and correspond to the initial text in English, without adding or withdrawing any elements. Those having validated a text will be acknowledged on the national website. National teams are entirely responsible for the publication and quality of translations.

e. Integration of the translations in Orphanet database

If the Orphanet international website is already translated in the national language, the ONT is in charge of registering the translation in the Orphanet knowledge base. In this case, the translated summary texts are published online in the disease page in the corresponding version of Orphanet web interface.

If the Orphanet web interface is not yet available in national language, the OCT is in charge of the publication of the translated disease definition or abstract. For this the ONT must deliver the translation in the adapated template to the OCT[1].

In this case, the translated texts are published online on the international Orphanet website, as a PDF document located at the bottom of the disease page, in the "Detailed information"

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[1] This template contains the Orphanet logo, the disease name, the ORPHAcode and all the relevant sections of the abstract. It also contains the names of the expert reviewers having collaborated to the abstract production, the last update date, the translation date and the name of the medical validators of the translation. If relevant, information regarding the sponsor of the translation can be included in the template. Finally, it contains a disclaimer and a link to the Orphanet website

section, under “Summary information”.

III. Characteristics of the data

For abstracts,

- The name(s) indicated at the end of the disease summary text on the Orphanet website correspond(s) to the expert reviewer(s) that validated the text in English.
- The date indicated after the name of the expert reviewer(s) corresponds to the last update of the published text in English.

For disease definitions,

- No date and no name of expert reviewer(s) is indicated at the end of the disease page on the Orphanet website.

For abstracts and disease definitions,

- Abbreviations are defined in the text at their first occurrence.
- Gene symbols and names follow the HGNC nomenclature (*HUGO Gene Nomenclature Committee*).

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

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