

Procedural document: Creation and Update of Disease Summary Texts for the Orphanet Encyclopaedia for Professionals

www.orpha.net

www.orphadata.org

Table of contents

I. Introduction	3
1. Purpose/objectives.....	3
2. Disclaimer	3
3. Range of application.....	3
4. References	4
5. Definitions	4
6. Filing and updates	4
II. METHODOLOGY	5
1. Flowchart.....	5
2. Description	6
2.1 Identification of priority entries for the creation or update of disease summary texts.....	6
2.2 Text preparation and expert identification:.....	6
a) Text preparation	6
b) External medical expert reviewer selection	7
2.3 Internal validation.....	7
2.4 External validation.....	7
2.5 Curation	7
2.6 Publication	7
2.7 Quality control	8
2.8 Translation	8
III. Characteristics of the data	9

I. Introduction

1. Purpose/objectives

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

This document aims to outline the Orphanet procedures for the creation or update of disease summary texts.

2. Disclaimer

- This publication is part of the project / joint action ‘677024 / RD-ACTION’ 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 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 involved in the creation and update of disease summary texts for the Orphanet Encyclopaedia for Professionals. The present procedure applies to all disorder entities of the Orphanet database.

4. References

[PubMed – NCBI](#): Bibliographical search engine

[GeneReviews](#): Clinical genetics information

[Hugo Gene Nomenclature Committee \(HGNC/HUGO\)](#): Gene nomenclature and locus information

[Uniprot](#): Protein nomenclature information

[OMIM](#): Online Catalog of Human Genes and Genetic Disorders

[RareCareNet](#): Information on rare cancers

[Neuromuscular Disease Center](#): Information on neuromuscular diseases

[Orphanet standard operating procedures](#)

5. Definitions

Abstracts: 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.

External medical expert reviewer: Expert contributing to the revision of the information on a disease for Orphanet.

Expert validator: External medical expert reviewer who has the right to validate the information on a disease allowing it to be published on Orphanet. He is referred to as “Expert Reviewer” on the Orphanet website.

Internal medical reviewer: Internal medical doctor reviewing all the texts produced by the editorial teams for editorial and medical consistency, at each major version of the document, and who validates the publication after the revision process is finalised.

Medical writer: Information scientist producing the textual information according to the present procedure.

Orphanet disease summary texts: These are short definitions or abstracts produced internally by Orphanet editorial teams in English and then translated into several different languages.

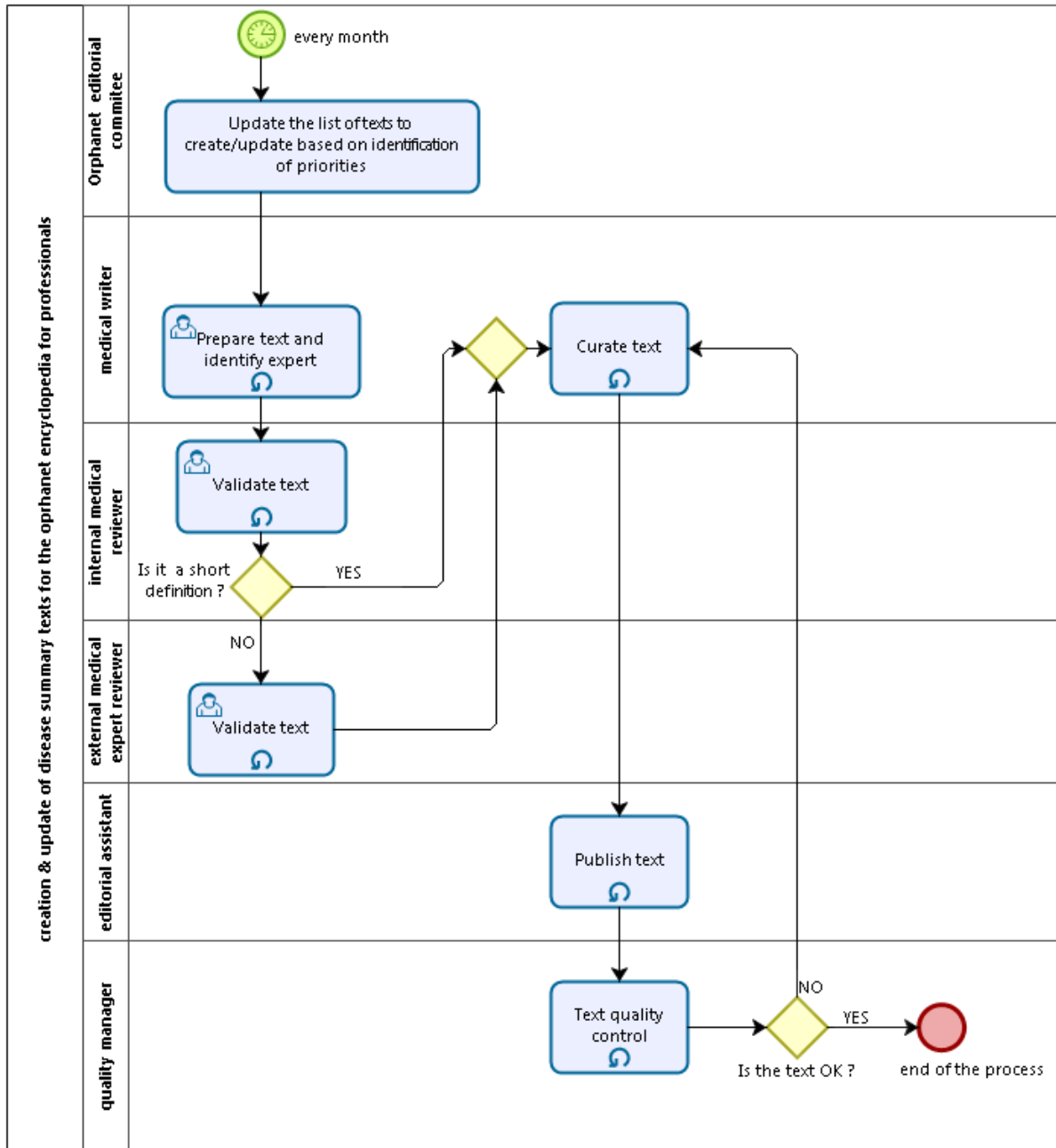
Short definitions: Very short texts (corresponding to the Disease definition section), stating which group of diseases the entry belongs to and listing the major clinical characteristics that define the disease and differentiate it from others in the same group.

6. Filing and updates

This procedure is updated annually and as often as necessary by someone designated by the director of operations. The most up-to-date version is available on the Orphanet website: http://www.orpha.net/orphacom/cahiers/docs/GB/eproc_Orphanet_DiseaseSummaryText_V01.pdf

II. METHODOLOGY

1. Flowchart



2. Description

2.1 Identification of priority entries for the creation or update of disease summary texts

Orphanet's aim is to produce textual information for every single rare disorder in the database. However, due to the considerable number of entries present, prioritisation is necessary in order to organise the production of the editorial team. Texts are thus created or updated based on a list of priorities decided upon by the Orphanet editorial team, which is composed of the chief medical officer, the internal medical reviewer and the medical writers.

The priorities are based both on the need to produce or update a text (due to lack of updated information in Orphanet or for the needs of an ongoing project or partnership) and on the opportunities available to produce high-quality and up-to-date information (recent publications of good review articles or guidelines). Therefore, priorities are identified based on the following criteria:

- 1) Texts that have not been updated in the last 5 years.
- 2) Entries that have no texts.
- 3) Entries for diseases with a relatively higher prevalence (>1/100,000).
- 4) Entries for diseases with an orphan designation/drug available.
- 5) Entries for which a definition is required for an ongoing project or partnership.
- 6) Entries for which detailed and recent information is available on Orphanet, particularly external review articles, clinical practice guidelines, or internal articles for the general public.
- 7) Entries for which an internal text (i.e. disability factsheets, general public articles, emergency guidelines) is planned.
- 8) Entries for which texts were requested from internal or external Orphanet users.
- 9) Entries that have generated recent interest among the public or the medical community.

2.2 Text preparation and expert identification:

a) Text preparation

The information collected during the bibliographical research is compiled under the following sections (depending on the type of text, see definitions) by the medical writer:

- **Disease definition:** This section provides a brief description of the entry (not exceeding 3-4 sentences) including which group of disorders the entry belongs to and its principle clinical characteristics that distinguish it from other entries in the same group.
- **Epidemiology:** This section states the most recent epidemiological information on a disease as defined by the *Epidemiology of rare diseases: Procedural document*. Priority is given to worldwide prevalence figures (point prevalence, prevalence at birth) when available, then to European ones, then to those of specific countries. Incidence figures are also reported when known.
- **Clinical description:** This section provides information about the age of onset of the disease as well as the detailed description of the clinical presentation of the disease.
- **Aetiology:** This section describes the aetiology and/or the physiopathology of the disease. For

genetic diseases, the causal gene (as per the HGNC nomenclature) and locus are always noted when the relationship has been established. For multifactorial diseases, the genetic background is also explained when established.

- **Diagnostic methods:** This section describes the principle tools used to reach a diagnosis, with a brief description of the key findings (e.g. laboratory results, imaging findings). The means by which a diagnosis is confirmed are stated. For genetic diseases, genetic screening is mentioned if applicable.
- **Differential diagnosis:** This section lists the main differential diagnoses of the disease.
- **Antenatal diagnosis:** This section is only completed when an antenatal diagnosis is possible and the tools used to reach such a diagnosis are described.
- **Genetic counseling:** This section is only completed for hereditary diseases and describes the mode of transmission as well as the genetic counselling to be given, in order to inform affected families of the genetic risk of transmission.
- **Management and treatment:** This section provides a brief description of the management and treatment options and includes any orphan drug availability, if appropriate. Please note that tradenames are never cited in the Orphanet disease summary texts, only the international non-proprietary names are used.
- **Prognosis:** This section briefly describes the prognosis of the disease in regards to functional consequences, quality of life, life-expectancy, and likelihood of recurrence after treatment.

b) External medical expert reviewer selection

During the process of creating/updating a disease summary text, the medical writer also proposes one or several external medical expert reviewers, based on a series of criteria (soon to be made available), to be selected by the internal medical reviewer. One expert validator is designated with the right to validate the final version of the text. Criteria are intended to prove that the expert has a specific, undisputable clinical expertise on the given disease or group of diseases.

2.3 Internal validation

The draft version of the disease summary text, as well as the external medical expert reviewer(s) proposed, are reviewed by the internal medical reviewer for medical accuracy and proofreading, before being submitted to the selected external medical expert reviewer(s).

2.4 External validation

Abstracts are reviewed and validated by the external medical expert reviewer(s). Short definitions are validated by the internal medical reviewer before publication.

2.5 Curation

The consistency with the Orphanet database of rare diseases, in regards to the nomenclature, classification, epidemiological and natural history information, and genetic data, is checked before publication, in order to ensure the intrinsic quality of the information contained in the text and to update the Orphanet database as needed.

2.6 Publication

The text is published on the Orphanet website by the editorial assistant.

2.7 Quality control

After publication, the disease summary texts produced during a cycle of production are sent to the quality manager for a process of post-release quality control, in order to ensure the consistency of content format throughout the encyclopaedia, the consistency of information with the database elements, and the quality of English writing. The necessary corrections are then carried out.

2.8 Translation

The quality-controlled, compiled document is sent to the Orphanet partner country teams for translation in other languages (see [Orphanet Standard Operating Procedures](#), part IV)

III. Characteristics of the data

- All disease summary texts are written in US English.
- Texts have a strict character limit of 4,000 characters (spaces included).
- Abbreviations are defined in the text at their first occurrence.
- Gene symbols and names follow the HGNC nomenclature. Protein names follow UniProtKB nomenclature.
- The date of the last update (month, year) is noted at the end of the disease summary text.
- The name of the expert validator(s) is/are listed after the date of the last update when the disease summary text has undergone an external review.

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

Editor of this procedural document: Medical writer, Natasha Barr - This procedural document has been approved by:
Ana Rath - Quality control: Charlotte Gueydan

The correct form when quoting this document is:

« Procedural document on Creation and Update of Disease Summary Texts for the Orphanet Encyclopaedia for
Professionals, Orphanet, January 2017 Version 01.1

http://www.orpha.net/orphacom/cahiers/docs/GB/eproc_Orphanet_DiseaseSummaryText_V01.pdf