orphanet

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Procedural document:

Collection and dissemination of disease information for health professionals and the general public

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La science pour la santé
From science to health

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I. Introduction

1. Purpose/objectives

Orphanet aims to be the reference resource for rare diseases, and as such developed a policy concerning the collection, evaluation and dissemination of quality information produced by others. This information encompasses review articles, clinical practice guidelines (CPGs), anesthesia guidelines, practical genetics guides, clinical genetics review, guidance for genetic testing, articles for general public (all available through the "Detailed information" box on the Orphanet website's disease pages) and websites containing added-value information on rare diseases (through the "Other websites" link on the same disease page in the "Additional information" box).

This document describes how Orphanet collects, assesses and gives access to this content intended for health professionals and for lay readers on the Orphanet website.

2. Disclaimer

- This procedure 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 procedure 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 availability of a link from the Orphanet website to other sites does not indicate endorsement of those sites by Orphanet, and Orphanet accepts no responsibility for the validity or accuracy of their content.

3. Range of application

Information is collected for all rare diseases belonging to the Orphanet nomenclature of rare diseases. Resources in all languages can be disseminated and are made available from the rare disease pages of the Orphanet website in all languages.

4. References

- AGREE II evaluation tool
- HONCode principles
- Orphanet Standard Operating Procedures

5. Abbreviation

AWMF: Arbeitsgemeinschaft der Wissenschaftlichen Medizinischen Fachgesellschaften

CPG: Clinical Practice Guideline

EJHG: European Journal of Human Genetics

ERN: European Reference Network

INCA: Institut National du Cancer

IS: Information Scientists

OCT: Orphanet Coordinating Team

PNDS : Protocole national de diagnostic et de soins

6. Definitions

AGREE II: The **A**ppraisal of **G**uidelines for **R**esearch and **E**valuation (AGREE[1]) is an international tool to assess the quality and reporting of practice guidelines.

Coordinator of acquisition of external content (hereafter named "coordinator"): Member of Orphanet coordinating team in charge of the quality assessment and dissemination by Orphanet of the different types of articles and websites of interest collected by the information scientists of the Orphanet network.

European Reference Networks (ERNs): Virtual networks involving healthcare providers across Europe that aim to tackle complex or rare diseases and conditions requiring highly specialised treatment and a concentration of knowledge and resources[2].

Expert: A medical doctor or researcher with prominent 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.

External content: Refers to the information produced and published by organisations external to Orphanet (e.g. learned society, scientific journal, patient association, governmental institution, etc.).

HON Code certification: Ethical standard aimed at offering quality health information. It demonstrates the intent of a website to publish transparent information. It guides site managers in setting up a minimum set of mechanisms to provide quality, objective and transparent

[1] 2010 version
[2] European Reference Network handout, ISBN 978-92-79-65469-5

medical information tailored to the needs of the audience.

Information scientists (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.

Orphanet coordinating team (OCT): French US14 Inserm-based team coordinating the Orphanet Network, producing the Orphanet nomenclature in English 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.

7. Filing and updates

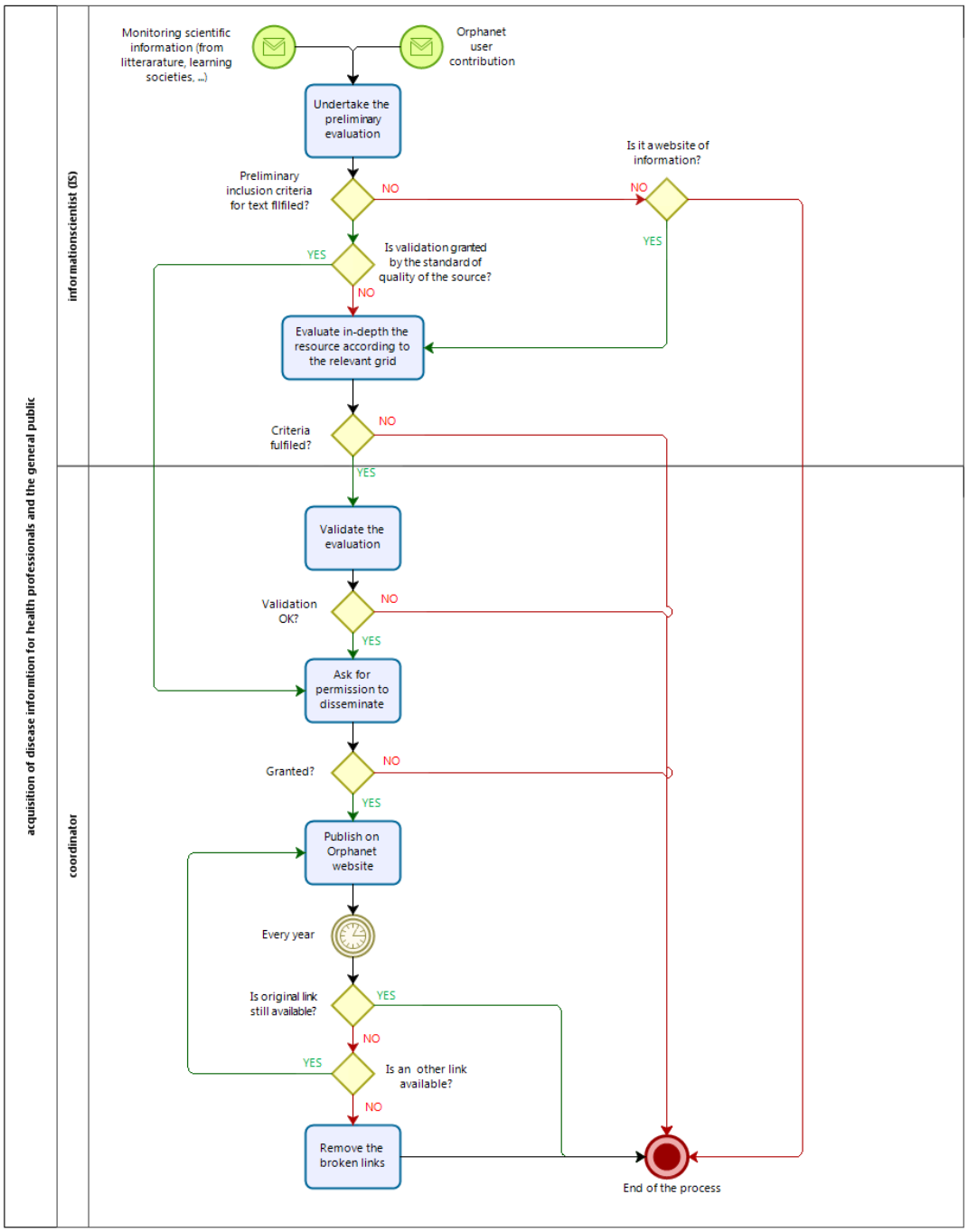
This document is updated by the coordinator of acquisition of external content as often as necessary and at least once a year. The most up-to-date version is available on the Orphanet website:
https://www.orpha.net/orphacom/cahiers/docs/GB/Acquisition_of_disease_information_R1_Wcont_EP_02.pdf

Type of text	Definition	Intended audience	Sources	Descriptive data provided by Orphanet
Anesthesia guidelines	Recommendations for the anaesthetic management of patients suffering from rare diseases	Anesthesists	OrphanAnesthesia project	Language; year of publication; (PDF format when applicable)
Articles for general public	Tackle various aspects of rare diseases	General public	Medical societies, patient organisations, research networks, reference centers	Language; year of publication; (PDF format when applicable)
Clinical genetics reviews	Review articles centered on the genetic aspects of diseases, including paragraphs on diagnosis, management and genetic counseling	Health professionals	GeneReviews®	Language; year of publication
Clinical practice guidelines (CPGs)	Recommendations to standardise the process of diagnosing and treating rare diseases	Health professionals, patients and healthcare administrators	Peer-reviewed articles; publication from medical societies, patient organisations, research networks, reference centres, etc.	Language; year of publication; (PDF format when applicable)
Guidance for genetic testing	Disease-specific guidelines regarding the clinical utility of genetic testing	Clinicians and geneticists	Peer-reviewed articles from the European Journal of Human Genetics	Language; year of publication; (PDF format when applicable)
Practical genetics	Peer-reviewed articles providing clinical, genetic and management information	Clinical geneticists	Peer-reviewed articles from the European Journal of Human Genetics	Language; year of publication; (PDF format when applicable)
Review articles	Peer-reviewed articles providing a clear, clinically-oriented and up-to-date description of rare diseases	Health professionals and scientists	Peer-reviewed articles from scientific journals	Language; year of publication; (PDF format when applicable)
Other websites of interest	Website displaying added-value information on rare diseases	All audiences	Medical societies, patient organisations, research networks, reference centers, etc.	Language, targeted public, access conditions, type of publisher and type of information;

Table 1: Definitions of the types of collected information

II. Methodology

1. Flowchart



[3]

Figure 1: Workflow for the collection, evaluation and dissemination of relevant articles and websites on Orphanet

[3] Powered by Bizagy Modeler

2. Description

a) Identification

Identification of medical information is carried out either directly by an information scientist of an Orphanet national team through scientific monitoring (literature[see Annex for the list of journals], Pubmed requests, learned societies etc) or following a spontaneous contribution from an Orphanet user or an organisation (ERNs, French National Rare Disease Networks).

b) Evaluation

Selected medical information undergoes a series of evaluation to assess its quality before dissemination.

Preliminary evaluation

The first step is to select articles that fulfil minimum quality criteria. These criteria are:

- Date of document: less than 5 years-old;
- Writers: expert of the disease. Single authors are not accepted in the case of CPGs;
- Category of document: consistent with one of the categories of texts defined by Orphanet (see definitions of article types);
- Topic: relevance of medical content in terms of addressed topics;
- Focus: on a rare disease or a group of rare diseases;
- Accessibility: full-text electronically available and for free (except for CPGs for which a link to the abstracts is sufficient whatever the full-text accessibility status);
- "Review article" type of text should be published in a peer-reviewed scientific journal.

If the preliminary criteria are fulfilled, it can

- Be published on the Orphanet website if the document is in the framework of a collaboration with Orphanet (i.e. with ERNs, other expert networks, learned societies, patient organisations, etc) or governmental sources for which their general standard of quality is granted;
- Go through an in-depth evaluation (see below).

If these preliminary criteria are not fulfilled, the documents are considered for the section "other website of interest" (see below) and undergo an in-depth evaluation based on inclusion/exclusion criteria derived from the HONCode principles (see Annex).

In-depth evaluation

A quick bibliographic search is carried out to check whether a similar article (in the same language) is available and fulfils the preliminary criteria. If yes, a rapid evaluation (based on the detailed

criteria in Annex) of the two articles is carried out to identify the most valuable, and then performed in-depth evaluation on the best one.

In case the quick evaluation is not sufficient to decipher between the articles, in-depth evaluation is performed for both to weigh the pros and cons.

The information scientist fills in the relevant form (see Annex) and provides his/her conclusions on the main aspects: topics covered, writing quality, relevance of authors, possible weaknesses and limitations. It allows a third party (including the final validator) to unambiguously understand the quality of the article (with strengths and weaknesses) without having to read it. Furthermore, the comments are useful for future comparison with another article on the same disease.

c) Validation

If the criteria of the preliminary evaluation are fulfilled and the text published by Orphanet's partners or governmental sources for which their general standard of quality is granted, the validation is automatically approved.

If the text underwent an in-depth evaluation, the evaluation sheet is checked by the coordinator, who quickly reviews the consistency of the evaluation sheet and the source and validates the added value of the source.

Table 2 summarises the validation steps in regard to the type of text.

d) Dissemination

If relevant, the IS requests permission from the copyright owner(s) before dissemination on Orphanet website.

e) Quality control

Identification of broken links is carried out by the coordinating team once a year, by running a script, completed by partially manual checking. Broken links are either corrected or removed (if correction is not possible) from the Orphanet website.

Table 2 : Validation steps of the different types of text according to their sources (NA= Not Applicable)

Type of text	Sources	Validation after quick evaluation is sufficient	Validation tools for in-depth evaluation
Anesthesia guidelines	OrphanAnesthesia	Yes	NA
Articles for general public	Medical societies, patient organisations, research networks, reference centers, etc.	No	Evaluation grid, see Annex
Clinical genetics review	GeneReviews	Yes	NA
Clinical practice guidelines	PNDS	Yes	NA
	ERNs guidelines	Yes	NA
	INCA guidelines	Yes	NA
	AWMF guidelines	Yes	NA
	Others	No	Evaluation grid for CPGs based on AGREE II evaluation tool, see Annex
Guidance for genetic testing	EJHG	Yes	NA
Other website of interest	Medical societies, patient organisations, research networks, reference centers, etc.	No	Evaluation with inclusion/exclusion criteria derived from the HONCode principles, see Annex
Practical Genetics	EJHG	Yes	NA
Review article	STArt Oncology	Yes	NA
	Others	No	Evaluation grid for "Review articles", see Annex

III. Annexes

1. Evaluation grids

☞ For Review articles:

Similar document, in the same language, is already linked on Orphanet			☐ Yes ☐ No
If yes, new information about: ☐ Epidemiology ☐ Classification ☐ Diagnosis ☐ Treatment ☐ Other:			
Author(s)			
Specialists of the condition	☐ Yes	☐ No	
Cover all medical specialties involved	☐ Yes	☐ No	☐ Not applicable
Expert reviewer(s) for abstract on Orphanet	☐ Yes	☐ No	
Clarity of presentation			
Contains introduction	☐ Yes	☐ No	
Contains abstract	☐ Yes	☐ No	
Article is well written	☐ Yes	☐ No	
Explanations are concise	☐ Yes	☐ No	
Main topics and conclusions are easy to find	☐ Yes	☐ No	
Scope, target population			
Topics covered by the article: ☐ Epidemiology ☐ Pathology ☐ Etiology/genetics ☐ Clinical Presentation ☐ Diagnosis criteria ☐ Differential diagnosis ☐ Clinical course and prognosis ☐ Genetic counseling ☐ Therapeutic considerations ☐ Ethics ☐ Cost efficiency ☐ Other:			
Population coverage is exhaustive	☐ Yes	☐ No, specify what population is not covered (pediatric,…):	
Geographic coverage ☐ International ☐ US	☐ Europe	☐ Other:	
All therapeutic options are equally described	☐ Yes	☐ No.	
Does the article explain why (described in another article,…)?			☐ Yes ☐ No
Contains information of importance to the patients? (e.g. change in morbidity/ mortality; Quality of life; disability information)	☐ Yes	☐ No	
Contains methodology about literature search and reference selection?			☐ Yes ☐ No
Contains description of evidence used to establish the key statements?			☐ Yes ☐ No
Other review available in literature?	☐ Yes	☐ No	
If yes, overall rating of the present review is ☐ higher ☐ equivalent ☐ lower ☐ different			
Comment:			
Assessment: ☐ Good ☐ Satisfactory ☐ Poor			
Comment:			

Additional detailed criteria to assess the overall quality of review articles are as follows:

- ✓ Relevance of authors: especially if there is only one author, he/she should be specialist of the disease or he/she has authored other publications on the disease. If there are several authors, they should cover all domains of expertise of the disease.

- ✓ Clarity of presentation, writing quality: an introduction and an abstract are not decisive elements, but are a plus for clarity and quick reading.
- ✓ Relevance of scope and target populations: the largest coverage of topic the best. An article addressing only one topic (e.g. only therapy) is not retained, with exceptions: if no other textual information is available on Orphanet (the rule "better than nothing" applies), if the topic corresponds to the dominant aspect of the disease. Inclusion of information for patient is not decisive but a plus for selection as it is rarely present in articles.
- ✓ A methodology section on bibliographic search is not decisive but is a plus as it reflects data robustness.

☞ For Clinical Practice Guidelines (adapted from the AGREE II evaluation tool):

Scope and purpose

Population coverage is exhaustive Yes ☐ No ☐

If no, what population is missing (pediatric,…):

Geographic coverage (area of release): ☐ International ☐ US ☐ Europe ☐ Other:

Covered topics: ☐ Prevention ☐ Diagnosis ☐ Genetic testing ☐ Therapeutic ☐ Emergency ☐ Other:

Overall objectives of the guidelines are described (AGREE II Item 1) Yes ☐ No ☐

Health questions are specifically described (AGREE II Item 2) Yes ☐ No ☐

Clear description of target patient population (AGREE II Item 3) Yes ☐ No ☐

Editorial independence

External funding Not described ☐ Yes ☐ No ☐

Evidence that potential biases due to the funding body were taken into account (AGREE II Item 22) Yes ☐ No ☐

Conflicts of interest of guideline development group members are recorded (AGREE II Item 23) Yes ☐ No ☐

Conflicts of interest of guideline development group members are addressed (AGREE II Item 23) Yes ☐ No ☐

Stakeholder involvement

Guideline development group includes individuals from all relevant professional groups (implies that the group is clearly described) (AGREE II Item 4) Yes ☐ No ☐

Patients' preferences are taken into account (AGREE II Item 5) Yes ☐ No ☐

Clear description of target users of the guidelines (AGREE II Item 6) Yes ☐ No ☐

Rigour of development

Systematic methods used to search for evidence are described (AGREE II Item 7) Yes ☐ No ☐

Criteria for selecting evidence are described (AGREE II Item 8) Yes ☐ No ☐

Methods used to assess the strength of evidence are clearly described (GRADE method,…) (AGREE II Item 9) Yes ☐ No ☐

Methods used to reach consensus are clearly described (Delphi technique,…)(AGREE II Item 10) Yes ☐ No ☐

Benefits and risks have been considered in formulating the recommendations (AGREE II Item 11) Yes ☐ No ☐

Evidence used in developing guidelines is referenced adequately (linked to the recommendations) (AGREE II Item 12) Yes ☐ No ☐

Guidelines have been externally reviewed prior to publication (AGREE II Item 13) Yes ☐ No ☐

A procedure for updating the guidelines is provided (AGREE II Item 14) Yes ☐ No ☐

Clarity of presentation

Recommendations are specific and unambiguous (AGREE II Item 15) Yes ☐ No ☐

The different management options are clearly presented (AGREE II Item 16) Yes ☐ No ☐

Major recommendations can be found easily (AGREE II Item 17) Yes ☐ No ☐

Applicability

Facilitators and barriers to the guideline's application are described (need for specific structure/material, special funding mechanism)(AGREE II Item 18) Yes ☐ No ☐

Tools on how to put the recommendations into practice are provided (quick guide, check list, algorithm, how-to directions,…)(AGREE II Item 19) Yes ☐ No ☐

Resources and costs implications have been considered (AGREE II Item 20) Yes ☐ No ☐

Other guidelines available Yes ☐ Yes, anterior version ☐ No ☐ Already linked on Orphanet ☐

If yes, overall rating of the present guidelines is ☐ higher ☐ equivalent ☐ lower ☐ different

Explain why:

Reviewer:
Date of review dd/mm/yy):
Assessment by topic:

Scope and purpose	☐ satisfactory	☐ insufficient
Editorial independence	☐ satisfactory	☐ insufficient
Stakeholder involvement	☐ satisfactory	☐ insufficient
Rigour of development	☐ satisfactory	☐ insufficient
Clarity of presentation	☐ satisfactory	☐ insufficient
Applicability	☐ satisfactory	☐ insufficient

Overall assessment: ☐ Good ☐ Satisfactory ☐ Poor
Comment:

Note: Regarding relevance of scope and purpose: the largest coverage of topic the best. Only documents addressing at least diagnosis and therapy are retained. However, documents that do not cover all aspects of a disease, for instance when no other recommendation is available (in the same language), or when the topic correspond to the dominant aspect of the disease, are retained.

☞ **For "Articles for the general public":**

The following conditions must all be present to continue the evaluation:

☐ The document targets the general public
☐ The document concerns a rare disease
☐ The document can be consulted free of charge on Internet
☐ The document does not contain an advertisement insert

General and methodological information:

Publisher:
☐ learned society ☐ national institution ☐ company *(e.g. pharmaceutical)*
☐ patient association ☐ expert group ☐ other
☐ not indicated Specify:

Production is financially supported by:
☐ learned society ☐ national institution ☐ company *(e.g. pharmaceutical)*
☐ patient association ☐ expert group ☐ other
☐ not indicated Specify:

The(s) writer(s) has/have appropriate knowledge of the disease ☐ yes ☐ no
He/she/they is/are from:
☐ learned society ☐ national institution ☐ expert group
☐ patient association ☐ other ☐ not indicated Specify:

Revision of the document has been carried out by experts of the disease:
☐ clinicians from expert centres ☐ patient association(s) ☐ institutions
☐ other ☐ none, or not indicated
Specify:

The document contains an invitation to financial donation ☐ yes ☐ no

The document contains a disclaimer indicating that the information cannot replace professional medical care ☐ yes ☐ no

The document mentions trade names, especially drug trade names ☐ yes ☐ no
If yes, are all the existing brands mentioned? ☐ yes ☐ no

The document contains patients/families testimonies ☐ yes ☐ no

Presentation and content *(the general public texts produced by Orphanet can serve as reference for the evaluations)*

Evaluation of the topics:	very insufficient ----------> very good			
Disease definition	☐1	☐2	☐3	☐4
Genetics and/or physiopathology data	☐1	☐2	☐3	☐4
Clinical aspects	☐1	☐2	☐3	☐4
Treatments	☐1	☐2	☐3	☐4
Daily life	☐1	☐2	☐3	☐4
Social/family issues	☐1	☐2	☐3	☐4

The document is clearly presented *(information is easy to find, summaries are present, …)*
☐1 ☐2 ☐3 ☐4

The length of the document is adapted? ☐1 ☐2 ☐3 ☐4

There is appropriate balance between positive and negative aspects of the disease
☐1 ☐2 ☐3 ☐4

The document is in compliance with medical ethics rules and patients' rights
☐1 ☐2 ☐3 ☐4

The statements are in agreement with the available scientific data
☐1 ☐2 ☐3 ☐4

The illustrations are adapted *(useful for understanding, and not shocking)**
☐1 ☐2 ☐3 ☐4

The level of language is adapted to the general public *(level of popularization, and not shocking)*
☐1 ☐2 ☐3 ☐4

Links to other relevant sources of information are provided*
☐1 ☐2 ☐3 ☐4

(do not tick if absent)*

Is there a similar document already available on Orphanet?
☐ no
☐ yes, in another language
☐ yes, in the same language. If yes, the present document has an added-value:
- ☐ no (it is less interesting). Specify**:
- ☐ yes (it is more interesting). Specify**:
- ☐ it is different. Specify**:

*(**e.g. document more up-to-date, more topics are treated, the target population is different, clarity of information, complementary information provided,…)*

Additional detailed criteria to assess the overall quality of review articles are as follows:

- ✓ Article is retained only if at least one expert of the disease has written and/or revised the document.
- ✓ Article is not retained if the text contains drug trade names or if invitations for donations, or patient testimonies are over-stressed.
- ✓ Pictures of patients or families that are not useful for understanding the content are considered as "not relevant" illustrations.

☞ **For "Other Website of interest"**

Inclusion criteria*	Exclusion criteria*
The source and the entity responsible for maintaining the site are clearly stated	A website of a national patient organisation of a country belonging to the Orphanet consortium as they are already registered in the Orphanet database (see Exception 1).
The target audience is clearly mentioned or obvious; the information is appropriate to the audience level.	A website of an expert center or a research network that is already listed in the Orphanet database (see Exception 2).
The website site provides accurate, science-based information that complements the information found on Orphanet. The site mentions the involvement of health professionals in the production of the content (as writers, or reviewers) or is from an academic or national institution. The site does not massively reproduce information from other websites.	A website mainly constituted by a forum/blog.
The primary purpose of the site is educational.	A commercial or industry website (see Exception 2).
Dates of creation/updates are mentioned.	A website of a patient advocacy group.
The site is available consistently.	Non-relevant information, and/or spelling mistakes or any other criteria indicating a lack of seriousness of the website.
	A website that displays advertising.

At least three inclusion criteria are needed to approve the publication and one exclusion criteria is sufficient to refuse it.

*The inclusion/exclusion criteria are derived from the HON Code principles. These criteria are expected to be found in a high-quality medical and health website certified by HONCode.

Exception 1: Exceptions can be made if some specific information (for instance a patient guide) is available on the website but located on a page that is difficult to retrieve. In this case, a link to the relevant specific page is added as other website, even if the website is already linked in another Orphanet section.

Exception 2: Exceptions can be made (*e.g.* with pharmaceutical companies) when the website provides objective added-value information, not commercially oriented.

2. List of monitored journals

Am. J. Human Genetics
Am. J. Med. Genet.
Ann. Int. Med.
Annals of neuro.
Annals of the Rheum. Dis.
Arch. Pediatr.
Arthritis and Rheumatology
Arthritis Care & Research
Blood
Bone
Brain
Circulation
Diabetes
Eur. Heart Journal
European Journal of Haematology
Eur. J. of Hum. Genet.
Eur. J. of Med. Genet.
Expert Opin Orphan Drugs
Gastroenterology
Gene Therapy
Genet Med
Gut
Hepatology
Hum. Mol. Genet.
Hum. Mutat.
J. clin. Endocrino. and Metabo.
J. Clin. Inv.
J. Invest. Dermato.
J. of Med. Genet.
JAMA
JAMA Neurology
Lancet
Lancet Inf. Disease
Lancet Neuro.
Lancet Oncology
Molecular therapy
Nature
Nature Genetics
Nature Medicine
Neurology
OJRD
Pediatric res.
Pediatrics
Plos Genetics
PNAS
Progr. Retin. Eye Res.
Sci.Transl. Med.
Science
Stem cells
The New England Journal of Medecine
Translational Science of Rare Diseases
BMC Medicine
British Journal of Haematology
Bulletin du Cancer
Cell Reports
Cell Stem Cell
Clinical Genetics
Cochrane Reviews (CDSR)
Current Rheumathology Reports
European Journal of Internal Medicine
Familial Cancer
Genome Medicine
Human Genetics
International Journal of RD & Orphan Drugs
Journal of Allergy and Clinical Immunology
Journal of the American Society of Nephrol
Journal of Neuromuscular Diseases
Journal of Rare Diseases: Rsch & Treatment
Journal of Rare Disorders: Diagno & Therapy
Intractable & RD Research
Molecular Genetics and Metabolism
Molecular Syndromology
Muscle & Nerve
Nature Neuroscience
Nature Reviews Clinical Oncology
Nature Reviews Endocrinology
Nature Reviews Immunology
Nature Reviews Nephrology
Nature Reviews Rheumathology
Neuromuscular Disorders
Orphan Drugs: Research and Reviews
Revue de Médecine Interne
Stem Cell Reports
Stem Cells Translational Medicine
The Journal of Rare Disorders

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

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