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User guide: How to make a suggestion using the Orphanet curation platform

www.orpha.net

www.orphadata.org

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I. Introduction

1. What is the curation platform?

The Orphanet curation platform (OCP) is a community-driven knowledge curation platform for rare disorders based on Orphanet scientific content.

The OCP will allow users (the wider expert community) to propose corrections or modifications for each entry and to meet virtually. Edit and suggestions can be reviewed by the curators to accept or further query the submitter as part of the quality control. Changes validated in the OCP will be incorporated to the Orphanet database and ultimately to the Orphanet Rare Disease Ontology (ORDO).

The curation platform creates an interactive knowledge curation process that enables input of new knowledge in Orphanet by 'expert-crowd sourcing', significantly reducing the resource intense curation processes while maintaining curatorial control over the high quality of the Orphanet data. It will allow for traceability and transparency of data validation.

It is a great way of engaging the community and is a virtual meeting room for experts.

Orphanet is a unique resource, gathering and improving knowledge on rare diseases so as to improve the diagnosis, care and treatment of patients with rare diseases. As this initiative became a European endeavour, supported by grants from the European Commission, Orphanet has gradually grown to a Consortium of 40 countries, within Europe and across the globe.

Orphanet works towards meeting three main goals:

- Improve the visibility of rare diseases in the fields of healthcare and research by maintaining the Orphanet rare disease nomenclature (ORPHANumbers): providing a common language to understand each other across the rare disease field
- Provide high-quality information on rare diseases and expertise, ensuring equal access to knowledge for all stakeholders: orientating users and actors in the field in the mass of information online

- Contribute to generating knowledge on rare diseases: piecing together the parts of the puzzle to better understand rare diseases

The OCP is co-developed by Orphanet and the Garvan Institute in Australia with the support of the Orphanet Australia country coordinator, Hugh Dawkins. It is a pilot project developed to allow curators and experts to visually explore the Orphanet scientific data through an easy-to-use web interface. It enables to connect with contributing expert in a more dynamic and efficient manner.

2. What will you find on the curation platform?

The curation platform website (<https://curation.orphanet.org>) banner contains five buttons (Figure 1):

- (1) **“Disorders”**: to browse the Orphanet clinical classification to search for a disorder and suggest the creation of a new disorder (button “New Disorder”).
- (2) **“Genes”**: to browse genes, and their associated disorders, registered in the Orphanet database.
- (3) **“Human Phenotype Ontology”** (HPO): to browse phenotypes using HPO’s standardized vocabulary of phenotypic abnormalities encountered in human disease.
- (4) **“Suggestions”**: to access to all suggestions of modification or creation made on the curation platform.
Note: This button is displayed only when logged in.
- (5) **“Account”**: to log in/out, modify your account settings, and edit your expertise.

On the homepage, you will also find:

- (6) Two search boxes to search for a disorder, gene, or phenotype using the full name or part of the name (autocomplete search, see section 3. Search for a disease) of the searched entity.
- (7) A Classification browser displaying the top level disorder classifications.

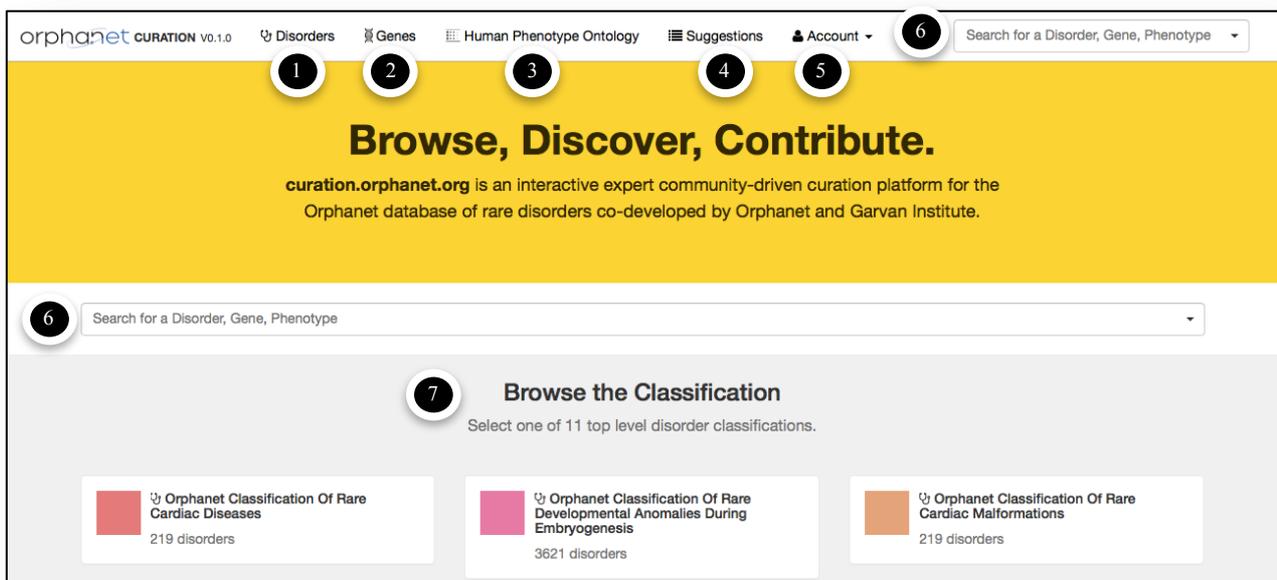


Figure 1

II. Use the curation platform

In the current version of the platform, you can contribute to the improvement of summary texts and nomenclature information of Orphanet. To make suggestions, you must create an account and declare your expertise.

1. Account registration

In order to be able to post a suggestion of modification or new content on the [curation platform](#), registration is necessary. To register (Figure 2):

(1) From the [curation platform](#) homepage, click on “Account” and choose “Register” from the drop-down list.

(2) Fill in the registration form with the requested personal information then click “Register”.

Note: For the username only lowercase letters and no spaces are allowed.

(3) At the next registration window, specify your area(s) of expertise by clicking on the corresponding disorder(s), or group(s) of disorders, in the Orphanet classification(s).

Note: This step is not required for registration but is recommended for experts in order to enable the Orphanet team to select appropriate external medical expert reviewers when processing suggestions that need external expertise.

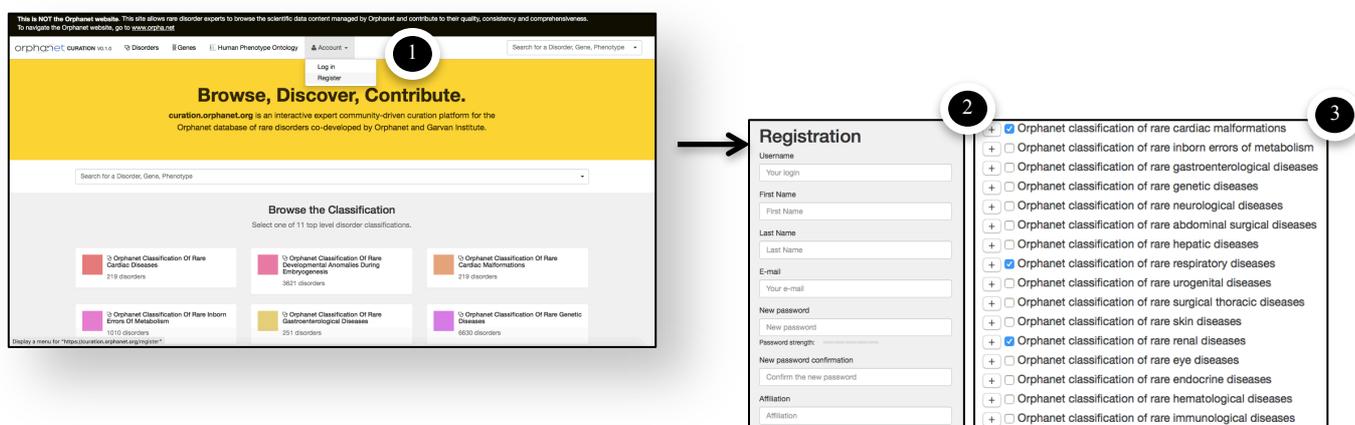


Figure 2

After completing the above steps, your account is immediately created and you will receive an email confirming its creation. You can immediately start using the curation platform.

Note: You can post suggestions immediately but cannot access discussions on a suggestion unless you have been invited to the discussion by an Orphanet curator or another expert already in the discussion.

2. Change user profile

If you need to change your personal account information (Figure 3):

- (1) Click on the “Account” button displayed on the website banner and from the drop-down list select “Settings”.
- (2) Change the desired information in the user settings window.
- (3) Click “Save”.

The screenshot shows a user interface for changing profile information. On the left, a navigation menu under the 'Account' header includes 'Settings', 'Expertise', 'Password', and 'Log out'. The 'Settings' option is highlighted with a circled '1'. An arrow points from this menu to a settings form on the right. The form contains the following fields: 'First Name' with the value 'John', 'Last Name' with 'Smith', 'E-mail' with 'john.smith@email.com', and 'Affiliation' with 'Saint Mary's Hospital'. At the bottom of the form is a blue 'Save' button, which is circled with a '3'. A circled '2' is located in the top right corner of the form's border.

Figure 3

If you need to change your areas of expertise (Figure 4):

- (1) Click on the “Account” button displayed on the website banner and from the drop-down list select “Expertise”.
- (2) Check the desired boxes of the classification (corresponding to the level “disorder”) for which you wish to declare an expertise. To uncheck a box, click one more time on the box.
- (3) Click “Save”.

The screenshot shows a user interface for selecting areas of expertise. On the left, a navigation menu under the 'Account' header includes 'Settings', 'Expertise', 'Password', and 'Log out'. The 'Expertise' option is highlighted with a circled '1'. An arrow points from this menu to a form on the right. The form displays a list of Orphanet classifications with checkboxes. The checked items are: 'Disorder of amino acid and other organic acid metabolism', 'Disorder of biogenic amine metabolism and transport', and 'Disorder of lipid metabolism'. At the bottom of the form is a blue 'Save' button, which is circled with a '3'. A circled '2' is located in the top right corner of the form's border.

Figure 4

3. Search for a disease

You can find the disorder you want to make a suggestion on by entering the name of the disease in one of the two search boxes of the curation platform homepage (Figure 5(1)).

Note 1: The search engine features an autocomplete function to provide suggestions while you type in either search field. The Orphanet databases of disorders, genes, and phenotypes are simultaneously searched and the entity type (a) is also listed in the autocomplete suggestion. To access the disorder, click on the autocomplete suggestion followed by the mention “Disorder”.

Note 2: As the search engine is still being improved, searching for a disease via the search boxes might occasionally return no results, even if the disorder is registered in the Orphanet database. In this case, the disorder should be searched via the classification.

Note 3: Searching via Orpha number is not possible at this time.

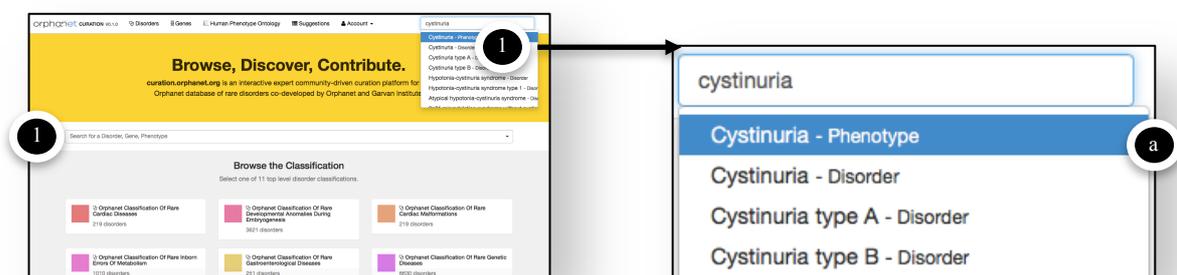


Figure 5

You can also find a disorder by browsing the classification (Figure 6 (1)) by clicking on one of the Orphanet top level disorder classifications.

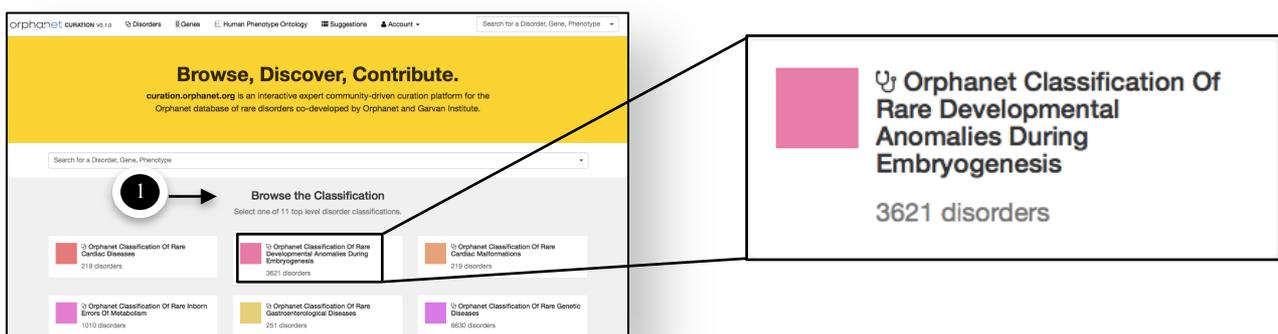


Figure 6

Once you have clicked on an Orphanet clinical classification, you can access all the “children” of the clinical classification group either by (Figure 7):

- (1) clicking on the chevron next to the name of the group in the left panel; or
- (2) scrolling down to the “Children” section of the page.

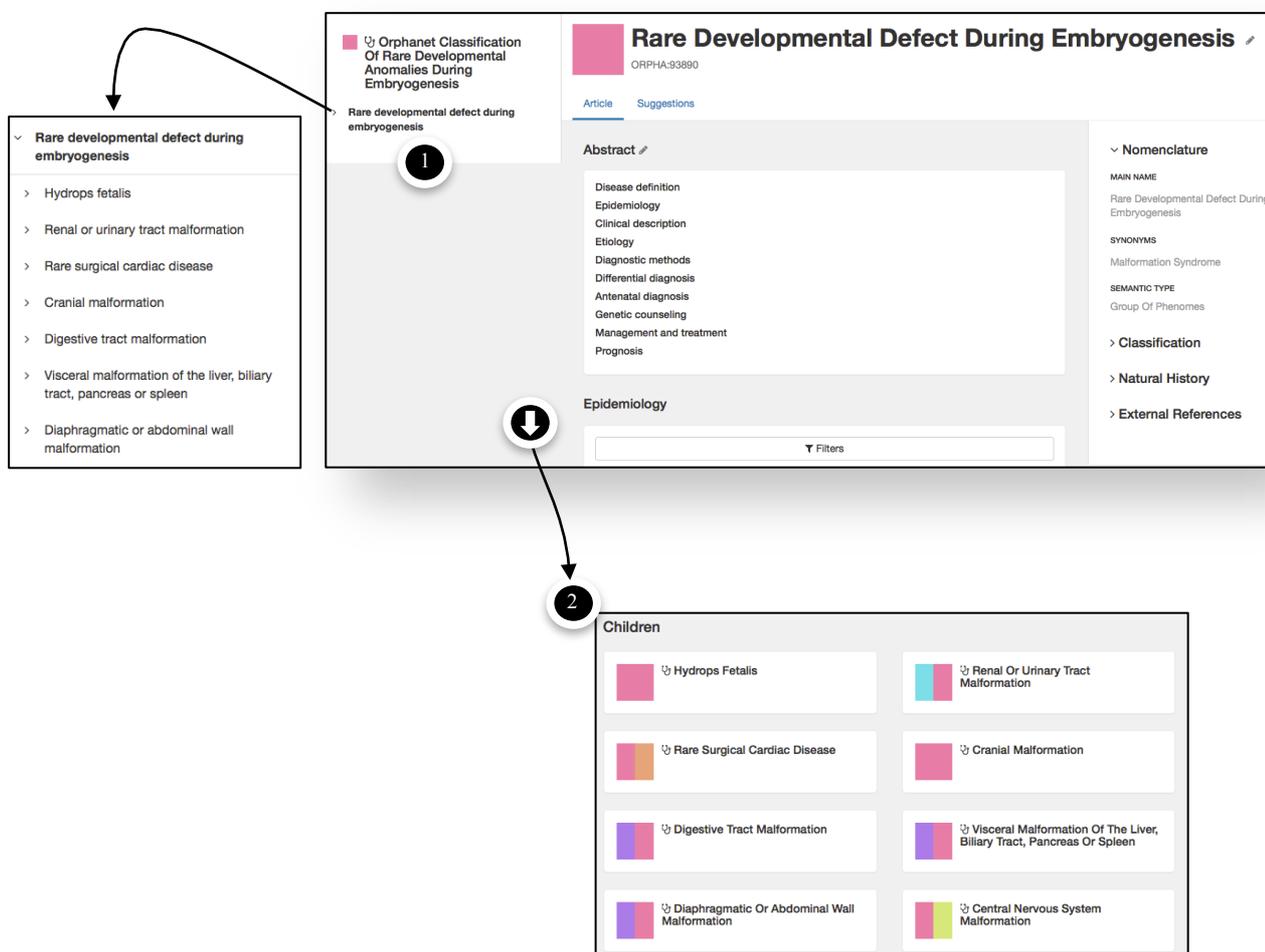


Figure 7

Once the disease found, click on its name. The disease page will have the following elements (Figure 8):

orphanet CURATION v0.1.0 **Disorders** [Genes](#) [Human Phenotype Ontology](#) [Suggestions](#) [Account](#)

Orphanet Classification Of Rare Genetic Diseases

- Rare genetic disease
 - Rare genetic developmental defect during embryogenesis
 - Rare developmental defect with connective tissue involvement
 - Marfan syndrome**
 - Desbuquois syndrome
 - Lethal Larsen-like syndrome
 - Cutis laxa
 - Autosomal dominant Larsen syndrome
 - Pseudoxanthoma elasticum
 - Grange syndrome
 - Pseudodiastrophic dysplasia
 - Ehlers-Danlos syndrome

- Genetic digestive tract malformation
- Genetic visceral malformation of the liver, biliary tract, pancreas or spleen
- Genetic respiratory or mediastinal
- Biological anomaly without phenotypic characterization
- Chromosomal anomaly
- Rare genetic tumor
- Rare genetic skin disease
- Rare inborn errors of metabolism
- Rare genetic cardiac disease
- Rare genetic renal disease
 - FRAXF syndrome
- Rare genetic eye disease
- Rare genetic hepatic disease
- Rare genetic respiratory disease
- Rare genetic urogenital disease
- Rare genetic endocrine disease
 - Sea-blue histiocytosis
- Rare genetic hematologic disease
 - Rare genetic gastroenterological disease
 - Rare genetic bone disease

Marfan Syndrome

ORPHA:558

[Article](#) [Suggestions](#) 1

Abstract

Disease definition
Marfan syndrome is a systemic disease of connective tissue characterized by a variable combination of cardiovascular, musculo-skeletal, ophthalmic and pulmonary manifestations.

Epidemiology
The prevalence is estimated at 1/5,000 and there is no difference between sexes.

Clinical description
Symptoms can appear at any age and vary greatly between individuals even within the same family. Cardiovascular involvement is characterized by 1) progressive dilation of the aorta accompanied by an increased risk of aortic dissection, which affects prognosis; the aortic dilation can result in a leaky aortic valve; and 2) mitral insufficiency, which can be complicated by arrhythmias, endocarditis or cardiac insufficiency. Skeletal involvement is often the first sign of the disease and can include dolichostenomelia (excessive length of extremities), large size, arachnodactyly, joint hypermobility, scoliotic deformations, acetabulum protrusion, thoracic deformity (pectus carinatum or pectus excavatum), dolichocephaly of the anteroposterior axis, micrognathism or malar hypoplasia. Ophthalmic involvement results in axile myopia, which can lead to retinal detachment and lens displacement (ectopia or luxation) are characteristic signs. Ocular complications, particularly lens ectopia, can lead to blindness. Cutaneous signs (vergetures), a risk of pneumothorax and dural ectasia can also occur.

Epidemiology

Geography	Type	Mean Value	Class of Prevalence	Source	Validation Status
Europe	Annual incidence	25.0	1-5 / 10 000	16325700[PMID],ORPHANET	Not yet validated
Worldwide	Point prevalence	15.0 / 100,000	1-5 / 10 000	20301510[PMID]	Not yet validated
Europe	Point prevalence	20.0 / 100,000	1-5 / 10 000	[EXPERT]	Validated

Genotype

- Disease-causing germline mutation(s) in **FBN1** Assessed
- Disease-causing germline mutation(s) in **TGFBR2** Assessed

Phenotype

- Pectus Excavatum Frequent (79-90%)
- Limitation Of Joint Mobility Occasional (29-5%)
- Abnormality Of The Aortic Valve Occasional (29-5%)
- Fatigue Very frequent (99-90%)
- Chest Pain Occasional (29-5%)
- Dural Ectasia Frequent (79-90%)

Children

- Marfan Syndrome Type 1
- Marfan Syndrome Type 2

Parents

- Rare Developmental Defect With Connective Tissue Involvement
- Marfan And Marfan-Related Disorder

Suggestion pencils

Summary text

Nomenclature section

Clinical classifications the disorder belongs in

Epidemiological data

Genes associated

Phenotypes

Included groups

Inclusive group(s)

Classification panel

Figure 8

4. Make a suggestion

You can suggest a new content (only for summary texts, nomenclature, and/or new disorder at this time) by clicking on one of the pencil icons (Figure 8) to open a suggestion form.

Note: *You can view all the suggestions that have previously been posted for the disorder by clicking on the “Suggestions” tab (Figure 8(1)) of the disease page. Each suggestion is treated independently therefore multiple suggestions for the same disease may not be processed simultaneously by the Orphanet team.*

- **To make a suggestion using the summary text form (Figure 9):**

- (1) Click on the pencil icon next to the word “Abstract”. A pop-up window opens. It contains sections that must be filled in with the modified or new information.
- (2) Enter the new text in each section (one or several) for which you want to make a suggestion. *(Note: In case of an update, the original text will be displayed by default in each section. You can modify it directly to submit your suggested changes.)*

Don't forget the field “Description REQUIRED” is mandatory. Please enter here the scientific references (PMID, etc.) supporting the new content.

- (3) Click “Send” to finalize your suggestion.

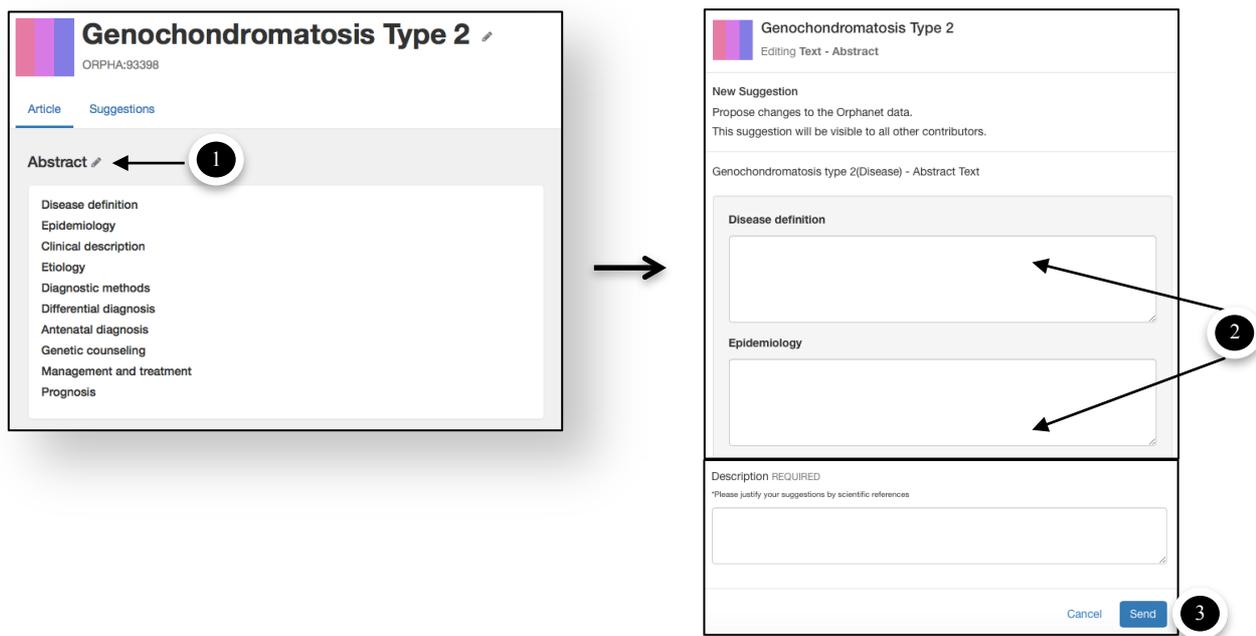


Figure 9

- **To make a suggestion using the nomenclature form (main name and/or synonym) (Figure 10):**

- (1) Click on the pencil icon next to “Synonym”, “Main name”, or the disease name on the disease page. A pop-up window opens with sections that must be filled in.
- (2) Write the new value in the box “Please suggest a new value”. In case of suggestion of a new synonym, don't forget to click “Add Synonym”.

Don't forget the field “Description REQUIRED” is mandatory. Please enter here the scientific references (PMID, etc.) supporting the new content.

(3) Click “Send”. Your suggestion has been sent for curation by the Orphanet team.

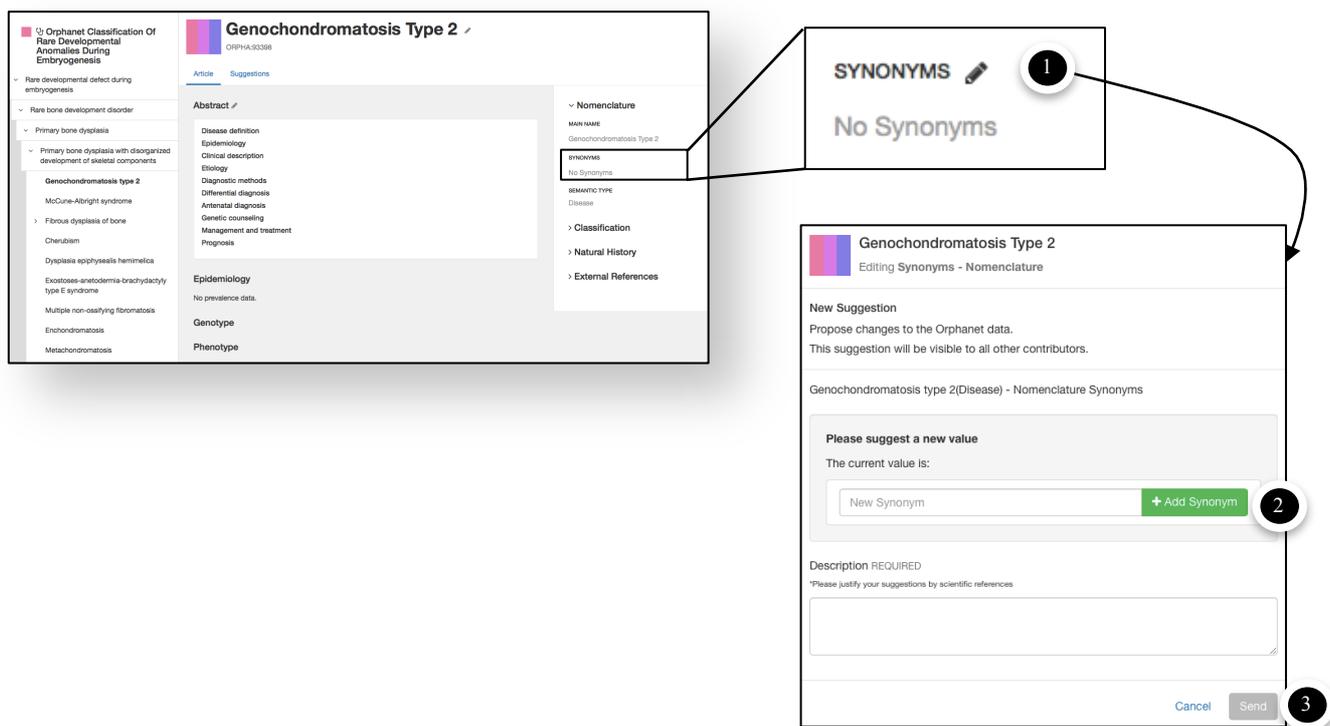


Figure 10

• To make a suggestion using the new disorder form (Figure 11):

- (1) Click on the button “Disorders” on the Orphanet website banner to access the Disorders page.
- (2) Click on the button “New Disorder”. A pop-up window opens with sections to fill in.
- (3) Write the name of the new disorder in the box “New Disorder REQUIRED”.

Don’t forget the field “Description REQUIRED” is mandatory. Please enter here the scientific references (PMID, etc.) supporting the new content.

(4) Click “Send”. Your suggestion has been sent for curation by the Orphanet team.

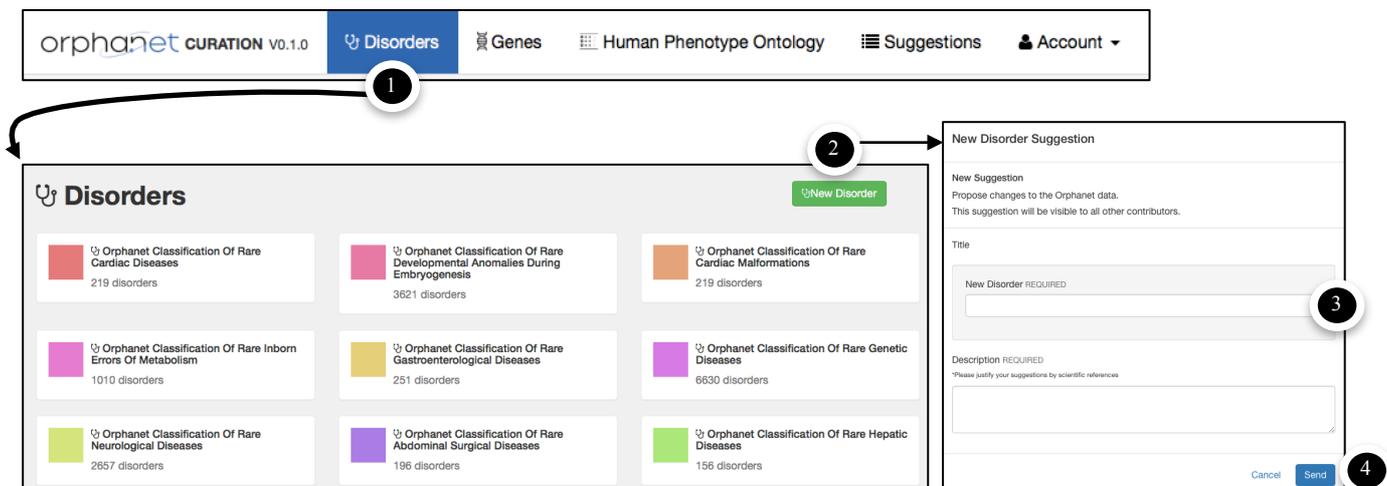


Figure 11

For your information, the suggestion will be curated by the Orphanet team (Figure 12). During curation, the Orphanet team may contact you as an expert to take part in the reviewing process of your suggestion(s) or suggestions from other contributors. In this case you will receive an email inviting you to the discussion where you will be able to post comments. Once the reviewing process of your suggestion is completed, you will receive an email informing you of the conclusion reached by the Orphanet team about your suggestion.

5. Suggestion life cycle

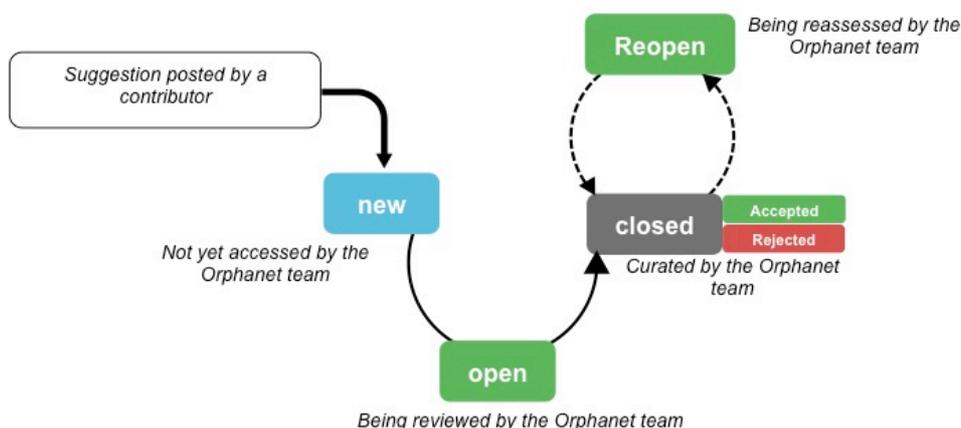


Figure 12. Life cycle of a suggestion on the curation platform.

- **To post a comment to the discussion** (Figure 13):

- (1) Write your text in the comment box.
- (2) Click “Post”.

Figure 13

- **To invite an expert to join a discussion:**

Any expert already participating in an ongoing discussion will be able to invite new expert(s) to take part in the same discussion. This function is currently being developed.

III. Annexes

1. Understanding the color code used in the curation platform

The same color code is used in all sections of the curation platform website, i.e. Disorders, Genes, and Human Phenotype Ontology sections. Every entity in the Orphanet database is classified under one or more clinical categories and each category is represented by a different color on the curation platform.

For example: The disease **Cystinuria** has three colors (Figure 14 (4)) since it is classified under:

- (1) “rare genetic disease” (violet),
- (2) “rare inborn errors of metabolism” (pink), and
- (3) “rare renal diseases” (blue).

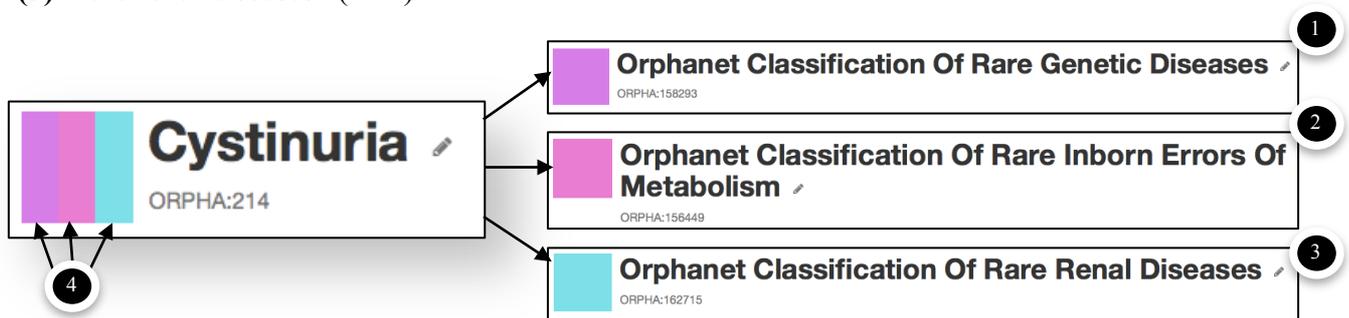


Figure 14

2. Definitions

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

Clinical classification: The Orphanet clinical classifications of rare disorders are based on clinical criteria. Disorders are classified by body system (e.g. respiratory, digestive) in order to correspond to medical domains that are mostly organized in such way. Additional criteria (pathological, etiological, etc.) are used in details of the classification when relevant in clinical practice. Every Orphanet disease is classified under one or more categories. Indeed, Orphanet classifications are multi-hierarchical, polyparental classifications.

Contributor: Any user registered on the curation platform. They can make a suggestion of creation or modification(s) for the Orphanet database of rare disorders (see Figure 15).

Curation platform: Interactive management platform for the Orphanet database of rare disorders that allows experts on rare diseases to update and validate the information provided by Orphanet.

External medical expert reviewer: Expert contributing to the revision of the information on a disorder 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 the Orphanet website. He/She is referred to as “Expert Reviewer” on the Orphanet website.

Internal medical reviewer: Medical doctor of the Orphanet team reviewing, for editorial and medical consistency, all summary text information produced by the Orphanet editorial teams and who validates the publication once the revision process is finalized.

Medical writer: Orphanet information scientist producing the textual information on a disorder according to the procedure called “[Creation and Update of Disease Summary Texts for the Orphanet Encyclopaedia for Professionals](#)”.

Orphanet curator: Member of the Orphanet team (see Figure 15) interacting with experts on the curation platform in order to update and/or create information for the Orphanet database according to the corresponding procedure. The Orphanet curator can also make suggestions on the curation platform.

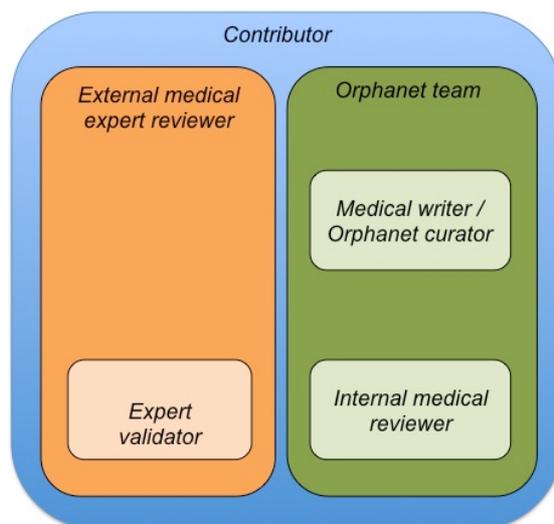


Figure 15. Overview of the actors involved in the creation and update of disease summary texts for the Orphanet Encyclopaedia for Professionals.

Orphanet disease summary texts: Disease definitions or abstracts produced by Orphanet editorial teams in English. They are translated into several different languages.

Status (of suggestion): Any suggestion posted on the curation platform has one of four statuses. “**New**”: suggestion that has not yet been accessed for curation by the Orphanet team; “**Open**”: suggestion being reviewed by the Orphanet team; “**Closed**”: suggestion curated and for which a verdict has been added by the Orphanet team; “**Reopen**”: suggestion being reassessed after being closed by an Orphanet team.

Suggestion (curation platform): Proposition for modification (i.e. update, creation, deletion) of the content in the Orphanet database made by a contributor on the curation platform. Suggestions are processed by the Orphanet team (Figure 12) and/or can be evaluated by Orphanet curators and external medical expert reviewers via discussions on the curation platform. (See also “Status (of suggestion)”.)

The Orphanet team thanks you very much for your cooperation.

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Reference documentation: For general information related to the Orphanet mission, please read the document “[Orphanet Standard Operating Procedures](#)”.

For information about the procedure followed by the Orphanet editorial team for the creation and update of Orphanet summary information, please read the document “[Creation and Update of Disease Summary Texts for the Orphanet Encyclopaedia for Professionals](#)”.

This document is updated as often as necessary and at least once a year by the Orphanet editorial team. The most up-to-date version is available on the [Orphanet website](#).

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