



Orphanet Curation Platform

ERN Conference, Brussels-Belgium, 26-27 April 2017

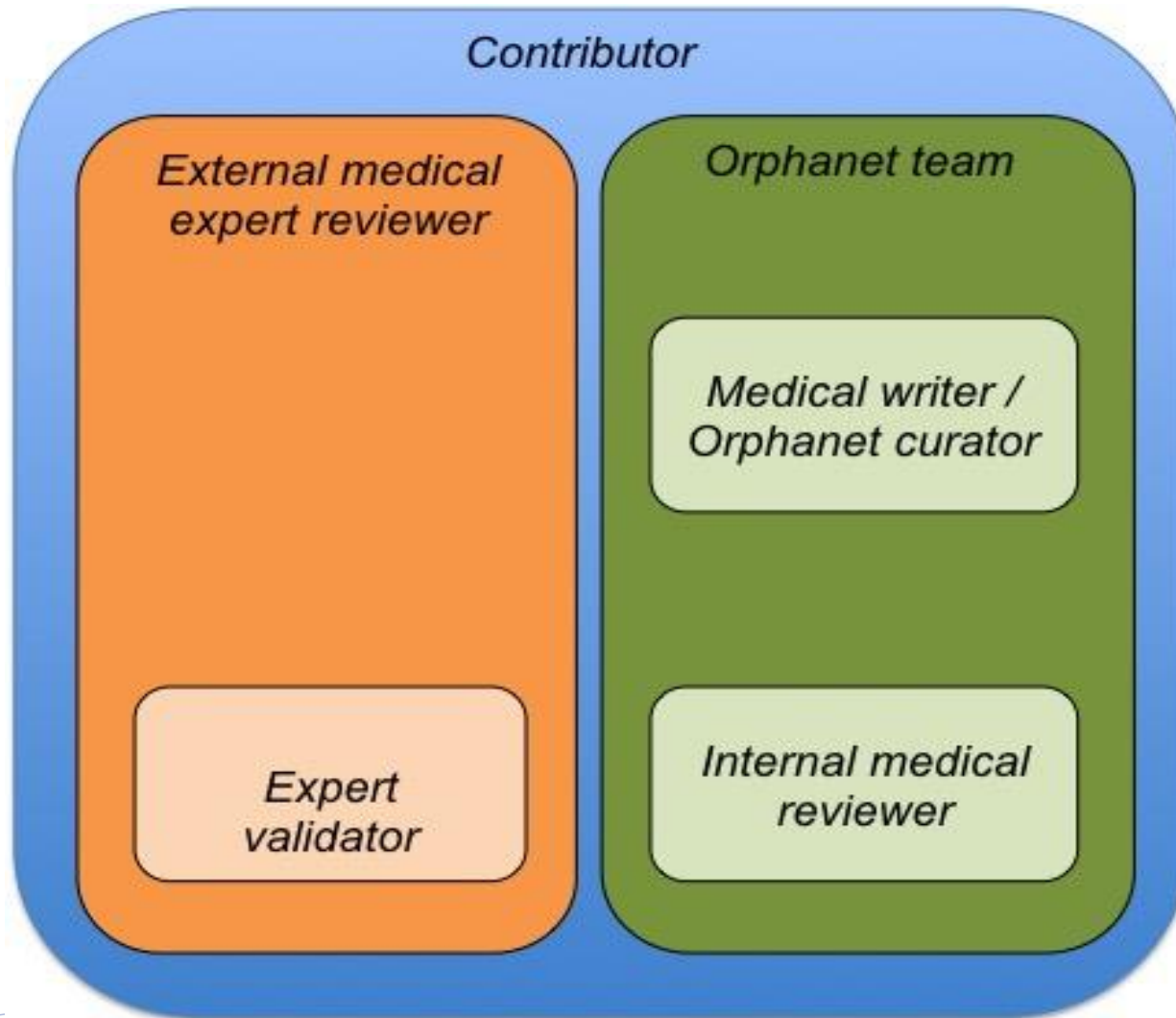
Montserrat ALFARO, MD
Internal Medical Reviewer
Orphanet - France

What is the Orphanet Curation Platform?

- It is a community-driven curation platform for rare disorders based on Orphanet's scientific content.
- It is a pilot project being co-developed by Orphanet and the Garvan Institute (Australia), with the support of the Orphanet Australia's country coordinator.
- Goals:
 - Allow users to visually explore the Orphanet scientific data
 - Enable contributing experts and the Orphanet team to connect in a more dynamic and efficient manner
 - Facilitate communication between the contributing experts and the Orphanet team in order to maintain the Orphanet database with the most up-to-date information

Orphanet Curation Platform

« Key Players »



Getting Started

<http://curation.orphanet.org>

orphanet CURATION v0.1.0

🔗 Disorders

🧬 Genes

📋 Human Phenotype Ontology

👤 Account ▾

Search for a Disorder, Gene, Phenotype ▾

Browse, Discover, Contribute.

curation.orphanet.org is an interactive expert community-driven curation platform for the Orphanet database of rare disorders co-developed by Orphanet and Garvan Institute.

Search for a Disorder, Gene, Phenotype ▾

Browse the Classification

Select one of 11 top level disorder classifications.



🔗 Orphanet Classification Of Rare Cardiac Diseases

219 disorders



🔗 Orphanet Classification Of Rare Developmental Anomalies During Embryogenesis

3621 disorders



🔗 Orphanet Classification Of Rare Cardiac Malformations

219 disorders



🔗 Orphanet Classification Of Rare Inborn Errors Of Metabolism

1010 disorders



🔗 Orphanet Classification Of Rare Gastroenterological Diseases

251 disorders



🔗 Orphanet Classification Of Rare Genetic Diseases

6630 disorders



🔗 Orphanet Classification Of Rare Neurological Diseases

2657 disorders



🔗 Orphanet Classification Of Rare Abdominal Surgical Diseases

196 disorders



🔗 Orphanet Classification Of Rare Hepatic Diseases

156 disorders

Create an Account

orphanet CURATION V0.1.0 [Disorders](#) [Genes](#) [Human Phenotype Ontology](#) [Account](#)


[Log in](#)
[Register](#)

Browse, Discover, Contribute.


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Select one of 11 top level disorder classifications.



Orphanet Classification Of Rare Cardiac Diseases
219 disorders



Orphanet Classification Of Rare Developmental Anomalies During Embryogenesis
3621 disorders



Orphanet Classification Of Rare Cardiac Malformations
219 disorders

orphanet CURATION V0.1.0 [Disorders](#) [Genes](#) [Human Phenotype Ontology](#) [Suggestions](#) [Account](#)

valtest2 / Expertise

Expertise settings for valtest2

- ☒ Orphanet classification of rare cardiac diseases
 - ☐ Rare cardiac disease
 - ☐ Cardiomyopathy
 - ☐ Arrhythmic right ventricular cardiomyopathy
 - ☐ Hypertrophic cardiomyopathy
 - ☐ Rare familial disorder with hypertrophic cardiomyopathy
 - ☒ Non-familial hypertrophic cardiomyopathy
 - ☐ Dilated cardiomyopathy
 - ☐ Restrictive cardiomyopathy
 - ☐ Unclassified cardiomyopathy
 - ☐ Idiopathic giant cell myocarditis
 - ☐ Rare cardiac tumor
 - ☐ Rare cardiac rhythm disease
 - ☐ LMNA-related cardiocutaneous progeria syndrome
 - ☐ Orphanet classification of rare developmental anomalies during embryogenesis
 - ☐ Orphanet classification of rare cardiac malformations

Registration

Username

First Name

Last Name

E-mail

Your e-mail is required.

New password

Your password is required.

Password strength:

New password confirmation

Affiliation

Warning: You will register to the Orphanet database curation platform.

This platform has the explicit aim to allow experts on rare diseases to update and validate the informations provided by Orphanet. For that reason, we kindly ask you to provide your background when registering, by telling us your affiliation and your domain of expertise. Once registered, you will be able to complete this information in your profile, what we encourage you to do.

Please note that any personal query or demand for information will be answered by this platform. You can consult the list of personal information services at [helplines page in Orphanet](#)

Thank you for your commitment to the quality of the Orphanet database.

[Register](#)

How to Search for a Disease?

- 1) Using one of the search engines
- 2) Browsing the classification

The screenshot shows the Orphanet Curation website interface. At the top, there is a navigation bar with links for 'Disorders', 'Genes', 'Human Phenotype Ontology', and 'Account'. A search bar is located in the top right corner, labeled 'Search for a Disorder, Gene, Phenotype'. A red arrow with the number '1' points to this search bar. Below the navigation bar is a large yellow banner with the text 'Browse, Discover, Contribute.' and a description of the platform. Below the banner is another search bar, also labeled 'Search for a Disorder, Gene, Phenotype', with a red arrow and the number '1' pointing to it. Below the search bar is a section titled 'Browse the Classification' with a subtitle 'Select one of 11 top level disorder classifications.' A red arrow with the number '2' points to this section. The section contains a grid of 9 cards, each representing a different classification category with a color-coded icon, the category name, and the number of disorders.

Classification Category	Number of Disorders
Orphanet Classification Of Rare Cardiac Diseases	219 disorders
Orphanet Classification Of Rare Developmental Anomalies During Embryogenesis	3621 disorders
Orphanet Classification Of Rare Cardiac Malformations	219 disorders
Orphanet Classification Of Rare Inborn Errors Of Metabolism	1010 disorders
Orphanet Classification Of Rare Gastroenterological Diseases	251 disorders
Orphanet Classification Of Rare Genetic Diseases	6630 disorders
Orphanet Classification Of Rare Neurological Diseases	2657 disorders
Orphanet Classification Of Rare Abdominal Surgical Diseases	196 disorders
Orphanet Classification Of Rare Hepatic Diseases	156 disorders

Browsing the Classification

The screenshot displays the Orphanet website interface. On the left, a navigation menu is shown, highlighting the path: Orphanet Classification Of Rare Developmental Anomalies During Embryogenesis > Rare developmental defect during embryogenesis > Craniosynostosis. The main content area shows the details for 'Rare Developmental Defect During Embryogenesis' (ORPHA:93890). The page includes a search bar, navigation tabs for 'Article' and 'Suggestions', and a list of related terms under the 'Abstract' section. The right sidebar contains sections for 'Nomenclature', 'Classification', 'Natural History', and 'External References'.

Orphanet Classification Of Rare Developmental Anomalies During Embryogenesis

- ▼ Rare developmental defect during embryogenesis
 - › Hydrops fetalis
 - › Renal or urinary tract malformation
 - › Rare surgical cardiac disease
 - ▼ Cranial malformation
 - Cleidocranial dysplasia
 - › **Craniosynostosis**
 - Acalvaria
 - Craniodiaphyseal dysplasia
 - Craniofacial dyssynostosis
 - Hypomandibular faciocranial dysostosis

orphanet CURATION v0.1.0 Disorders Genes Human Phenotype Ontology Suggestions Account Administration

Search for a Disorder, Gene, Phenotype

Rare Developmental Defect During Embryogenesis ORPHA:93890

Article Suggestions

Abstract

- Disease definition
- Epidemiology
- Clinical description
- Etiology
- Diagnostic methods
- Differential diagnosis
- Antenatal diagnosis
- Genetic counseling
- Management and treatment
- Prognosis

Epidemiology

Filters

▼ Nomenclature

MAIN NAME

Rare Developmental Defect During Embryogenesis

SYNONYMS

Malformation Syndrome

SEMANTIC TYPE

Group Of Phenomes

› Classification

› Natural History

› External References

Browsing the Classification

orphanet CURATION V0.1.0

Disorders Genes Human Phenotype Ontology Suggestions Account Administration

Search for a Disorder, Gene, Phenotype

Orphanet Classification Of Rare Developmental Anomalies During Embryogenesis

Rare developmental defect during embryogenesis

Rare Developmental Defect During Embryogenesis

ORPHA:93890

Article Suggestions

Abstract

Disease definition
Epidemiology
Clinical description
Etiology
Diagnostic methods
Differential diagnosis
Antenatal diagnosis
Genetic counseling
Management and treatment
Prognosis

Epidemiology

Filters

Nomenclature

MAIN NAME

Rare Developmental Defect During Embryogenesis

SYNONYMS

Malformation Syndrome

SEMANTIC TYPE

Group Of Phenomes

Classification

Natural History

External References

How to Post a Summary Text Suggestion?

orphanet CURATION v0.1.0

Disorders Genes Human Phenotype Ontology Suggestions Account Administration

Search for a Disorder, Gene, Phenotype

Orphanet Classification Of Rare Developmental Anomalies During Embryogenesis

- Rare developmental defect during embryogenesis
 - Hydrops fetalis
 - Renal or urinary tract malformation
 - Rare surgical cardiac disease
 - Cranial malformation

Cleidocranial dysplasia

- Craniosynostosis
- Acalvaria
- Craniodiaphyseal dysplasia
- Craniofacial dyssynostosis
- Hypomandibular faciocranial dysostosis
- Delayed membranous cranial ossification

Cleidocranial Dysplasia ORPHA:1452

Article Suggestions

Abstract

Disease definition
Cleidocranial dysplasia (CCD) is a rare genetic developmental abnormality of bone characterized by hypoplastic or aplastic clavicles, persistence of wide-open fontanels and sutures and multiple dental abnormalities.

Epidemiology
The prevalence of CCD is 1/1,000,000, with higher rates in groups with a founder effect. The disorder is found in many ethnic groups and no sex predilection has been reported. It may be underdiagnosed because of the number of relatively mild cases.

Clinical description
There is an extremely wide range of clinical manifestations (even within the same family) from isolated dental anomalies to severe malformations with functional repercussions. The main clinical signs are hypoplasia or aplasia of the clavicles with narrow, sloping shoulders that can be approximated anteriorly, delayed fusion of cranial sutures with large, wide-open fontanels at birth that may persist throughout life, and a wide spectrum of dental anomalies including abnormal dentition, uniform or chaotic supernumerary teeth (hyperdontia) in the primary and secondary dentition resulting in crowding and malocclusion, retention of deciduous teeth, delayed eruption of secondary dentition and failure to shed the primary teeth. The dental manifestations may affect articulation and mastication. Other signs include broad flat forehead, hypertelorism, midface hypoplasia,

Nomenclature

MAIN NAME
Cleidocranial Dysplasia

SYNONYMS
Cleidocranial Dysostosis

SEMANTIC TYPE
Malformation Syndrome

Classification

Natural History

External References

Cleidocranial Dysplasia
Editing Text - Abstract

New Suggestion
Propose changes to the Orphanet data.
This suggestion will be visible to all other contributors.

Cleidocranial dysplasia(Malformation syndrome) - Abstract Text

Disease definition

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The prevalence of CCD is 1/1,000,000, with higher rates in groups with a founder effect. The disorder is found in many ethnic groups and no sex predilection has been reported. It may be underdiagnosed because of the number of relatively mild cases.

Management and treatment

Management of dental anomalies is very important with the aim of achieving optimal function and esthetics. Options include removal of retained deciduous, supernumerary and abnormal permanent teeth. Dental surgery should be considered for unerupted teeth and orthodontics for malocclusion. Speech therapy may be required. Antibiotics are recommended for recurrent infections. Because of the role of RUNX2 in bone maintenance and

Prognosis

The malformations and complications of CCD rarely cause significant disability. The prognosis is generally good.

Description REQUIRED
*Please justify your suggestions by scientific references

Cancel Send



How to Post a Nomenclature/Synonym Suggestion?

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

Search for a Disorder, Gene, Phenotype


Orphanet Classification Of Rare Developmental Anomalies During Embryogenesis

- Rare developmental defect during embryogenesis
 - Hydrops fetalis
 - Renal or urinary tract malformation
 - Rare surgical cardiac disease
 - Cranial malformation
 - Cleidocranial dysplasia**
 - Craniosynostosis
 - Acalvaria
 - Craniodiaphyseal dysplasia
 - Craniofacial dysynostosis
 - Hypomandibular faciocranial dysostosis
 - Delayed membranous cranial ossification

Cleidocranial Dysplasia  

ORPHA:1452

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

Abstract 

Disease definition

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Nomenclature

MAIN NAME 
Cleidocranial Dysplasia

SYNONYMS 
Cleidocranial Dysostosis

SEMANTIC TYPE
Malformation Syndrome

Classification

Natural History

External References

How to Post a New Disorder Suggestion?

orphanet CURATION v0.1.0

Disorders Genes Human Phenotype Ontology Suggestions Account Administration

Disorders

- Orphanet Classification Of Rare Diseases
219 disorders
- Orphanet Classification Of Rare Errors Of Metabolism
1010 disorders
- Orphanet Classification Of Rare Neurological Diseases
2657 disorders
- Orphanet Classification Of Rare Respiratory Diseases
272 disorders

New Disorder Suggestion

New Suggestion
Propose changes to the Orphanet data.
This suggestion will be visible to all other contributors.

Title

New Disorder REQUIRED

Description REQUIRED
*Please justify your suggestions by scientific references

Cancel Send

Search for a Disorder, Gene, Phenotype

New Disorder

Orphanet Classification Of Rare Cardiac Informations
9 disorders

Orphanet Classification Of Rare Genetic Diseases
30 disorders

Orphanet Classification Of Rare Hepatic Diseases
6 disorders

Orphanet Classification Of Rare Surgical Diseases
108 disorders

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
 - Pseudodistrophic 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



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

Filters

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	15.07 / 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**

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

Nomenclature

MAIN NAME

Marfan Syndrome

SYNONYMS

MFS

SEMANTIC TYPE

Disease

Classification

- Orphanet Classification Of Rare Genetic Diseases
- Orphanet Classification Of Rare Developmental Anomalies During Embryogenesis
- Orphanet Classification Of Rare Systemic And Rheumatological Diseases

Suggestion pencils

Summary text

Nomenclature section

Clinical classifications the disorder belongs in

Epidemiological data

Genes associated

- > 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

Epidemiology

Filters

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 (75-90%)

Limitation Of Joint Mobility

Occasional (20-5%)

Abnormality Of The Aortic Valve

Occasional (20-5%)

Fatigue

Very frequent (90-95%)

Chest Pain

Occasional (20-5%)

Dural Ectasia

Frequent (75-90%)

Children

Marfan Syndrome Type 1

Marfan Syndrome Type 2

Parents

Rare Developmental Defect With Connective Tissue Involvement

Marfan And Marfan-Related Disorder

Epidemiological data

Genes associated

Phenotypes

Included groups

Inclusive group(s)

Discussing a Suggestion

orphanet CURATION v0.1.0

Disorders

Genes

Human Phenotype Ontology

Suggestions

Account

Administration

Search for a Disorder, Gene, Phenotype

Suggestions / #17:214 Ovarian fibrothecoma(Disease) - Abstract - Text

Ovarian fibrothecoma(Disease) - Abstract - Text Svitlana Havrylenko - 2/23/17 6:50 PM



Svitlana Havrylenko

PMID: 27876070 PMID: 25556284 PMID: 24763236



Ovarian fibrothecoma (Disorder)

ORPHA:314478

Change To: Text

Ovarian fibrothecoma is a rare benign sex cord-stromal neoplasm, characterized by predominant occurrence at post- or peri-menstrual age, unilateral location (in 90% cases) and mixed features of both fibroma (microscopically, intersecting bundles of spindle-shaped collagen producing cells) and thecoma (microscopically, perifollicular thecal-resembling cells; usually hormon-inactive). Clinically it can be either asymptomatic, or present itself with pelvic or abdominal pain and/or distension. In case of estrogen-producing forms, a menorrhagia is present. Large tumors (>10cm) are often associated with pleural effusion and ascytes (the Meig's syndrome triad). In rare cases, an elevated cancer antigen 125 (CA125) is observed, which returns to normal after tumor excision.

Status open

Close and Add Verdict

DISCUSSION

Members

Montserrat Alfaro

Svitlana Havrylenko

+ Invite Curator

+ Invite Experts



Montserrat Alfaro 3/10/17 12:25 PM



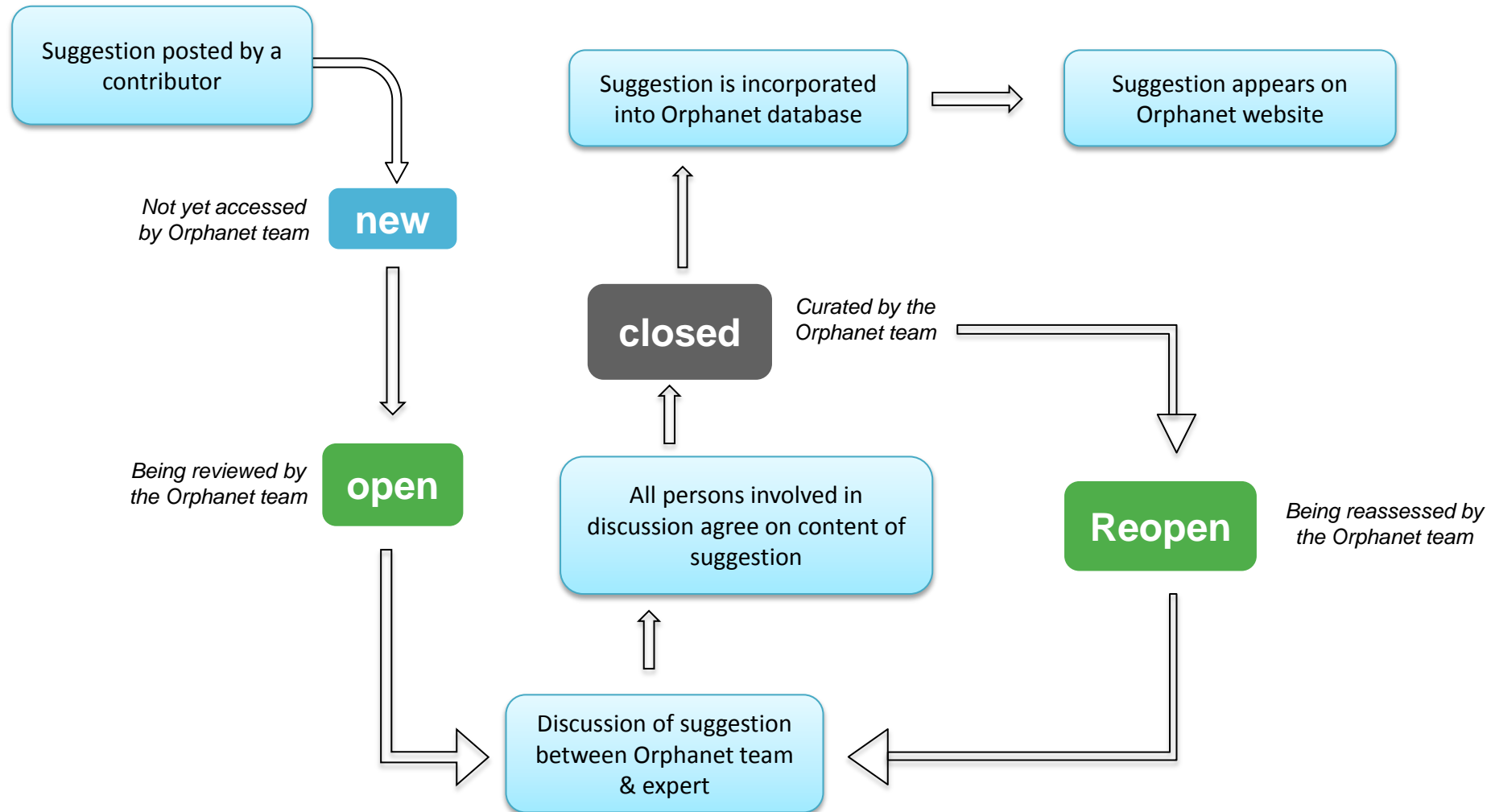
Svitlana, please consider the following observations: 1) the predominant occurrence at post- or peri-menstrual age is indicative of epidemiology and this should not be included in the definition, 2) the percentage of cases in which the tumor is unilateral should not be mentioned. It suffices to write for example "usually", "typically", "frequently", "predominantly" etc. (although this is also not obligatory), 3) the detailed description of the microscopic characteristics of the fibroma and thecoma could be deleted and it could just be specified that the tumor has "mixed features of both fibroma and thecoma", 4) symptoms can be just listed without explaining the circumstance in which that symptom occurs, 5) in the symptoms listed I think it is important to include post-menopausal bleeding. Thanks!



Post

Type @ to invite / mention other users.

Life Cycle of a Suggestion





Thank you!