Orphanet Curation Platform

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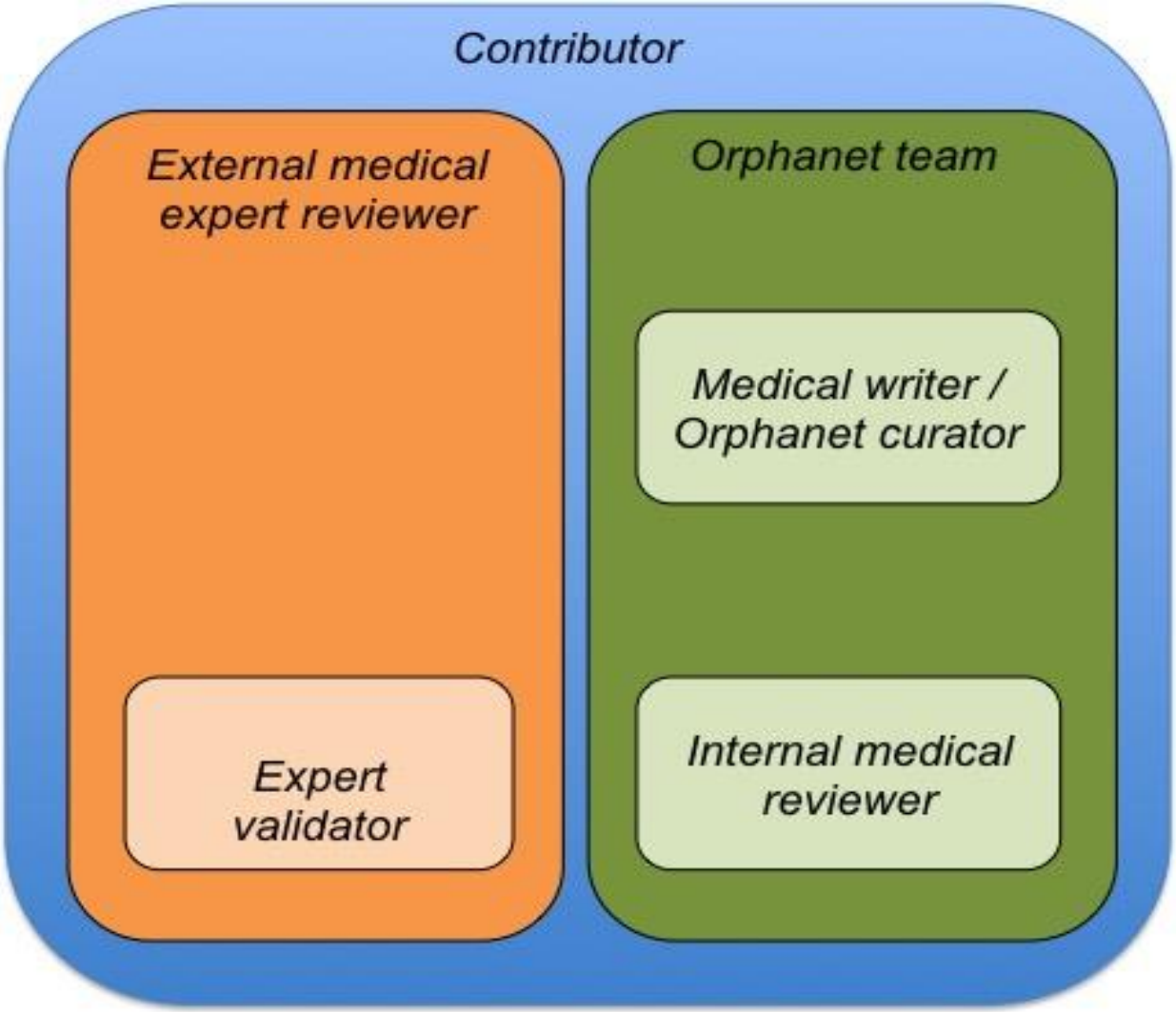
Internal Medical Reviewer

Orphanet - France

What is the Orphanet Curation Platform?

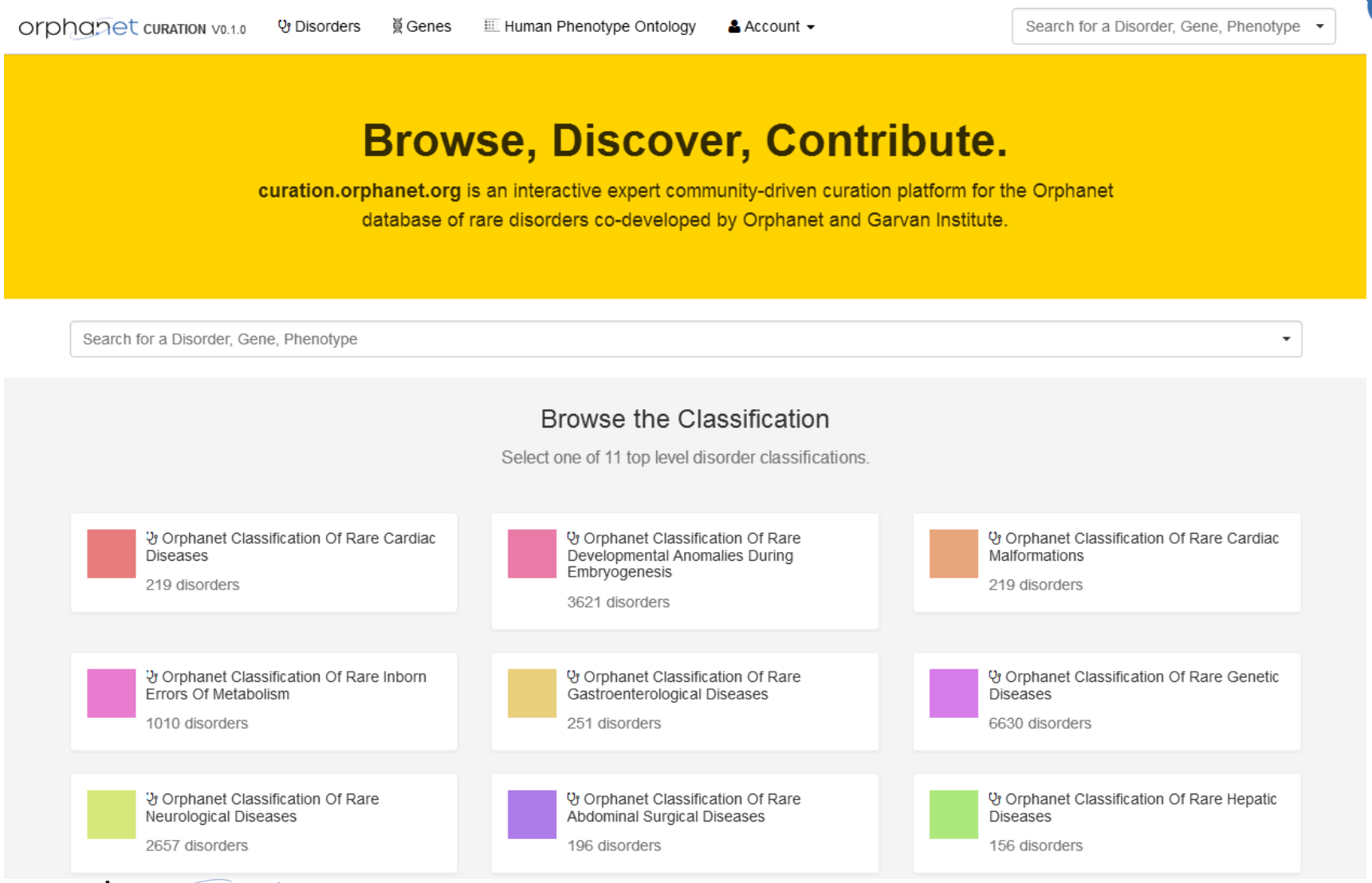
- It is a community-driven curation platform for rare disorders based on Orphanet's scientific content.
- It is a pilot project being co-developped by Orphanet and the Garvan Institute (Australia), with the support of the Orphanet Australia's country coordinator.
- Goals:
  - Allow users to visually 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 »

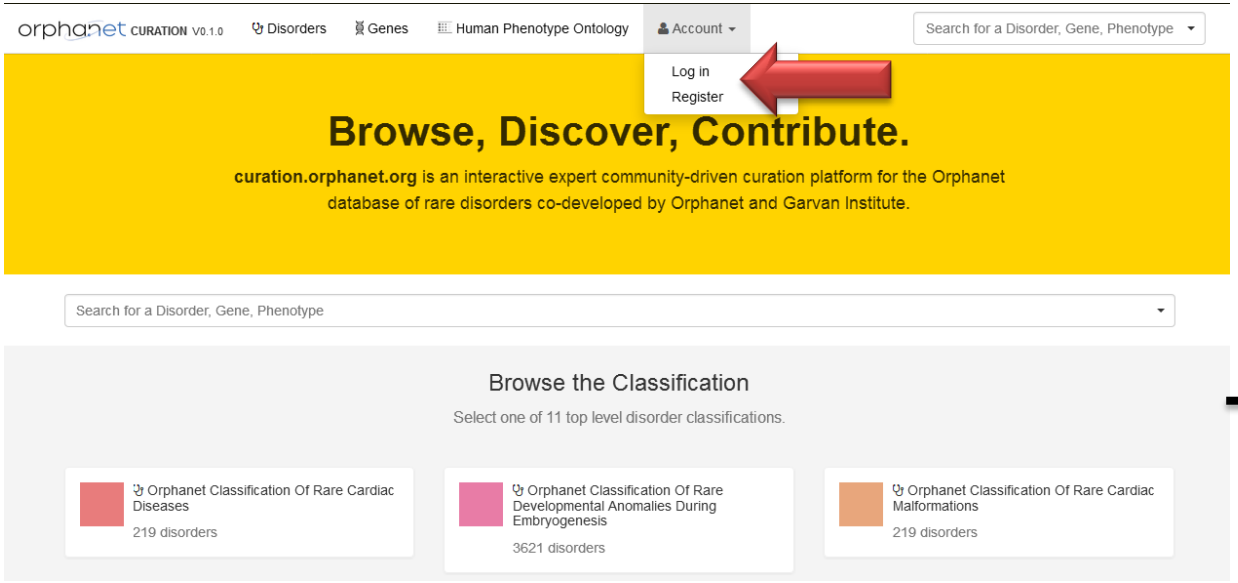


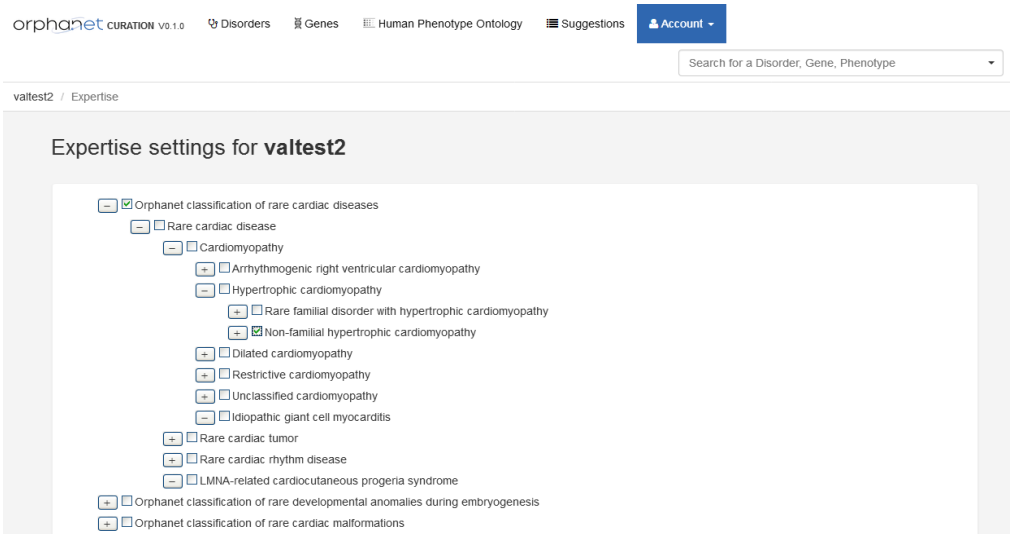
Getting Started

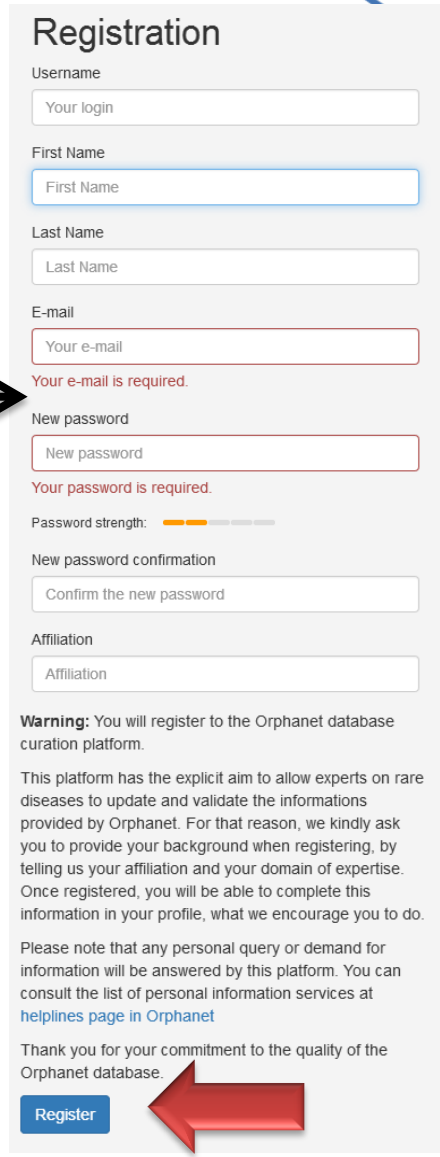
http://curation.orphanet.org



Create an Account





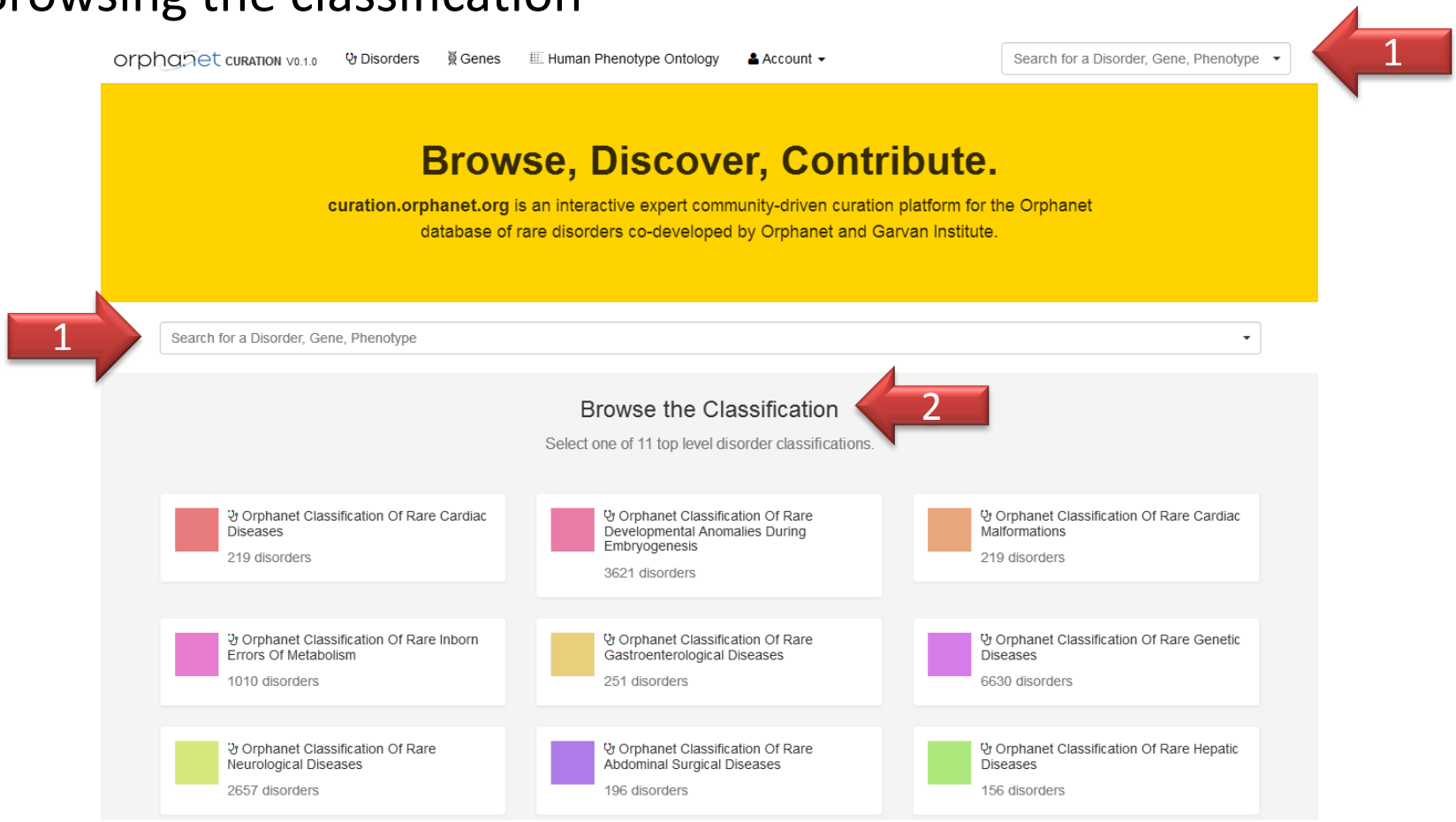


orphanet

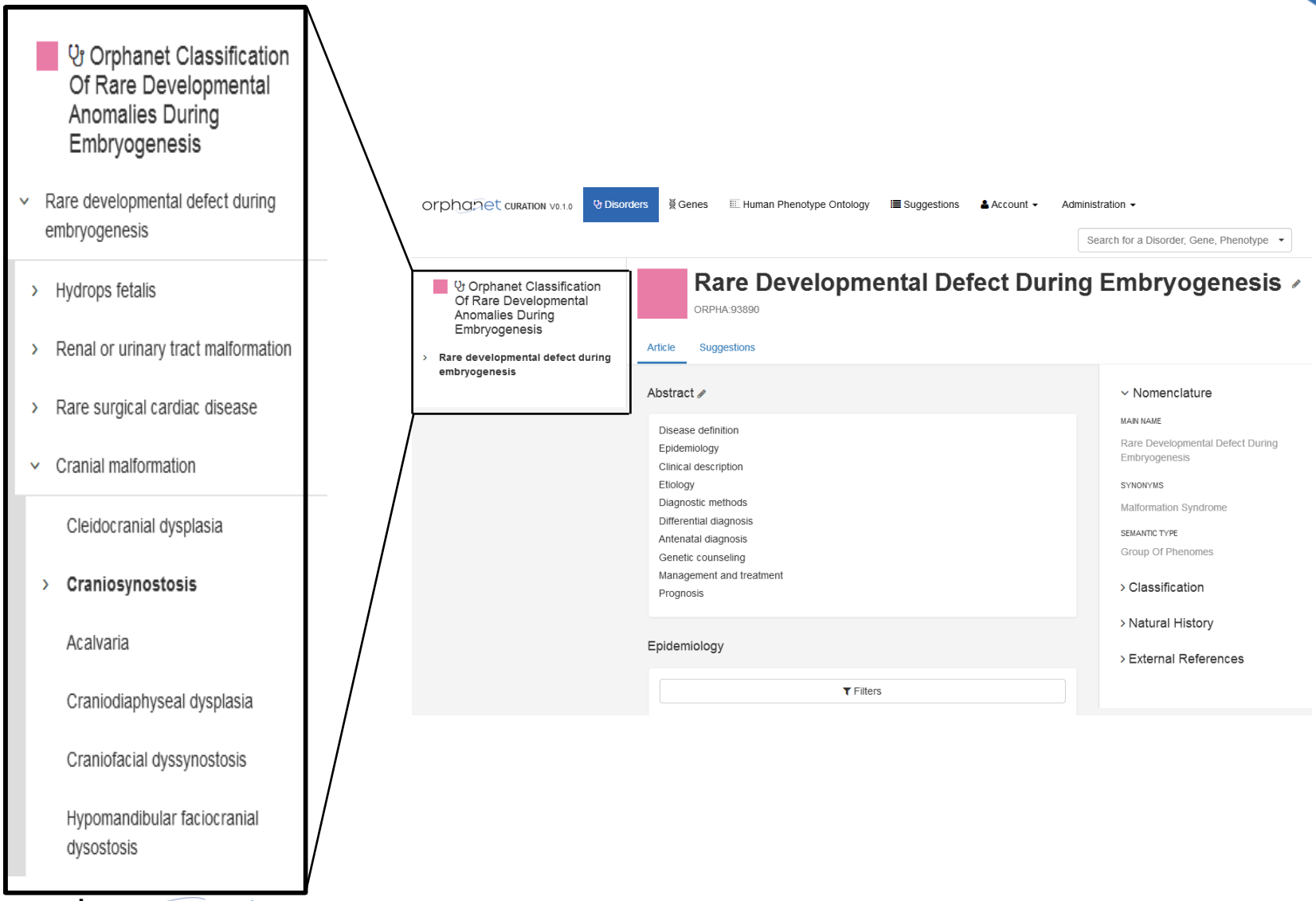
www.orpha.net

How to Search for a Disease?

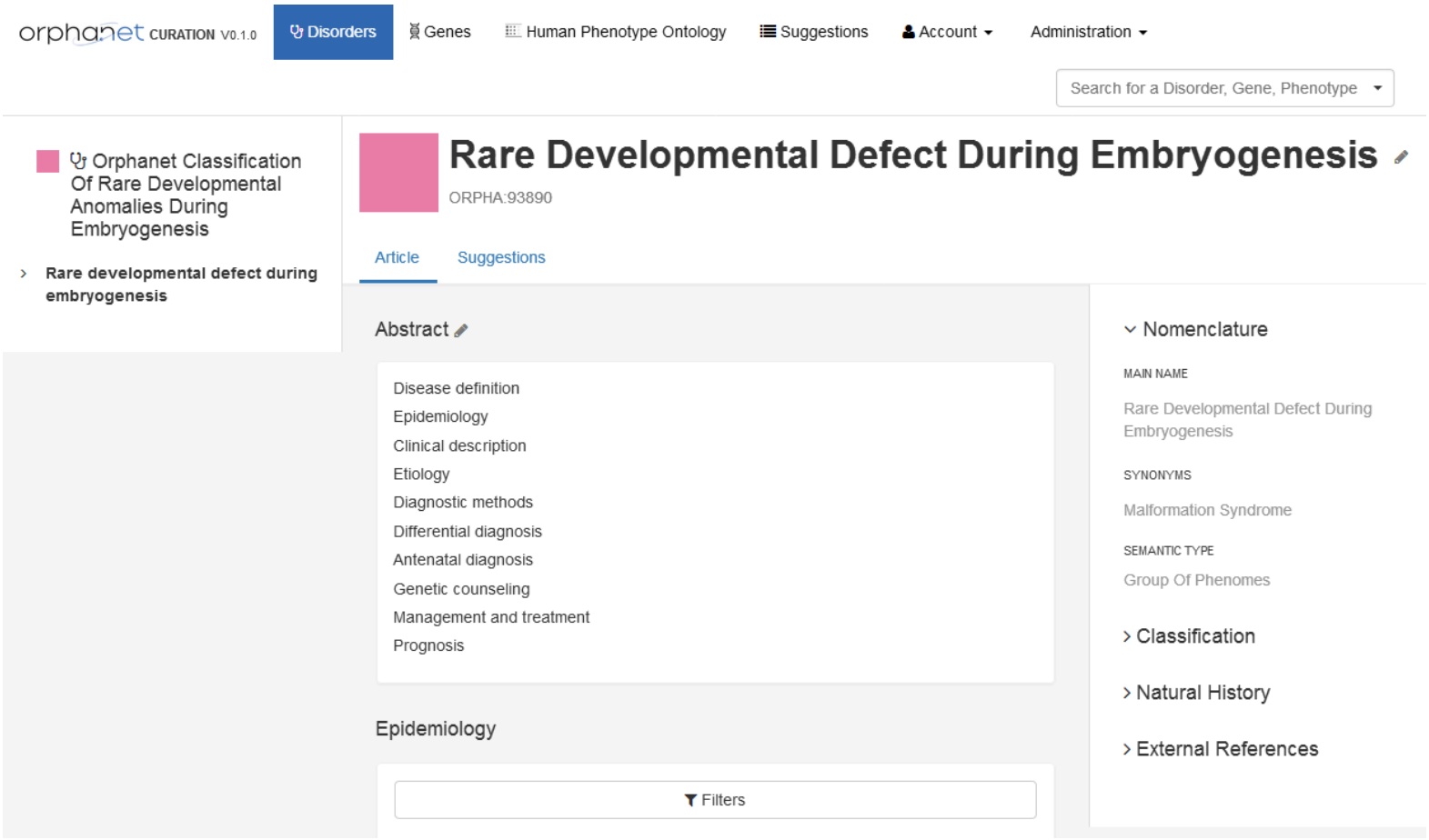
1) Using one of the search engines
2) Browsing the classification



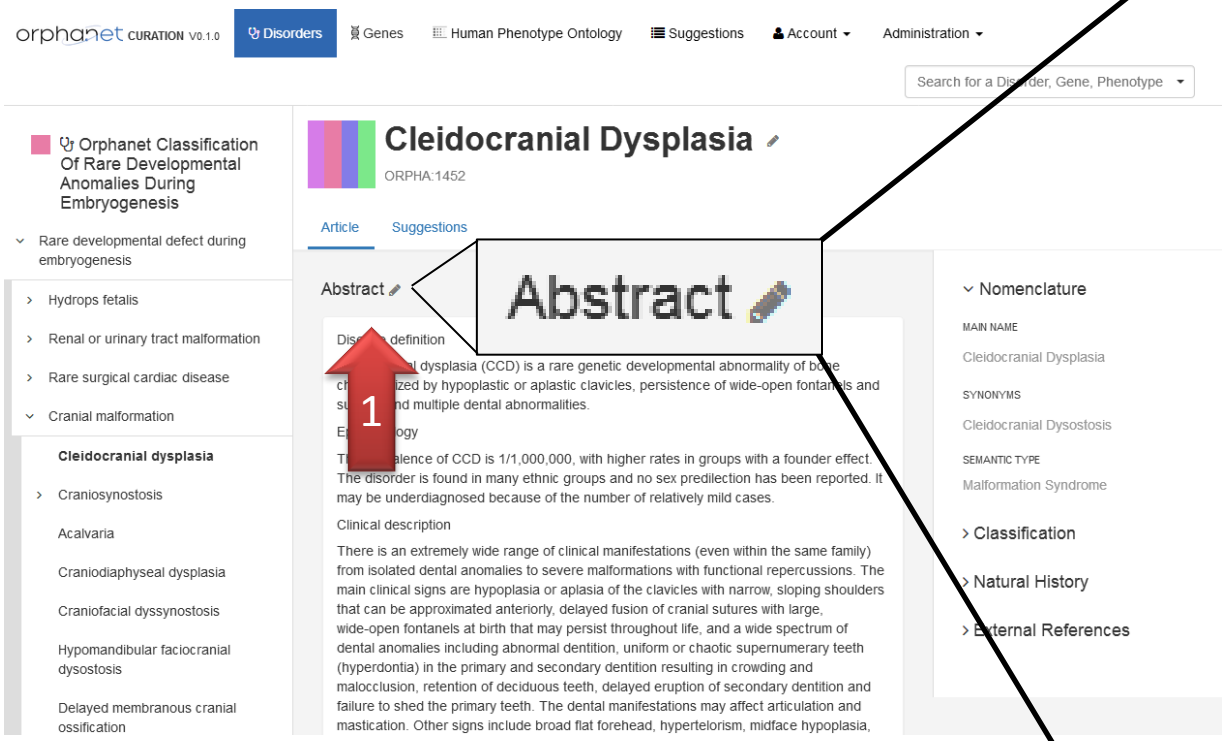
Browsing the Classification

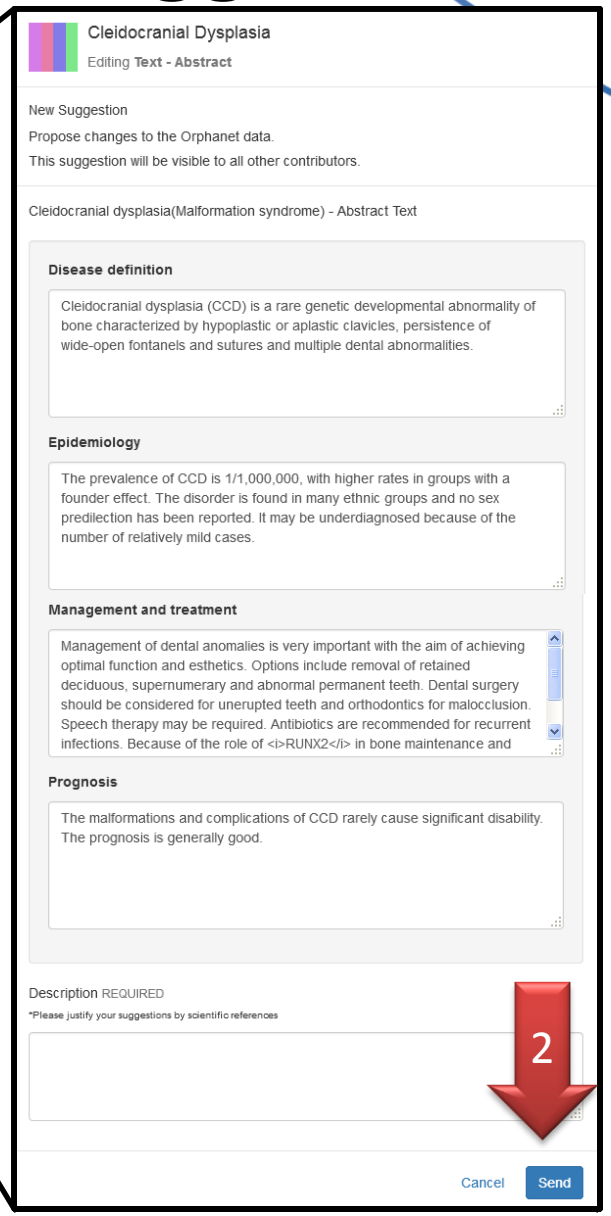


Browsing the Classification

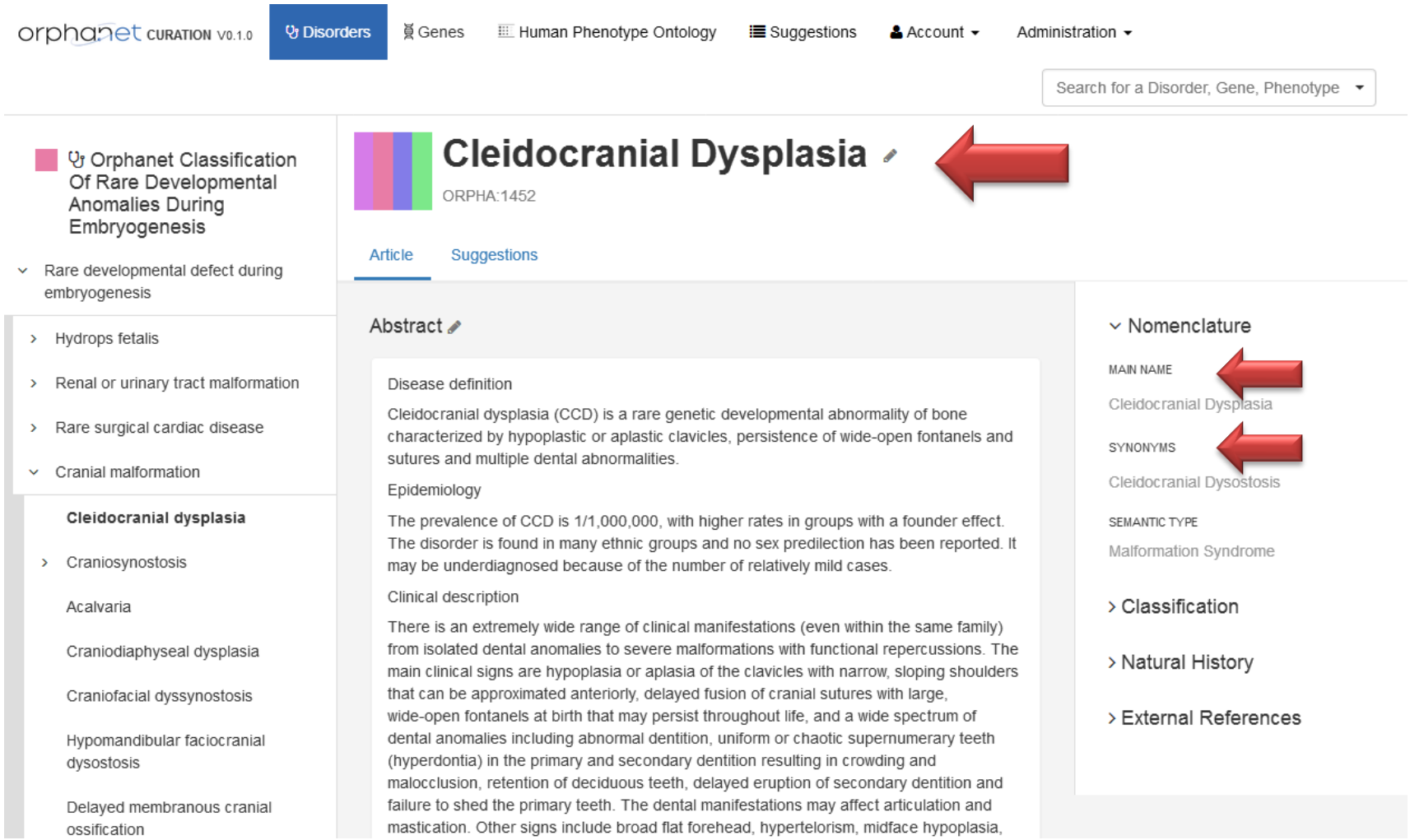


How to Post a Summary Text Suggestion?

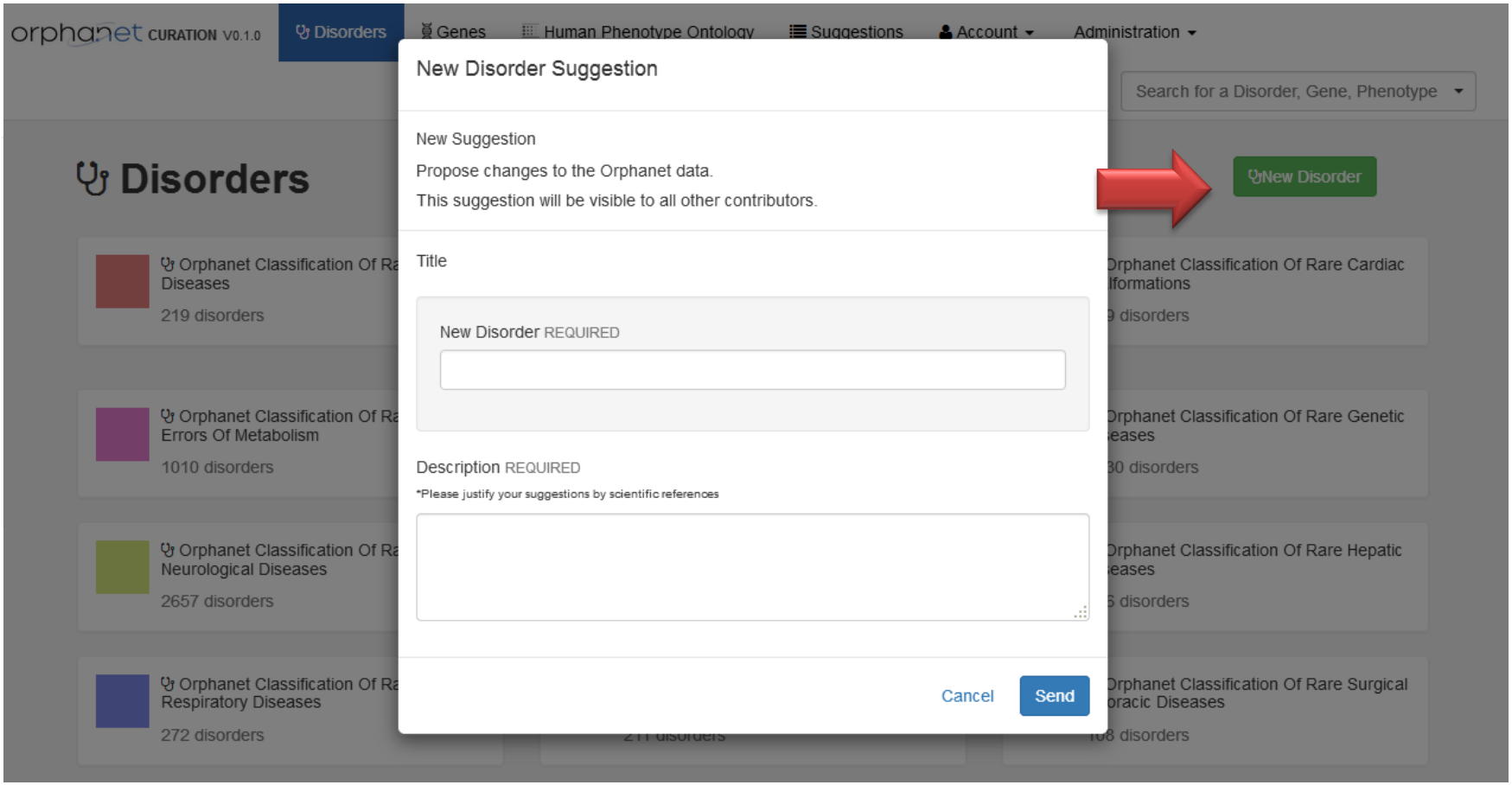


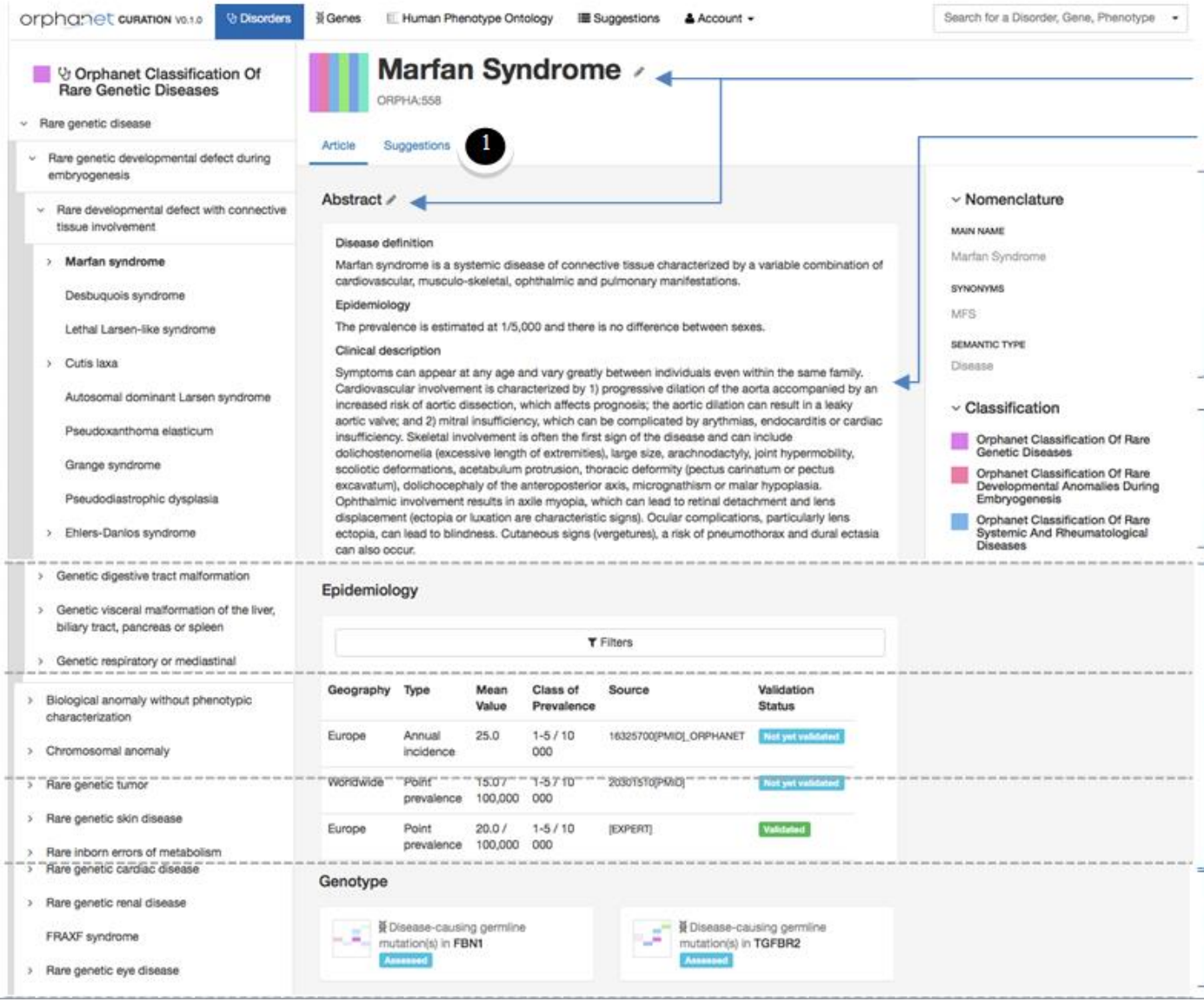


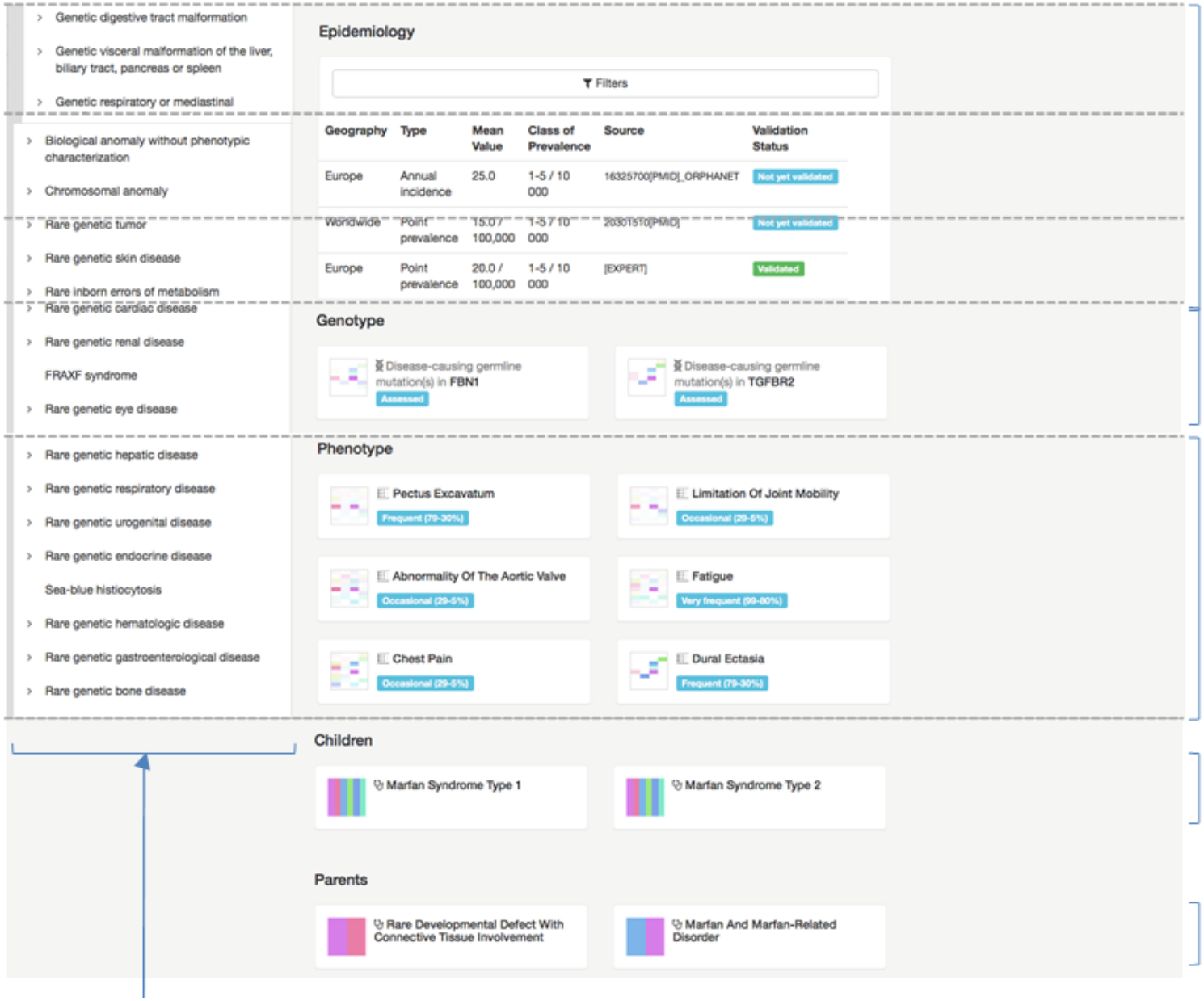
How to Post a Nomenclature/Synonym Suggestion?



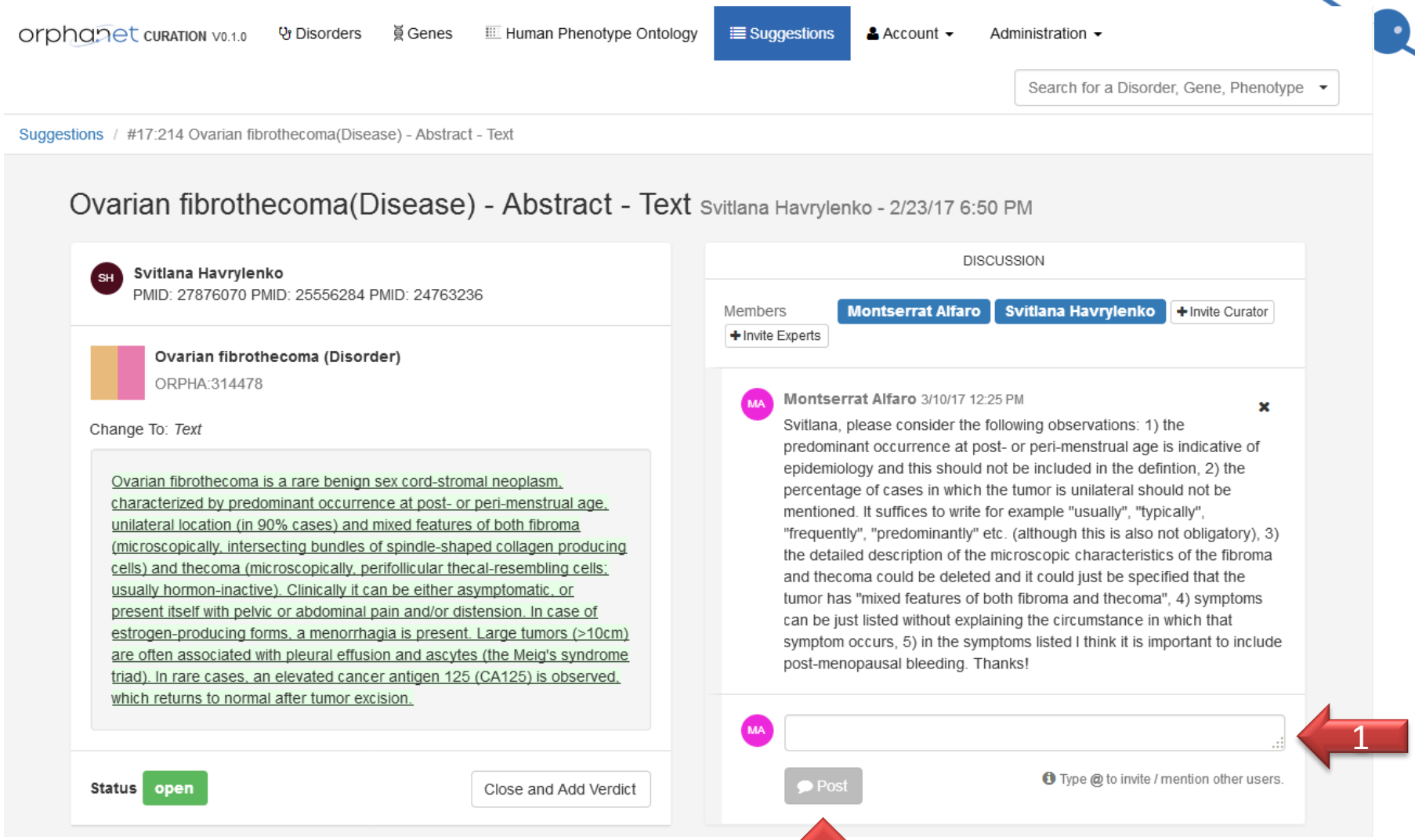
How to Post a New Disorder Suggestion?



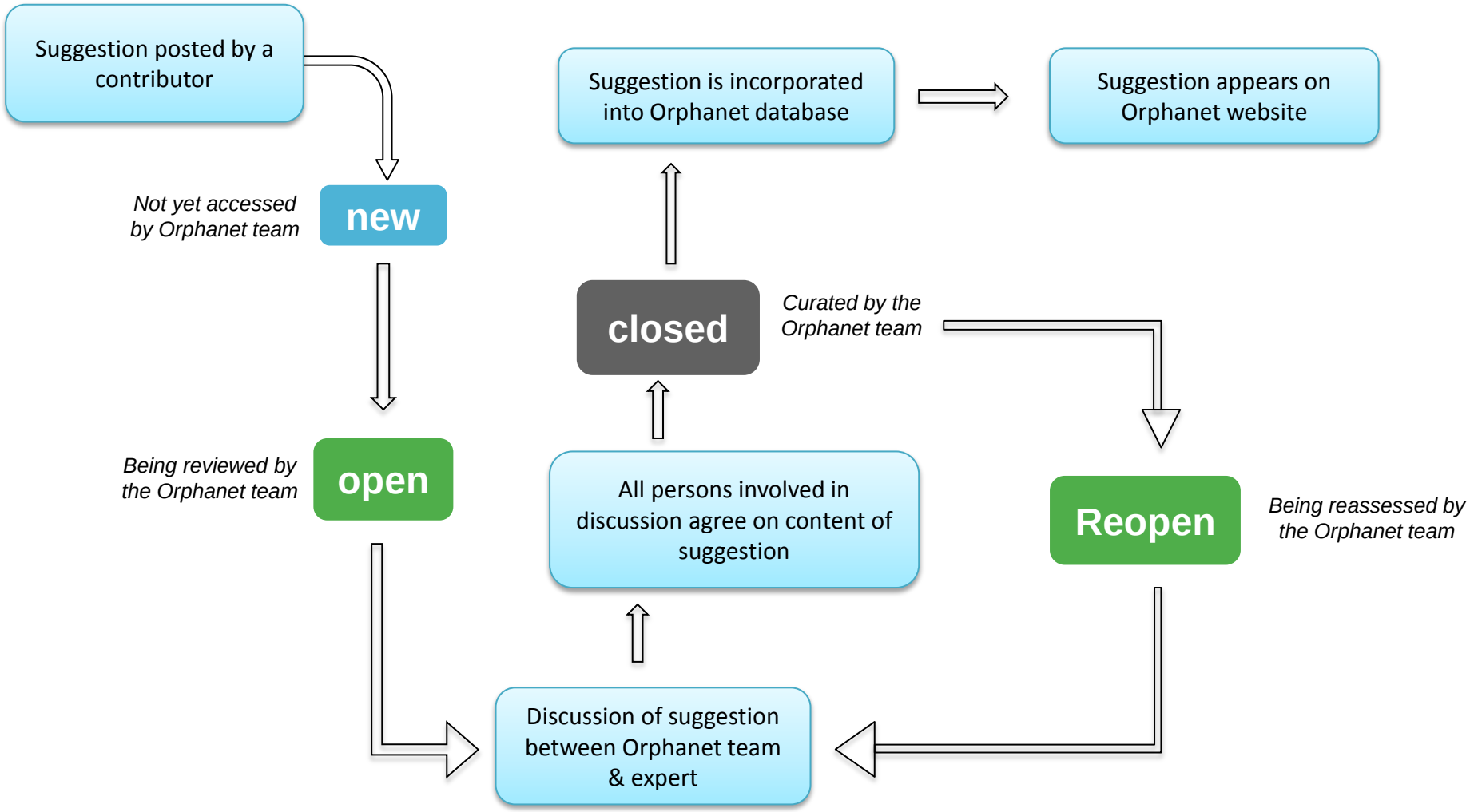

orphanet CURATION V0.1.0
Disorders
Genes
Human Phenotype Ontology
Suggestions
Account
Search for a Disorder, Gene, Phenotype
Orphanet Classification Of Rare Genetic Diseases
Rare genetic disease
Rare genetic developmental defect during embryogenesis
Rare developmental defect with connective tissue involvement
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
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 arythmias, 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.
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
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
Assessed
Disease-causing germline mutation(s) in TGFBR2
Assessed
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
15.07 / 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
Assessed
Disease-causing germline mutation(s) in TGFBR2
Assessed
Phenotype
Pectus Excavatum
Frequent (79-30%)
Limitation Of Joint Mobility
Occasional (29-5%)
Abnormality Of The Aortic Valve
Occasional (29-5%)
Fatigue
Very frequent (99-80%)
Chest Pain
Occasional (29-5%)
Dural Ectasia
Frequent (79-30%)
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



Life Cycle of a Suggestion



Thank you!