orphanet

orphadata

ON REQUEST PRODUCTS DESCRIPTION

SEPTEMBER 2019

IRDiRC RECOGNIZED RESOURCES

THE HUMAN VARIOME PROJECT RECOMMENDED SYSTEM

Global Alliance for Genomics & Health
Collaborate. Innovate. Accelerate.

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Cystic fibrosis

Suggest an update

Disease definition

Cystic fibrosis (CF) is a genetic disorder characterized by the production of sweat with a high salt content and mucus secretions with an abnormal viscosity.

Epidemiology

It is the most common genetic disorder among Caucasian children. The incidence varies between populations: the condition is considerably less common in Asian and African populations than in the white populations of Europe and North America, with variation within each country. The exact prevalence in Europe is unknown, but estimates range between 1/8,000 and 1/10,000 individuals.

Clinical description

...

Management and treatment

Treatment of cystic fibrosis remains purely symptomatic, revolving around bronchial drainage, antibiotics for respiratory infections, pancreatic analysis and administration of vitamins and calorific supplements for digestive and nutritional problems. These cost-effective treatments have significantly improved the prognosis for cystic fibrosis patients: in the 1960's the majority of patients died before 5 years of age, whereas the current average life-span exceeds 35 years and life-expectancy is 40 years. Symptomatic treatment of the disease should improve with the development of etiological treatments with complementary benefits (pharmacological approaches or gene therapy), neonatal testing and multidisciplinary management.

Detailed information

Article for general public

Svenska (2016)

Français (2006, pdf)

Deutsch (2014, pdf)

Professionals

> Summary information

Greek (2006, pdf)

> Emergency guidelines

Português (2009, pdf)
Deutsch (2014, pdf)
Italiano (2009, pdf)
Español (2019, pdf)
Français (2018, pdf)

> Clinical practice guidelines

English (2014)
Deutsch (2013)
Français (2017, pdf)

> Guidance for genetic testing

English (2009, pdf)

> Clinical genetics review

English (2017)

Textual information

Additional information

Expert resources

Further information on this disease

> Classification(s) (6)
> Gene(s) (5)
> Disability
> Clinical signs and symptoms
> Publications in PubMed

Health care resources for this disease

> Expert centres (447)
> Diagnostic tests (450)
> Patient organisations (79)
> Orphan drug(s) (85)

Research activities on this disease

> Research projects (179)
> Clinical trials (151)
> Registries/biobanks (52)
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Orphan drugs

I. Textual Information

Orphanet provides textual information on rare diseases. Information can be presented in the form of a definition, an abstract, or through linked articles (in peer-reviewed journals, or produced by learned societies). Orphanet definitions or abstracts are unique and written in English by a member of the editorial team and reviewed by an invited, world-renowned expert. Linked external articles are evaluated according to a set of quality criteria.

1. Articles

Description of the XML tags

- DisorderList count: total number of disorders, group of disorders and subtypes in the Xml file
- Orphanum: unique identifying number assigned by Orphanet to a given disease.
- ExpertLink: stable URL pointing to the specific page of the given disease on the Orphanet website.
- Disorder_Name: preferred name of a given disorders, group of disorders and subtypes.
- TextualInformationList count: number of associated articles linked to the disease
- TextType: Type of the associated article. Can be

Anesthesia guidelines
Article for general public
Clinical genetics review
Clinical practice guidelines
Diagnostic criteria
Disability factsheet
Emergency card
Emergency guidelines
Guidance for genetic testing
Multimedia
Practical genetics
Review article
Summary information

- URL: URL of the associated article
- Journal: Name of the journal of the associated article

Example

<DisorderList count="**XXXX**">

XXXX is the total number of diseases, groups or subtypes presented in this XML file

<OrphaNumber>586</OrphaNumber>
<Name lang="**en**">Cystic fibrosis</Name>

The concerned disease has 586 as its ORPHAnumber and Cystic fibrois as preferred term

<TextualInformation count="**6**">

6 articles are associated to the concerned disease

<TextType id="**226**">
<Name lang="**en**">Article for general public</Name>
<Journal>Socialstyrelsen</Journal>
<URL> http://www.socialstyrelsen.se/rarediseases/cysticfibrosis</URL>

The associated article is an article for general public available via the socialstyrelsen resource at the URL http://www.socialstyrelsen.se/rarediseases/cysticfibrosis

2. Abstracts

Description of the XML tags

- DisorderList count: total number of disorders, group of disorders and subtypes in the Xml file
- Orphanum: unique identifying number assigned by Orphanet to a given disease.
- ExpertLink: stable URL pointing to the specific page of the given disease on the Orphanet website.
- Disorder_Name: preferred name of a given disorder(s), group of disorders and subtypes.
- TextsectionList count: number of abstract sections filled for the concerned disease. In Orphanet, abstracts are divided into 10 ordered sections, and the definition section is mandatory.

Section label	Order
Disease definition	1
Epidemiology	2
Clinical description	3
Etiology	4
Diagnostic methods	5
Differential diagnosis	6
Antenatal diagnosis	7
Genetic counseling	8
Management and treatment	9

Prognosis	10

- **Textsection Name:** name of the selected section
- **Textsection Content:** content of the selected section

Example

<DisorderList count="**XXXX**">

XXXX is the total number of diseases, groups or subtypes presented in this XML file

<OrphaNumber>586</OrphaNumber>
<Name lang="**en**">Cystic fibrosis</Name>

The concerned disease has 586 as its ORPHAnumber and Cystic fibrosis as preferred term

<TestSectionList count="**10**">

The 10 different sections of the abstract are filled

<TextSection Type id="**16913**">
<Name lang="**en**">Epidemiology</Name>
<Order>3</Order>
<Contents> It is the most common genetic disorder among Caucasian children. The incidence varies between populations: the condition is considerably less common in Asian and African populations than in the white populations of Europe and North America, with variation within each country. The exact prevalence in Europe is unknown, but estimates range between 1/8,000 and 1/10,000 individuals</Contents>

The third section of the abstract is the epidemiology section and its content is: « It is the most common genetic disorder among Caucasian children. The incidence varies between populations: the condition is considerably less common in Asian and African populations than in the white populations of Europe and North America, with variation within each country. The exact prevalence in Europe is unknown, but estimates range between 1/8,000 and 1/10,000 individuals. »

3. Publications in PuMed

Description of the XML tags

- **DisorderList count**: total number of disorders, group of disorders and subtypes in the Xml file
- **Orphanum**: unique identifying number assigned by Orphanet to a given disease.
- **ExpertLink**: stable URL pointing to the specific page of the given disease on the Orphanet website.
- **Disorder_Name:** preferred name of a given disorders, group of disorders and subtypes.
- **Source:** the unique source in the file is “medline”

- **Reference**: the PubMed query to find information on the concerned disease

Example

<DisorderList count="**XXXX**">

XXXX is the total number of diseases, groups or subtypes presented in this XML file

<OrphaNumber>586</OrphaNumber>
<Name lang="**en**">Cystic fibrosis</Name>

The concerned disease has 586 as its ORPHAnumber and Cystic fibrois as preferred term

<Source> Medline<Source>

The source is Medline

<Reference>http://www.ncbi.nlm.nih.gov/sites/entrez?Db=pubmed&Cmd=Search&Term=%22cystic+fibrosis%22%5BMeSH+Terms%5D+OR+%28%28Cystic+fibrosis%5BText+Word%5D+OR+mucoviscidosis%5BTW%5D%29+NOT+medline%5BSB%5D%29 </Reference>

The associated information is available at http://www.ncbi.nlm.nih.gov/sites/entrez?Db=pubmed&Cmd=Search&Term=%22cystic+fibrosis%22%5BMeSH+Terms%5D+OR+%28%28Cystic+fibrosis%5BText+Word%5D+OR+mucoviscidosis%5BTW%5D%29+NOT+medline%5BSB%5D%29

II. Expert resources

DATA COLLECTION:

The directory of expert resources contains:

- expert centers and networks of expert centres;
- patient organisations and umbrella organisations/federations/alliances (as networks of patient organisations);
- patient registries and network of patient registries;
- biobanks and networks of biobanks;
- variant databases;
- medical laboratories;
- research projects and networks of research projects;
- clinical trials and networks of clinical trials.

SCOPE OF THE COLLECTION:

Data collection takes place in the Orphanet's network countries and is currently ongoing. Thus the database cannot be regarded as comprehensive. If an expert resource is not listed in a region or a country, it may not have been identified or may have refused to be listed. It is also possible that the expert resource has not yet been set up for some rare diseases.
Patient organisations not based in an Orphanet country can be considered for inclusion if they are an alliance and/or are members of EURORDIS AND they have legal status.

Research projects, clinical trials, patient registries, biobanks and variant databases are also collected if they are founded by a member agency of the IRDiRC consortium (International Rare Diseases Research Consortium) located in a country for which there isn't an Orphanet national team.

1. Expert centers and networks of expert centres

Orphanet provides information on centres of expertise or networks of centres of expertise dedicated to the medical management and/or genetic counselling for one particular rare disease or a group of rare diseases. Medical management centres should deliver a service of indisputably higher quality than a standard hospital service in the relevant speciality. It comprises centres that are officially designated by the health authorities in the country and centres that are not officially designated but which fulfil the Orphanet eligibility criteria, adapted from the recommendations of the European Union Committee of Experts on Rare Diseases. Genetic counselling centres are those organising genetic counselling consultations for all genetic diseases or for a particular genetic disease/group of diseases.

a. Expert centres

Description of the XML tags

- **ExpertCentreList count**: total number of expert centres in the XML file
- **ExpertLink**: stable URL pointing to the specific page of the expert centre on the Orphanet website
- **Lang:** ISO 639 code for language names
- **Name:** Name of the expert centre
- **ValidationDate:** The last update date of the expert centre
- **ExpertCentreStatusFlag:** attributes of the expert centre. Can be:
    - specialised for "Adult", for "Child"
    - a "Center of reference"
    - a centre for "Genetic counselling", for "Medical management".
- **Departement_Service_Lab_PatientOrganisation_Acronym:** Acronym of the department/service name of the expert centre
- **Departement_Service_Lab_PatientOrganisation_Name:** Name of the department/service of the expert centre
- **Status:** Status of the department/service. Can be "public", "Private non-for-profit" or "Private for-profit"
- **Hosting_Institution:** Name of the hosting institution of the department/service of the expert centre
- **Town:** Town of the hosting institution
- **Country Name:** Name of the country where the expert centre is located
- **DisorderList count**: Total number of disorders, group of disorders and subtypes concerned by the expert centre
- **Orphanum**: Unique identifying number assigned by Orphanet to a given disease
- **ExpertLink**: Stable URL pointing to the specific page of the given disease on

the Orphanet website.

- **Disorder_Name:** Preferred name of a given disorder(s), group of disorders and subtypes.

Example

<ExpertCentreList count="**XXXX**">

XXXX is the total number of expert centres presented in this XML file

<ExpertCentre id="**33745**">

The unique identifier of the expert centre is 33745

<ExpertLink lang="**en**">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=65157 </ExpertLink>

The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=65157

<Name>Centre de Ressources et de Compétences Mucoviscidose et affections liées à une anomalie de CFTR - CRCM</Name>
<LanguageOfName>fr</LanguageOfName>

The name of this entry is Centre de Ressources et de Compétences Mucoviscidose et affections liées à une anomalie de CFTR – CRCM in french (fr)

<Name_en>Ressources and Competence Centre for cystic fibrosis and diseases related to an abnormal CFTR - CRCM</Name_en>

The english name of this entry is Ressources and Competence Centre for cystic fibrosis and diseases related to an abnormal CFTR – CRCM

<ExpertCentreStatusFlagList count="**5**">
<ExpertCentreStatusFlag id="**38**">
<Name lang="**en**">Center of reference</Name>
</ExpertCentreStatusFlag><ExpertCentreStatusFlag id="**42**">
<Name lang="**en**">Adult clinic</Name>
</ExpertCentreStatusFlag><ExpertCentreStatusFlag id="**40**">
<Name lang="**en**">Medical management clinic</Name>
</ExpertCentreStatusFlag><ExpertCentreStatusFlag id="**41**">
<Name lang="**en**">Child clinic</Name>

This expert centre is a center of reference, specialized in medical management for Adult and Child.

<ValidationDate>2018-03-09 00:00:00.0</ValidationDate>

The last update date of the expert centre was the 09/03/2018

<InstitutionList count="**1**">
<Institution id="**37179**"><Department_Service_Lab_PatientOrganisationAcronym/>
<Department_Service_Lab_PatientOrganisationName>Service de Pédiatrie</Department_Service_Lab_PatientOrganisationName>
<Status id="**13**"><Name lang="**en**">Public</Name></Status>
<Address id="**127**"><Hosting_Institution>CHBA Centre hospitalier Bretagne Atlantique - CH Chubert</Hosting_Institution>

<Town><Name>VANNES</Name></Town>
<Country id="**75**"><Name lang="**en**">FRANCE</Name></Country>

The expert centre is located in the "Service de Pédiatrie" of the "Public" hosting institution named "CHBA Centre hospitalier Bretagne Atlantique - CH Chubert" located at "VANNES" in "FRANCE"

<DisorderList count="**2**">
<OrphaNumber>586</OrphaNumber>
<Name lang="**en**">Cystic fibrosis</Name>
<OrphaNumber>48</OrphaNumber>
<Name lang="**en**">Congenital bilateral absence of vas deferens</Name>

The expert centre operates for two diseases: Cystic fibrosis (orphanumber 586) and Congenital bilateral absence of vas deferens (orphanumber 48)

b. Network of expert centres

Description of the XML tags

- **NetworkList count**: total number of networks in the XML file
- **ExpertLink**: stable URL pointing to the specific page of a network on the Orphanet website
- **Lang:** ISO 639 code for language names
- **Name:** Name of the network
- **URL:** Website of the network
- **Geocoverage:** Geographical coverage of the network. Can be "Global", "Regional", "National" or "European"
- **ValidationDate:** The last update date of the network
- **DisorderList count**: total number of disorders, group of disorders and subtypes concerned by the network
- **ExpertCentre count**: total number of expert centres involved in the network

Example

<NetworkList count="**228**">

228 is the total number of networks of expert centres presented in this XML file

<Network id="**113119**">

The unique identifier of the network is 113119

<ExpertLink lang="**en**">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=113119</ExpertLink>

The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=113119

<Name_en> Ressources and Competence Centre for cystic fibrosis and diseases related to

an abnormal CFTR - CRCM </Name>

The name of the entry in English is "Ressources and Competence Centre for cystic fibrosis and diseases related to an abnormal CFTR - CRCM"

<Url>http://muco-cftr.fr/</Url>

The URL of the network is http://muco-cftr.fr

<GeoCoverage id="**15**"><Name lang="**en**">European</Name></GeoCoverage>

The network performs its activities at European level

<ValidationDate>2018-07-06 00:00:00.0</ValidationDate>

The last update date of the network was the 07/06/2018

<PersonList count="**1**">
<Person id="**49712**">
<Country id="**22**"><Name lang="**en**">BELGIUM</Name></Country>
<PersonActivityList count="**0**">
<PersonActivity><PersonFunction id="**39**"><Name lang="**en**">Coordinator of expert centre network</Name></PersonFunction></PersonActivity>

The network is coordinate by a person living in France

<DisorderList count="**1**">
<Disorder id="**49**">
<OrphaNumber>**586**</OrphaNumber>
<Name lang="**en**">**Cystic fibrosis**</Name></Disorder></DisorderList>

The network operates for one disease: Cystic fibrosis (orphanumber 586)

<ExpertCenter count="**26**">

The network made up of 26 expert centres

2. Patient organisations and umbrella organisations/federations/alliances (as networks of patient organisations)

Orphanet provides information on patient organisations, umbrella organisations, federations and alliances dedicated to one particular rare disease or to a group of rare diseases.

a. Patient organisation

Description of the XML tags

- **PatientOrganisationList count**: total number of patient organisations in the XML file.
- **ExpertLink**: stable URL pointing to the specific page of a patient organisation on the Orphanet website
- **Lang:** ISO 639 code for language names
- **Name:** Name of the patient organisation

- **URL:** Website of the patient organisation
- **Country ISO:** ISO code of the country where the patient organisation is located
- **Country Name:** Name of the country where the patient organisation is located
- **Geocoverage:** Geocoverage of the patient organisation. Can be "Global", "Regional", "National" or "European"
- **ValidationDate:** The last update date of the patient organisation
- **DisorderList count**: total number of disorders, group of disorders and subtypes concerned by the patient organisation
- **Orphanum**: unique identifying number assigned by Orphanet to a given disease.
- **ExpertLink**: stable URL pointing to the specific page of the given disease on the Orphanet website
- **Disorder_Name:** preferred name of a given disorder(s), group of disorders and subtypes.

Example

<PatientOrganisationList count="**XXXX**">

XXXX is the total number of patient organisations presented in this XML file

<PatientOrganisation id="**188**">

The unique identifier of the patient organisation is 188

<ExpertLink lang="**en**">**http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=8493**</ExpertLink>

The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=8493

<Name lang="**en**">**Vaincre la Mucoviscidose**</Name>

The name of the entry in English is "Vaincre la mucoviscidose"

<URL>**http://www.vaincrelamuco.org/**</URL>

The URL of the patient organisation's website is http://www.vaincrelamuco.org/

<Country><ISO>**FR**</URL>
<Name lang="**en**">FRANCE</Name>

The ISO Code and the name of the country of the patient organisation is FR for France

<ValidationDate>**2018-06-07 00:00:00.0**</ValidationDate>

The last update date of the patient organisation was the 07/06/2018

<GeoCoverage id="**1135**"><Name lang="**en**">**National**</Name></GeoCoverage>

The patient organisation operates at National level

<DisorderList count="**1**">
<Disorder id="**49**">
<OrphaNumber>**586**</OrphaNumber>
<Name lang="**en**">**Cystic fibrosis**</Name></Disorder></DisorderList>

The patient organisation operates for one disease: Cystic fibrosis (orphanumber 586)

b. Network of patient organisations

Description of the XML tags

- **NetworkList count**: total number of networks in the XML file.
- **ExpertLink**: stable URL pointing to the specific page of a network on the Orphanet website
- **Lang:** ISO 639 code for language names
- **Name:** Name of the network
- **URL:** Website of the network
- **Geocoverage:** Geographical coverage of the network. Can be "Global", "Regional", "National" or "European"
- **ValidationDate:** The last update date of the network
- **DisorderList count**: total number of disorders, group of disorders and subtypes concerned by the network
- **PatientOrganisation count**: total number of patient organisations involved in the network

Example

<NetworkList count="**XXXX**">

XXXX is the total number of patient organisations presented in this XML file

<Network id="**81635**">

The unique identifier of the network is 81635

<ExpertLink lang="**en**">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=257907</ExpertLink>

The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=257907

<Name>CFE: Cystic Fibrosis Europe</Name>

The name of the entry in English is " CFE: Cystic Fibrosis Europe"

<Url>http://www.cf-europe.eu</Url>

The URL of the network is http://www. cf-europe.eu

<GeoCoverage id="**15**"><Name lang="**en**">European</Name></GeoCoverage>

The network performed at the European level

<ValidationDate>2018-07-06 00:00:00.0</ValidationDate>

The last update date of the network was the 07/06/2018

<PersonList count="**1**">
<Person id="**49712**">
<Country id="**22**"><Name lang="**en**">BELGIUM</Name></Country>

<PersonActivityList count="**0**">
<PersonActivity><PersonFunction id="**38**"><Name lang="**en**">Coordinator of patient organisation network</Name></PersonFunction></PersonActivity>

The network is coordinate by a person living in Belgium

<DisorderList count="**1**">
<Disorder id="**49**">
<OrphaNumber>**586**</OrphaNumber>
<Name lang="**en**">**Cystic fibrosis**</Name></Disorder></DisorderList>

The network operates for one disease: Cystic fibrosis (orphanumber 586)

<PatientOrganisation count="**26**">

The network made up of 26 patient organisations.

<PatientOrganisation id="**31387**">

See the description of the patient organisation product

3. Patient registries and network of patient registries

Orphanet provides information on patient registries: systematic collections of clinical data for clinical research explicitly focused on a particular rare disease or group of diseases governed by an identified body. Single patient registries or networks of patient registries (national or international) must be based in one of the countries in the Orphanet consortium. Registries outside the Orphanet consortium countries can be considered if they fulfill Orphanet's inclusion criteria.

a. Patient registry

Description of the XML tags

- **Registry_BiobankList count**: total number of patient registries in the XML file.
- **ExpertLink**: stable URL pointing to the specific page of the patient registry on the Orphanet website
- **Lang:** ISO 639 code for language names
- **Name:** Name of the patient registry
- **Registry_BiobankType Flag:** Type of the expert resource. Can be only "Patient registries/Databases" in this product
- **URL:** Website of the patient registry
- **Country:** Name of the country where the patient registry is located
- **Geocoverage:** Geocoverage of the patient registry. Can be "Global", "Regional", "National", "Not defined" or "European"
- **ValidationDate:** The last update date of the patient registry
- **DisorderList count**: total number of disorders, group of disorders and subtypes concerned by the patient registry
- **Orphanum**: unique identifying number assigned by Orphanet to a given disease.
- **Disorder_Name:** preferred name of a given disorder(s), group of disorders and subtypes.

Example

<Registry_BiobankList count="**XXXX**">
XXXX is the total number of patient registry presented in this XML file

<Registry_Biobank id="**88000**">
The unique identifier of the patient registry is 88000

<ExpertLink lang="**en**">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=284084 </ExpertLink>
The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=284084

<Name_en> EIMD: European registry and network for intoxication type metabolic diseases</Name>
The English name of this entry is EIMD: European registry and network for intoxication type metabolic diseases

<Registry_BiobankType id="**15234**">
<Name lang="**en**">Patient registries/Databases</Name>
This expert resource is a patient registry/database

<ValidationDate>2018-03-09 00:00:00.0</ValidationDate>
The last update date of the patient registry was the 09/03/2018

<GeoCoverage id="**21**"><Name lang="**en**">Global</Name></GeoCoverage>
The coverage of the patient registry is "Global"

<Country id="**75**"><Name lang="**en**">FRANCE</Name></Country>
The patient registry is located in "FRANCE"

<DisorderList count="1">
<OrphaNumber>664</OrphaNumber>
<Name lang="**en**"> Ornithine transcarbamylase deficiency </Name>
The patient registry operates for Ornithine transcarbamylase deficiency (orphanumber 664)

b. Network of patient registries

Description of the XML tags

NetworkList count: total number of networks in the XML file.

ExpertLink: stable URL pointing to the specific page of a network on the Orphanet website.

Lang: ISO 639 code for language names.

Name: Name of the network

URL: Website of the network

Geocoverage: Geographical coverage of the network. Can be "Global", "Regional", "National" or "European"

ValidationDate: The last update date of the network

DisorderList count: total number of disorders, group of disorders and subtypes concerned by the network.

Registry-Biobank count: total number of biobanks involved in the network

Example

<NetworkList count="**XXXX**">

XXXX is the total number of network of patient registries presented in this XML file

<Network id="**132258**">

The unique identifier of the network is 1325258

<ExpertLink lang="**en**">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2529127</ExpertLink>

The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=529127

<Name_en> Marfan Europe Network</Name>

The name of the entry in English is " Marfan Europe Network"

<Url> https://www.marfan.eu/</Url>

The URL of the network is http:// www.marfan.eu/

<ValidationDate>2018-07-06 00:00:00.0</ValidationDate>

The last update date of the network was the 07/06/2018

<PersonList count="**1**">
<Person id="**49712**">
<Country id="**22**"><Name lang="**en**">BELGIUM</Name></Country>
<PersonActivityList count="**0**">
<PersonActivity><PersonFunction id="**41**"><Name lang="**en**">Coordinator of patient registry network</Name></PersonFunction></PersonActivity>

The network is coordinated by a person living in Belgium

<DisorderList count="**1**">
<Disorder id="**20632**">
<OrphaNumber>**284993**</OrphaNumber>
<Name lang="**en**"> **Marfan and Marfan-related disorders**</Name></Disorder>

The network operates for Marfan and Marfan-related disorders (orphanumber 284993)

<Registry-Biobank count="**11**">

The network is made up of 11 patient registries.

4. Biobanks and networks of biobanks

Orphanet provides information on biobanks and network of biobanks: any kind of systematic, open-for-collaboration register of biological specimen for clinical research with a clear orientation towards the field of rare diseases.

a. Biobanks

Description of the XML tags

- **Registry_BiobankList count**: total number of biobanks in the XML file
- **ExpertLink**: stable URL pointing to the specific page of the biobank on the Orphanet website
- **Lang:** ISO 639 code for language names
- **Name:** Name of the biobank
- **Registry_BiobankType Flag:** Type of the expert resource. Can be only "Biobanks" in this product
- **ValidationDate:** The last update date of the biobank
- **URL:** Website of the biobank
- **Country:** Name of the country where biobank is located
- **Geocoverage:** Geographical coverage of the biobank. Can be "Global", "Regional", "National", "Not defined" or "European"
- **DisorderList count**: total number of disorders, group of disorders and subtypes concerned by the expert centre
- **Orphanum**: unique identifying number assigned by Orphanet to a given disease
- **Disorder_Name:** preferred name of a given disorder(s), group of disorders and subtypes.

Example

<Registry_BiobankList count="**XXXX**">

XXXX is the total number of biobank presented in this XML file

<Registry_Biobank id="**70624**">

The unique identifier of the biobank is 70624

<ExpertLink lang="**en**">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=218452 </ExpertLink>

The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=218452

<Name_en> Galliera Genetic Bank</Name>

The English name of this entry is Galliera Genetic Bank

<Registry_BiobankType id="**15234**">
<Name lang="**en**">Biobank</Name>

This expert resource is a Biobank

<ValidationDate>2018-03-09 00:00:00.0</ValidationDate>

The last update date of the biobank was the 09/03/2018

<GeoCoverage id="**15**"><Name lang="**en**">Not defined</Name></GeoCoverage>

The coverage of the biobank is "Not defined"

<Country id="**75**"><Name lang="**en**">FRANCE</Name></Country>

The biobank is located in "FRANCE"

<DisorderList count="**2**">
<OrphaNumber>586</OrphaNumber>
<Name lang="**en**">Cystic fibrosis</Name>
<OrphaNumber>48</OrphaNumber>
<Name lang="**en**">Congenital bilateral absence of vas deferens</Name>

The biobank operates for only diseases: Cystic fibrosis (ORPHAnumber 586) and Congenital bilateral absence of vas deferens (ORPHAnumber 48)

b. Network of biobanks

Description of the XML tags

- **NetworkList count**: total number of networks in the XML file
- **ExpertLink**: stable URL pointing to the specific page of a network on the Orphanet website
- **Lang:** ISO 639 code for language names
- **Name:** Name of the network
- **URL:** Website of the network
- **Geocoverage:** Geographical coverage of the network. Can be "Global", "Regional", "National" or "European"
- **ValidationDate:** The last update date of the network
- **DisorderList count**: total number of disorders, group of disorders and subtypes concerned by the network
- **Registry-Biobank count**: total number of biobanks involved in the network

Example

<NetworkList count="**XXXX**">

XXXX is the total number of network of biobanks presented in this XML file

<Network id="**792579**">

The unique identifier of the network is 792579

<ExpertLink lang="**en**">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=255828</ExpertLink>

The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=255828

<Name_en>EuroBioBank: European Network of DNA, Cell and Tissue banks for rare diseases</Name>

The name of the entry in English is "EuroBioBank: European Network of DNA, Cell and Tissue banks for rare diseases"

<Url>http:// www.eurobiobank.org/</Url>

The URL of the network is http:// www.eurobiobank.org

<ValidationDate>2018-07-06 00:00:00.0</ValidationDate>

The last update date of the network was the 07/06/2018

<PersonList count="**1**">
<Person id="**49712**">
<Country id="**22**"><Name lang="**en**">BELGIUM</Name></Country>
<PersonActivityList count="**0**">
<PersonActivity><PersonFunction id="**41**"><Name lang="**en**">Coordinator of biobank network</Name></PersonFunction></PersonActivity>

The network is coordinated by a person living in Belgium

<DisorderList count="**1**">
<Disorder id="**49**">
<OrphaNumber>**586**</OrphaNumber>
<Name lang="**en**">**Cystic fibrosis**</Name></Disorder></DisorderList>

The network operates for one disease: Cystic fibrosis (orphanumber 586)

<Registry-Biobank count="**26**">

The network is made up of 26 biobanks.

5. Variant databases

Orphanet provides information on databases of variants described as causative for a particular rare disease or group of diseases, and having an online interface, governed by an identified body.

Description of the XML tags

- Registry_BiobankList count: total number of variant databases in the XML file
- ExpertLink: stable URL pointing to the specific page of the variant database on the Orphanet website.
- Lang: ISO 639 code for language names
- Name: Name of the variant database

- **Registry_BiobankType Flag:** Type of the expert resource. Can be only "variant databases" in this product
- **ValidationDate:** The last update date of the variant database
- **URL:** Website of the variant database
- **Country:** Name of the country where the variant database is located
- **Geocoverage:** Geocoverage of the variant database. Can be "Global", "Regional", "National", "Not defined" or "European"
- **DisorderList count**: total number of disorders, group of disorders and subtypes concerned by the variant database
- **Orphanum**: unique identifying number assigned by Orphanet to a given disease or given gene
- **Disorder_Name:** preferred name of a given disorder(s), group of disorders and subtypes
- **GeneList count**: total number of disorders, group of disorders and subtypes concerned by the variant database
- **Symbol:** Symbol of the concerned gene

Example

<Registry_BiobankList count="**XXXX**">

XXXX is the total number of variant databases presented in this XML file

<Registry_Biobank id="**1206**">

The unique identifier of the variant database is 1206

<ExpertLink lang="**en**">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=8876 </ExpertLink>

The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=8876

<Name_en> NF-France: genotype/phenotype database on neurofibromatosis 1 </Name>

The english name of this entry is NF-France: genotype/phenotype database on neurofibromatosis 1

<Registry_BiobankType id="**15246**">
<Name lang="**en**">Biobank</Name>

This expert resource is a variant database

<ValidationDate>2018-03-09 00:00:00.0</ValidationDate>

The last update date of the variant database was the 09/03/2018

<GeoCoverage id="**1135**"><Name lang="**en**">National</Name></GeoCoverage>

The coverage of the variant database is "National"

<Country id="**75**"><Name lang="**en**">FRANCE</Name></Country>

The variant database is located in "FRANCE"

<DisorderList count="2">
<OrphaNumber>586</OrphaNumber>
<Name lang="en">Cystic fibrosis</Name>
<OrphaNumber>48</OrphaNumber>
<Name lang="en">Congenital bilateral absence of vas deferens</Name>

The variant database operates for two diseases: Cystic fibrosis (orphanumber 586) and Congenital bilateral absence of vas deferens (orphanumber 48)

<GeneList count="2">
<OrphaNumber>123722</OrphaNumber>
<Symbol>NF1</Symbol>

The variant database operates for one gene which symbol is NF1

6. Medical laboratories

Orphanet provides information on diagnostic tests able to establish a diagnosis of a rare disease and that need a rare technical competence, or that is the best standard in a given country. Constitutional genetic tests are also registered for non-rare diseases, for diseases with a genetic susceptibility and for pharmacogenetics. Tests should be offered in a clinical setting.

Description of the XML tags

- **InstitutionList count**: total number of medical laboratories in the Xml product
- **Departement_Service_Lab_PatientOrganisation_Acronym:** Acronym of the department/service name of the medical laboratory
- **Departement_Service_Lab_PatientOrganisation_Name:** Name of the department/service of the medical laboratory
- **Hosting_Institution:** Name of the hosting institution of the department/service of the medical laboratory
- **Town:** Town of the hosting institution
- **Country:** country of the hosting institution
- **Accreditation_EqA List count:** Number of accreditation or EqA performed by the medical laboratory
- **QualityType_Name:** indicate if the quality data is an accreditation or an EqA
- **DiagnosticList count**: total number of diagnostic tests performed by the medical laboratory
- **ExpertLink**: stable URL pointing to the specific page of the diagnostic test on the Orphanet website
- **Name**: Name of the diagnostic test
- **Lang:** ISO 639 code for language names
- **DgsTestPurposeList count:** Number of purpose(s) associated to the diagnostic test
- **DgsTestPurposeLabel:** Name of the purpose of the diagnostic test. Can be :

Antenatal diagnosis

Newborn screening
Not defined
Pharmacogenetics
Post-natal diagnosis
Pre-implantation diagnosis
Pre-symptomatic diagnosis
Risk assessment
Somatic genetics

- DgsTestTechniqueList count: Number of technique(s) associated to the diagnostic test. Each technique is a combination of one specialty, one objective and one technique
- DgsTestSpecialityLabel: Name of the specialty of the diagnostic test. Can be:

Bacteriology
Biochemical genetics
Cytogenetics
Hematology
Imaging
Immunology
Molecular genetics
Mycology
Other
Parasitology
Pathology
Virology

- DgsTestObjectiveLabel: Name of the objective of the diagnostic test. Can be:

Analyte / Enzyme assay
Chromosomal instability
Deletion / Duplication analysis
Detection of chromosome alterations large in size
Detection of microdeletions/microduplications
Methylation analysis
Mutation scanning/screening and sequence analysis of selected exons
Not defined
Protein expression
Sequence analysis: entire coding region
Targeted mutation analysis
Uniparental disomy study

- DgsTestTechniqueLabel: Name of the technique of the diagnostic test. Can be:

Array based techniques
BS-Pyrosequencing
Chromosome breakage analysis
FISH
Immunohistochemistry
Karyotyping
M-FISH/SKY
Microsatellite analysis
MLPA based techniques
NGS sequencing (except WES)
Not defined
PCR based techniques
Sanger sequencing
Western Blot
Whole Exome Sequencing (WES)

- **DgsTestTechniqueList count:** Number of technique(s) associated to the diagnostic test
- **ValidationDate:** The last update date of the diagnostic test
- **DisorderList count**: total number of disorders, group of disorders and subtypes concerned by the diagnostic test
- **Orphanum**: unique identifying number assigned by Orphanet to a given disease.
- **Disorder_Name:** preferred name of a given disorder(s), group of disorders and subtypes
- **GeneList count**: total number of genes concerned by the diagnostic test
- **Symbol:** Symbol of the concerned gene

Example

<Institution List count="**XXXX**">

XXXX is the total number of medical laboratories presented in this XML file

<Institution id="**35510**"><Department_Service_Lab_PatientOrganisationAcronym/>
<Department_Service_Lab_PatientOrganisationName>Kindernephrologie Bonn</Department_Service_Lab_PatientOrganisationName>
<Address id="**127**"><Hosting_Institution>Universitäts-Kinderklinik Bonn</Hosting_Institution>
<Town><Name>BONN</Name></Town>
<Country id="**75**"><Name lang="**en**">GERMANY</Name></Country>

The medical laboratory is located in the "Kindernephrologie Bonn" of the hosting institution named "Universitäts-Kinderklinik Bonn" located at "BONN" in "GERMANY"

<ValidationDate>2018-03-09 00:00:00.0</ValidationDate>

The last update date on the medical laboratory'sinformation was the 09/03/2018

<Accreditation_EQAList count="2">
<Accreditation_EQA id="20840">
<QualityType id="12538">
<Name lang="en">External Quality Assesment</Name>
<Accreditation_EQA id="16012">
<QualityType id="280">
<Name lang="en">Accreditation</Name>
The medical laboratory has two quality data: one accreditation and one EqA

< DiagnosticTest List count="**12**">
12 is the total number of diagnostic tests performed by the medical laboratory

< DiagnosticTest id="**121117**">
The unique identifier of the diagnostic test is 121117

<ExpertLink lang="**en**">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=484255 </ExpertLink>
The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=484255

<Name> Molekulare Diagnostik des Carnitin-Palmitoyl-Transferase II-Mangels (CPT2-Gen)</Name>
<LanguageOfName>de</LanguageOfName>
The German name of this entry is Molekulare Diagnostik des Carnitin-Palmitoyl-Transferase II-Mangels (CPT2-Gen)

<DgsTestPurpose id="**154842**">
<DgsTestPurpose Label="**28741**">
<Name lang="**en**"> Post-natal diagnosis </Name>
The diagnostic test is performed on post-natal diagnosis purpose

<DgsTestSpeciality id="**154841**">
<DgsTestSpeciality Label id="**28468**">
<Name lang="en"> **Molecular genetics**</Name>
<DgsTestSpecialityObjective id="1**54841**">
<DgsTestSpecialityObjectiveLabel id="**28489**">
<Name lang="en">**Sequence analysis: entire coding region**</Name>
<DgsTestSpecialityObjectiveTechnique id="1**54841**">
<DgsTestSpecialityObjectiveTechniqueLabel id="**28650**">
<Name lang="en">**Sanger sequencing**</Name>
The diagnostic test is performed on Molecular genetics by Sanger on sequence analysis

<DisorderList count="1">
<OrphaNumber>157</OrphaNumber>
<Name lang="**en**">Carnitine palmitoyltransferase II deficiency</Name>
The diagnostic test operates for Carnitine palmitoyltransferase II deficiency (orphanumber157)

<GeneList count="**1**">
<Symbol>CPT2 </Symbol>
The diagnostic test operates for CPT2 gene

7. Research projects and networks of research projects

Orphanet provides information on ongoing and unpublished research projects explicitly focused on a rare disease or on a group of rare diseases and funded by a funding body with a scientific committee performing a competitive selection of research projects, or issued from the regular national research funding. Single-centre and national or international multicentric research projects are registered.

a. Research projects

Description of the XML tags

- **ResearchProjectList count**: total number of research projects in the XML file
- **ExpertLink**: stable URL pointing to the specific page of the research project on the Orphanet website
- **Lang:** ISO 639 code for language names
- **Name_en:** Name of the research project in English
- **ResearchProjectType Flag:** Type of the research project:

Animal model creation / study	In vitro functional study
Biomarker development	Induced pluripotent stem cells (iPS) creation / study
Biorepositories development/creation	Medical device / instrumentation development
Biotechnology innovation	Mutations search
CRISPR-Cas9 study	Natural history study
Databases & Registries development/creation	Observational clinical study
Diagnostic tool/protocol development	Ontology / bioinformatics study
Drug repurposing	Outcomes measures development
Epidemiological study	Pre-clinical cell therapy
Gene expression profile	Pre-clinical drug development / Drug delivery
Gene search	Pre-clinical gene therapy
Genotype-phenotype correlation	Pre-clinical vaccine development
Health economics study	Public health study (excluding health economics)
Health sociology study	Small molecule screening
Human physiopathology study	

- **URL, Protocol URL, PSOIEURL:** Website and descriptions of the research project in English and other language as appropriate
- **Country:** Name of the country where the research project is carried out
- **ValidationDate:** The last update date of the research project
- **InstitutionList Count:** number of institutions linked to the research project
- **Status:** Status of institution(s). Can be "public", "Private non-for-profit" or "Private for-profit"

- **Departement_Service_Lab_PatientOrganisation_Acronym:** Acronym of the department/service name of the hosting institution
- **Departement_Service_Lab_PatientOrganisation_Name:** Name of the department/service of the hosting institution
- **Hosting_Institution:** Name of the hosting institution of the department/service
- **Town:** Town of the hosting institution
- **Country Name:** Country of the hosting institution
- **DisorderList count**: total number of disorders, group of disorders and subtypes concerned by the patient registry
- **Orphanum**: unique identifying number assigned by Orphanet to a given disease.
- **Disorder_Name:** preferred name of a given disorder(s), group of disorders and subtypes.

Example

<ResearchProjectList count="**XXXX**">

XXXX is the total number of research projects presented in this XML file

<ResearchProject id="**61234**">

The unique identifier of the research project is 61234

<ExpertLink lang="**en**">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=172703 </ExpertLink>

The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=173703

<Name_en> Contribution to the study of the CFTR interactome: dynamics and role of a CFTR-containing complex in inflammation in cystic fibrosis </Name>

The English name of this entry is Contribution to the study of the CFTR interactome: dynamics and role of a CFTR-containing complex in inflammation in cystic fibrosis

<ReserachProjectType id="**14976**">
<Name lang="**en**"> In vitro functional study </Name>

The research project type is “In vitro functional study”

<Url/>
<ProtocolUrl>http://www.orpha.net/data/prj/FR/ID61234FR.pdf</ProtocolUrl>
<PSOIEUrl/>

The research project has only a description available at http://www.orpha.net/data/prj/FR/ID61234FR.pdf

<Country id="**75**"><Name lang="**en**">FRANCE</Name></Country>

The research project is located in “FRANCE”

<ValidationDate>2018-03-09 00:00:00.0</ValidationDate>

The last update date of the research project was the 09/03/2018

<InstitutionList count="**2**">

<Institution id="185">
<Status id="13"><Name lang="en">Public</Name></Status>
</Institution><Institution id="1066">
<Status id="15"><Name lang="en">Private non-for-profit</Name></Status>

The research project is linked to two institutions: one with public status and the second with Private non-for-profit status

<PersonFunction id="5"><Type>Rch</Type>
<Name lang="en">Investigator of research project</Name>
</PersonFunction><Institution id="185"><Department_Service_Lab_PatientOrganisationAcronym/><Department_Service_Lab_PatientOrganisationName>Etude des dysfonctions lymphocytaires T en immunopathologie rénale et en transplantation</Department_Service_Lab_PatientOrganisationName>
<UpperLevelOfAffiliationAcronym/><UpperLevelOfAffiliation>INSERM U 955 - Institut Mondor de Recherche Biomédicale</UpperLevelOfAffiliation>
<Address id="19"><Town>
<Name>CRÉTEIL</Name></Town>
<Country id="75"><Name lang="en">FRANCE</Name></Country>

The research project is linked to an investigator located in the service "Etude des dysfonctions lymphocytaires T en immunopathologie rénale et en transplantation » of the department « INSERM U 955 - Institut Mondor de Recherche Biomédicale », at « Créteil » in « France."

<PersonFunction id="45"><Type>Rch</Type>
<Name lang="en">Private non-profit funding body</Name>
</PersonFunction><Institution id="1066"><Department_Service_Lab_PatientOrganisationAcronym/><Department_Service_Lab_PatientOrganisationName>Vaincre La Mucoviscidose</Department_Service_Lab_PatientOrganisationName>
<Address id="287"><Town>
<Name>PARIS</Name></Town>
<Country id="75"><Name lang="en">FRANCE</Name>

The research project is linked to a Private non-profit funding body who is located on the service "Vaincre La Mucoviscidose » at « Paris » in « France".

<DisorderList count="1">
<OrphaNumber>586</OrphaNumber>
<Name lang="en">Cystic fibrosis</Name>

The research project operates for Cystic fibrosis (orphanumber 586)

b. Network of research projects

Description of the XML tags

- **NetworkList count**: total number of networks in the XML file

- **ExpertLink**: stable URL pointing to the specific page of a network on the Orphanet website
- **Lang:** ISO 639 code for language names
- **Name:** Name of the network
- **URL:** Website of the network
- **Country:** country of the coordinating team of the network
- **Geocoverage:** Geographical coverage of the network. Can be "Global", "Regional", "National" or "European"
- **ValidationDate:** The last update date of the network
- **Person_LastName:** Name of the funding body of the network
- **DisorderList count**: total number of disorders, group of disorders and subtypes concerned by the network
- **Researchproject count**: total number of research projects involved in the network

Example

<NetworkList count="**XXXX**">

XXXX is the total number of networks of research projects presented in this XML file

<Network id="**120740**">

The unique identifier of the network is 120740

<ExpertLink lang="**en**">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=482674</ExpertLink>

The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=482674

<Name_en> EURORETT: MeCP2 interaction with DNA and its role on chromatin organisation and subsequent changes of gene expression profile in Rett syndrome</Name>

The name of the entry in English is "EURORETT: MeCP2 interaction with DNA and its role on chromatin organisation and subsequent changes of gene expression profile in Rett syndrome"

<Url> http://www.erare.eu/financed-projects/eurorett </Url>

The URL of the network is http://www.erare.eu/financed-projects/eurorett

<ValidationDate>2018-07-06 00:00:00.0</ValidationDate>

The last update date of the network was the 07/06/2018

<Person id="**45206**">
<Firstname/><Lastname> ERA-Net for research programs on rare diseases - France </Lastname>
<PersonActivityList count="**0**"><PersonActivity><PersonFunction id="**58**"><Type>Net</Type><Name lang="**en**">Public funding body</Name>

The Public funding body of the network is ERA-Net for research programs on rare diseases - France

<DisorderList count="**1**">

<OrphaNumber>**778**</OrphaNumber>
<Name lang="**en**"> **Rett syndrome**</Name></Disorder>
The network operates for rett syndrome (orphanumber 778)

<Researchproject count="**11**">
The network is made of 11 research projects

8. Clinical trials and networks of clinical trials

Orphanet provides information on clinical trials focused on a rare disease or on a group of rare diseases. Trials comprise interventional studies aiming to evaluate a drug (or a combination of drugs or a biological product) to treat (or prevent) a rare disease or a group of rare diseases. Single-centre and national or international multicentric clinical trials are registered. The trials registered in Orphanet can be ongoing, recruiting, or finished.

a. Clinical trial

Description of the XML tags

- **ClinicalTrialList count**: total number of clinical trials in the XML file
- **ExpertLink**: stable URL pointing to the specific page of the clinical trial on t h e Orphanet website
- **Lang:** ISO 639 code for language names
- **Name_en:** Name of the clinical trial in English
- **ClinicalTrialType Flag:** Type of the clinical trial. Can be:

Drug clinical trial
Protocol clinical trial
Gene therapy clinical trial
Cell therapy clinical trial
Vaccine clinical trial
Medical device trial

- **ClinicalTrialStatus Flag:** status of the clinical trial. Can be "Terminated", "Multicentric" or ongoing
- **URL, Protocol URL, PSOIEURL:** Website and descriptions of the clinical trial in English and other language as appropriate
- **Country:** Name of the country where the research project performed
- **Geocoverage:** Geographical coverage of the clinical trial Can be "Global", "Regional", "National" or "European"
- **Phase:** phase of the clinical trial. Can be:

I
I - II
II
II - III
III

IV
bioequivalence

- **ValidationDate:** The last update date of the clinical trial
- **Detail:** specific code
- **PersonList Count:** number of legal entities linked to the clinical trial
- **PersonFunction:** Function of legal entitie(s). Can be "public funding body", "Private non-for-profit funding body" or "Private for-profit funding body"
- **DisorderList count**: total number of disorders, group of disorders and subtypes concerned by the patient registry
- **Orphanum**: unique identifying number assigned by Orphanet to a given disease
- **Disorder_Name:** preferred name of a given disorders, group of disorders and subtypes.
- **DrugActivityDisorderList:** number of substance or tradenames tested in the clinical trial
- **Substance_Tradename:** substance or tradename tested in the clinical trial

Example

<ClinicalTrial List count="**XXXX**">

XXXX is the total number of clinical trials presented in this XML file

< ClinicalTrial id="**61196**">

The unique identifier of the clinical trial is 61196

<ExpertLink lang="**en**">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=172160 </ExpertLink>

The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=172160

<Name_en> STIMUCO: Assessment of Quadriceps Muscle Electrostimulation Used as an Additional Procedure for Effort Retraining in Patients Suffering From Cystic Fibrosis Associated With Severe Pulmonary Dysfunction </Name>

The English name of this entry is STIMUCO: Assessment of Quadriceps Muscle Electrostimulation Used as an Additional Procedure for Effort Retraining in Patients Suffering From Cystic Fibrosis Associated With Severe Pulmonary Dysfunction

<ClinicalTrial Type id="**15102**">
<Name lang="**en**"> Medical device trial </Name>

The clinical trial type is "Medical device trial"

<ClinicalTrialStatus Flag id="**133**">
<Name lang="**en**"> Terminated </Name>

The clinical trial type is "Terminated"

<Url/> https://clinicaltrials.gov/ct2/show/NCT00391703
<ProtocolUrl>
<PSOIEUrl/>

The clinical trial has only a description available at https://clinicaltrials.gov/ct2/show/NCT00391703

<Country id="**75**"><Name lang="**en**">FRANCE</Name></Country>

The clinical trial is located in "FRANCE"

<ValidationDate>2018-03-09 00:00:00.0</ValidationDate>

The last update date of the clinical trial was the 09/03/2018

<Detail>NCT00391703</Detail>

The specific code of the clinical trial was the 09/03/2018

<Phase id="**84**">
<Name lang="**en**">IV</Name>

The clinical trial is in the phase IV

<Person id="**24743**">
<Firstname/>
<Lastname>VAINCRE LA MUCOVISCIDOSE (VLM)</Lastname>
<PersonFunction id="**14**">
<Name lang="**en**">Private non-profit funding body</Name>
</PersonFunction>

The clinical trial is linked to a Private non-profit funding body whose name is VAINCRE LA MUCOVISCIDOSE (VLM).

<DisorderList count="1">
<OrphaNumber>586</OrphaNumber>
<Name lang="**en**">Cystic fibrosis</Name>

The clinical trial operates for Cystic fibrosis (orphanumber 586)

<DrugActivityDisorderList count="**1**">
<DrugActivityDisorder><Substance id="**2614**">
<Name lang="**en**">Leuco-methylthioninium bis(hydromethanesulfonate)</Name>

The clinical trial operates for a substance named "Leuco-methylthioninium bis(hydromethanesulfonate) » (unique identifier 2614)

b. Network of clinical trials

Description of the XML tags

- **NetworkList count**: total number of networks in the XML file
- **ExpertLink**: stable URL pointing to the specific page of a network on the Orphanet website
- **Lang:** ISO 639 code for language names
- **Name_en:** English name of the network
- **URL:** Website of the network

- **Country:** country of the coordinating team of the network
- **Geocoverage:** Geographical coverage of the network. Can be "Global", "Regional", "National" or "European"
- **Phase:** phase of the network of clinical trials. Can be:

I
I - II
II
II - III
III
IV
bioequivalence

- **ValidationDate:** The last update date of the network
- **Person_LastName:** Name of the sponsor of the network
- **DisorderList count**: total number of disorders, group of disorders and subtypes concerned by the network
- **Orphanum**: unique identifying number assigned by Orphanet to a given disease
- **Disorder_Name:** preferred name of a given disorder(s), group of disorders and subtypes.
- **ClinicalTrial count**: total number of clinical trials involved in the network

Example

<NetworkList count="**XXXX**">

XXXX is the total number of networks of clinical trials presented in this XML file

<Network id="**95737**">

The unique identifier of the network is 120740

<ExpertLink lang="**en**">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=316468</ExpertLink>

The stable URL pointing to information on the Orphanet website of this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=316468

<Name_en> ENDEAVOR: A Randomized, Open-label, Phase 3 Study of Carfilzomib Plus Dexamethasone vs. Bortezomib Plus Dexamethasone in Patients With Relapsed Multiple Myeloma -Coordination </Name>

The name of the entry in English is "ENDEAVOR: A Randomized, Open-label, Phase 3 Study of Carfilzomib Plus Dexamethasone vs. Bortezomib Plus Dexamethasone in Patients With Relapsed Multiple Myeloma -Coordination"

<Url> http://clinicaltrials.gov/ct2/show/study/NCT01568866 </Url>

The URL of the network is http://clinicaltrials.gov/ct2/show/study/NCT01568866

<ValidationDate>2018-07-06 00:00:00.0</ValidationDate>

The last update date of the network was the 07/06/2018

<Person id="**32925**">

<Firstname/><Lastname> ONYX PHARMACEUTICALS INC.
</Lastname>
<PersonActivityList count="0"><PersonActivity><PersonFunction id="58"><Type>Net</Type><Name lang="en">Public funding body</Name>

The sponsor of the network is ONYX PHARMACEUTICALS INC.

<DisorderList count="1">
<Disorder id="8776">
<OrphaNumber>29073</OrphaNumber>
<Name lang="en"> Multiple myeloma</Name></Disorder>

The network operates for multiple myeloma (orphanumber 29073)

<ClinicalTrial count="2">

The network is made of 2 clinical trials

III. Orphan drugs

Orphanet provides an inventory of drugs at all stages of development for one particular rare disease or a group of rare diseases. This includes all the substances which have been granted an **orphan designation** for disease(s) considered as rare in Europe or the USA, whether they were further developed to become approved drugs with **marketing authorisation** (MA) or not.

Orphanet also includes drugs without an orphan designation as long as they have been granted a marketing authorisation with a specific indication for a rare disease or because they are tested in a clinical trial performed on a rare disease, but they do not have a regulatory status.

Orphan designation is a legal procedure that allows for the designation of a medicinal substance with therapeutic potential for a rare disease, before its first administration in humans or during its clinical development. The exact therapeutic indication is then defined at the time of **marketing authorisation**. This procedure has been established in Europe by the Regulation on Orphan Medicinal Products (EC) No 141/2000and in the US by the Orphan Drug Act.

Description of the XML tags

- **DrugRegulatroyStatusList count**: total number of networks in the XML file
- **Name:** English name of the designation. Can be “Orphan designation withdrawn”, “Orphan designation”, “Marketing authorization without orphan designation”, “Marketing authorisation with orphan designation” or “Marketing authorization withdrawn”
- **PSOIEURL:** information about the regulatory status
- **AdminZone:** administrative zone of regulatory status. Can be “Europe”, “USA” or “Switzerland”
- **ATCCode:** Anatomical Therapeutic Chemical (ATC) Code of the drug/tradename
- **Creationdateof MA_OD:** creation date of the marketing authorisation or the orphan designation

- **ValidationDate:** The last update date of the regulatory status
- **Person_LastName:** Name of the Sponsor of orphan designation or the Marketing authorisation holder
- **Person function:** Can be "Sponsor of orphan designation" or "Marketing authorisation holder"
- **DisorderList count**: total number of disorders, group of disorders and subtypes concerned by the network
- **Orphanum**: unique identifying number assigned by Orphanet to a given disease
- **Disorder_Name:** preferred name of a given disorder(s), group of disorders and subtypes.
- **DrugActivityDisorderList:** number of substance or tradename tested in the clinical trial
- **DrugTradeName:** name of the associated tradename
- **Substance_Code:** name of the associated substance
- **ChemicalName:** chemical name of the substance
- **Inn:** International Nonproprietary Names (INN) of the substance
- **ProductType:** Can be :

Blood-derived drug
Cell therapy product
Drug
Gene therapy product
Human/animal tissue/organ
Ingredient/substance
Other type of health product

- **ProductType:** Can be "Synthetic / extractive chemistry", "Biotchnology"
- **EUNumber**
- **Abstract:** Indication of the regulatory status

Example

<DrugRegulatoryStatusList count="**XXXX**">

XXXX is the total number of regulatory status presented in this XML file

<DrugRegulatoryStatus="**56474**">

The unique identifier of the regulatory status is 56474

<Name> Marketing authorization without orphan designation </Name>

The name of the entry in English is "Marketing authorization without orphan designation"

<PSOIUrl> http://www.ema.europa.eu/docs/en_GB/document_library/EPAR_-_Summary_for_the_public/human/000406/WC500022201.pdf </PSOIUrl>

More information is available using the URL

<AdminZone id="**1159**">
<Name lang="**en**">Europe</Name>

The administrative zone of the regulatory status is “Europe”

<ATCCode>L01XE01

The ATC Code is “L01XE01”

<CreationDateofMA_OD> 2001-11-07 00:00:00.0</ CreationDateofMA_OD>

The creation date of the status was the 07/11/2001

<Person id="**23244**">
<Firstname/><Lastname> NOVARTIS EUROPHARM LIMITED
</Lastname>
<PersonActivityList count="**0**"><PersonActivity><PersonFunction id="**58**"><Type>Eta</Type><Name lang="**en**"> Marketing authorisation holder </Name>

The Marketing authorisation holder is NOVARTIS EUROPHARM LIMITED

<DisorderList count="**1**">
<OrphaNumber>**3260**</OrphaNumber>
<Name lang="**en**"> **Idiopathic hypereosinophilic syndrome** </Name></Disorder>

The regulatory status operates for Idiopathic hypereosinophilic syndrome (orphanumber 3260)

<DrugTradeName id="528">
<Name lang="**en**"> GLIVEC</Name>
<Substance id="818">
<Code>Imatinib mesilate</Code>
<ChemicalName/>
<Inn>Imatinib</Inn>
<ProductType id="121">
<Name lang="en">Ingredient/substance</Name>
</ProductType>
<ProductionType id="199">
<Name lang="en">Synthetic / extractive chemistry</Name>
</ProductionType>
<Name lang="en">Imatinib mesilate</Name>

The regulatory status operates for GLIVEC which substance is “Imatinab mesylate”

<EU Number>EU/1/01/198/...

The EU Number is EU/1/01/198/…

<Abstract>Glivec is indicated for the treatment of :
 - adult and paediatric patients with newly diagnosed Philadelphia chromosome (bcr-abl) positive (Ph+) chronic myeloid leukaemia (CML) for whom bone marrow transplantation is not considered as the first line of treatment;
- adult and paediatric patients with Ph+ CML in chronic phase after failure of interferon-alpha therapy, or in accelerated phase or blast crisis;
- adult and paediatric patients with newly diagnosed Philadelphia chromosome positive acute lymphoblastic leukaemia (Ph+ ALL) integrated with chemotherapy;
- adult patients with relapsed or refractory Ph+ ALL as monotherapy;
- adult patients with myelodysplastic/myeloproliferative diseases (MDS/MPD) associated with platelet-derived growth factor receptor (PDGFR) gene re-arrangements;
- adult patients with advanced hypereosinophilic syndrome (HES) and/or chronic eosinophilic leukaemia (CEL) with FIP1L1-PDGFRα rearrangement.
 The effect of Glivec on the outcome of bone marrow transplantation has not been determined.

 Glivec is indicated for :
 - the treatment of adult patients with Kit (CD 117) positive unresectable

and/or metastatic malignant gastrointestinal stromal tumours (GIST).
 - the adjuvant treatment of adult patients who are at significant risk of relapse following resection of Kit (CD117)-positive GIST. Patients who have a low or very low risk of recurrence should not receive adjuvant treatment.
 - the treatment of adult patients with Kit (CD 117) positive unresectable and/or metastatic malignant gastrointestinal stromal tumours (GIST).
 - the treatment of adult patients with unresectable dermatofibrosarcoma protuberans (DFSP) and adult patients with recurrent and/or metastatic DFSP who are not eligible for surgery.

 This product is no longer an orphan medicine. It was originally designated an orphan medicine for the treatment of : chronic myeloid leukaemia (14/02/2001); malignant gastrointestinal stromal tumours (20/11/2001); dermatofibrosarcoma protuberans (26/08/2005); acute lymphoblastic leukaemia (26/08/2005); chronic eosinophilic leukaemia and the hypereosinophilic syndrome (28/10/2005); myelodysplastic/myeloproliferative diseases (23/12/2005). Upon request of the marketing-authorisation holder, Glivec has now been removed from the Community register of orphan medicinal products.</Abstract>

The is the designation of the regulatory status

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