



Orphadata Products : Product Description

V3

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Cystic fibrosis

[Suggest an update](#)

Textual information

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 \(2006, pdf\)](#)
 - [English \(2014\)](#)
 - [Deutsch \(2013\)](#)
 - [Français \(2017, pdf\)](#)
- > 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 \(2017\)](#)

Additional information

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 \(52\)](#)
- > [Networks \(43\)](#)

Expert resources

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 clinical entities (disorders, group of disorders or subtypes) in the Xml file
- **ORPHAcode:** a unique and time-stable numerical identifier attributed randomly by the database upon creation of the entity.
- **ExpertLink:** stable URL pointing to the specific page of the given clinical entity on the Orphanet website
- **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
- **Lang:** ISO 639 code for language names
- **Name:** preferred name of a given clinical entity
- **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

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

<DisorderList count="XXXX">

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

<ORPHAcode>586</ORPHAcode>

<Name lang="en">Cystic fibrosis</Name>

The concerned disease has 586 as its ORPHAcode and Cystic fibrosis 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 clinical entities (disorders, group of disorders or subtypes) in the Xml file
- **ORPHAcode:** a unique and time-stable numerical identifier attributed randomly by the database upon creation of the entity.
- **ExpertLink:** stable URL pointing to the specific page of the given clinical entity on the Orphanet website
- **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
- **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

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

<DisorderList count="XXXX">

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

<ORPHAcode>586</ORPHAcode>

<Name lang="en">Cystic fibrosis</Name>

The concerned disease has 586 as its ORPHAcode 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 PubMed

Description of the XML tags

- **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes) in the Xml file
- **ORPHAcodes:** a unique and time-stable numerical identifier attributed randomly by the database upon creation of the entity.
- **ExpertLink:** stable URL pointing to the specific page of the given clinical entity on the Orphanet website
- **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
- **Source:** the unique source in the file is “medline”
- **Reference:** the PubMed query to find information on the concerned disease

Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

<DisorderList count="XXXX">

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

<ORPHAcodes>586</ORPHAcodes>

<Name lang="en">Cystic fibrosis</Name>

The concerned disease has 586 as its ORPHAcodes and Cystic fibrosis 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;
- [medical laboratories](#);
- [national research projects](#) and multinational research projects;
- [national clinical trials](#) and multinational 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/or are members of RDI AND they have legal status.

Research projects, clinical trials, patient registries and biobanks 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 and should deliver genetic advice either for all genetic diseases or for a genetic disease/group of diseases, and be either officially designated either officially designated by the country's health authorities, a member of a European Reference Network (ERN), or comply with the Orphanet quality questionnaire for Genetic Counselling Units / Clinics based on peer reviewed literature about genetic counselling practices in Europe and the European Board of Medical Genetics



documentation for European unified competency standards for ensuring patient safety.

a. Expert centres

Description of the XML tags

- **ExpertCentreList count:** total number of expert centres in the XML file
- **ExpertCentre id:** Internal and numerical identifier of the expert center in the Orphanet database.
- **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 in local language
- **Name_en:** Name of the expert centre in English
- **LanguageofName:** name of the local language
- **ExpertCentreStatusFlagList count:** Number of expert centre's status
- **ExpertCentreStatusFlag Name:** Status of the expert centre that can be:
 - o "on-line" which is an automatic status meaning that the expert centre is published on Orphanet.
 - o and/or "Member of a European Reference Network (ERN)" if the expert centre belongs to an ERN
 - o and/or "Adult clinic" if the expert centre is specialized for adults
 - o and/or "Child clinic" if the expert centre is specialized for children
 - o and/or "Medical management clinic" if it is specialised in medical management.
 - o and/or "Officially designated Medical management" if the center is designated officially by a local health authority
 - o and/or "Genetic counselling clinic" if it is specialised in genetic counselling
 - o and/or "Officially designated Genetic counselling" if the center is designated officially by a local health authority
- **ValidationDate:** The last update date of the expert centre
- **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 clinical entities (disorders, group of disorders or subtypes) in the Xml file
- **ORPHAcodes:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.



- **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community

Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

<ExpertCentreList count="XXXX">

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

<ExpertCentre id="86703">

The internal identifier of the expert centre is 86703

<ExpertLink lang="en"> http://www.orphadata.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=281376 </ExpertLink>

The stable URL pointing to information on the Orphanet website of this entry is http://www.orphadata.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=281376.

<Name> [Radboudumc-Maastricht UMC+ Expertisecentrum voor Marfan Syndroom en Aanverwante Aandoeningen](#) </Name>

The name of this entry in local language is: “Radboudumc-Maastricht UMC+ Expertisecentrum voor Marfan Syndroom en Aanverwante Aandoeningen”.

<Name en> [Radboudumc-Maastricht UMC+ Center of Expertise for Marfan and Related Disorders](#) </Name en>

The English name of this entry is: “Radboudumc-Maastricht UMC+ Center of Expertise for Marfan and Related Disorders”.

<LanguageOfName>nl</LanguageOfName>

The local language is Dutch.

<ExpertCentreStatusFlagList count="8">

<ExpertCentreStatusFlag id="35">

<Name lang="en">on-line</Name>

<ExpertCentreStatusFlag id="39">

<Name lang="en">Genetic counselling clinic</Name>

<ExpertCentreStatusFlag id="42">

<Name lang="en">Adult clinic</Name>

<ExpertCentreStatusFlag id="40">

<Name lang="en">Medical management clinic</Name>

<ExpertCentreStatusFlag id="41">

<Name lang="en">Child clinic</Name>

<ExpertCentreStatusFlag id="492">

<Name lang="en">Officially designated Genetic counselling</Name>

<ExpertCentreStatusFlag id="491">



```
<Name lang="en">Officially designated Medical Management</Name>  
<ExpertCentreStatusFlag id="485">  
<Name lang="en">Member of a European Reference Network  
(ERN)</Name>
```

This expert centre is a clinic, specialized in medical management and genetic counselling for Adult and Child. Thus, it has the statuses “officially designated medical management” and “officially designated genetic counselling”, and is a member of a European Reference Network (ERN). The on-line status, only means that it is published (on-line) on the Orphanet website.

```
<ValidationDate>2025-05-21 00:00:00.0</ValidationDate>
```

The last update date of the expert centre was the 21/05/2021.

```
<InstitutionList count="1">  
<Institution id="30508">  
<Department Service Lab PatientOrganisationAcronym/>  
<Department Service Lab PatientOrganisationName> Sectie Klinische  
Genetica </Department Service Lab PatientOrganisationName>  
<Status id="13">  
<Name lang="en">Public</Name></Status>  
<Address id="12571">  
<Hosting Institution>Radboudumc - Radboud universitair medisch  
centrum </Hosting Institution>  
<Town><Name> NIJMEGEN </Name></Town>  
<Country id="161">  
<Name lang="en">NETHERLANDS</Name></Country>
```

The expert centre is located in the department/service “Sectie Klinische Genetica” of the “Public” hosting institution named “Radboudumc - Radboud universitair medisch centrum” located at “NIJMEGEN” in the “NETHERLANDS”.

```
<DisorderList count="6">  
<ORPHAcode>558</ORPHAcode>  
<Name lang="en">Marfan syndrome</Name>  
<ORPHAcode>286</ORPHAcode>  
<Name lang="en">Vascular Ehlers-Danlos syndrome</Name>  
<ORPHAcode>60030</ORPHAcode>  
<Name lang="en">Loeys-Dietz syndrome</Name>  
<ORPHAcode>91387</ORPHAcode>  
<Name lang="en">Familial thoracic aortic aneurysm and aortic  
dissection</Name>  
<ORPHAcode>285014</ORPHAcode>  
<Name lang="en">Rare disease with thoracic aortic aneurysm and  
aortic dissection</Name>  
<ORPHAcode>458718</ORPHAcode>  
<Name lang="en">Idiopathic spontaneous coronary artery  
dissection</Name>
```

The expert centre operates for five diseases/groups of diseases: Marfan syndrome



(ORPHAcode 558), Vascular Ehlers-Danlos syndrome (ORPHAcode 286), Loeys-Dietz syndrome (ORPHAcode 60030), Familial thoracic aortic aneurysm and aortic dissection (ORPHAcode 91387), Rare disease with thoracic aortic aneurysm and aortic dissection (ORPHAcode 285014) and Idiopathic spontaneous coronary artery dissection (ORPHAcode 458718).

b. Network of expert centres

Description of the XML tags

- **NetworkList count:** total number of networks in the XML file
- **Network id:** Internal and numerical identifier of the network in the Orphanet database.
- **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 in local language
- **Name_en:** Name of the network in English
- **LanguageOfName:** name of the local language
- **NetworkStatusFlagList count:** Number of network's status
- **NetworkStatusFlag Name:** Status of the network that can be:
 - o "on-line" which is an automatic status meaning that the network is published on Orphanet.
 - o and/or "Member of a European Reference Network (ERN)" if the network is an ERN
- **URL:** Website of the network
- **Geocoverage:** Geographical coverage of the network. Can be "Regional", "National", "European" or "International (beyond Europe)"
- **ValidationDate:** The last update date of the network
- **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes) in the Xml file
- **ORPHAcode:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.
- **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
- **ExpertCentre count:** total number of expert centres involved in the network

[For each expert centre participating in this network:](#)

- o **ExpertCentre id:** Internal and numerical identifier of the expert center in the Orphanet database.
- o **ExpertLink:** stable URL pointing to the specific page of the given expert centre on the Orphanet website
- o **Name:** name of the given expert centre in local language



- **Name_en**: name of the expert center in English
- **LanguageOfName**: name of the local language
- **Url**: website of the given expert center
- **Country_name**: country where the expert centre is located
- **Institution_Address_Town**: more specific location of the expert center in the country

Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

<NetworkList count="XXXX">

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

<Network id="113119">

The internal identifier of the network is 113119.

<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=442815</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=442815.

<Name > [Muco-CFTR - Filière de santé maladies rares : mucoviscidose et affections liées à une anomalie de CFTR](#) </Name>

The name of the entry in local language (French) is "Muco-CFTR - Filière de santé maladies rares : mucoviscidose et affections liées à une anomalie de CFTR".

<Name en> [Muco-CFTR - French rare diseases Healthcare Network : Cystic fibrosis and diseases related to an abnormal CFTR](#)</Name>

The name of the entry in English is: "Muco-CFTR - French rare diseases Healthcare Network: Cystic fibrosis and diseases related to an abnormal CFTR".

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

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

<GeoCoverage id="1135">

<Name lang="en">[National](#)</Name></GeoCoverage>

The network performs its activities at the National level.

<ValidationDate>[2019-09-27 00:00:00.0](#)</ValidationDate>

The last update date of the network was the 27/09/2019.

<DisorderList count="1">

<Disorder id="49">

<ORPHAcode>[586](#)</ORPHAcode>



<Name lang="en">Cystic fibrosis</Name></Disorder></DisorderList>

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

<Subtype>Cns</Subtype>

<ExpertCenter count="47">

The network made up of 47 expert centres.

For each expert centre participating in this network, you will find the following information in the xml file:

<ExpertCentre id="21692">

The internal identifier of the expert centre is 21692.

<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=67729</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=67729.

<Name>Centre de référence de la mucoviscidose et affections liées à une anomalie de CFTR - Site coordonnateur</Name>

The name of this entry in local language is: « Centre de référence de la mucoviscidose et affections liées à une anomalie de CFTR - Site coordonnateur ».

<Name en>National reference centre for cystic fibrosis and diseases related to an abnormal CFTR - Coordinator centre</Name en>

The English name of this entry is: "National reference centre for cystic fibrosis and diseases related to an abnormal CFTR - Coordinator centre".

<LanguageOfName>fr</LanguageOfName>

The local language is French (fr).

<Url><https://muco-cftr.fr/index.php></Url>

The URL of the network is <https://muco-cftr.fr/index.php>.

<Country id="75">

<ISO>FR</ISO>

<Name lang="en">FRANCE</Name>

</Country>

This expert centre is located in France (e.g. the 47 experts centers associated to the network are all located in France since the network has a national coverage).

<InstitutionList count="1">

<Institution id="6378">

<Address id="7706">



```
<Town><Name>PARIS</Name>  
</Town>  
</Address>  
</Institution>  
</InstitutionList>
```

More precisely, this expert centre is located in the town of Paris.

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.
- **PatientOrganisation id:** Internal and numerical identifier of the patient organisation in the Orphanet database.
- **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 organization in local language
- **LanguageOfName:** name of the local language
- **PatientOrganisationStatusFlagList count:** Number of patient organisation status
- **PatientOrganisationStatusFlag Name:** Status of the patient organization that can be:
 - o “on-line” which is an automatic status meaning that the patient organization is published on Orphanet.
 - o and/or “Member of a European Reference Network (ERN)” if the patient organization is an European Patient Advocacy Groups (ePAGs) belonging to an ERN
- **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 “Regional”, “National”, “European” or “International (beyond Europe)”
- **ValidationDate:** The last update date of the patient organisation
- **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes) in the Xml file



- **ORPHAcode:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.
- **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community

Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

<PatientOrganisationList count="XXXX">

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

<PatientOrganisation id="697">

The internal identifier of the patient organisation is 697.

<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>[Vaincre la Mucoviscidose](#)</Name>

The name of the entry in local language is "Vaincre la mucoviscidose".

<LanguageOfName>[fr](#)</LanguageOfName>

The local language is French.

<PatientOrganisationStatusFlagList count="1">

<PatientOrganisationStatusFlag id="35">

<Name lang="en">[on-line](#)</Name>

The patient organization has only one status: "on-line", which means that the patient organization is published on Orphanet's website.

<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.

<GeoCoverage id="1135">

<Name lang="en">[National](#)</Name>

</GeoCoverage>

The patient organisation operates at the National level.

<ValidationDate>[2025-06-23 00:00:00.0](#)</ValidationDate>

The last update date of the patient organisation was the 23/06/2025.

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

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

b. Network of patient organisations

Description of the XML tags

- **NetworkList count:** total number of networks in the XML file.
 - **Network id:** Internal and numerical identifier of the network in the Orphanet database.
 - **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 in local language
 - **LanguageofName:** Name of the local language
 - **NetworkStatusFlagList count:** Number of network's status
 - **NetworkStatusFlag Name:** Status of the network that can be:
 - o "on-line" which is an automatic status meaning that the network is published on Orphanet.
 - o and/or "Member of a European Reference Network (ERN)" if the network is an European Patient Advocacy Groups (ePAGs) belonging to an ERN
 - **URL:** Website of the network
 - **Geocoverage:** Geographical coverage of the network. Can be "Regional", "National", "European" or "International (beyond Europe)"
 - **ValidationDate:** The last update date of the network
 - **PersonList count:** total number of persons/contact points of the network
 - **Country:** country name and ISO code where the person is coordinating the network
 - **PersonFunction:** function of the person identified – coordinator of the patient organisations network
 - **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes) in the Xml file
 - **ORPHAcode:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.
 - **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
 - **PatientOrganisation count:** total number of patient organisations involved in the network
- [For each patient organisation participating in this network:](#)
- o **PatientOrganisation id:** Internal and numerical identifier of the patient organization in the Orphanet database.



- **ExpertLink:** stable URL pointing to the specific page of the given patient organisation on the Orphanet website
- **Name:** name of the given patient organisation in local language
- **LanguageOfName:** name of the local language
- **Url:** website of the given patient organisation

Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

<NetworkList count="XXXX">

XXXX is the total number of patients organisations networks presented in this XML file.

<Network id="81635">

The internal identifier of the network is 81635.

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

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

<LanguageOfName>en</LanguageOfName>

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

<NetworkStatusFlagList count="1">

<NetworkStatusFlag id="35">

<Name lang="en">on-line</Name>

The patient organization network has only one status: "on-line", which means that it is published on Orphanet's website.

<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 performs at the European level.

<ValidationDate>2023-03-15 00:00:00.0</ValidationDate>

The last update date of the network was the 15/03/2023.

<PersonList count="1">

<Person id="49712">

<Country id="22">

<ISO>BE</ISO>

<Name lang="en">BELGIUM</Name></Country>

<PersonActivityAssociationList count="0">

<PersonActivityAssociation>



```
<PersonFunction id="38">  
<Name lang="en">Coordinator of patient organisation  
network</Name></PersonFunction></PersonActivityAssociation>
```

The network is coordinated by a person living in Belgium.

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

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

```
<Subtype>Ass</Subtype>  
<PatientOrganisation count="17">
```

The network made up of 17 patient organisations.

[For each patient organisation participating in this network, you will find the following information in the xml file:](#)

```
<PatientOrganisation id="31387">
```

The internal identifier of the patient organization is 31387.

```
<ExpertLink lang="en"> http://www.orpha.net/consor/cgi-bin/OC\_Exp.php?lng=en&Expert=60119</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=60119.

```
<Name> Cystische Fibrose Hilfe Österreich </Name>
```

The name of this entry in local language is: « Cystische Fibrose Hilfe Österreich ».

```
<LanguageOfName>de</LanguageOfName>
```

The local language is German (de).

```
<Url>https://muco-cftr.fr/index.php </Url>
```

The local URL of the patient organisation is <http://www.cf-austria.at>.



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 registries

Description of the XML tags

- **PatientregistryList count:** total number of patient registries in the XML file.
- **Patientregistry id:** Internal and numerical identifier of the patient registry in the Orphanet database.
- **OrphaNumber:** Unique and time-stable numerical identifier of the patient registry in the Orphanet database.
- **ExpertLink:** stable URL pointing to the specific page of the patient registry on the Orphanet website
- **Lang:** ISO 639 code for language names
- **Name_en:** Name of the patient registry in English
- **Name:** Name of the patient registry in local language (language of the country where the registry is located)
- **LanguageofName:** Name of the local language
- **PatientregistryStatusFlagList count:** Number of patient registry's status
- **PatientregistryStatusFlag Name:** Status of the patient registry that can be:
 - o "on-line" which is an automatic status meaning that the patient registry is published on Orphanet.
 - o and/or "Terminated" if the patient registry is no longer active. If the status "Terminated" is not specified, the patient registry is still on-going.
 - o and/or "IRDiRC" if the patient registry is funded by a member of the IRDiRC consortium
 - o and/or "multicentric" if the patient registry is regional or national and involves several sites within the country
 - o and/or "Member of a European Reference Network (ERN)" if the patient registry belongs to an ERN
- **URL, Protocol URL, PSOIEURL:** Website and descriptions of the patient registry in English and other language as appropriate
- **Country:** ISO code and name of the country where the patient registry is located
- **Geocoverage:** Geocoverage of the patient registry. Can be "Regional", "National", "European" or "International (beyond Europe)"
- **Recruitment:** Recruitment status of the patient registry. Can be "yes" if the registry is still



- recruiting or “no” if it no longer recruiting.
- **BeginDate:** date on which the patient registry started
 - **EndDate:** date on which the patient registry ended or will end.
 - **ValidationDate:** The last update date of the patient registry by the Orphanet team
 - **PersonFunction_Name:** Name of the functions of stakeholders involved in the patient registry. Stakeholders are the funding body financing the patient registry and/or the persons in charge of the patient registry. Thus, 5 functions can be found in the xml file: “Public funding body”, “Private for-profit funding body”, “Private non-for-profit funding body”, “Manager of registry” and “Contact person of the registry”.
 - **Person_LastName:** Name of the funding body of the patient registry
 - **Institution_Departement_Service_Lab_PatientOrganisation_Name:** Name of the department(s)/service(s) of the funding body hosting institution and/or manager hosting institution and/or contact person hosting institution
 - **Hosting_Institution:** Name of the hosting institution(s) of the funding body and/or manager and/or contact person
 - **Town:** Town(s) of the hosting institution(s)
 - **Country ISO:** Country(ies) ISO of the hosting institution(s)
 - **Country Name:** Country(ies) of the hosting institution(s)
 - **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes) studied in the patient registry
 - **ORPHAcodes:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.
 - **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community

Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

<PatientregistryList count="XXXX">

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

<PatientRegistry id="175491">

<OrphaNumber=" 719572">

The internal identifier of the patient registry is 175491. Its OrphaNumber is 719572.

<ExpertLink lang="en"> http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=719572 </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=719572.



<Name> PIN'SNP : Cohorte prospective nationale de patients suivis pour un syndrome néphrotique idiopathique ayant débuté à l'âge pédiatrique</Name>

<Name en> PIN'SNP: National prospective cohort of patients followed for idiopathic nephrotic syndrome that began in childhood</Name en>

<LanguageofName>fr</LanguageOfName>

The name of this entry in local language is « PIN'SNP : Cohorte prospective nationale de patients suivis pour un syndrome néphrotique idiopathique ayant débuté à l'âge pédiatrique ». The title in English is: « PIN'SNP: National prospective cohort of patients followed for idiopathic nephrotic syndrome that began in childhood ». The local language is “fr” thus, French.

<PatientRegistryStatusFlagList count="2">

<PatientRegistryStatusFlag id="35">

<Name>on-line</Name>

<PatientRegistryStatusFlag id="37">

<Name>multicentric</Name>

The patient registry has 2 statuses: it is “online”, meaning it is published on the Orphanet website and it is “multicentric”, meaning that it is recruiting in several centers within the country.

<Url><https://www.snephroped.org/recherche/pin-snp/></Url>

<ProtocolUrl> </ProtocolUrl>

<PSOIEUrl> </PSOIEUrl>

The patient registry has one URLs: <https://www.snephroped.org/recherche/pin-snp/>

<Country id="75">

<Iso>FR</Iso>

<Name lang="en">FRANCE</Name>

</Country>

The country where the patient registry is located is France.

<Geocoverage id="1135">

<Name lang="en">National</Name>

This patient registry geocoverage is National.

<Recruitment id="96">

<Name lang="en">yes</Name>

This patient registry is still recruiting (Recruitment = “yes”).

<BeginDate> 2020-01-01 00:00:00.0 </BeginDate>

<EndDate> </EndDate>

<ValidationDate>2025-12-16 00:00:00.0</ValidationDate>

The patient registry started on the 1st January 2020 and has no end date for



now. It was last updated by the Orphanet team on the 16/12/2025.

```
<PersonActivityAssociation>
<PersonFunction id="31">
  <Name lang="en">Manager of registry</Name>
</PersonFunction>
<Institution id="13284">
<Department Service Lab PatientOrganisationName>Service de
Pédiatrie médicale - spécialités
médicales</Department Service Lab PatientOrganisationName
>
<Address id="14305">
<Hosting Institution>CHU de Limoges - Hôpital de la femme,
de la mère et de l'enfant</Hosting Institution>
<Town>
<Name>LIMOGES</Name></Town>
<Country id="75">
<ISO>FR</ISO>
<Name lang="en">FRANCE</Name>
```

The patient registry is managed by a “Manager of registry” located in the service « Service de Pédiatrie médicale - spécialités médicales » of the hosting institution named « CHU de Limoges - Hôpital de la femme, de la mère et de l'enfant », which is located in the town of « Limoges » in the country « France ».

```
<PersonFunction id="50">
  <Name lang="en">Public funding body</Name>
</PersonFunction>
<Person id="41933">
<LastName>ASSISTANCE PUBLIQUE - HÔPITAUX DE
PARIS</LastName>
</Person>
<Institution id="22606">
<Department Service Lab PatientOrganisationName>Service de
Néphrologie
pédiatrique</Department Service Lab PatientOrganisationNa
me>
<Address id="24">
<Hosting Institution>AP-HP.Nord - Université Paris Cité -
Hôpital Robert-Debré</Hosting Institution>
<Town>
<Name>PARIS</Name></Town>
<Country id="75">
<ISO>FR</ISO>
<Name lang="en">FRANCE</Name>
```

The patient registry is funded by a « Public funding body » named « Assistance Publique – Hôpitaux de Paris », located in the service « Service de Néphrologie pédiatrique » of the hosting institution named « AP-HP.Nord - Université Paris Cité - Hôpital Robert-Debré », which is located in the town of « Paris » in the country « France ».



```
<PersonFunction id="32">  
  <Name lang="en">Contact person of registry</Name>  
</PersonFunction>  
<Institution id="22606">  
  <Department Service Lab PatientOrganisationName>Service de  
  Néphrologie  
  pédiatrique</Department Service Lab PatientOrganisationNa  
  me>  
  <Address id="24">  
  <Hosting Institution>AP-HP.Nord - Université Paris Cité -  
  Hôpital Robert-Debré</Hosting Institution>  
  <Town>  
  <Name>PARIS</Name></Town>  
  <Country id="75">  
  <ISO>FR</ISO>  
  <Name lang="en">FRANCE</Name>
```

The patient registry also has a “Contact person of registry” located in the service « Service de Néphrologie pédiatrique» of the hosting institution named « AP-HP.Nord - Université Paris Cité - Hôpital Robert-Debré», which is located in the town of « Paris » in the country « France ».

```
<DisorderList count="1">  
  <Disorder id="22226">  
  <ORPHAcode>357502</ORPHAcode>  
  <Name lang="en">Idiopathic nephrotic  
  syndrome</Name></Disorder></DisorderList>
```

The patient registry operates for one diseases/group of diseases: Idiopathic nephrotic syndrome (ORPHAcode 357502).

b. Network of patient registries

Description of the XML tags

- **NetworkList count:** total number of networks in the XML file
- **Network id:** Internal and numerical identifier of the network in the Orphanet database.
- **OrphaNumber:** Unique and time-stable numerical identifier of the network of patient registries in the Orphanet database.
- **ExpertLink:** stable URL pointing to the specific page of the network of patient



- registries on the Orphanet website
- **Lang:** ISO 639 code for language names
 - **Name:** Name of the network in local language
 - **Name_en:** Name of the network in English
 - **LanguageOfName:** name of the local language
 - **NetworkStatusFlagList count:** Number of network's status
 - **NetworkStatusFlag Name:** Status of the network that can be:
 - o "on-line" which is an automatic status meaning that the network is published on Orphanet.
 - o and/or "Terminated" if the network is no longer active. If the status "Terminated" is not specified, the network is still on-going.
 - o and/or "IRDiRC" if the network is funded by a member of the IRDiRC consortium
 - o and/or "Member of a European Reference Network (ERN)" if all patient registries included in the network belong to ERNs
 - **URL, Protocol URL, PSOIEURL:** Website and descriptions of the network in English and other language as appropriate
 - **Country:** ISO code and name of the country where the network is coordinated
 - **Geocoverage:** Geographical coverage of the network. Can be "European" or "International (beyond Europe)".
 - **ValidationDate:** The last update date of the network by the Orphanet team
 - **PersonFunction_Name:** Name of the functions of stakeholders involved in the network of patient registries. Stakeholders are the funding body financing the network and/or the coordinator of the network. Thus, 4 functions can be found in the xml file: "Public funding body", "Private for-profit funding body", "Private non-for-profit funding body" and "Coordinator of patient registry network".
 - **Person_LastName:** Name of the funding body of the network
 - **Institution_Departement_Service_Lab_PatientOrganisation_Name:** Name of the department(s)/service(s) of the funding body hosting institution and/or coordinator of the patient registries network
 - **Hosting_Institution:** Name of the hosting institution(s) of the funding body and/or coordinator
 - **Town:** Town(s) of the hosting institution(s)
 - **Country ISO:** Country(ies) ISO of the hosting institution(s)
 - **Country Name:** Country(ies) of the hosting institution(s)
 - **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes) studied in the network
 - **ORPHAcodes:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.
 - **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
 - **PatientRegistry count:** total number of patient registries involved in the network

[For each patient registry involved in the network, you will find its whole dataset described in part 3.a\) of this document.](#)



Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

<NetworkList count="XXXX">

XXXX is the total number of patient registry networks presented in this XML file.

<Network id="165480">

<OrphaNumber="685656">

The internal identifier of the network is 165480 and its OrphaNumber is 685656.

<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=685656</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=685656.

<Name>[ERN \[GUARD-Heart\] - Heart-Core Registry](#)</Name>

<Name en>[ERN \[GUARD-Heart\] - Heart-Core Registry](#)</Name en>

<LanguageOfName>en</LanguageOfName>

The name of the entry in English is " ERN [GUARD-Heart] - Heart-Core Registry".

<NetworkStatusFlagList count="4">

<NetworkStatusFlag id="35">

<Name lang="en">[on-line](#)</Name>

<NetworkStatusFlag id="463">

<Name lang="en">[IRDiRC](#)</Name>

<NetworkStatusFlag id="493">

<Name lang="en">[Member of a European Reference Network \(ERN\)](#)</Name>

The patient registry network has 3 statuses: "on-line", meaning that it is published on Orphanet ; "IRDiRC" meaning that the network is funded by an IRDiRC member; "Member of a European Reference Network (ERC)", meaning that all patient registries of this network belong to ERNs.

<Url><https://guardheart.ern-net.eu/disease-registries/heart-core-registry/></Url>

<ProtocolUrl>

<PSOIEUrl>

The network is described in one URL, which is <https://guardheart.ern-net.eu/disease-registries/heart-core-registry/>.

<Country id="161">

<ISO>[NL](#)</ISO>

<Name lang="en">[NETHERLANDS](#)</Name>



The network is coordinated in the Netherlands (ISO code: NL).

```
<Geocoverage id="15">  
<Name lang="en">European</Name>
```

The network's geocoverage is "European", meaning that it includes patient registries located in several European countries.

```
<ValidationDate>2024-09-27 00:00:00.0</ValidationDate>
```

The last update date of the network was the 27/09/2024.

```
<PersonActivityAssociation>  
<PersonFunction id="58">  
  <Name lang="en">Public funding body</Name>  
</PersonFunction>  
<Person id="51178">  
  <LastName>EUROPEAN COMMISSION - 3HP - 3RD HEALTH  
PROGRAMME</LastName>  
</Person>  
<Institution id="27901">  
<Department Service Lab PatientOrganisationName>DG SANTE -  
Directorate General for Health and Food  
Safety</Department Service Lab PatientOrganisationName>  
<Address id="15783">  
<Hosting Institution>European Commission</Hosting Institution>  
<Town>  
<Name>BRUSSELS</Name></Town>  
<Country id="22">  
<ISO>BE</ISO>  
<Name lang="en">BELGIUM</Name>
```

The network is funded by a Public funding body, which is the «European Commission – 3HP - 3rd health programme» located in the service « DG Santé – Directorate General for Health and Food Safety» of the hosting institution named « European Commission», which is located in the town of «Brussels» in the country «Belgium».

```
<PersonFunction id="40">  
  <Name lang="en">Coordinator of patient registry network</Name>  
</PersonFunction>  
<Institution id="26458">  
<Department Service Lab PatientOrganisationName>Polikliniek  
Cardiologie</Department Service Lab PatientOrganisationName>  
<Address id="7812">  
<Hosting Institution>Amsterdam UMC, locatie  
AMC</Hosting Institution>  
<Town>  
<Name>AMSTERDAM</Name></Town>  
<Country id="161">  
<ISO>NL</ISO>  
<Name lang="en">NETHERLANDS</Name>
```



The network is coordinated by a “Coordinator of patient registry network” located in the service «Polikliniek» of the hosting institution named « Amsterdam UMC, locatie AMC», which is located in the town of «Amsterdam » in the country «Netherlands ».

```
<DisorderList count="1">  
<Disorder id="12948">  
<ORPHAcode>97929</ORPHAcode>  
<Name lang="en">Rare cardiac  
disease</Name></Disorder></DisorderList>
```

The network operates for one group of diseases: Rare cardiac diseases (ORPHAcode 97929).

```
<PatientRegistry count="7">  
7 patient registries are part of this network.
```

[For each patient registry participating in this network, you will find the following information in the xml file:](#)

```
<PatientRegistry id="165477">  
<OrphaNumber=" 685651">
```

The internal identifier of the patient registry is 165477. Its OrphaNumber is 685651.

```
<ExpertLink lang="en"> http://www.orpha.net/consor/cgi-  
bin/OC\_Exp.php?lng=en&Expert=685651 </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=685651.

```
<Name> ERN \[GUARD-Heart\] - Catecholaminergic Polymorphic  
Ventricular Tachycardia Registry</Name>  
<Name en> ERN \[GUARD-Heart\] - Catecholaminergic Polymorphic  
Ventricular Tachycardia Registry </Name en>  
<LanguageofName>en</LanguageOfName>
```

The name of this entry in local language which is the same as English language is: « ERN [GUARD-Heart] - Catecholaminergic Polymorphic Ventricular Tachycardia Registry ».

```
<PatientRegistryStatusFlagList count="3">  
<PatientRegistryStatusFlag id="35">  
<Name>on-line</Name>  
<PatientRegistryStatusFlag id="463">  
<Name>IRDIRC</Name>  
<PatientRegistryStatusFlag id="485">  
<Name>Member of a European Reference Network (ERN)</Name>
```



The patient registry has 3 statuses: it is “online”, meaning it is published on the Orphanet website; it is “IRDiRC”, meaning that it is funded by a member of the IRDiRC consortium and it is “Member of a European Reference Network (ERN)”, meaning that it belongs to an ERN.

```
<Url>https://guardheart.ern-net.eu/condition-specific-registries/</Url>
```

```
<ProtocolUrl> </ProtocolUrl>
```

```
<PSOIEUrl>https://guardheart.ern-net.eu/disease-registries/heart-core-registry/</PSOIEUrl>
```

The patient registry has two different URLs: <https://guardheart.ern-net.eu/condition-specific-registries/> and <https://guardheart.ern-net.eu/disease-registries/heart-core-registry/>

```
<Country id="161">
```

```
<Iso>NL</Iso>
```

```
<Name lang="en">NETHERLANDS</Name>
```

```
</Country>
```

The country where the patient registry is located is the Netherlands.

```
<Geocoverage id="15">
```

```
<Name lang="en">European</Name>
```

This patient registry geocoverage is European.

```
<Recruitment id="96">
```

```
<Name lang="en">yes</Name>
```

This patient registry is still recruiting (Recruitment = “yes”).

```
<BeginDate> 2020-03-01 00:00:00.0 </BeginDate>
```

```
<EndDate> </EndDate>
```

```
<ValidationDate>2024-09-27 00:00:00.0</ValidationDate>
```

The patient registry started on the 1st March 2020 and has no end date for now. It was last updated by the Orphanet team on the 27/09/2024.

```
<PersonActivityAssociation>
```

```
<PersonFunction id="31">
```

```
<Name lang="en">Manager of registry</Name>
```

```
</PersonFunction>
```

```
<Institution id="26458">
```

```
<Department Service Lab PatientOrganisationName>Poliklini  
ek Cardiologie
```

```
</Department Service Lab PatientOrganisationName>
```

```
<Address id="7812">
```

```
<Hosting Institution>Amsterdam UMC,  
AMClocatie
```

```
<Town>
```

```
<Name>AMSTERDAM</Name></Town>
```



```
<Country id="161">  
<ISO>NL</ISO>  
<Name lang="en">NETHERLANDS</Name>
```

The patient registry is managed by a “Manager of registry” located in the service « Polikliniek Cardiologie» of the hosting institution named « Amsterdam UMC, locatie AMC», which is located in the town of « Amsterdam » in the country « Netherlands ».

```
<PersonFunction id="32">  
<Name lang="en">Contact person of registry</Name>  
</PersonFunction>  
<Institution id="26458">  
<Department Service Lab PatientOrganisationName>Poliklini  
ek  
Cardiologie  
</Department Service Lab PatientOrganisationName>  
<Address id="7812">  
<Hosting Institution>Amsterdam UMC, locatie  
AMC</Hosting Institution>  
<Town>  
<Name>AMSTERDAM</Name></Town>  
<Country id="161">  
<ISO>NL</ISO>  
<Name lang="en">NETHERLANDS</Name>
```

The patient registry also has a “Contact person of registry” located in the service « Polikliniek Cardiologie» of the hosting institution named « Amsterdam UMC, locatie AMC», which is located in the town of « Amsterdam » in the country « Netherlands ».

```
<DisorderList count="1">  
<Disorder id="3525">  
<ORPHACode>3286</ORPHACode>  
<Name lang="en">Neuromuscular  
disease</Name></Disorder></DisorderList>
```

The patient registry operates for one diseases/group of diseases: Catecholaminergic polymorphic ventricular tachycardia (ORPHACode 3286).



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

- **BiobankList count:** total number of biobanks in the XML file
- **Biobank id:** Internal and numerical identifier of the biobank in the Orphanet database.
- **OrphaNumber:** Unique and time-stable numerical identifier of the biobank in the Orphanet database.
- **ExpertLink:** stable URL pointing to the specific page of the biobank on the Orphanet website
- **Lang:** ISO 639 code for language names
- **Name_en:** Name of the biobank in English
- **Name:** Name of the biobank in local language (language of the country where the registry is located)
- **LanguageofName :** Name of the local language
- **BiobankStatusFlagList count:** Number of biobank's status
- **BiobankStatusFlag Name:** Status of the biobank that can be:
 - o "on-line" which is an automatic status meaning that the biobank is published on Orphanet.
 - o and/or "IRDiRC" if the biobank is funded by a member of the IRDiRC consortium
 - o and/or "multicentric" is the biobank is regional or national and involves several sites within the country
- **URL, Protocol URL, PSOIEURL:** Website and descriptions of the biobank in English and other language as appropriate
- **Country:** ISO code and name of the country where the biobank is located
- **Geocoverage:** Geocoverage of the biobank. Can be "Regional", "National", "European" or "International (beyond Europe)"
- **BeginDate:** date on which the biobank started
- **ValidationDate:** The last update date of the biobank by the Orphanet team
- **PersonFunction_Name:** Name of the functions of stakeholders involved in the biobank. Stakeholders are the funding body financing the biobank and/or the persons in charge of the biobank. Thus, 5 functions can be found in the xml file: "Public funding body", "Private for-profit funding body", "Private non-for-profit funding body", "Manager of biobank" and "Contact person of biobank".
- **Person_LastName:** Name of the funding body of the biobank
- **Institution_Departement_Service_Lab_PatientOrganisation_Name:** Name of the department(s)/service(s) of the funding body hosting institution and/or manager



- hosting institution and/or contact person hosting institution
- **Hosting_Institution:** Name of the hosting institution(s) of the funding body and/or manager and/or contact person
- **Town:** Town(s) of the hosting institution(s)
- **Country ISO:** Country(ies) ISO of the hosting institution(s)
- **Country Name:** Country(ies) of the hosting institution(s)
- **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes) studied in the biobank
- **ORPHAcode:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.
- **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
- **GeneList count:** total number of genes involved/studied in the biobank
- **Gene id:** Internal and numerical identifier of the gene in the Orphanet database.
- **Gene OrphaNumber:** Unique and time-stable numerical identifier of the gene in the Orphanet database.
- **Symbol:** symbol of the concerned gene.

Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

```
<Biobank id="164732">  
<OrphaNumber=" 683491">
```

The internal identifier of the biobank is 164732. Its OrphaNumber is 683491.

```
<ExpertLink lang="en"> http://www.orpha.net/consor/cgi-bin/OC\_Exp.php?lng=en&Expert=683491 </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=683491.

```
<Name> RITA-Ireland Biobank </Name>  
<Name en> RITA-Ireland Biobank </Name en>  
<LanguageofName>en</LanguageOfName>
```

The name of this entry in local language and in English is the same as the language used is English: « RITA-Ireland Biobank ».

```
<BiobankStatusFlagList count="1">  
<BiobankStatusFlag id="35">  
<Name>on-line</Name>
```

The biobank has the automatic status “on-line”, meaning it is published on Orphanet.



```
<Url> https://www.hiqa.ie/areas-we-work/health-information/data-collections/irish-national-rare-kidney-disease-registry </Url>
```

```
<ProtocolUrl> </ProtocolUrl>
```

```
<PSOIEUrl> </PSOIEUrl>
```

The local URL of the biobank is <https://www.hiqa.ie/areas-we-work/health-information/data-collections/irish-national-rare-kidney-disease-registry>.

```
<Country id="101">
```

```
<Iso>IE</Iso>
```

```
<Name lang="en">IRELAND</Name>
```

```
</Country>
```

The country where the biobank is located is Ireland.

```
<Geocoverage id="1135">
```

```
<Name lang="en">National</Name>
```

This biobank geocoverage is National.

```
<BeginDate>2012-01-01 00:00:00.0</BeginDate>
```

```
<ValidationDate>2024-06-25 00:00:00.0</ValidationDate>
```

This biobank was created on January 2012. The last update of the biobank data by the Orphanet team was on the 25/06/2024.

```
<PersonActivityAssociation>
```

```
<PersonFunction id="52">
```

```
<Name lang="en">Private non-profit funding body</Name>
```

```
</PersonFunction>
```

```
<Person id="42909">
```

```
<LastName>MEATH FOUNDATION</LastName>
```

```
<Institution id="34578">
```

```
<Department Service Lab PatientOrganisationName>Meath Foundation </Department Service Lab PatientOrganisationName>
```

```
<Address id="16298">
```

```
<Hosting Institution>Tallaght University Hospital</Hosting Institution>
```

```
<Town>
```

```
<Name>DUBLIN</Name></Town>
```

```
<Country id="101">
```

```
<ISO>IE</ISO>
```

```
<Name lang="en">IRELAND</Name>
```

The biobank is funded by a “Private non-profit funding body” named “Meath Foundation” located in the service «Meath Foundation» of the hosting institution named «Tallaght University Hospital», which is located in the town of « Dublin » in the country « Ireland ».

```
<PersonActivityAssociation>
```

```
<PersonFunction id="50">
```

```
<Name lang="en">Public funding body</Name>
```

```

</PersonFunction>
<Person id="28704">
<LastName>SFI - SCIENCE FOUNDATION IRELAND</LastName>
<Institution id="26904">
<Department Service Lab PatientOrganisationName>Science
Foundation Ireland
</Department Service Lab PatientOrganisationName>
<Address id="17226">
<Hosting Institution>Science Foundation Ireland
</Hosting Institution>
<Town>
<Name>DUBLIN</Name></Town>
<Country id="101">
<ISO>IE</ISO>
<Name lang="en">IRELAND</Name>

```

The biobank is also funded by a “Public funding body” named “SFI - SCIENCE FOUNDATION IRELAND” located in the service «Science Foundation Ireland » of the hosting institution of the same name, which is located in the town of « Dublin » in the country « Ireland ».

```

<PersonFunction id="34">
<Name lang="en">Contact person of biobank/collection</Name>
</PersonFunction>
<Institution id="42073">
<Department Service Lab PatientOrganisationName> Trinity Health
Kidney Centre - Trinity College Dublin
</Department Service Lab PatientOrganisationName>
<Address id="10312">
<Hosting Institution> St James's Hospital </Hosting Institution>
<Town>
<Name>DUBLIN</Name></Town>
<Country id="101">
<ISO>IE</ISO>
<Name lang="en">IRELAND</Name>

```

The biobank is managed by a “Contact person of biobank/collection” located in the service « Trinity Health Kidney Centre - Trinity College Dublin » of the hosting institution named « St James's Hospital », which is located in the town of « Dublin » in the country « Ireland ».

```

<PersonFunction id="33">
<Name lang="en">Manager of biobank/collection</Name>
</PersonFunction>
<Institution id="34581">
<Department Service Lab PatientOrganisationName> rinity Health
Kidney Centre </Department Service Lab PatientOrganisationName>
<Address id="16298">
<Hosting Institution> Tallaght University Hospital
</Hosting Institution>
<Town>

```



```
<Name>DUBLIN</Name></Town>  
<Country id="101">  
<ISO>IE</ISO>  
<Name lang="en">IRELAND</Name>
```

The biobank is managed by a “Manager of biobank/collection” located in the service « rinity Health Kidney Centre » of the hosting institution named « Tallaght University Hospital », which is located in the town of « Dublin » in the country « Ireland ».

```
<DisorderList count="3">  
<Disorder id="747">  
<ORPHAcode>375</ORPHAcode>  
<Name lang="en">Anti-glomerular basement membrane  
disease</Name></Disorder>  
<Disorder id="10704">  
<ORPHAcode>52759</ORPHAcode>  
<Name lang="en">Vasculitis</Name></Disorder>  
<Disorder id="12170">  
<ORPHAcode>93126</ORPHAcode>  
<Name lang="en">Pauci-immune  
glomerulonephritis</Name></Disorder>  
</DisorderList>
```

The biobank operates for three diseases/group of diseases : Anti-glomerular basement membrane disease (ORPHAcode 375), Vasculitis (ORPHAcode 52759) and Pauci-immune glomerulonephritis (ORPHAcode 93126).

```
<GeneList count="0">  
No gene (0) is involved in this biobank
```

b. Network of biobanks

Description of the XML tags

- **NetworkList count:** total number of networks in the XML file
- **Network id:** Internal and numerical identifier of the network in the Orphanet database.
- **OrphaNumber:** Unique and time-stable numerical identifier of the network of biobanks in the Orphanet database.
- **ExpertLink:** stable URL pointing to the specific page of the network of biobanks on the Orphanet website
- **Lang:** ISO 639 code for language names
- **Name:** Name of the network in local language
- **Name_en:** Name of the network in English
- **LanguageOfName:** name of the local language
- **NetworkStatusFlagList count:** Number of network’s status
- **NetworkStatusFlag Name:** Status of the network that can be:



- “on-line” which is an automatic status meaning that the network is published on Orphanet.
 - and/or “IRDiRC” if the network is funded by a member of the IRDiRC consortium
- **URL, Protocol URL, PSOIEURL:** Website and descriptions of the network in English and other language as appropriate
- **Country:** ISO code and name of the country where the network is coordinated
- **Geocoverage:** Geographical coverage of the network. Can be “European” or “International (beyond Europe)”.
- **ValidationDate:** The last update date of the network by the Orphanet team
- **PersonFunction_Name:** Name of the functions of stakeholders involved in the network of biobanks. Stakeholders are the funding body financing the network and/or the coordinator of the network. Thus, 4 functions can be found in the xml file: “Public funding body”, “Private for-profit funding body”, “Private non-for-profit funding body” and “Coordinator of biobank network”.
- **Person_LastName:** Name of the funding body of the network
- **Institution_Departement_Service_Lab_PatientOrganisation_Name:** Name of the department(s)/service(s) of the funding body hosting institution and/or coordinator of the biobank network
- **Hosting_Institution:** Name of the hosting institution(s) of the funding body and/or coordinator
- **Town:** Town(s) of the hosting institution(s)
- **Country ISO:** Country(ies) ISO of the hosting institution(s)
- **Country Name:** Country(ies) of the hosting institution(s)
- **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes) studied in the network
- **ORPHAcodes:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.
- **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
- **GeneList count:** total number of genes involved/studied in the network
- **Gene id:** Internal and numerical identifier of the gene in the Orphanet database.
- **Gene OrphaNumber:** Unique and time-stable numerical identifier of the gene in the Orphanet database.
- **Symbol:** symbol of the concerned gene.
- **Biobanks count:** total number of biobanks involved in the network

[For each biobank involved in the network, you will find its whole dataset described in part 4.a\) of this document. Please note that at the moment, only one network of biobanks is registered in Orphanet.](#)



Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

`<NetworkList count="XXXX">`

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

`<Network id="79579">`

`<OrphaNumber=" 255828">`

The internal identifier of the network is 79579 and its OrphaNumber is 255828.

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

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

`<LanguageOfName>en</LanguageOfName>`

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

`<NetworkStatusFlagList count="1">`

`<NetworkStatusFlag id="35">`

`<Name lang="en">on-line</Name>`

The biobanks network only has the "on-line" status.

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

`<ProtocolUrl>`

`<PSOIEUrl>`

The network is described in one URL, which is <http://www.eurobiobank.org>.

`<Country id="148">`

`<ISO>MT</ISO>`

`<Name lang="en">MALTA</Name>`

The network is coordinated in Malta (ISO code: MT).

`<Geocoverage id="21">`

`<Name lang="en">International (beyond Europe)</Name>`

The network's geocoverage is "International (beyond Europe)", meaning that it is conducted in several countries, including country(ies) outside Europe.



<ValidationDate>2023-04-25 00:00:00.0</ValidationDate>

The last update date of the network was the 25/04/2023.

<PersonActivityAssociation>

<PersonFunction id="41">

<Name lang="en">Coordinator of biobank network</Name>

</PersonFunction>

<Institution id="28334">

<Department Service Lab PatientOrganisationName>Molecular
Genetics

Laboratory

</Department Service Lab PatientOrganisationName>

<Address id="18000">

<Hosting Institution>Universita ta Malta</Hosting Institution>

<Town>

<Name>MALTA</Name></Town>

<Country id="148">

<ISO>MT</ISO>

<Name lang="en">MALTA</Name>

The network is coordinated by a “Coordinator of of biobank network” located in the service « Molecular Genetics» of the hosting institution named « Universita ta Malta», which is located in « Malta».

<DisorderList count="33">

<Disorder id="10468">

<ORPHAcode>68329</ORPHAcode>

<Name lang="en">Rare maxillo-facial surgical
disease</Name></Disorder></DisorderList>

The network operates for 33 diseases/groups of diseases including Rare maxillo-facial surgical disease (ORPHAcode 68329). *The other 32 diseases/groups of diseases are not presented in this document.*

<GeneList count="0">

No gene (0) is involved in this network of biobank.

<Biobank count="12">

12 biobanks are part of this network.

For each biobank participating in this network, you will find the following information in the xml file:

<Biobank id="66882">

<OrphaNumber=" 204507">

The internal identifier of the biobank is 66882. Its OrphaNumber is 204507.

<ExpertLink lang="en"> http://www.orphadata.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=204507 </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=204507.

<Name> [MYOBANK-AFM: banque de tissus pour la recherche de personnes atteintes de maladies neuromusculaires ou rares \(Partenaire EuroBioBank\)](#) </Name>

<Name en> [MYOBANK-AFM: research dedicated human tissue bank from neuromuscular or rare diseases affected people \(EuroBioBank partner\)](#) </Name en>

<LanguageOfName>fr</LanguageOfName>

The name of this entry in local language (French) is: « MYOBANK-AFM: banque de tissus pour la recherche de personnes atteintes de maladies neuromusculaires ou rares (Partenaire EuroBioBank)» and in English is: « MYOBANK-AFM: research dedicated human tissue bank from neuromuscular or rare diseases affected people (EuroBioBank partner)».

<BiobankStatusFlagList count="1">

<BiobankStatusFlag id="35">

<Name>[on-line](#)</Name>

The biobank has the automatic status “on-line”, meaning it is published on Orphanet.

<Url> <http://www.institut-myologie.org/recherche/myobank/> </Url>

<ProtocolUrl> </ProtocolUrl>

<PSOIEUrl> </PSOIEUrl>

The local URL of the biobank is <http://www.institut-myologie.org/recherche/myobank/>.

<Country id="75">

<Iso>[FR](#)</Iso>

<Name lang="en">[FRANCE](#)</Name>

</Country>

The country where the biobank is located is France.

<Geocoverage id="1135">

<Name lang="en">[National](#)</Name>

This biobank geocoverage is National.

<BeginDate> </BeginDate>

<ValidationDate>[2024-07-16 00:00:00.0](#)</ValidationDate>

This biobank has no specified begindate. Its last update date by the Orphanet team was on the 16/07/2024.

<PersonActivityAssociation>

```

<PersonFunction id="33">
  <Name lang="en">Manager of biobank/collection</Name>
</PersonFunction>
<Institution id="35775">
  <Department Service Lab PatientOrganisationName>Centre de
  recherche en myologie
</Department Service Lab PatientOrganisationName>
  <Address id="28569">
  <Hosting Institution>Faculté de Santé - Site Pitié
  Salpêtrière</Hosting Institution>
  <Town>
  <Name>PARIS</Name></Town>
  <Country id="75">
  <ISO>FR</ISO>
  <Name lang="en">FRANCE</Name>

```

The biobank is managed by a “Manager of biobank/collection” located in the service «Centre de recherche en myologie» of the hosting institution named « Faculté de Santé – Site Pitié Salpêtrière», which is located in the town of « Paris » in the country « France ».

```

<DisorderList count="1">
  <Disorder id="10521">
  <ORPHAcode>68381</ORPHAcode>
  <Name lang="en">Neuromuscular
  disease</Name></Disorder></DisorderList>

```

The biobank operates for one diseases/group of diseases: Neuromuscular disease (ORPHAcode 68381).

```

<GeneList count="0">

```

No gene (0) is involved in this biobank.

5. 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
- **Institution id:** Internal and numerical identifier of the medical laboratory in the Orphanet database.
- **OrphaNumber:** Unique and time-stable numerical identifier of the medical laboratory in



the Orphanet database.

- **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

For each diagnostic test performed by the medical laboratory:

- o **Diagnosticid:** Internal and numerical identifier of the diagnostic test in the Orphanet database.
- o **ExpertLink:** stable URL pointing to the specific page of the diagnostic test on the Orphanet website
- o **Name:** Name of the diagnostic test
- o **Lang:** ISO 639 code for language names
- o **DgsTestPurposeList count:** Number of purpose(s) associated to the diagnostic test
- o **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

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

- **ValidationDate:** The last update date of the diagnostic test by the Orphanet team
- **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes) in the Xml file
- **ORPHAcodes:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.
- **ExpertLink:** stable URL pointing to the specific page of the given clinical entity on the Orphanet website
- **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
- **GeneList count:** total number of genes concerned by the diagnostic test
- **Gene id:** Internal and numerical identifier of the gene in the Orphanet database.
- **Gene OrphaNumber:** Unique and time-stable numerical identifier of the gene in the Orphanet database.
- **Symbol:** Symbol of the concerned gene
- **Accreditation_EqA List count:** Number of accreditation or EqA associated with this particular diagnostic test
- **QualityType_Name:** indicate if the quality data is an accreditation or an EqA

Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

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

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

```
<Institution id="54">
```

```
<OrphaNumber>7637</OrphaNumber>
```

```
<Department Service Lab PatientOrganisationAcronym/>
```

```
<Department Service Lab PatientOrganisationName>Unité de  
génétique
```

```
biologique</Department Service Lab PatientOrganisationName>
```

```
<UpperLevelOfAffiliation>Laboratoire de génétique moléculaire-  
Tripode 1er étage</UpperLevelOfAffiliation>
```

```
<Address id="426">
```

```
<Hosting Institution>CHU de Bordeaux - Groupe Hospitalier  
Pellegrin</Hosting Institution>
```

```
<Town><Name>BORDEAUX</Name></Town>
```

```
<Country id="75">
```

```
<Name lang="en">FRANCE</Name></Country>
```



The medical laboratory which, internal identifier is 7637, is located in the “Unité de génétique biologique” of the “Laboratoire de génétique moléculaire” within the hosting institution named “CHU de Bordeaux - Groupe Hospitalier Pellegrin” located at “BORDEAUX” in “FRANCE”.

```
<Accreditation EQAList count="24">  
  <Accreditation EQA id="35479">  
    <QualityType id="12538">  
      <Name lang="en">External Quality Assesment</Name>  
      <QualityStatus id="3">  
        <Name lang="en">Online/Validated</Name>
```

The medical laboratory has twenty-four quality data. The first one described here is an External Quality Assesment (EqA).

```
<ValidationDate>2025-07-08 00:00:00.0</ValidationDate>
```

The last update date on the medical laboratory’s information was the 08/07/2025.

```
< DiagnosticTest List count="37">
```

37 is the total number of diagnostic tests performed by this medical laboratory.

[For each diagnostic test performed by the medical laboratory, you will find the following information in the xml file:](#)

```
< DiagnosticTest id="76489">
```

The internal identifier of the diagnostic test is 76489.

```
<ExpertLink lang="en"> http://www.orpha.net/consor/cgi-bin/OC\_Exp.php?lng=en&Expert=240591 </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=240591.

```
<Name> Diagnostic du syndrome de l'X fragile \(gène FMR1\) </Name>  
<LanguageOfName>fr</LanguageOfName>
```

The French (fr) name of this entry is “Diagnostic du syndrome de l'X fragile (gène FMR1)”.

```
<DiagnosticTestTypeFlagList count="1">  
  <DiagnosticTestTypeFlag id="15486">  
    <Name lang="en">Diagnostic</Name>
```

The flag “Diagnostic” is automatically attributed to diagnostic tests in the Orphanet database.

```

<DgsTestPurposeList count="3">
  <DgsTestPurpose id="111315">
    <DgsTestPurposeLabel id="28727">
      <OrphaNumber>453356</OrphaNumber>
      <Name lang="en">Antenatal diagnosis</Name>
    </DgsTestPurpose id="243500">
      <DgsTestPurposeLabel id="28734">
        <OrphaNumber>453357</OrphaNumber>
        <Name lang="en">Pre-implantation diagnosis</Name>
      </DgsTestPurpose id="44046">
        <DgsTestPurposeLabel id="28741">
          <OrphaNumber>453358</OrphaNumber>
          <Name lang="en">Post-natal diagnosis</Name>
        </DgsTestPurpose id="44046">

```

The diagnostic test has three purposes for which it is performed: “Antenatal diagnosis”, “Pre-implantation diagnosis” and “Post-natal diagnosis”.

```

<DgsTestTechniqueList count="1">
<DgsTestTechnique id="243499">
  <DgsTestSpeciality id="243499">
    <DgsTestSpecialityLabel id="28468">
      <OrphaNumber>453319</OrphaNumber>
      <Name lang="en">Molecular genetics</Name>
    </DgsTestSpecialityObjective id="243499">
      <DgsTestSpecialityObjectiveLabel id="28475">
        <OrphaNumber>453320</OrphaNumber>
        <Name lang="en">Targeted mutation analysis</Name>
      </DgsTestSpecialityObjectiveTechnique id="243499">
        <DgsTestSpecialityObjectiveTechniqueLabel id="28657">
          <OrphaNumber>453346</OrphaNumber>
          <Name lang="en">PCR based techniques</Name>
        </DgsTestSpecialityObjectiveTechnique id="243499">

```

The diagnostic test specialty is Molecular genetics, which objective is targeted mutation analysis by PCR based techniques.

```

<ValidationDate>2025-02-03 00:00:00.0</ValidationDate>

```

The last update date of the genetic test’s information was the 03/02/2025.

```

<DisorderList count="2">
  <Disorder id="120">
    <OrphaCode>908</OrphaCode>
    <Name lang="en">Fragile X syndrome</Name>
  </Disorder id="12199">
    <OrphaCode>93256</OrphaCode>
    <Name lang="en">Fragile X-associated tremor/ataxia syndrome</Name>
  </Disorder id="12199">

```

The diagnostic test operates for “Fragile X syndrome” (ORPHAcode 908) and “Fragile X-associated tremor/ataxia syndrome” (ORPHAcode 93256).



```
<GeneList count="1">  
<Gene id="16063">  
<OrphaNumber>121878</OrphaNumber>  
<Symbol> FMR1 </Symbol>
```

The diagnostic test operates for FMR1 gene, which two identifiers in the Orphanet database are id 16063 and the time-stable OrphaNumber 121878.

```
<Accreditation EQAList count="3">
```

This specific diagnostic test has three EqA (not detailed here but in the xml file, the level of information displayed is the same as for the medical laboratory EqA).

6. Research projects

Orphanet provides information on ongoing (or terminated if it started after 2010) 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. National and multinational research projects are registered. National research projects take place in a single country, they can be multicentric if several collaborative sites within the country are involved. Multinational research projects are conducted in at least two different countries and can be “European” or “International (beyond Europe)”.

a. National research projects

Description of the XML tags

- **ResearchProjectList count:** total number of research projects in the XML file
- **ResearchProject id:** Internal and numerical identifier of the research project in the Orphanet database.
- **OrphaNumber:** Unique and time-stable numerical identifier of the research project in the Orphanet database.
- **ExpertLink:** stable URL pointing to the specific page of the research project on the Orphanet website
- **Name:** Title of the research project in local language (language of the country where the project is conducted)
- **Name_en:** Title of the research project in English
- **Lang:** ISO 639 code for language names
- **ResearchProjectType Flag count:** number of research project’s types attributed to the project. Types reflect the main objectives of the research project (described below)
- **ResearchProjectType Flag name:** Name of the research project’s types. Please note that



each research project is at least flagged “Research project” + one other type amongst the following 29 types:

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	

- **ResearchProjectStatus count:** Number of research project’s status attributed to the project (described below)
- **ResearchProjectStatus Name:** Status of the research project that can be:
 - o “on-line” which is an automatic status meaning that the national research project is published on Orphanet.
 - o and/or “Terminated” if the project’s funding has ended. If the status “Terminated” is not specified, the project is on-going.
 - o and/or “Not heritable” is used when it is impossible to determine with certainty which disorders from a group of disorders are concerned by the project and/or if the project is only interested in drawing general conclusions on the clinical entity(ies).
 - o and/or “IRDiRC” if the project is funded by a member of the IRDiRC consortium,
 - o and/or “multicentric” if the project is conducted in several sites.
- **URL, Protocol URL, PSOIEURL:** Website and descriptions of the research project in English and other language as appropriate
- **Country:** ISO code and name of the country where the research project is carried out
- **DrugTradeNameResearchProjectAssociationList count:** total number of drug tradenames involved/studied in the research project
- **DrugTradeNameResearchProjectAssociationList Name:** Name of the tradename
- **SubstanceResearchProjectAssociationList count:** total number of substances involved/studied in the research project
- **SubstanceResearchProjectAssociationList Name:** Name of the substance
- **BeginDate:** date on which the research project started
- **EndDate:** date on which the research project ended (if the research project has the status ‘Terminated’) or will end (if it does not have the status ‘Terminated’ and is therefore still ongoing)



- **ValidationDate:** The last update date of the research project by the Orphanet team
- **PersonFunction_Name:** Name of the functions of stakeholders involved in the research projects. Stakeholders are the funding body financing the project and/or the principal investigator of the research project. Thus, 5 functions can be found in the xml file: “Public funding body”, “Private for-profit funding body”, “Private non-for-profit funding body”, “Investigator of research project” and “Coordinator of research project” (for multicentric national research project).
- **Person_LastName:** Name of the funding body of the multinational research project
- **Institution_Departement_Service_Lab_PatientOrganisation_Name:** Name of the department(s)/service(s) of the funding body hosting institution and/or coordinator hosting institution and/or investigator hosting institution
- **Hosting_Institution:** Name of the hosting institution(s) of the funding body and/or investigator and/or coordinator
- **Town:** Town(s) of the hosting institution(s)
- **Country ISO:** Country(ies) ISO of the hosting institution(s)
- **Country Name:** Country(ies) of the hosting institution(s)
- **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes) studied in the research project
- **ORPHAcodes:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.
- **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
- **GeneList count:** total number of genes involved/studied in the research project
- **Gene id:** Internal and numerical identifier of the gene in the Orphanet database.
- **Gene OrphaNumber:** Unique and time-stable numerical identifier of the gene in the Orphanet database.
- **Symbol:** symbol of the concerned gene.



Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

```
<ResearchProjectList count="XXXX">
```

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

```
<ResearchProject id="174079">  
<OrphaNumber> 712847 </OrphaNumber>
```

The research project has two identifiers in the orphanet database: ID 174079 and OrphaNumber 712847.

```
<ExpertLink lang="en"> http://www.orpha.net/consor/cgi-bin/OC\_Exp.php?lng=en&Expert=712847 </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=712847.

```
<Name > PREDA : Développement d'une approche thérapeutique des bêta-hémoglobinopathies utilisant la reprogrammation post-transcriptionnelle de BCL11A </Name>
```

The name of this entry in local language (language of the country where the project is conducted) is « PREDA : Développement d'une approche thérapeutique des bêta-hémoglobinopathies utilisant la reprogrammation post-transcriptionnelle de BCL11A ».

```
<Name en> PREDA: Development of a therapeutic approach for beta-hemoglobinopathies using post-transcriptional reprogramming of BCL11A </Name en>
```

The English name of this entry is "PREDA: Development of a therapeutic approach for beta-hemoglobinopathies using post-transcriptional reprogramming of BCL11A".

```
<ResearchProjectTypeFlagList count="3">  
<ResearchProjectTypeFlag id="26949">  
<Name lang="en"> Biotechnology innovation </Name>  
<ResearchProjectTypeFlag id="14964">  
<Name lang="en"> Gene expression profile </Name>  
<ResearchProjectTypeFlag id="14946">  
<Name lang="en"> Research project </Name>
```

The research project has three research types representing the main goals of the project. All research projects automatically have the type "Research project" and this research project also has the types "Biotechnology innovation" and "Gene expression profile".



<ResearchProjectStatusFlagList count="3">

The research project has three statuses associated.

<ResearchProjectStatusFlag id="35">

<Name lang="en"> on-line </Name>

<ResearchProjectStatusFlag id="493">

<Name lang="en"> Not heritable </Name>

<ResearchProjectStatusFlag id="463">

<Name lang="en"> IRDiRC </Name>

All research projects have the automatic status “on-line” meaning that they are published on the Orphanet website. Moreover, the research project has the status “Not heritable”, meaning that it concerns a group of diseases but it is impossible to determine with certainty which disorders from this group are concerned by the project. The research project is funded by a member of the IRDiRC consortium. Finally, since the project is not under the status “Terminated”, it means that it is still on-going.

<Url/> <https://anr.fr//projet-ANR-23-CE18-0032> <Url/>

<ProtocolUrl>

[https://storage.orphanet.app/RCH/FR/55c53d1a40e57f5dfb0a63259ff4585](https://storage.orphanet.app/RCH/FR/55c53d1a40e57f5dfb0a63259ff4585bf9a08246.pdf)

[bf9a08246.pdf](https://storage.orphanet.app/RCH/FR/55c53d1a40e57f5dfb0a63259ff4585bf9a08246.pdf) <ProtocolUrl>

<PSOIEUrl/>

[https://storage.orphanet.app/RCH/FR/55c53d1a40e57f5dfb0a63259ff4585](https://storage.orphanet.app/RCH/FR/55c53d1a40e57f5dfb0a63259ff4585bf9a08246.pdf)

[bf9a08246.pdf](https://storage.orphanet.app/RCH/FR/55c53d1a40e57f5dfb0a63259ff4585bf9a08246.pdf) <PSOIEUrl/>

The research project has three different URLs describing the project.

<Country id="75">

<ISO>FR<ISO>

<Name lang="en">FRANCE</Name>

The research project is located in “FRANCE”, ISO code of France is FR.

<DrugTradeNameResearchProjectAssociationList count="0"> </DrugTradeNameResearchProjectAssociationList>

<SubstanceResearchProjectAssociationList count="0"> </SubstanceResearchProjectAssociationList>

No Drug tradename or substance is studied/involved in this research project.

<BeginDate>2023-12-01 00:00:00.0</BeginDate>

The research project started on the 01/12/2023.

<EndDate>2026-12-01 00:00:00.0</EndDate>

The research project is expected to be completed by the 01/12/2026.

<ValidationDate>2025-08-13 00:00:00.0</ValidationDate>



The last update date of the research project was the 13/08/2025.

```
<PersonFunction id="43">  
  <Name lang="en">Public funding body</Name>  
</PersonFunction>  
<Person id="25194">  
  <Firstname/>  
  <Lastname/>ANR - AGENCE NATIONALE DE LA RECHERCHE </Lasttname>  
</Person>  
<Institution id="24584">  
<Department Service Lab PatientOrganisationName>Agence  
Nationale de la  
Recherche</Department Service Lab PatientOrganisationName>  
<Address id="15861">  
<Hosting Institution>ANR - Agence Nationale de la  
Recherche</Hosting Institution>  
<Town>  
<Name>PARIS</Name></Town>  
<Country id="75">  
<ISO>FR</ISO>  
<Name lang="en">FRANCE</Name>
```

The research project is funded by a public funding body named “ANR – Agence Nationale de la Recherche” located in the Department and Hosting institution of the same name “ANR – Agence Nationale de la Recherche”, in the town of “PARIS” in the country “FRANCE”.

```
<PersonActivityAssociation>  
<PersonFunction id="5">  
  <Name lang="en">Investigator of research project</Name>  
</PersonFunction>  
<Institution id="38164">  
<Department Service Lab PatientOrganisationName>Laboratoire des  
Maladies Rénales  
Héréditaires</Department Service Lab PatientOrganisationName>  
<Address id="21245">  
<Hosting Institution>IMAGINE - Institut des Maladies  
Génétiques</Hosting Institution>  
<Town>  
<Name>PARIS</Name></Town>  
<Country id="75">  
<ISO>FR</ISO>  
<Name lang="en">FRANCE</Name>
```

The research project is linked to an investigator located in the service « Laboratoire des Maladies Rénales Héréditaires » in the hosting institution named « IMAGINE - Institut des Maladies Génétiques », which is located in the town of « PARIS » in the country « FRANCE ».



```
<DisorderList count="1">  
<ORPHAcode> 68364 </ORPHAcode>  
<Name lang="en"> Hemoglobinopathy </Name>
```

The research project operates for the group of diseases Hemoglobinopathy (ORPHAcode 68364).

```
<GeneList count="1">  
<Gene id="68364">  
<OrphaNumber> 139145 </OrphaNumber>  
<Symbol> BCL11A</Symbol>
```

One gene is studied in this research project. It has two identifiers in the orphanet database: ID 68364 and OrphaNumber 139145. The gene symbol is "BCL11A".

b. Multinational research projects

Description of the XML tags

- **NetworkList count:** total number of multinational research projects in the XML file
- **Network id:** Internal and numerical identifier of the multinational research project in the Orphanet database.
- **OrphaNumber:** Unique and time-stable numerical identifier of the multinational research project in the Orphanet database.
- **ExpertLink:** stable URL pointing to the specific page of the multinational research project on the Orphanet website
- **Name_en:** Title of the multinational research project in English
- **Lang:** ISO 639 code for language names
- **NetworkStatusFlag count:** Number of multinational research project's status attributed to the project (described below)
- **NetworkStatusFlag Name:** Status of the multinational research project that can be:
 - o "on-line" which is an automatic status meaning that the multinational research project is published on Orphanet.
 - o and/or "Terminated" if the project's funding has ended. If the status "Terminated" is not specified, the project is on-going.
 - o and/or "Not heritable" is used when it is impossible to determine with certainty which disorders from a group of disorders are concerned by the project and/or if the project is only interested in drawing general conclusions on the clinical entity(ies).
 - o and/or "IRDiRC" if the project is funded by a member of the IRDiRC consortium,
 - o and/or "Member of a European Reference Network (ERN)" if the project is an observational prospective study involving ERN members in at least two Member



States

- **URL, Protocol URL, PSOIEURL:** Website and descriptions of the multinational research project in English and other language as appropriate
- **Country:** ISO code and name of the country where the multinational research project is carried out
- **Geocoverage:** Geographical coverage of the multinational research project. Can be “European” or “International (beyond Europe)”.
- **DrugTradeNameNetworkAssociationList count:** Total number of drug tradenames involved/studied in the multinational research project
- **DrugTradeNameNetworkAssociationList Name:** Name of the tradename
- **SubstanceNetworkAssociationList count:** Total number of substances involved/studied in the research project
- **SubstanceNetworkAssociationList Name:** Name of the substance
- **ValidationDate:** The last update date of the multinational research project by the Orphanet team
- **PersonFunction_Name:** Name of the functions of stakeholders involved in the multinational research projects. Stakeholders are the funding body financing the project and/or the coordinator of the multinational research project. Thus, 4 functions can be found in the xml file: “Public funding body”, “Private for-profit funding body”, “Private non-for-profit funding body” and “Coordinator of research network”.
- **Person_LastName:** Name of the funding body of the multinational research project
- **Institution_Departement_Service_Lab_PatientOrganisation_Name:** Name of the department(s)/service(s) of the funding body hosting institution and/or coordinator hosting institution
- **Hosting_Institution:** Name of the hosting institution(s) of the funding body and/or coordinator
- **Town:** Town(s) of the hosting institution(s)
- **Country ISO:** Country(ies) ISO of the hosting institution(s)
- **Country Name:** Country(ies) of the hosting institution(s)
- **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes) studied in the multinational research project
- **ORPHAcodes:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.
- **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
- **GeneList count:** total number of genes involved/studied in the research project
- **Gene id:** Internal and numerical identifier of the gene in the Orphanet database.
- **Gene OrphaNumber:** Unique and time-stable numerical identifier of the gene in the Orphanet database.
- **Symbol:** symbol of the concerned gene.
- **Researchproject count:** total number of countries involved in the multinational research project

[For each country participating in this multinational research project:](#)

- o **Researchproject id:** Internal and numerical identifier of the given research



project/country in the Orphanet database.

- **OrphaNumber:** Unique and time-stable numerical identifier of the research project in the Orphanet database.
- **ResearchProjectType Flag count:** number of research types attributed to each country participating in the multinational project. Types reflect the main objectives of the research project (described in part 6.a “Description of the XML Tags” of this document)
- **ResearchProjectType Flag name:** Types of research conducted in each country participating. Please note that each research project is at least flagged “Research project” + one other type
- **Country:** ISO code and name of the countries participating in the multinational research project
- **BeginDate:** date on which the research project started in each participating country
- **EndDate:** date on which the research project will end/has ended in each participating country



Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

<NetworkList count="XXXX">

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

<Network id=" 168082">

<OrphaNumber="693497">

The multinational research project has two identifiers in the database: ID 168082 and OrphaNumber 693497.

<ExpertLink lang="en"> http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=693497 </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=693497.

<Name en> [IMPACT TB: Implementing proven community-based active case finding interventions in Vietnam and Nepal](#) </Name>

The name of the entry in English is “IMPACT TB: Implementing proven community-based active case finding interventions in Vietnam and Nepal”.

<NetworkStatusFlagList count="3">

The multinational research project has three statuses associated.

<NetworkStatusFlag id="35">

<Name lang="en"> [on-line](#) </Name>

<NetworkStatusFlag id="133">

<Name lang="en"> [Terminated](#) </Name> </NetworkStatusFlag>

<NetworkStatusFlag id="463">

<Name lang="en"> [IRDIRC](#) </Name></NetworkStatusFlag>

All multinational research projects have the automatic status “on-line” meaning that they are published on the Orphanet website. The multinational research project is funded by a member of the IRDiRC consortium and is terminated.

<Url/> <https://impacttbproject.org/> <Url/>

<ProtocolUrl><https://storage.orphenet.app/NETRCH/GB/5b82146c2d5cdf70aa44c60abc0fef27c0ebee7.pdf></ProtocolUrl>

<PSOIEUrl/><https://storage.orphenet.app/NETRCH/GB/5b82146c2d5cdf70aa44c60abc0fef27c0ebee7.pdf></PSOIEUrl/>

The multinational research project abstract is described in three different URLs.

```
<Country id="77">
<ISO>GB</ISO>
<Name lang="en">UNITED KINGDOM</Name>
```

The multinational research project is coordinated in United-Kingdom (ISO code: GB).

```
<Geocoverage id="21">
<Name lang="en">International (beyond Europe)</Name>
```

The multinational research project geocoverage is “International (beyond Europe)”, meaning that it is conducted in several countries, including countries beyond Europe.

```
<DrugTradeNameResearchProjectAssociationList count="0"> </DrugTradeNameResearchProjectAssociationList>
<SubstanceNetworkAssociationList count="0">
</SubstanceNetworkAssociationList>
```

No Drug tradename or Substance is studied/involved in this multinational research project.

```
<ValidationDate>2025-01-24 00:00:00.0</ValidationDate>
```

The last update date of the research project was the 24/01/2025.

```
<PersonActivityAssociation>
<PersonFunction id="58">
<Name lang="en">Public funding body</Name>
</PersonFunction>
<Person id="46881">
<Firstname/>
<Lastname/>EUROPEAN COMMISSION - HORIZON 2020 </Lasttname>
</Person>
<Institution id="24448">
<Department Service Lab PatientOrganisationName>DG Research -
Directorate General for
Research</Department Service Lab PatientOrganisationName>
<Address id="15783">
<Hosting Institution> European Commission </Hosting Institution>
<Town>
<Name>Brussels</Name></Town>
<Country id="22">
<ISO>BE</ISO>
<Name lang="en">BELGIUM</Name>
```

The research project is funded by the European Commission through the programme Horizon 2020. The European Commission is a public funding body. The European Commission department/service involved is the DG Research – Directorate General for Research, located in Brussels, Belgium.



```
<PersonActivityAssociation>
<PersonFunction id="4">
<Name lang="en">Coordinator of research network</Name>
</PersonFunction>
<Institution id="46118">
<Department Service Lab PatientOrganisationName>Liverpool School of
Tropical Medicine</Department Service Lab PatientOrganisationName>
<Address id="15783">
<Hosting Institution> Liverpool School of Tropical
Medicine</Hosting Institution>
<Town>
<Name>LIVERPOOL</Name></Town>
<Country id="77">
<ISO>GB</ISO>
<Name lang="en">UNITED KINGDOM</Name>
```

The multinational research project is coordinated by a coordinator of research network, located at the Liverpool School of Tropical Medicine in Liverpool, United Kingdom (ISO code GB).

```
<DisorderList count="1">
<Disorder id="863">
<ORPHAcode>3389</ORPHAcode>
<Name lang="en"> Tuberculosis </Name></Disorder>
```

The multinational research project operates Tuberculosis (ORPHAcode 3389).

```
<GeneList count="0">
```

No gene (0) is studied in the project.

```
<Subtype>Rch<Subtype>
<Researchproject count="3">
```

Three countries (local “research projects”) are participating in this multinational research project.

[For each country participating in this multinational research project, you will find the following information in the xml file:](#)

```
<ResearchProject id="168060">
<OrphaNumber>693475</OrphaNumber>
```

The first country/research project participating has two identifiers in the orphanet database: ID 168060 and OrphaNumber 693475.

```
<ResearchProjectTypeFlagList count="4">
<ResearchProjectTypeFlag id="15060">
<Name lang="en"> Public health study (excluding health
economics) </Name>
```



```
<ResearchProjectTypeFlag id="15054">  
<Name lang="en"> Health economics study</Name>  
<ResearchProjectTypeFlag id="15036">  
<Name lang="en"> Epidemiological study </Name>  
<ResearchProjectTypeFlag id="14946">  
<Name lang="en">Research project</Name>  
</ResearchProjectTypeFlag>
```

This participating country has 4 research types: the mandatory “Research project” type along with “Public health study (excluding health economics)”, “Health economics study” and “Epidemiological study”.

```
<Country id="191">  
<ISO>SE</ISO>  
<Name lang="en">SWE DEN</Name>
```

The first country participating to the multinational research project is Sweden.

```
<BeginDate>2017-01-01 00:00:00.0</BeginDate>  
<EndDate>2019-12-31 00:00:00.0</EndDate>
```

In Sweden, the study started on the 01/01/2017 and ended on the 31/12/2019.

7. 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. National clinical trials take place in a single country, they can be multicentric if several collaborative sites within the country are involved. Multinational clinical trials are conducted in at least two different countries and can be “European” or “International (beyond Europe)”. The trials registered in Orphanet can be ongoing, recruiting, or terminated.

a. National clinical trial

Description of the XML tags

- **ClinicalTrialList count:** total number of national clinical trials in the XML file
- **ClinicalTrial id:** Internal and numerical identifier of the clinical trial in the Orphanet database.
- **OrphaNumber:** Unique and time-stable numerical identifier of the clinical trial in the Orphanet database.
- **ExpertLink:** stable URL pointing to the specific page of the clinical trial on the Orphanet website
- **Name:** Title of the national clinical trial in local language (language of the country where



the trial is conducted)

- **Name_en:** Title of the national clinical trial in English
- **Lang:** ISO 639 code for language names
- **ClinicalTrialType Flag:** Please note that each clinical trial is at least flagged “Clinical Trial” + one other type amongst the following:

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 national clinical trial that can be:
 - o “on-line” which is an automatic status meaning that the national clinical trial is published on Orphanet.
 - o and/or “Terminated” if the trial is completed. If the status “Terminated” is not specified, it means that the trial is still on-going.
 - o and/or “Not heritable” is used when it is impossible to determine with certainty which disorders from a group of disorders are concerned by the trial and/or if the trial is only interested in drawing general conclusions on the clinical entity(ies).
 - o and/or “IRDiRC” if the trial is sponsored by a member of the IRDiRC consortium,
 - o and/or “multicentric” if the trial is conducted in several sites in the country.
- **URL, Protocol URL, PSOIEURL:** Website and descriptions of the clinical trial in English and other language as appropriate
- **Country:** ISO code and name of the country where the clinical trial is conducted
- **Geocoverage:** Geographical coverage of the clinical trial. Can only be “National” for national clinical trials
- **Phase:** phase of the clinical trial. Can be:

I
I - II
II
II - III
III
IV
bioequivalence

- **Recruitment:** indicates if the clinical trial is currently recruiting or not

Drugs evaluated by the clinical study can be tradenames and/or substances depending on what is indicated in the study protocol. The xml file contains the following information:

- **DrugTradeNameClinicalTrialAssociationList count:** Total number of drug tradenames evaluated by the national clinical trial, as indicated in the study protocol
- **DrugTradeNameClinicalTrialAssociationList Name:** Name of the tradename, as indicated



- in the study protocol
- **SubstanceList Name:** Name of the substance associated with the tradename if a tradename is evaluated by the clinical trial
 - **SubstanceClinicalTrialAssociationList count:** Total number of substances evaluated by the national clinical trial, as indicated in the study protocol
 - **SubstanceClinicalTrialAssociationList Name:** Name of the substance as indicated in the study protocol
-
- **BeginDate:** date on which the national clinical trial started
 - **EndDate:** date on which the national clinical trial was completed (if its status 'Terminated') or is expected to be completed (if it does not have the status 'Terminated' and is therefore still ongoing)
 - **ValidationDate:** The last update date of the clinical trial by the Orphanet team
 - **Detail:** EudraCT number which, is a unique identifier of the trial or, if the EudraCT number is not available, another unique identifier of official trial database is indicated
 - **PersonList Count:** number of legal entities linked to the clinical trial
 - **Person_LastName:** Name of the legal entity of the national clinical trial
 - **PersonFunction:** Name of the functions of stakeholders involved in the national clinical trial. Stakeholders are the sponsor and funding body financing the project and if relevant, the investigators of the clinical trial (can have several investigators if the trial is multicentric). Thus, 6 functions can be found in the xml file: "Sponsor of clinical trial", "Public funding body", "Private for-profit funding body", "Private non-for-profit funding body", "Principal investigator of clinical trial" and "Investigator of clinical trial".
 - **Institution_Departement_Service_Lab_PatientOrganisation_Name:** Name of the department(s)/service(s) of the sponsor and funding body hosting institution and investigator hosting institution
 - **Hosting_Institution:** Name of the hosting institution(s) of the sponsor, funding body and investigator
 - **Town:** Town(s) of the hosting institution(s)
 - **Country ISO:** Country(ies) ISO of the hosting institution(s)
 - **Country Name:** Country(ies) of the hosting institution(s)
 - **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes) in the Xml file targeted by the clinical trial
 - **ORPHAcode:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.
 - **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
 - **GeneList count:** total number of genes involved in the clinical trial
 - **Gene id:** Internal and numerical identifier of the gene in the Orphanet database.
 - **Gene OrphaNumber:** Unique and time-stable numerical identifier of the gene in the Orphanet database.
 - **Symbol:** symbol of the concerned gene.



Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

<ClinicalTrial List count="XXXX">

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

<ClinicalTrial id=" 167846 ">

<OrphaNumber=" 692873 ">

The national clinical trial has two identifiers in the database: ID 167846 and OrphaNumber 692873.

<ExpertLink lang="en"> http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=692873 </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=692873.

<Name> *E-MERGE: Comparaison d'un traitement par mépolizumab à la stratégie thérapeutique conventionnelle pour l'induction de la rémission au cours de la granulomatose éosinophilique avec polyangéite (Syndrome de Churg-Strauss). Etude prospective, en double-aveugle, contrôlée, randomisée contre la stratégie thérapeutique conventionnelle - FR* </Name>

The name of this entry in local language (language of the country where the trial is conducted) is: "E-MERGE: Comparaison d'un traitement par mépolizumab à la stratégie thérapeutique conventionnelle pour l'induction de la rémission au cours de la granulomatose éosinophilique avec polyangéite (Syndrome de Churg-Strauss). Etude prospective, en double-aveugle, contrôlée, randomisée contre la stratégie thérapeutique conventionnelle - FR".

<Name en> *E-MERGE: Evaluation of MEpolizumab-based Regimen Compared to Conventional Therapeutic Strategy for Remission Induction in Patients with Eosinophilic Granulomatosis with Polyangiitis. Prospective, Randomized, Controlled, Double-blind Study - FR* </Name>

The English name of this entry is: "E-MERGE: Evaluation of MEpolizumab-based Regimen Compared to Conventional Therapeutic Strategy for Remission Induction in Patients with Eosinophilic Granulomatosis with Polyangiitis. Prospective, Randomized, Controlled, Double-blind Study - FR".

<ClinicalTrialTypeFlagList count="2">

The clinical trial has two types.

<ClinicalTrialTypeFlag id="15066">



```
<Name lang="en"> Clinical trial </Name> </ClinicalTrialTypeFlag>  
<ClinicalTrialTypeFlag id="15072">  
<Name lang="en"> Drug clinical trial </Name> </ClinicalTrialTypeFlag>
```

The national clinical trial has the mandatory type “clinical trial” and is a “drug clinical trial”.

```
<ClinicalTrialStatusFlagList count="3">
```

The clinical trial has three statuses associated.

```
<ClinicalTrialStatusFlag id="35">  
<Name lang="en"> on-line </Name> </ClinicalTrialStatusFlag>  
<ClinicalTrialStatusFlag id="37">  
<Name lang="en"> multicentric </Name> </ClinicalTrialStatusFlag>  
<ClinicalTrialStatusFlag id="463">  
<Name lang="en"> IRDiRC </Name> </ClinicalTrialStatusFlag>
```

All clinical trials have the status “on-line” meaning that they are published on the Orphanet website. The national clinical trial is multicentric, thus conducted in several sites within the country and is sponsored by a member of the IRDiRC consortium.

```
<Url/> https://clinicaltrials.gov/study/NCT05030155  
<ProtocolUrl/>  
<PSOIEUrl/> https://euclinicaltrials.eu/search-for-clinical-trials/?lang=en&EUCT=2024-513653-75-00
```

The clinical trial has two descriptions available at <https://clinicaltrials.gov/study/NCT05030155> and <https://euclinicaltrials.eu/search-for-clinical-trials/?lang=en&EUCT=2024-513653-75-00>.

```
<Country id="75">  
<ISO>FR</ISO>  
<Name lang="en">FRANCE</Name>  
</Country>
```

The clinical trial is located in “FRANCE (FR)”.

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

The clinical trial geocoverage is National as that is the case for each national clinical trial in the XML file.

```
<Phase id="72">  
<Name lang="en">III</Name>
```

This clinical trial is a phase III trial.

```
<Recruitment id="102">  
<Name lang="en">no</Name>
```

This clinical trial is not recruiting.



<DrugTradeNameClinicalTrialAssociationList count="0"> </DrugTradeNameResearchProjectAssociationList>

<SubstanceClinicalTrialAssociationList count="1">

<SubstanceResearchProjectAssociationList id=" 850819">

<Substance id="1135">

<Name lang="en"> Mepolizumab</Name>

No drug under its tradename is evaluated by the clinical study. The clinical trial evaluates a substance named “Mepolizumab” (identifier is 1135) as indicated in the study protocol.

<BeginDate>2022-05-30 00:00:00.0</BeginDate>

The clinical trial started on the 30/05/2022.

<EndDate>2026-01-30 00:00:00.0</EndDate>

The clinical trial is expected to be completed by the 30/01/2026.

<ValidationDate>2025-01-17 00:00:00.0</ValidationDate>

The clinical trial was last updated on the 17/01/2025.

<Detail>2025-521340-37-00</Detail>

The EUCT number of this clinical trial is “2024-513653-75-00”.

<PersonActivityAssociation>

<PersonFunction id="2">

<Name lang="en">Principal investigator of clinical trial</Name>

</PersonFunction>

<Institution id="6627">

<Department Service Lab PatientOrganisationName>Service de Médecine

Interne</Department Service Lab PatientOrganisationName>

<Address id="7706">

<Hosting Institution>AP-HP. Centre - Université Paris Cité - Hôpital Cochin - Port-Royal</Hosting Institution>

<Town>

<Name>PARIS</Name></Town>

<Country id="75">

<ISO>FR</ISO>

<Name lang="en">FRANCE</Name>

The clinical trial is conducted by a “Principal investigator of clinical trial” located in the service « Service de Médecine Interne» in the hosting institution named « AP-HP. Centre - Université Paris Cité - Hôpital Cochin - Port-Royal », which is located in the town of « PARIS » in the country « FRANCE».



```
<PersonActivityAssociation>
<PersonFunction id="3">
  <Name lang="en">Investigator of clinical trial</Name>
</PersonFunction>
<Institution id="45139">
<Department Service Lab PatientOrganisationName>Service de
Médecine
Interne</Department Service Lab PatientOrganisationName>
<Address id="11638">
<Hosting Institution>CH de Perpignan</Hosting Institution>
<Town>
<Name>PERPIGNAN</Name></Town>
<Country id="75">
<ISO>FR</ISO>
<Name lang="en">FRANCE</Name>
```

As the clinical trial is multicentric, it is conducted by another “Investigator of clinical trial” located in the service « Service de Médecine Interne» in the hosting institution named «CH de Perpignan », which is located in the town of « PERPIGNAN» the country « FRANCE».

```
<PersonFunction id="10">
  <Name lang="en">Sponsor of clinical trial </Name>
</PersonFunction>
<Person id="11690">
  <Firstname/>
  <Lastname/>AP-HP (DRCD)</Lasttname>
</Person>
<Institution id="14875">
<Department Service Lab PatientOrganisationName>Délégation à la
Recherche Clinique et à l'Innovation de l'AP-
HP</Department Service Lab PatientOrganisationName>
<Address id="81">
<Hosting Institution>AP-HP.Nord - Université Paris Cité -
Hôpital Saint-Louis</Hosting Institution>
<Town>
<Name>PARIS</Name></Town>
<Country id="75">
<ISO>FR</ISO>
<Name lang="en">FRANCE</Name>
```

The clinical trial is sponsored by a legal entity named “AP-HP (DRCD)” located in the Department/Service named “Délégation à la Recherche Clinique et à l'Innovation de l'AP-HP”, in the Hosting Institution “AP-HP.Nord - Université Paris Cité - Hôpital Saint-Louis”, which is located in the town of « PARIS » in the country « FRANCE»

```
<DisorderList count="1">
<Disorder id="745">
<ORPHAcodes>183</ORPHAcodes>
<Name lang="en"> Eosinophilic granulomatosis with polyangiitis</Name>
```



The clinical trial is focused on Eosinophilic granulomatosis with polyangiitis (ORPHAcode 183).

<GeneList count="0">

No gene is involved in this clinical trial.

b. Multinational clinical trials

Description of the XML tags

- **NetworkList count:** total number of multinational clinical trials in the XML file
- **Network id:** Internal and numerical identifier of the multinational clinical trial in the Orphanet database.
- **OrphaNumber:** Unique and time-stable numerical identifier of the multinational clinical trial in the Orphanet database.
- **ExpertLink:** stable URL pointing to the specific page of the multinational clinical trial on the Orphanet website
- **Name_en:** Title of the multinational clinical trial in English
- **Lang:** ISO 639 code for language names
- **NetworkStatusFlagList count:** Number of multinational clinical trial's status attributed to the project (described below)
- **NetworkStatusFlagList Name:** Status of the multinational clinical trial that can be:
 - o "on-line" which is an automatic status meaning that the multinational clinical trial is published on Orphanet.
 - o and/or "Terminated" if the trial is completed. If the status "Terminated" is not specified, it means that the trial is still on-going.
 - o and/or "Not heritable" is used when it is impossible to determine with certainty which disorders from a group of disorders are concerned by the trial and/or if the trial is only interested in drawing general conclusions on the clinical entity(ies).
 - o and/or "IRDiRC" if the trial is sponsored by a member of the IRDiRC consortium,
 - o and/or "Member of a European Reference Network (ERN)" if the trial is involving ERN members in at least two Member States
- **URL, Protocol URL, PSOIEURL:** Website and descriptions of the multinational clinical trial in English and other language as appropriate
- **Country:** ISO code and name of the country where the multinational clinical trial is coordinated
- **Geocoverage:** Geographical coverage of the multinational clinical trial. Can be "European" or "International (beyond Europe)".



- **Phase:** phase of the multinational clinical trial. Can be:

I
I - II
II
II - III
III
IV
bioequivalence

Drugs evaluated by the clinical study can be tradenames and/or substances depending on what is indicated in the study protocol. The xml file contains the following information:

- **DrugTradeNameNetworkAssociationList count:** Total number of drug tradenames evaluated by the multinational clinical trial, as indicated in the study protocol
- **DrugTradeNameNetworkAssociationList Name:** Name of the tradename, as indicated in the study protocol
- **SubstanceList Name:** Name of the substance associated with the tradename if a tradename is evaluated by the clinical trial
- **SubstanceNetworkAssociationList count:** Total number of substances evaluated by the multinational clinical trial, as indicated in the study protocol
- **SubstanceNetworkAssociationList Name:** Name of the substance as indicated in the study protocol

- **ValidationDate:** The last update date of the multinational clinical trial by the Orphanet team
- **PersonList Count:** number of legal entities linked to the multinational clinical trial
- **Person_LastName:** Name of the legal entity of the multinational clinical trial
- **PersonFunction_Name:** Name of the functions of stakeholders involved in the multinational clinical trial. Stakeholders are the sponsor and funding body financing the project and if relevant, the coordinator of the multinational clinical trial. Thus, 5 functions can be found in the xml file: “Sponsor of multinational clinical trial”, “Public funding body”, “Private for-profit funding body”, “Private non-for-profit funding body” and “Coordinator of multinational clinical trial”.
- **Institution_Departement_Service_Lab_PatientOrganisation_Name:** Name of the department(s)/service(s) of the sponsor hosting institution, funding body hosting institution and coordinator hosting institution
- **Hosting_Institution:** Name of the hosting institution(s) of the sponsor, funding body and/or coordinator
- **Town:** Town(s) of the hosting institution(s)
- **Country ISO:** Country(ies) ISO of the hosting institution(s)
- **Country Name:** Country(ies) of the hosting institution(s)
- **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes) studied in the multinational clinical trial



- **ORPHAcode:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.
- **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
- **GeneList count:** total number of genes involved/studied in the multinational clinical trial
- **Gene id:** Internal and numerical identifier of the gene in the Orphanet database.
- **Gene OrphaNumber:** Unique and time-stable numerical identifier of the gene in the Orphanet database.
- **Symbol:** symbol of the concerned gene.
- **ClinicalTrial count:** total number of countries participating in the multinational clinical trial

[For each country participating in this multinational clinical trial:](#)

- o **Clinicaltrial id:** Internal and numerical identifier of the given clinical trial/site in the Orphanet database.
- o **OrphaNumber:** Unique and time-stable numerical identifier of the clinical trial in the Orphanet database.
- o **ClinicalTrialType Flag count:** number of clinical trial types attributed to each country participating in the multinational trial and reflecting the overall multinational trial types.
- o **ClinicalTrialType Flag name:** Please note that each clinical trial is at least flagged “Clinical Trial” + one other type amongst the following:

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

- o **Country:** ISO code and name of the country participating in the multinational clinical trial
- o **Recruitment:** indicates if the clinical trial is currently recruiting or not
- o **BeginDate:** date on which the clinical trial started in each participating country
- o **EndDate:** date on which the clinical trial is expected to be completed/ has been completed in each participating country



Example

Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

<NetworkList count="XXXX">

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

<Network id=" 174069">
<OrphaNumber=" 712829">

The multinational clinical trial has two identifiers in the database: ID 174069 and OrphaNumber 712829.

<ExpertLink lang="en"> http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=712829</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=712829.

<Name en> [RAGNAR: Phase II Study Of Tarlatamab Alone Or In Combination With Chemotherapy In Advanced Neuroendocrine Carcinomas Of The Digestive System Or Unknown Primary Origin](#)</Name>

The name of the entry in English is “RAGNAR: Phase II Study Of Tarlatamab Alone Or In Combination With Chemotherapy In Advanced Neuroendocrine Carcinomas Of The Digestive System Or Unknown Primary Origin”.

<NetworkStatusFlagList count="1">
<NetworkStatusFlag id="35">
<Name lang="en">on-line</Name>

As for all multinational clinical trials in the xml file, there is the automatic status “on-line” meaning that they are published on the Orphanet website. Moreover, as this multinational clinical trial does not have the status “Terminated”, it means that it is still on-going.

<Url/> <https://clinicaltrials.gov/study/NCT07061080></Url/>
<ProtocolUrl>
<PSOIEUrl/> <https://euclinicaltrials.eu/search-for-clinical-trials/?lang=en&EUCT=2024-514327-42-00></PSOIEUrl/>

The multinational clinical trial is described in two different URLs.

<Country id="68">
<ISO>ES</ISO>
<Name lang="en">SPAIN</Name>

The multinational clinical trial is coordinated in Spain (ISO code: ES).



```
<Geocoverage id="15">  
<Name lang="en"> European</Name>
```

The multinational clinical trial geocoverage is “European”, meaning that it is conducted in several countries across Europe.

```
<Phase id="60">  
<Name lang="en"> II </Name>
```

The multinational clinical trial is a phase II study.

```
<DrugTradeNameNetworkAssociationList count="0">  
<SubstanceNetworkAssociationList count="1">  
  <SubstanceNetworkAssociation id="885814">  
    <Substance id="5684">  
      <Name lang="en">Taratamab</Name>
```

No drug under its tradename is evaluated by this multinational clinical study. The clinical trial evaluates a substance named “Taratamab” (identifier is 5684) as indicated in the study protocol

```
<ValidationDate>2025-09-01 00:00:00.0</ValidationDate>
```

The last update date of this multinational clinical trial was the 01/09/2025.

```
<PersonFunction id="57">  
  <Name lang="en">Sponsor of multinational clinical trial</Name>  
</PersonFunction>  
<Person id="67664">  
  <Firstname/>  
  <Lastname/>GRUPO ESPAÑOL DE TUMORES NEUROENDOCRINOS</Lasttname>  
</Person>  
<Institution id="47073">  
<Department Service Lab PatientOrganisationName>GRUPO ESPAÑOL  
DE TUMORES NEUROENDOCRINOS</Department Service Lab PatientOrganisationName  
>  
<Address id="28918">  
<Hosting Institution>GRUPO ESPAÑOL DE TUMORES  
NEUROENDOCRINOS</Hosting Institution>  
<Town>  
<Name>BARCELONA</Name></Town>  
<Country id="68">  
<ISO>ES</ISO>  
<Name lang="en">SPAIN</Name>
```

The multinational clinical trial is sponsored by a legal entity named “Grupo Español De Tumores Neuroendocrinos” located in the Department/Service and Hosting Institution



of the same name in the town of “Barcelona” in the country “Spain”.

```
<PersonActivityAssociation>
<PersonFunction id="55">
  <Name lang="en">Coordinator of multinational clinical
  trial</Name>
</PersonFunction>
<Institution id="38624">
<Department Service Lab PatientOrganisationName>Tumores
  Gastrointestinales y
  Endocrinos</Department Service Lab PatientOrganisationName>
<Address id="22055">
<Hosting Institution>VHIO - Vall d'Hebron Institute of
  Oncology</Hosting Institution>
<Town>
<Name>BARCELONA</Name></Town>
<Country id="68">
<ISO>ES</ISO>
<Name lang="en">SPAIN</Name>
```

The multinational clinical trial is coordinated by a “Coordinator of multinational clinical trial” located in the service « Tumores Gastrointestinales y Endocrinos» of the hosting institution named « VHIO - Vall d'Hebron Institute of Oncology», which is located in the town of « Barcelona » in the country « Spain».

```
<DisorderList count="1">
  <Disorder id=" 14664">
  <ORPHAcode> 100092 </ORPHAcode>
  <Name lang="en"> Gastroenteropancreatic neuroendocrine neoplasm
  </Name></Disorder>
```

The multinational clinical trial operates for the disease “Gastroenteropancreatic neuroendocrine neoplasm” (ORPHAcode 100092).

```
<GeneList count="0">
```

No gene (0) is studied in the trial.

```
<Subtype>Rc1<Subtype>
<Researchproject count="2">
```

Two countries are participating in this multinational clinical trial.

[For each country participating in this multinational clinical trial, you will find the following information in the xml file:](#)

```
<ClinicalTrial id=" 174070">
<OrphaNumber> 712830</OrphaNumber>
```

The first country/clinical trial participating has two identifiers in the Orphanet



database: ID 174070 and OrphaNumber 712830.

```
< ClinicalTrialTypeFlagList count="2">  
  < ClinicalTrialTypeFlag id="15066">  
    <Name lang="en"> Clinical Trial </Name>  
  < ClinicalTrialTypeFlag id="15072">  
    <Name lang="en">Drug clinical trial </Name>  
  </ ClinicalTrialTypeFlag>
```

This participating country has two types: the mandatory “Clinical trial” type along with “Drug clinical trial”. All participating countries have the same types, reflecting the overall multinational clinical trial purpose.

```
<Country id="75">  
  <ISO>FR</ISO>  
  <Name lang="en">FRANCE</Name>
```

The first country participating to this multinational clinical trial is France (ISO code: FR).

```
<Recruitment id="102">  
  <Name lang="en">no</Name>
```

The trial is no longer recruiting.

```
<BeginDate> 2025-08-01 00:00:00.0</BeginDate>  
<EndDate> 2028-12-31 00:00:00.0</EndDate>
```

In France, the study started on the 01/08/2025 and is expected to be completed by the 31/12/2028.

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 141/2000](#) and in the US by the [Orphan Drug Act](#).

Description of the XML tags



- **DrugRegulatoryStatusList 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
- **DrugTradeNameRegulatoryStatusAssociationList count:** total number of drug tradenames concerned by the regulatory status
- **DrugTradeName_Name:** Name of the tradename
- **SubstanceList count:** total number of substance associated to the drug tradename.
- **ProductType:** Can be :

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

- **ProductionType:** Can be “Synthetic / extractive chemistry”, “Biotechnology”
- **Substance_Code:** name of the substance
- **SubstanceChemicalName:** chemical name of the substance
- **SubstanceRegulatoryStatusAssociationList count:** total number of substance concerned by the regulatory status
- **Creationdateof MA_OD:** creation date of the marketing authorisation or the orphan designation
- **ValidationDate:** The last update date of the regulatory status by the Orphanet team
- **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 clinical entities (disorders, group of disorders or subtypes) in the Xml file
- **ORPHAcodes:** unique numerical identifier of the clinical entity (disorders, groups of disorders or subtypes). More information on <http://www.orphacode.org>.
- **Disorder_Name:** preferred name of a given clinical entity. The most generally accepted name according to the literature, and as adopted by the medical community
- **Abstract:** Indication of the regulatory status

Example



Note: The text in red represents information not to consider in the XML code; the text in blue represents information to be considered in the XML code for analysis; and the text in black provides an interpretation of the XML code.

<DrugRegulatoryStatusList count="XXXX">

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

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

The name of the entry in English is “Marketing authorization without orphan designation”.

<PSOIUrl>

<https://www.ema.europa.eu/en/medicines/human/EPAR/orkambi></PSOIUrl>

More information is available using the URL:
<https://www.ema.europa.eu/en/medicines/human/EPAR/orkambi>.

<AdminZone id="1159">

<Name lang="en">Europe</Name>

The administrative zone of the regulatory status is “Europe”.

<ATCCode>R07AX30</ATCCode>

The ATC Code is “R07AX30”.

<DrugTradeNameDrugRegulatoryStatusAssociationList count="1">

<DrugTradeNameDrugRegulatoryStatusAssociation id="451432">

<DrugTradeName id="2963">

<Name lang="en">ORKAMBI</Name>

<SubstanceList count="1">

<Substance id="2739">

<Code/>

<ChemicalName/>

<ActiveIngredient/>

<ProductType id="121">

<Name lang="en">Ingredient/substance</Name>

</ProductType>

<ProductionType id="199">

<Name lang="en">Synthetic / extractive chemistry</Name>

</ProductionType>

<Name lang="en">Lumacaftor / Ivacaftor</Name>

The regulatory status operates for ORKAMBI which substance is “Lumacaftor / Ivacaftor”, which is classified as an “Ingredient/substance” and produced through “Synthetic/extractive chemistry”.

<CreationDateofMA OD> 2015-11-19 00:00:00.0</ CreationDateofMA OD>

The creation date of the status was the 19/11/2015.

<OrphaDesignationMA Number>EU/1/15/1059</OrphaDesignationMA Number>



The EU Number is EU/1/15/1059.

```
<Person id="51022">  
<Firstname/><Lastname> ERTEX PHARMACEUTICALS (IRELAND) LIMITED  
</Lastname>  
<PersonActivityList count="0">  
<PersonFunction id="11">  
<Type>Eta</Type>  
<Name lang="en"> Marketing authorisation holder </Name>
```

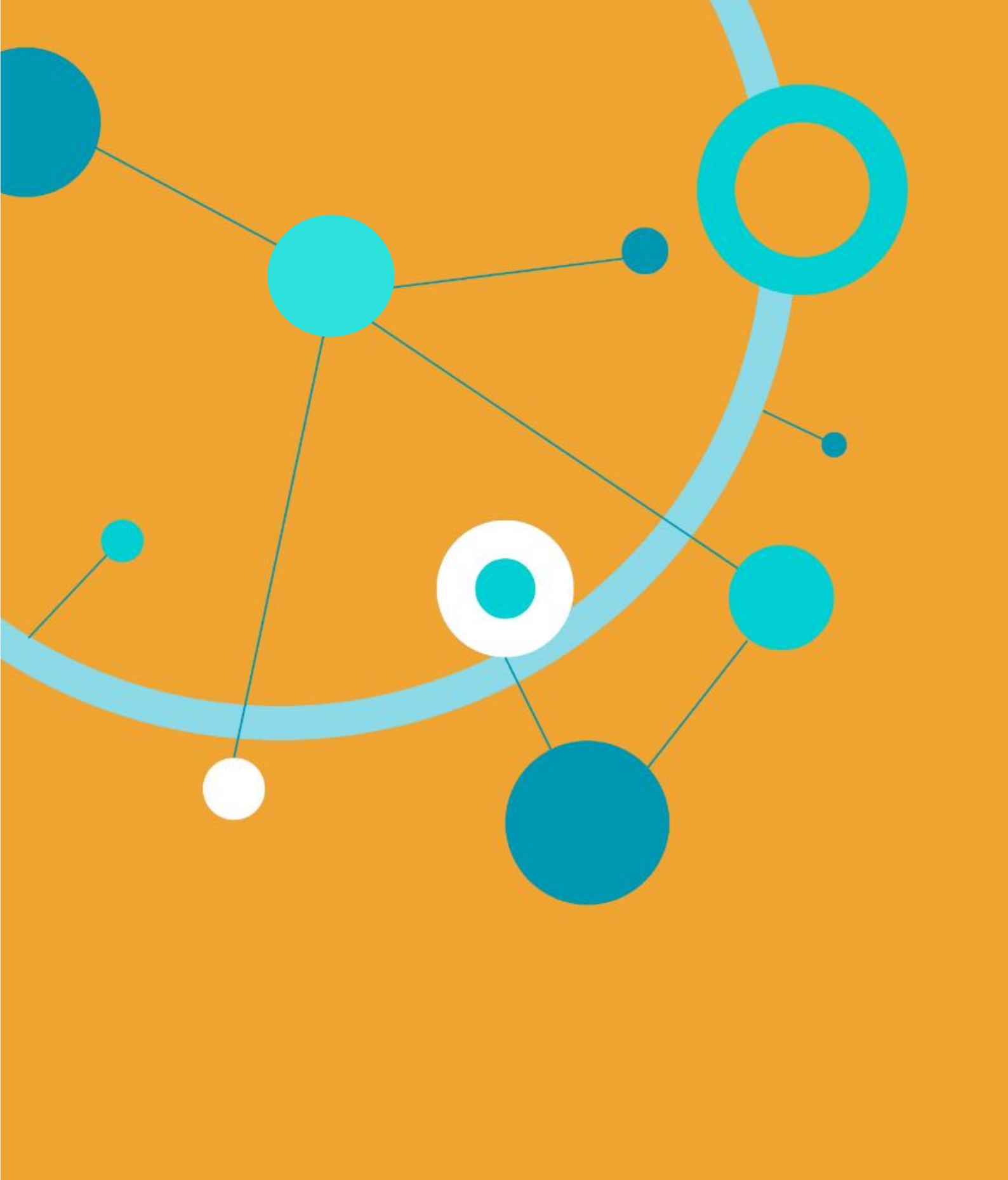
The Marketing authorisation holder is ERTEX PHARMACEUTICALS (IRELAND) LIMITED.

```
<DisorderList count="1">  
<ORPHAcode>586</ORPHAcode>  
<Name lang="en"> Cystic fibrosis </Name></Disorder>
```

The regulatory status operates for Cystic fibrosis (ORPHAcode 586).

```
<Abstract>Orkambi tablets are indicated for the treatment of <b>cystic fibrosis</b> (<b>CF</b>) in patients aged 6 years and older who are homozygous for the <i>F508del</i> mutation in the <i>CFTR</i> gene.<br>Orkambi granules are indicated for the treatment of cystic fibrosis (CF) in patients aged 1 year and older who are homozygous for the F508del mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) gene.</Abstract>
```

This is the designation of the regulatory status.



Authors : Orphadata Team

ORPHADATA PRODUCTS : PRODUCT DESCRIPTION, V3, March 2026

<https://www.orphadata.com/docs/OrphadataOnRequestProductsDescription.pdf>