SCIENSANO RD TEAM – ORPHANET AND OD4RD PROJECTS
JANUARY 2024 NEWSLETTER

1. ORPHANET: THE INTERNATIONAL PORTAL FOR RARE DISEASES AND ORPHAN DRUGS

A) NEW ORPHANET BELGIUM COORDINATOR

Since November 2023, **the Orphanet Belgium team has a new country coordinator, Prof. Dr. Olivier Devuyst,** who takes over the role held by Elfriede Swinnen. Olivier Devuyst is a nephrologist, head of clinic and head of the Institute of Rare Diseases (IMR) at the Cliniques universitaires Saint-Luc (CUSL). He is Full Professor of Medicine at the UCLouvain Medical School (Belgium) and teaches nephrology at the University of Zurich (Switzerland) where he co-directs the university program on RDs. Prof. Dr. Devuyst has coordinated several RDs European programs and is one of the founding members of the European Reference Network on Rare Kidney Diseases (ERKNet).

What are the roles of an Orphanet country coordinator?

In every participating country of the Orphanet network, a country coordinator is nominated by the management board. Each country coordinator agrees to advance the objectives of the project and to be responsible for the collection of data on expert resources at a national level. He/she organizes the governance of the project at national level, including liaison with learned societies, health authorities and patient organisations, and the build-up of the Orphanet team if applicable. The country coordinator is responsible for data quality management about expert resources in the country. He/she acts as the national contact point for the health authorities on RDs. He/she is a professional well established in the field of RDs, with a strong interest for public health and research issues. The country coordinator participates in the Orphanet Management Board meetings, edits the national web pages of Orphanet, contributes to the dissemination of national initiatives in the field of RDs via Orphanews and the OrphaNetWork internal newsletter, and participates to the annual meeting.

B) NEW ORPHANET BELGIUM WEBSITE

A **new version of the Orphanet Belgium site** was launched in 2023. **Do not hesitate to send us information concerning your conferences, symposiums, patient associations'activities, etc., so that we can relay the information on our national site.** Contact: **orphanetbelgium@sciensano.be**

C) IT ISSUES CONCERNING THE ORPHANET DATABASE

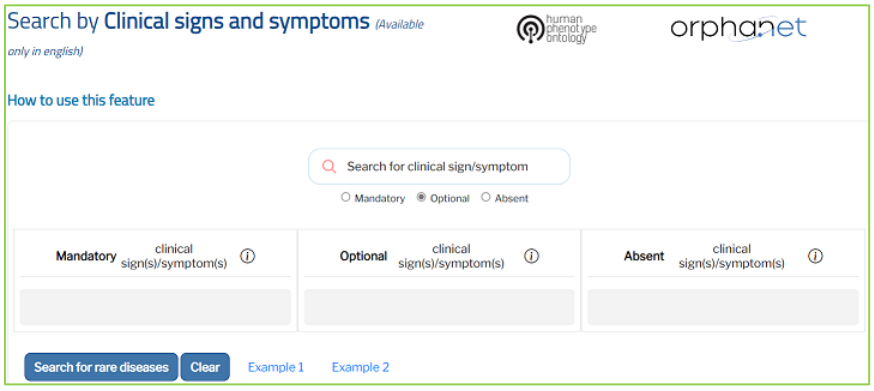
Orphanet provides access to a **directory of expert resources by disease**, such as expert centres, medical laboratories and diagnostic tests, patient organisations, research projects, clinical trials, registries and biobanks in each country of the Orphanet consortium. Through Sciensano, Belgium contributes to this international project.

Due to technical problems affecting the IT tools made available by the Orphanet network coordinating team (France, Inserm), **requests to modify data cannot be processed since the end of November 2023**. According to the latest information received from Inserm, the problem should be solved by the end of Jan 2024. However, your modification requests are taken into account and will be processed as soon as possible. We thank you for your understanding.
Contact to register/update a RD activity in the Orphanet database: orphanetbelgium@sciensano.be

D) SEARCH FUNCTION FOR RDs BY CLINICAL SIGNS AND SYMPTOMS

Orphanet provides a **tool for searching rare disease by clinical signs or symptoms.** The clinical description of rare diseases, based on cases published in biomedical literature, uses the phenotypic abnormalities referenced in the Human Phenotype Ontology (HPO). Each phenotypic abnormalities are presented by order of frequency of occurrence in the patient population.

This tool (available only in English) is dedicated to searching for a rare disease by clinical signs or symptoms. It must been used as a search facility to find information in Orphanet and not as a way to assist making a diagnosis. In the search bar, you can enter the clinical sign/symptom you are looking for and select the corresponding HPO term. It is possible to select several clinical signs/symptoms. In this case you can choose the Mandatory/Optional/Absent options to combine them. You can then drag and drop the clinical sign/symptom from one category to another. The combination of the defined clinical signs/symptoms will generate a graphical representation of the corresponding rare disease(s) based on Orphanet's knowledge.



E) RDK™: THE NEW APP TO REDUCE DIAGNOSTIC ERRANCY IN PEOLE LIVING WITH RARE DISEASES

RDK™ (Rare Disease Knowledge app), also based on HPO terms, was recently developed to accelerate the diagnosis and care of rare diseases. **RDK is a medical device software, available in French and English, providing an assistance tool that allows users to identify potential rare diseases based on clinical symptoms and guide patients to specialized expert centres, overall helping to shorten the diagnostic process.**

The RDK tool is co-developed by Orphanet, Tekkare® and As We Know®. While primarily dedicated to physicians, this device is freely available to all (without the need for prior registration) and allows:
- to search by sign or symptom, helping the healthcare professional make the first step to identifying a potential RD;
- to find the expert centres able to diagnose and advise on care of the RD(s) identified (geographical coverage is currently limited to France);
- to access up-to-date knowledge on RDs including data from Orphanet, as well as best practice tools such as emergency guidelines and national diagnostic and care protocols, which enable patients to be monitored long-term.

This new tool combines Orphanet's data and expertise on rare diseases with Tekkare's technical know-how and represents a wealth of knowledge, putting information on more than 6,200 rare diseases right in your pocket.

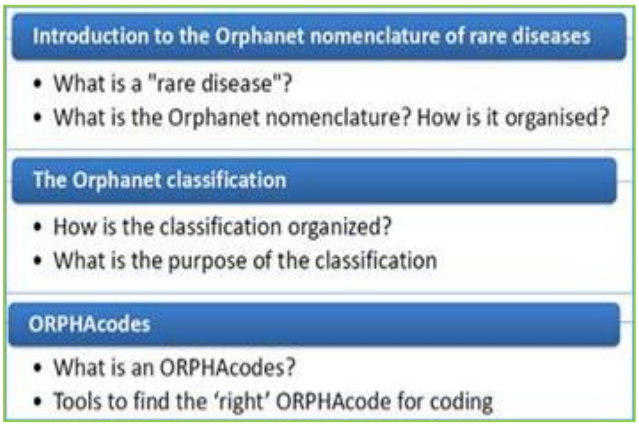
2. OD4RD: EUROPEAN PROJECT "ORPHANET DATA FOR RARE DISEASES"

Rare diseases are poorly represented in the medical terminologies commonly used. There is no specific terminology dedicated to these conditions, with the exception of the **multilingual standardized specific nomenclature developed by Orphanet**. Orphanet created and continuously updates the nomenclature and the corresponding classification of RDs, essential in improving their visibility in health and research information systems. Each clinical entity referenced in Orphanet is assigned a unique, time-stable and non-reusable numerical identifier, the **ORPHAcode**, around which the rest of the data present in the Orphanet database is structured.

One of the main objectives of the **OD4RD project**, co-funded by the EU4Health programme of the European Commission, is to **advance the implementation of ORPHAcodes in hospitals hosting European Reference Network (ERN) centres and to use the expertise of ERNs to further improve the Orphanet nomenclature**. This should make it possible to establish a **common language, with codes specifically created for rare diseases**, to effectively monitor and report on rare diseases (including undiagnosed cases) at European level.

A) TRAININGS ON RD-CODING

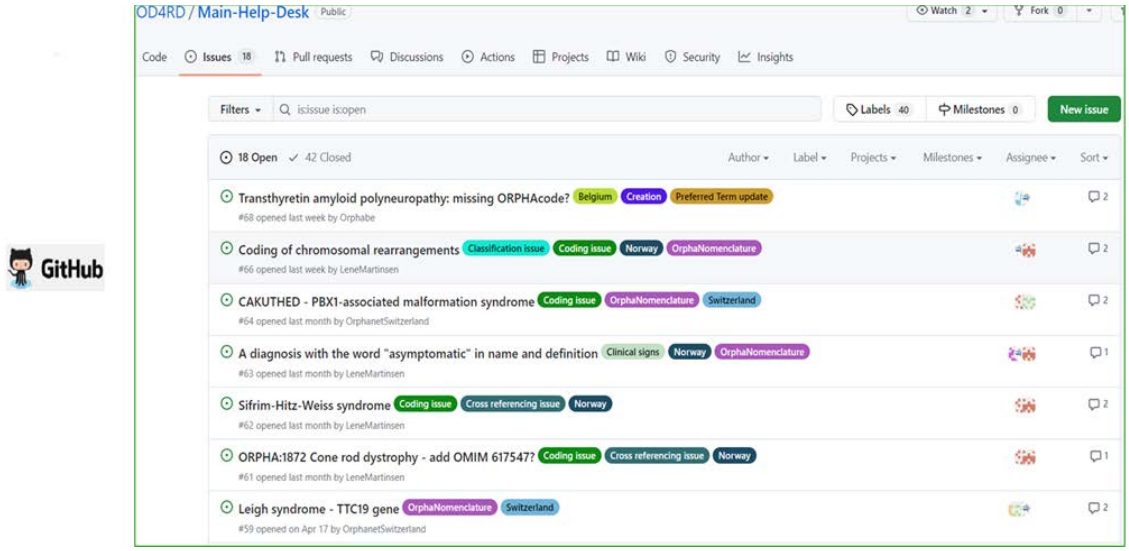
What do we offer? **Basic training on the coding of RDs based on the nomenclature and classification developed by Orphanet**, in several languages (French, Dutch and/or English) to clinicians, coders, hospital information managers, statistical services and other stakeholders involved in the field. This training takes around 2 hours, online or on-site, and is structured in 3 main parts (see below). **Quizzes** (one basic, one with practical cases) are available to check understanding of the basic principles. **More in-depth trainings** can be organised, on request.
Contact: Orphacodes.Belgium@sciensano.be



This training is highly recommended for Rare Disease Functions. UCL Saint-Luc, UZ Gent, UZ Leuven, Liège University hospital and IPG-GHdC already participated in these training courses. For other rare disease functions, discussions are underway.

Please note that it is strongly recommended to **use ORPHAcodes for sending registrations to the Central Registry Rare Diseases (CRRD) hosted at Sciensano**. If you have questions regarding registration in the CRRD you can contact us via '**crrd@sciensano.be**'.

B) HELPDESK ON RD-CODING



An **OD4RD-code GitHub repository** is dedicated to **addressing issues related to the Orphanet nomenclature content and implementation in the health information systems** (e.g. difficulties to identify the correct ORPHAcode for a diagnosis, missing ORPHAcode, mapping with other terminologies, technical difficulties related to the integration of ORPHAcodes into local IT systems, etc.), in order to support clinicians and coders in their daily practice when dealing with ORPHAcodes. The access is free: everyone can read the questions and answers posted on this platform, and ask questions after creating an account.

You can use this GitHub to post your issues on RD-coding to the Orphanet coordinating team (only in English). However, please do not hesitate to contact your **OD4RD national hub** (in FR, NL or EN) at the following email address: **Orphacodes.Belgium@sciensano.be**. We will act as a first level of support (with advanced support from the Inserm-Orphanet nomenclature team when needed).

A **frequently asked questions (FAQ) page** was developed based on the most frequently received questions from users. It provides standardized answers among 8 themes: alignments of ORPHAcodes with other terminologies, coding recommendations, education and communication, epidemiology of rare diseases, ORPHAcodes and nomenclature, Orphanet classification, tools developed by Orphanet and collaborations between Orphanet and the ERNs. **We invite you to consult this page rich in interesting information on the coding of rare diseases.**

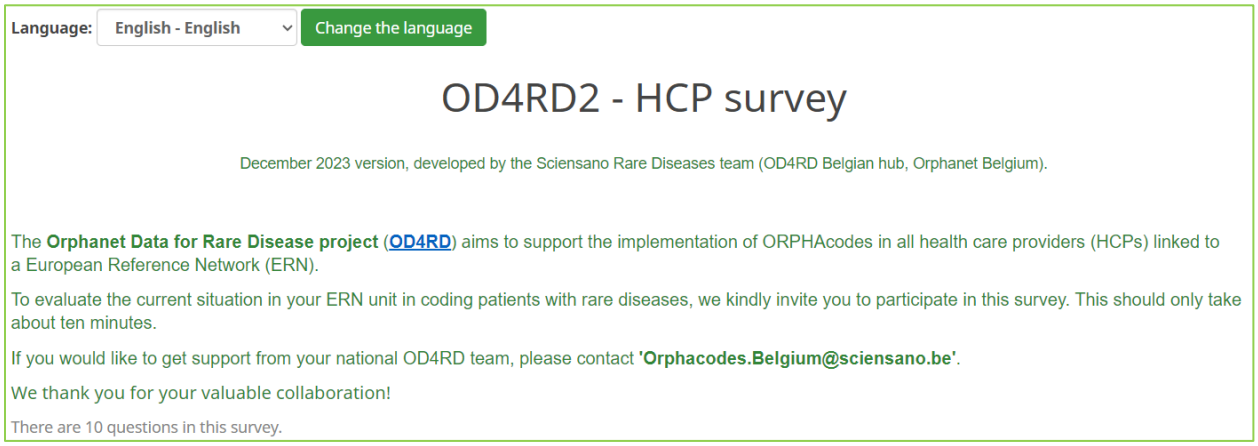
C) MATERIAL AVAILABLE ON RD-CODING

New communication material on RD-coding was developed in the context of the OD4RD project. Do not hesitate to share this material with your colleagues active in the RD field.

1. WHY ORPHAcoding vs other generic terminologies - Booklet. If your firewall does not allow access to the booklet, you can also access this information in PDF format
2. Making IT easy: ORPHAcode implementation in health information systems - Flyer
3. What is the Orphanet Network of National Hubs - Flyer

D) HCP SURVEY ON RD-CODING

As medical (co-)coordinator of a unit (service/department) member of an ERN, we invite you to participate in a survey on the coding method used for your RD patients. The purpose is to analyze the current situation in terms of coding of RD patients in the health information systems of the Member States. It will be launched simultaneously in the 20 countries participating in the OD4RD2 project. This survey should take less than 10 minutes to complete and the Belgian version is available in 3 languages (EN, FR, NL).



Please click here to **access the Lime survey**.

The **deadline for participation is January 31, 2024**. We will then analyze the data collected and write a report on the results for the European Union which is funding the project. The analysis of the responses will allow us to have data on current practices in our country, as well as on the need for support to facilitate the implementation of ORPHAcodes in each hospital/centre caring for people living with a rare disease.

Invitations to participate were sent by the Orphanet Belgium team to each (co-)coordinator of a unit (service/department) member of an ERN, as well as to the RD coordinators. It may happen that the invitation reached several contact persons working in the same hospital within units that are members of the same ERN. In this case, we kindly ask you to coordinate your response to provide us with only one response/ERN where possible.

At the time of writing this newsletter, the participation rate is still quite low, despite the high number of Belgian RD experts contacted (280 emails were sent on December 18, 2023 to representatives of 135 Belgian ERN-units and to the RD coordinators). We invite you to take the time to participate in this survey and take the opportunity to give your feedback on areas for improvement in the RD-coding field.

3. EVENTS AROUND RARE DISEASES

A) RD DAY 2024

Like every year, on the last day of February, thousands of events are organized around the world to raise awareness about rare diseases and the challenges faced by the patients, their families and caregivers. This year, the RD Day will take place on **Thursday February 29.** The key concept of the 2024 Belgian campaign revolves around the message conveyed by the hashtags **#partagetescouleurs, #deeljekleuren, #raresensemble, #samenzeldzaam, #rarediseaseday** and the sharing of the colours of rare diseases (pink, blue, green, purple) throughout the country.

The Sciensano RD team supports the actions initiated by RaDiOrg, the Belgian umbrella association for people with rare diseases. Actions will be implemented within our institution (communication on the Intranet, decoration of our office, organization of an information stand, nail painting workshop, etc.). **We invite you to consult the Rare Disease Day website** to discover various tips and tricks for organizing your own action, involving those around you (family, friends, colleagues, mayor of your town, director of your children's school, etc.) and have access to communication material (posters, visuals for social networks, mailing headers, visual elements for your email signatures, etc.).

B) ECRD 2024

The **European Conference on Rare Diseases & Orphan products (ECRD)** is recognised globally as the largest, patient-led rare disease policy event. It is organized every two years by EURORDIS, the non-profit alliance of over 1000 rare disease patient organisations from 74 countries. Orphanet is co-organiser of this conference. The **12th edition** will take place on **15-16 May 2024 at The Square in Brussels and online**.

Registration is now open with a limited number of 300 places available to join in person. Early-bird fees are available for online participation until 02 February. The **call for posters is open until the 23rd of February**.

We strongly encourage the registration and submission of posters by Belgian healthcare professionals and researchers, especially given the special attention paid to our country which currently holds the EU Presidency.