



Orphanet Belgium National Team Highlights (2023)

Institution / Team composition

Hosted by: Sciensano, the Scientific Institute of Public Health; endorsed by the Ministry of Health.



Team composition:



New Country Coordinator: Prof. Dr. Olivier Devuyt (Nov 2023 -)
Nephrologist, Full Professor of Medicine at the UCLouvain Medical School (BE) and at the University of Zurich (CH).
Clinical activities at Saint-Luc Academic Hospital, Brussels (BE).



PM - Information Scientist: Annabelle Calomme (2017 -)
Management of the database content (collection, analysis, registration and update of Belgian data); management of the national website (FR); member of the OD4RD national hub.



Translator terminology, encyclopedia: Kim Van Roey (2017 -)
Dutch translation and validation of the Orphanet nomenclature, classification and encyclopedia of RD; management of the national website (NL); member of the OD4RD national hub.

National Coordinating Board: FPS Public Health, NIHDI (financer), Sciensano.

External partnerships:

- « RaDiOrg », the umbrella patient organisation for RD;
- College of Human Genetics.

Partnerships within Sciensano:

- Belgian Genetic Test Database (BGTD);
- Belgian Central Registry Rare Diseases (CRRD), RD specific registries;
- Reference centres/labs for rare infectious diseases.

Orphanet Belgium website:

- French version: <https://orphanet.site/belgique>
- Dutch version: <https://orphanet.site/belgie>



Highlights

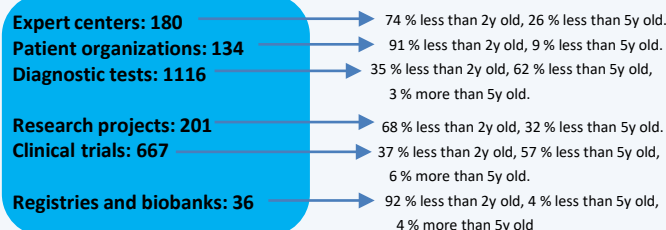
Estimation of the number of Belgians living with a RD: at least half a million. Belgium has been part of the Orphanet network since 2001. Belgium participates in the 24 ERNs.

Participation of our team in the Orphanet task force for new diagnostic tests model (2022-2023) and in the Orphanet working team for the creation and completion of multinational clinical trials (2023-2024).

National RD plan: contribution to its writing (2013); Orphanet Belgium is described in Action 17; no new national plan planned.

Codification: participation as a pilot-country in **OD4RD-WP4**; organisation of trainings on Orphanet nomenclature and codification in hospitals/ERN-centres and for general practitioners (2022-2025).

Expert Resources: Belgian data (Nov 2023)



Translation activity data: 6729 texts (2321 abstracts/4408 definitions) translated into Dutch. 28628 terms (preferred/synonyms/keywords): 25643 for active clinical entities / 108 for functional entities / 3 for pharmacological entities / 2874 for inactive entities. Translation of HPO terms to Dutch.

Awareness/dissemination activities: communication through meetings, national rare diseases website and conferences (College Human Genetics, ESHG congress, Rare Diseases Day, university hospitals seminars, etc.).

Challenges



- Workload vs staff available:** human resources constraints, translation activity, involvement in new Orphanet projects and task forces.
- Sustainability:** the Orphanet Belgium team will continue to be partially financed by NIDHI in 2024 but a new framework agreement between Sciensano and NIHDI must be negotiated at the end of 2024 to obtain new sustainable structural financing.
- Belgium is a **multilingual country** (Dutch, French, German) and an **"institutional lasagna"**: increases the workload (material to create and communication to master in several languages), complicates the implementation of projects at national level.
- Designation of centres of expertise:** no legal basis for the recognition of expertise in Belgium (federal versus regional level?).
- ORPHAcoding:** lack of support from the national health authorities and policy-makers to stimulate the use of ORPHAcodes (no legal framework and no financial support for use in Belgian hospitals); ongoing transition to SNOMED CT® as the common national reference terminology in all Belgian electronic health records; resistance of some clinicians to adopt ORPHAcodes (no time or resources for double coding).
- Achieve the completeness of the database/identification of new expert resources/annual update:** low level of response from professionals and institutions.

Plan for next year and Take away message



- Maintain and increase the content of the Orphanet database** with "clean" and up-to-date Belgian data.
- Continue the translation effort into Dutch.**
- Continue to promote the Orphanet database** among Belgian healthcare professionals (RD clinical experts, researchers), patient organisations, etc.
- Improve the follow-up of the national strategy on rare diseases** with regional/federal health authorities.
- Continue our participation in OD4RD2:** organisation of new training sessions on RD coding for experts, maintaining an efficient helpdesk, recruitment of a new collaborator to help deliver trainings.
- Work proactively towards the interoperability of ORPHAcodes**, in collaboration with the Belgian Rare Diseases Registry and other Belgian RD registries.
- Continue our collaboration with the FPS-Terminology centre** to promote the use of ORPHAcodes as a complement/subset of the SNOMED nomenclature.
- Improve the content of the Orphanet Belgium website**, by strengthening our collaboration with patient associations (publication of their initiatives, etc.).