



Belgian Team Highlights, 2021

Institution /Team composition

Hosted by Sciensano, Scientific Institute of Public Health

Endorsed by Ministry of Health

Team composition:

Information Scientist: Annabelle Calomme

Translator Terminology, Encyclopedia: Kim Van Roey

Country Coordinator: Elfriede Swinnen

National Coordinating Board: Health Authorities, Sciensano, Belgian Terminology Centre

Additional Partnerships:

* Umbrella Patient Organisation « RaDiOrg »

* College of Human Genetics

* Partnerships @ Sciensano: Belgian Genetic Test Database (BGTD), Service responsible for Reference Labs Rare Infections

Belgian websites:

* Dutch version:

<http://www.orpha.net/national/BE-NL/index/homepage/>

* French version:

<http://www.orpha.net/national/BE-FR/index/page-d-accueil/>

Highlights

Statistics Database of resources:



Translation to Dutch: 4454 texts (3141 Def & 1313 Abs)
27077 terms

➔ **SnomedCT Belgian NL release:** Integration of 3128 rare disease related terms

Awareness/dissemination: communication through meetings and conferences (Workgroup on Molecular Genetics, ESHG congress, RaDiOrg members day, Rare Diseases Day, at university hospitals)

Challenges

Challenging registration of Belgian expert centres : no specific “expertise centres” are recognized (yet). Only for some disease (groups) there are reference centres. Additionally, centres participating in ERNs may be registered.

“Validation” of translated Orphanet terms and selection of preferred terms due to differing opinions and the terminology being continuously updated.

Maintain an up-to-date database despite disruptions in manpower.

Find the time to educate experts on the possibilities of Orphadata, ORDO ...



Elfriede



Annabelle



Kim

Take away message

Finalisation phase of the **update of all genetic tests** in collaboration with the BGTD manager. This work led to the creation of around 700 genes as well as the creation of 1 rare disease and the validation of 3 gene-disease relationships not yet registered in the Orphanet database.

Finalisation phase of the **registration of all centres belonging to an ERN** (call 1).

Sufficient funding and collaboration between stakeholders is essential to ensure a complete and accurate database for all Orphanet users.

Translation of Orphanet terminology and encyclopedia is not a one-time task but a **continuous activity** as these Orphanet resources are continuously being adapted to current medical knowledge.

Synergy with other rare disease projects carried out at Sciensano that are benefitting from the Orphanet team’s expertise and its contacts with Belgian rare disease field.