ORPHANETWORK DIRECT GRANT

The international rare disease and orphan drug database bridging healthcare & research

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RÉPUBLIQUE FRANÇAISE
MINISTÈRE DES SOLIDARITÉS ET DE LA SANTÉ

Inserm
La science pour la santé
From science to health

Co-funded by the Health Programme of the European Union

OVERVIEW

Project description

INSERM, US14 - Orphanet now coordinates the OrphaNetWork Direct Grant
co-funded by the 3rd EU Health Programme

Rare diseases (RD) are considered a challenge for Europe. RD have been identified as one of the paradigmatic fields in which actions conducted at the European level constitute the adequate response to the specific challenges they represent:

- **Poor recognition leading to diagnostic delay and inappropriate management including adapted social services;**
- **Poor health outcomes;**
- **Social burden;**
- **Limited knowledge on natural history and pathophysiology leading to an insufficient development of new therapies.**

Amongst the key actions developed with the support of the European Commission to address key priorities in the field of RD is Orphanet, a European knowledge base dedicated to RD and orphan drugs. Orphanet is accessible from the portal www.orpha.net, and re-usable data is provided via the platform www.orphadata.org, or through the Orphanet Rare Disease Ontology (ORDO). Orphanet has also be recognised, by the EC , as having a de facto monopoly in its field.

The objectives of the project are:

1. Provide the RD community with **interoperability tools**, in particular concerning an inventory of RD, to allow for semantic interoperability between countries and between the domains of health and research;
2. Provide **high-quality information** on RD, in particular through an encyclopedia in several languages;
3. Provide a **directory of expert services** in order to help patients, physicians and stakeholders find expertise on a particular disease in Europe and beyond, and to produce data needed to support policy actions;
4. Further **develop and sustain Orphanet** as the reference knowledge base on RD by establishing and consolidating collaboration within the Orphanet pan-European network and with European Reference Networks (ERNs) for the production, improvement and dissemination of knowledge on rare diseases : this will support the creation of a consistent, expertise ecosystem for rare diseases in Europe.

The **main expected outcome** of this project is to consolidate Orphanet as the European database for rare diseases, positioning it as the international reference for information and data on rare diseases. **Other expected outcomes** will be to provide the rare diseases community with a standard nomenclature enabling rare diseases patients to be detected and brought to the best expertise and to clinical research; to provide the rare disease community with a tool for semantic interoperability between health information systems, registries, biobanks and other data repositories; to build as stable yet dynamic European rare diseases ecosystem as the result of the close cooperation between Orphanet and European Reference Networks.

PARTICIPANTS

Partners

- Institut national de la santé et de la recherche médicale (INSERM), France
- Medical University of Vienna (MUW), Austria
- Sciensano, Belgium
- Bulgarian Association for Promotion of Education and Science (BAPES), Bulgaria
- Children's Hospital Zagreb, Medical School University of Zagreb (KDB), Croatia
- Charles University, Second Faculty of Medicine (CUNI), Czech Republic
- University of Tartu (UT), Estonia
- Rinnekoti Foundation, Finland
- Medizinische Hochschule Hannover (MHH), Germany
- Ministry of Human Capacities (HU-MHC), Hungary
- Health Service Executive (HSE), Ireland
- Ospedale Pediatrico Bambin Gesù (OPBG), Italy
- Vilnius University Hospital Santaros Klinikos (VUHSK), Lithuania
- Department of Health (DHIR), Malta
- Academicsch ziekenhuis Leiden-Leids Universitair Medisch Centrum (LUMC), the Netherlands
- Oslo University Hospital (OUS-HF), Norway
- Instytut Pomnick Centrum Zdrowia Driecka (IPCZD), Poland
- Directorate-General of Health (DGS), Portugal
- Institute of molecular genetics and genetic engineering- Belgrade University (IMGGE), Republic of Serbia
- Universitatea de Medicina si Farmacie Iasi (UMF), Romania
- Consorcio Centro De Investigacion Biomedica En Red (CIBER), Spain
- Karolinska University Hospital, Sweden
- Public Health England (PHE), United Kingdom
- University Medical Center Ljubljana (UMCL), Slovenia

Collaborating Partners

Ministry of Health, Argentina
Center of Medical Genetics and Primary Health Care , Armenia
Office Population Health genomics, Department of Health Western Australia
McGill University, Canada
Georgian Rare Diseases Foundation, Georgia
Chaim Sheba Medical Center, Israel
Children Hospital, Latvia
Translational Research Center for Medical Innovation (Foundation for Biomedical Research and Innovation at Kobe), Japan
Institut National d'Hygiène, Morocco
Hôpitaux Universitaires de Genève HUG, Switzerland
Farhat HACHED University Hospital, Tunisia

PLANNED ACTIVITES

7 Sub-projects (workpackages)

Coordination of the project	Orphanet-Inserm US14 will manage the project and will make sure that it is implemented as planned. The Management Board meets every two months by conference call and annually during a face to face meeting.
Dissemination of the project	To ensure that the results of the project are made available to the target groups, all results will be released via www.orpha.net and news and updates will be regularly published in the OrphaNews newsletters. Furthermore dissemination activities will be carried out via booths at RD related conferences.
Evaluation of the project	To verify that the project is being implemented as planned and reaches the objectives and end-users expectations evaluation reports will be issued and user surveys will be launched annually online
Availability of the Disease nomenclature and interoperability tools	The rare disease nomenclature, alignments with other terminologies and genetic databases will be maintained and updated, information will be made available on orpha.net, orphadata.org , ORDO and through the Orphanet Report Series. Production procedures are made available here.
Availability Information on rare diseases and orphan drugs	Definitions and up-to-date information on rare diseases and orphan drugs will be provided and made available on orpha.net, orphadata.org and ORDO and in the Orphanet Report Series. Work procedures are made available here.
Availability of the Expert resources catalogue	The catalogue of expert services in 24 participating MS plus Norway and Serbia will be maintained and updated. The information will available on orpha.net, orphadata.org and in the Orphanet Report Series. Work procedures are made available here.
Consolidation of Orphanet as the reference European database for RD	Internal quality of the database content, capacity of the network, and cooperation at country level and with ERNs towards Orphanet sustainability will be ensured.

Target

Actions in this project are intended to meet the needs of patients and their relatives, healthcare professionals, researchers, industry and policy makers.

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