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Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases

JA2015 - GPSD [705038]

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Project abstract

Rare diseases (RD) have been identified as one of the paradigmatic fields in which actions conducted at the European level constitute the adequate response to their specific problems: 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. The low prevalence and the specificity of RD make that a global, multi-stakeholder approach, intended to gather specific expertise and to build shared strategies is necessary to address these issues.
The general objectives of RD-Action are to:
▪ Support the further development and sustainability of the Orphanet database, the biggest global repository of information on RD
▪ Contribute to solutions to ensure an appropriate codification of RD in health information systems
▪ Continue implementation of the priorities identified in Council Recommendation 2009/C151/02 and the Commission Communication (COM 2008 679) on RD, with a view to ensuring the sustainability of the recommended priority actions and to support the work of the Commission Expert Group on Rare Diseases (CEGRD).
This JA will expand and consolidate the achievements of the former JAs on RD supported by the European Commission: the Orphanet JA and the EUCERD JA. More precisely, this proposal has the ambition to help member states to implement the recommended measures adopted or to be adopted by the CEGRD and to produce the data necessary for countries to do so. Interactions between the production of data at the Orphanet database level and the implementation of policy priorities including codification will be strengthened during this JA.RD-Action large geographical coverage is key to success as it will promote the transfer of European recommendations into national policies and the collection of information and concerns from MS to the CEGRD, thus to the European Commission.

Summary of context, overal objectives,strategic, relevance and contribution of the action

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Methods and means

Methods and means:
The governance of the action is organised into three levels in order to gather specific expertise and to build shared strategies to address the specific issues of RD:
- a General assembly,composed of one member per designated authority, which is the decision-making body of the consortium in charge of review and steer the project.
- An Executive committee composed by the 7 sub-projects leaders in charge of the supervision, communication and cross-talk promotion
- Sub-projects teams in charge of executing and monitoring the tasks and establish cross-talks with the other Workpackages (WPs).
- Workgroups established ‘à la carte’ according to the need identified

Fig.1 RD-ACTION governance and stakeholders interactions

The executive committee of RD-ACTION composed of all Workpackage leaders has met every two months to discuss the advancements of the project and the potential mitigating actions to be taken in order to ensure on-time delivery of the deliverables. All meeting reports are available on the internal website, here

All partners of the action are kept informed on all progresses, issues and identified risks through the internal newsletter, RD-ACTIONews which is sent every two months and through the internal website.
The General public can also keep-up with the project progresses through the public website, www.rd-action.eu, where news are regularly posted as well as all material produced by the different WPs and also through Orphanews, the RD-ACTION public newsletter which is issued every two weeks. An informative RD-ACTION leaflet for all audiences is also available on www.rd-action.eu. Orphanet Report Series covering topics relevant to RD are also regularly published on www.orpha.net.

Work performed during the reportingperiod

Achievemtns 2015-2018:
- Organisation of the ECRD 2016. The conference has been held in Edinburgh from 26 to 28 May, it brings together over 80 speakers and more than 800 participants, covering six themes of content over two days: from the latest research, to developments in new treatments, to innovations in healthcare, social care and support at the European, national and regional levels. The focus in 2016 was on Game Changers in Rare Diseases.
- EUROPLAN National conferences/workshops were organised in 19 countries to promote the implementation of national plans for rare diseases and facilitate the integration of EU rare disease policies and recommendations into the national system, (http://www.rd-action.eu/europlan-rd-national-plans-conferences-reports/)
- Organisation of 10 workshops (see below)
- Publication of Literature narrative review "Sustainable Health Systems for Rare Diseases" and publication of six policy briefs on Resilience, Primary prevention, Actions on educational programmes, Health systems financial sustainability, integrated care and Patient Empowerement (http://www.rd-action.eu/leaflet-and-documents/)

- Orphanet database evolutions in order to adapt to the community needs and to be transparent and traceable
The annual update of the information of expert resources linked to the professionals registered in the Orphanet database was launched at the end of May 2016, in May 2017 and in May 2018.
Release of the Orphanet curation platform at curation.orpha.net
- Organisation of the Orphanet evaluation
1. Organisation of the Annuals Orphanet users satisfaction survey
2. Launching of Orphanet stakeholders satisfaction and utility survey
3. Setting up of an internal evalutaion process
- Facilitation of Orphanet sustainability through decentralisation of editorial activites, production of a Sustainability plan and Analysis of compatible legal models. The OrphaNetWork Operating committe was also set up in order to

reach tne same level of corporate culture and knowledge across the Orphanet Network.
- For Steering, maintain and promoting adoption of Orphacodes across MS (http://www.rd-action.eu/leaflet-and-documents/)
1. Review of the technical existing implementations of RD coding to have a clear overview of specifications strategy of the required resources for coding RD consostently across MS
2. preparation of Standard procedures and guide for the coding with Orphacodes to facilitate the implementation of Orpha Codification into Health Inofrmation Systems
3. Preparation of a master file and implementation manual for statisticla reporting to ensure data sharing and exploitation at the EU level and ensure consistency in codification with Oprhacodes amongts MS
4. Specifications for an integrated coding application with Orphacodes to facilitate the practicla implementation of Oprhacodes in MS
5. Recommendations for routine maintenance of codification resources for RD for standardisation of long-term maintenance of the codification system in MS
6. A final workshop set the scene of RD coding situation in Europe and focused on the WP5 legacy and the future perspectives of Orphacodes' use. Starting from the resources developed during the RD-action, the issue of RD coding was tackled from the perspectives of all the relevant stakeholders involved: WHO, Orphanet, JRC, ERNs. Examples of orphacodes' use at regional/national level (Generalitat Valenciana-Spain, Portugal, Ireland) have been presented as well. The agenda and the presentations are available (http://www.rd-action.eu /wp-content/uploads/2018/07/WS-Codification_final_presentations.zip).

- Establishment of a Task Force on Interoperable data-sharing within the ERN framework. The scope of the TF is to define, by the time of the launch of the first wave of ERNs, the elements needed to provide a long term vision in the form of a European interoperability roadmap for data sharing in the framework of operations of ERNs. More immediate goals have been to build

The main output achieved so far and their potential impact and use by target group (including benefits)

All major RD-ACTION outputs are available on the website: www.rd-action.eu

The actions of the project are intended to meet the needs of patients and their relatives, healthcare professionals,researchers, industry and policy makers. The projects targets experts and policy-makers to support them in their work delivering recommendations and position documents on RD policy. Patients are not only ultimately taking benefit from these policies, but are involved, through their patient organizations, in them.
The JA works with experts, patients, policy-makers, people involved in

codification and in registries, industrials, and all the stakeholders in the field.

As far as Orphanet database is concerned, it is a strategic element in national policies for RD. The nomenclature and classification of RD are the basis for codification in health information systems. The Orphanet encyclopedia is intended to help professionals in their clinical practice, but patients and their relatives also
benefit from it. The high-quality directory of expert resources and patient organizations is of help both for health professionals and for patients, improving referrals and patient orientation. Directory of resources related to research promotes networking and collaborations. Industrials can identify experts and resources by this mean. Directory of orphan designations and drugs linked to their indications and to rare diseases is also an important piece of information for patients, healthcare professionals, researchers and pharma industry. Production of reports (ORS) containing compiled data and analysis is of help for policy-makers, and for experts in the CEGRD. Data is delivered through the Orphanet website, which has 82,000 pages views/day from more than 200 countries, as well as massive datasets for free re-use in machine-readable formats through Orphadata (approximately 17,000 downloads/month) [figures from the "Orphanet-2015 activity report"]. The Orphanet website is currently available in 7 languages, but the textual information is available in many more languages.
The IT evolution of the database during this JA will allow to translate the website in virtually every EU language,depending on MS translation capability.

Through the development of coding guidelines and master file all routine users (collectors as well as users) of data on rare diseases will be guided in how to collect data in a standardized way thereby allowing a more reliable interpretation of the collected data. Patient groups, decision makers as well as politics will benefit as they will be able to compare more reliable data and identify patients better once the standardized way of coding is used. Investigators in clinical research will benefit from a reliable identification of RD patients in health information systems and will be able to capture data from the clinical setting. People involved in the codification will be guided in the coding process, which will make it more easy and reliable.

The project is in perfect line with the objectives of the a third Programme for the Union's action in the field of health (2014-2020), in particular those concerning RD. The aims of this project include improving coordination of the action of health professionals, patient organizations and stakeholders in areas in which the
European level provides an added value, in particular ERNs, cross-border healthcare and genetic testing, interoperability between databases and registries, and consistent codification of patients suffering from RDs. Moreover the project is exploring the issues of resilience and sustainability of health systems for RDs.
We are providing efficient support implementing EC recommendations to both the CEGRD and to MS, we will disseminate best practice guidelines and provide practical guidelines and instruments for RD codification in health information

systems in order to achieve better and safer care for European citizens suffering from rare diseases. This support includes pr

Achieved outcomes compared to the expected outcomes

Major results achieved during the three years of the action and their outcomes are presented in table 1

Dissemination and evaluation activitiescarried out so far and their major results

RD-ACTION is using a wide variety of dissemination methods and tools to raise awareness, inform, engage and promote the outcomes of the Joint Action. Dissemination is flexible, and will take advantage of media channels, formal materials, conferences, workshops and face-to-face meetings to share information amongst relevant stakeholders.
All partners are engaged in the sharing of information within their networks. The overall strategic approach is flexible and open to adaptation in line with constant evaluation and feedback of the methods And a dissemination plan was made available in April 2016 on www.rd-action.eu.

Online media are the most versatile and the most frequently used channel of dissemination.
- Project website (see below)
- Websites of RD-ACTION partners: all partners of the Joint Action have included at least a link to the RD-ACTION website.
- Orphanews reports the latest developments in the field of rare diseases and orphan drugs, and therefore communicate on the outcomes of RD-ACTION. The content of the newsletter is comprehensive of all updates in the rare disease community. Each newsletter is organised thematically, which enables more focused communication of the work packages' outcomes. This also makes it easier for stakeholders to access information particularly relevant to their field.
OrphaNews is freely available and electronic. It is designed to suit all stakeholders in the rare disease and orphan drugs community and to be easily accessible. OrphaNews is sent to subscribers (more than 12.800)
twice monthly, and all past issues are archived online at www.orpha.net.

- Partners' newsletters: those partners with newsletters are encouraged to include information on the Joint Action according to the target group of the newsletter in order to inform on the development of the project and outcomes.

When communicating about RD-ACTION in other newsletters, partners must include the link to www.rd-action.eu to channel interested parties back to the website.

-Social media: the WP6 "Policy Development for Rare Diseases and Integration has a specific twitter account: @RareDiseaseEU.
This twitter account has over 900 followers. Using social media creates an engaging presence to enable interested parties to exchange information and further disseminate updates within their own networks.

- Materials: materials used to inform stakeholders of updates are made available on the RD-ACTION website and are be disseminated via the media channels aforementioned where relevant (leaflet, Orphanet Report series, posters and speakers presentations at congresses and press releases.
In order to keep the scientific community informed during year 1-2, RD-ACTION has been presented in nearly 20 events.

Conference and workshops having take place:
ECRD 2016 (http://www.rare-diseases.eu/)
3 Workshops: Sustainable Health Systems for Rare Diseases
Workshop: Exchanging Data for Virtual Care in the Framework of ERNs
Workshop: Using Standards and Embedding Good Practices to Enable Interoperable Data-Sharing in ERNs
Workshop: Indicators and Outcomes for ERNs
Workshop:How can ERNs generate, appraise and utilise clinical practice guidelines, to enhance the impact of consensus guidelines in national health systems?
Workshop:Creating a Sustainable Environment for Holistic & Innovative Care for Rare Diseases & Complex Conditions
Workshop: How ERNs can add value to clinical research in rare diseases and highly specialised domains
WP5 FInal Workshop
19 EUROPLAN National conferences

Dissemination activities related to specific WP progresses are also regularly carried out.

Evaluation activities carried out so far: Survey of the joint action partners' satisfaction, survey of the Orphanet users' satisfaction and after each congress, meeting, training, workshop organised satisfaction of the participant is assessed in order to improve the further events. Orphanet stakeholders survey. Joint ACtion Evaluation report

Work package

Work Package 1: Coordination

Start month: 1
End month: 36
Work Package Leader: INSERM

Task 1.1 : Organisation of the Joint-action kick-off meeting.
Leader (lead applicant): Ana Rath [Inserm]
Start date: M1 End date: M6
It implies preparing a detailed workplan containing a detailed description of all activities of the project, milestones and deliverables to be approved during the kick off meeting.
Task 1.2 : Monitoring of the activities and overall quality of the project
Leader (lead applicant): Ana Rath [Inserm] Contributors: WPs leaders
Start date: M1 End date: M36
The Steering committee composed of WPs leaders will monitor the progress achieved (compliance with the milestones and timetable validated during the kick-off meeting) and address the possible difficulties and opportunities arising during the project. They will also process the information coming from the evaluators, the international advisory board and from the CEGRD. Dissemination issues will also be discussed and validated by the Steering Committee.
Task 1.3: Ensure communication and information exchange amongst Joint action participants
Leader (lead applicant): Sylvie Maiella [Inserm], Contributors: WPs leaders/project managers
Start date: M1 End date: M36
An internal newsletter will be edited every two months in order to ensure a smooth communication among WPs. Where necessary in between newsletters, emails and conference calls will be organized. It aims to inform the partners on the conclusions of the Monitoring meeting. It will also ensure circulation of information, among all partners, relating to each team’s activities and outputs in order to facilitate the acquisition of comprehensive knowledge regarding the big consortium. Two annual meetings will be organised after the kick-off in Y2 and Y3, in order to allow Management Board to meet. A private space intended to allow JA partners to share documents will be created in the JA website.
Task 1.4 : Intermediary for all communication with the Chafea and the DG SANTE
Leader (Inserm): Ana Rath [Inserm]
Start date: M1 End date: M36
The project coordinator will act as the official representative towards the Chafea and DG SANTE. The project coordinator will provide them two interims and a final report, assisted by the Project Manager and the Financial Officer.

Work Package 2: Dissemination

Start month: 1
End month: 36
Work Package Leader: EURORDIS

Task 2.1 : To set up and maintain the Joint Action dissemination tools
Task Leader (lead applicant): Sylvie Maiella [Inserm] Contributors: All partners
Start date: M1 End date: M36

• A website containing information on the action will publish information on the partners of the JA, on the JA progress from the different stakeholders and, at the end of the contract, the layman brochure of the final report.
• A leaflet promoting and explaining the JA activities will be prepared and translated in all partners languages
• The State of the Art 'Report/Resource' will be disseminated/promoted via JA tools

Task 2.2: To produce the Orphanews newsletter
Task Leader (lead applicant): Ana Rath [INSERM], Contributors: all
Start date: M1 End date: M36

Twenty electronic issues of the twice-monthly Newsletter of the Rare Diseases Community - Orphanews - will be produced per year. This will amount to a total of 60 issues during the whole duration of the JA. A section dedicated to this JA will be included in Orphanews in order to communicate timely the progresses to the 15,000 registered readers. The Editorial Board of the new version of Orphanews will be composed at least of WP2 contributors.

Task 2.3: To hold the reference European Conference on Rare Diseases and OrphanProducts in 2016
Leader: EURORDIS, Contributors: All partners
Start date: M17 End date: M17

The European Conference on Rare Diseases and Orphan Products (ECRD) which will be held in May 2016 in Edinburgh will be the 8th ECRD. The ECRD is a major dissemination tool and represents an important platform for policy promotion that covers all areas of relevance for the Rare Disease Community at large. As such, this Conference is considered as a priority event by all stakeholders active in the field of rare diseases and has been officially mentioned in the “Commission Communication on Rare Diseases: Europe’s challenges”, in the Governance and Monitoring Chapter. The ECRD 2016 Edinburgh will involve all stakeholders relevant to the Rare Disease Community at large. The Program Committee in charge of developing the Conference’s program comprises of a representative from all main stakeholders groups, patients, national decision-makers/national authorities, European policy makers, Members of the CEGRD industry representatives, researchers, academics, learned societies and medical experts. In 2016, more than 800 participants are expected to attend the ECRD (in Berlin in 2014 there were 768 participants representing 43 countries).
The program of the Conference, including all the different sessions within the

specific themes, is focused around the main structuring measures impacting on the rare diseases field that are taken and implemented at national and European levels, such as the ones deriving from the CEGRD recommendations, the Council Recommendation on rare diseases, as well as from other pieces of legislation, e.g. the Cross-Border Healthcare Directive and the various national transposition laws.

Task 2.4: To support national and European integration through national workshops
Leader: EURORDIS, Contributors:, ISS, UNEW.
Start date: M9 End date: M36

This task aims at supporting the development of the content of the national workshops to be organised by National Alliances, in close collaboration and with the support of national authorities, at least one in every Member State. The overarching goal of the national workshops and their substantial added-value is that they will ultimately facilitate the integration of rare diseases-related activities at national and European levels.
These workshops have the specific objectives:
1. To disseminate at national level the JA activities and the Recommendations discussed and adopted by the CEGRD, and previously by the EUCERD. As well as to facilitate their appropriation by different actors at national and local levels.
2. To accompany the implementation of these recommendations in the specific national context

Work Package 3: Evaluation

Start month: 1
End month: 36
Work Package Leader: MUW

Task 3.1 : Evaluation of the Joint action achievements
Leader (lead applicant): Medical University of Vienna; Contributors (applicants involved): Eurordis, INSERM, UNEW
Start date: M1 End date: M36

Overall Joint action evaluation will be based on indicators measuring the process, output, outcome and impact. (please refer also to section 2.2)
In particular:
- European Conference on Rare Diseases (ECRD). Process indicators will include: well defined steps necessary in the overall preparation of the conference (i.e., the nomination of a program and an organising committee, meetings and conference calls of these committees, the development of a conference website and of other information tools like stakeholder-tailored flyers), the range of different themes and topics of the ECRD including accompanying satellite meetings and tutorials, as well as the coverage of the different stakeholders participating in the program committee and the conference. Output indicators will include the number of invited lectures, the number of oral presentations and posters selected from submitted abstracts, and the production of further information material like newsletters or an online conference report. Outcome indicators comprise, inter alia, the total number

of participants, as well as by stakeholder groups. Impact indicators will include the degree of dissemination of the final conference report, the coverage of the conference in classical media as well as in social media. On-site participant satisfactory surveys for each session, as well as a more general online participant satisfactionory survey will provide information on the impact of the conference, and information on the general organisation of the conference and the quality of the content.
• Conference on sustainable health systems for RD. Process indicators will include: well defined steps necessary in the overall preparation of the conference, preparatory steps conducted (literature review, analyses, preparatory workshop, eventually establishement f specific working groups). Output indicators will include one analysis on epidemiological data on RD and one review on sustainable health systems carried out, policy briefs delivered. Outcome indicators comprise, inter alia, the total number of participants, as well as by stakeholder groups. Impact indicators will include the degree and quality of the dissemination of the conference conslusions and, inter alia, the workshops participants' satisfaction (86 expected), by means of survey during and/or after the meeting
- To evaluate the testing phase of the master-file with Orpha-codes and the related guidelines, a set of common indicators for all participating countries will be developed including process indicators (like, inter alia, compatibility with and easy integration into existing health information and coding systems and applicability of the guidelines) and output indicators (like, for instance, the number of single RD entities registered using the master file, the number of more specific codings and the ratio of correct and incorrect coding entries). The workshop addressing the information about the strategies and tools to implement the Orpha codes in the European countries will be evaluated by means of a set of specifically developed indicators including outcome indicators (for instance the number of participants and the number of Member States represented), output indicators (like information materials provided in the workshop intended for the distribution within the Member States and/or a workshop report) and – if applicable – impact indicators (like decisions on the further implementation of Orpha codes in individual Member States). The assessment by indicators will be accompanied by an online user satisfactory survey for all participants.
- All workshops organized in the context of the Policy Development for RD and Integration with other relevant initiatives will be evaluated by online participant satisfactory surveys

Work Package 4: Orphanet, the European database for rare diseases

Start month: 1
End month: 36
Work Package Leader: INSERM

Task 4.1 : Coordination of the Orphanet consortium
Task Leader (lead applicant): Ana Rath (INSERM) Contributors (applicants involved): All WP4 members

Start date: M1 End date: M36

26 associated partners in this JA and 14 collaborating partners are involved in the Orphanet consortium activities. The completeness, consistency and quality of the final database largely depend on the efficacy of the coordination of these partners. The INSERM is coordinating the Orphanet consortium since 2000. The coordination of Orphanet consortium activities includes:

1. Ensure smooth communication and information exchange related to Orphanet database activity through a Supplement edited with the RD-action newsletter, and bi-monthly management board conference calls.
2. Provide day to day technical support to the partners
3. Perform the annual Orphanet user's survey and the Orphanet annual activity report

Task 4.2 : Maintain and expand the rare diseases database
Task Leader (lead applicant): Annie OLRY (INSERM) Contributors (applicants involved): All WP4 and WP5 members
Start date: M1 End date: M36

During this task, the inventory and classification of RD annotated with genes cross-referenced with other resources will be expanded and maintained. A definition for all RD to be included in the content model of ICD11 and SNOMED CT will be produced and the professional encyclopaedia of RD will be further populated and updated.
The aim of the Orphanet nomenclature and classification is to provide the RD community, from healthcare to research, with a well-structured hierarchy specific for RD with different degrees of granularity so as to allow linking data coming from healthcare (i.e. clinical diagnosis in health records) to data coming from research (i.e. genetic entities in databases). The Orphanet nomenclature is at the centre of a rich network of relations inside the Orphanet database and its ontological expression (ORDO, for Orphanet rare diseases ontology), comprised of genes interrelated with other resources (HGNC, OMIM, UniProt, ensembl, Reactome, IUPHAR, Genatlas), epidemiological data (prevalence, incidence, age of onset, age of death, geographical distribution), medical terminologies (MeSH, MedDRA, SNOMED CT, ICD10, UMLS) and resources (OMIM, and, in the near future, HPO). It is therefore considered as a standard nomenclature for rare diseases, and promoted as such by the IRDiRC. This nomenclature is intended to multiple uses (codification in health information systems, registries, research databases…). Therefore this task is interrelated with WP5 for it provides the core nomenclature to be adapted to patient codification needs.
The Orphanet database is completed by producing textual information for each rare disease. In the context of this JA, a new, decentralized organization will be established with two goals: a) to have a definition for every RD in the Orphanet database, and to expand and update the encyclopaedia, and b) to disseminate high-quality articles produced by others in order to provide complete, useful and timely information to physicians and patients, as well as to other actors in the field of information, such as help-lines and national contact points. In order to achieve the first goal, the core editorial activities will be progressively transferred from the

central facility at the INSERM to other participating countries, starting at M18 with Ireland, The Netherlands and Slovakia, and to others that manifest the interest and the possibility to assume this task. This transfert will be facilitated by the tools developed in Task 4.3. Translations of all or part of the website will be encouraged as a national effort in each country. To achieve the second goal, partners in WP4 will contribute identifying and assessing relevant articles according to Orphanet's quality standards that will be published in the website for transparency.

Task 4.3 : Develop the necessary tools to track changes of the Orpha nomenc

Work Package 5: Steering, maintaining and promoting the adoption of Orphacodes across MS

Start month: 1
End month: 36
Work Package Leader: DIMDI

Task 5.1: To define and set the necessary strategy and tools to implement the Orpha codes in the European countries.
Task Leader: Remy Choquet, [BNDMR, APHP,France] - Contributors: All WP5 contributors
Start date: M1 End date: M36

Some MS have already started the work of introducing the Orphacode in their registries or health information systems, and others have expressed their interest adopting them. Different approaches have already been implemented and start producing results, raising problems and bringing solutions that are of interest for all MS. A coding nomenclature alone is not enough to guarantee that the patient data will be comparable from a member state to the other. Along with the right and quality assessed nomenclature of rare diseases (Orphanet), it is required to provide the coders with the right instructions and clear objectives of coding. Also, given the nature of the rare diseases patients and the celerity of new discoveries, it is required to handle uncertainty in diagnoses and frequent updates of the nomenclature.
All MS use morbidity and mortality recording systems. Morbidity recording systems utilize, for the generality of diseases and for the majority of countries, ICD classification. Only in a few countries other systems like SNOMED CT are utilized. The Orpha code classification is specifically dedicated to rare diseases and is used only in few countries. Taking into account these ongoing experiences, the contexts, the prerequisites, the methods to implement specific monitoring systems of RD patients will be defined.
In this part of the work, we will use a bottom-up approach to reach a consensus in defining guidelines to implement RD monitoring in MS. Starting from the existing experiences, a set of rules and guidelines will be produced in order to support the MS in implementing RD monitoring systems and the use of Orpha codes.
A steering group, comprised from the contributing institutions, will be set up by the task leader. The steering group will identify the common denominator of already

existing approaches in the different countries and based on that will define:
• A complete review of current coding systems actually in place in member states and actual plans. This review should give a clear overview of possible strategies and planning as to identify RD patients in each MS. This review will help in setting the master file. (deliverable D5.1)
• The level of granularity of Orphacodes that are essential for all systems working with Orpha codes (registries, centres of expertise, others). Additional level of detail may be used but does not have to be used right from the start. (deliverable D5.2)
• A data exploitation plan, with clear objectives to be addressed by the coding investment (deliverable D5.2)
The Steering group will work during the three-year length and will meet face-to-face at each JA annual meeting and will set up a series of distant meetings. A virtual working space will be shared. A workshop will be organized during year 3 in order to present the results of the whole WP and to promote their use in countries.

Task 5.2: Specification of the required resources for coding RD consistently across Europe
Task Leader: Stefanie Weber, [DIMDI , Germany], Contributors: All WP5 contributors
Start date: M13 End date: M24

Within the second year of the project a master file will be created and populated with preexisting data from countries. It will be further specified according to the guidelines and granularity results from year 1.
The master file will be defined bringing together the current experiences of implementation in countries already using the Orphacodes, and will probably combine the definition of a significant subset of codes needed for interpretation at the European level, their alignments with ICD-10 national extensions and the minimum multi-hierarchical classification structure derived from the Orphanet central resource. (Deliverable 5.3).
Guidelines on

Work Package 6: Policy Development for RD and Integration with other relevant initiatives

Start month: 1
End month: 36
Work Package Leader: UNEW

This WP will build on the work previously developed within the Eucerd Joint Action (EJA) intended to support the implementation of the EC recommendations on rare diseases at the MS level, by establishing position papers and recommendations issued from high-level multidisciplinary working groups in which all the stakeholders were represented. During this WP a methodology will be developed in order to prioritize areas for which there is still need to foster implementation of policies. A pre-selection of topics are proposed below as areas in which substantial unmet needs remain with respect to rare diseases and / or where a cycle of

ongoing or updating of current recommendations will be required over the period of the Joint Action. For all of these subject areas, synergies will be assured with other funded initiatives and their leadership engaged. These topics largely correspond to Operational Actions defined under the Commission Communication (COM 2008 679), where work remains to be carried out (Operational actions related to improving recognition and visibility of RD will primarily be addressed by WP4 and WP5).
These topics include, but are not limited to, the following:
Thematic Priority Proposed for WP6
European Reference Networks (ERNs)
Centres of Expertise and healthcare pathways
Objective of the Commission Communication underpinning this work:
5.1: Improving universal access to high-quality healthcare for rare diseases in particular through development of national/regional centres of expertise and establishing EU Reference network

Registries, databases and data collection (including quality, and access and sharing)
Objective of the Commission Communication underpinning this work: 5.11 : Registries and databases

Integration of RDs into Social Policies and Specialised Social Services
Objective of the Commission Communication underpinning this work: 5.2 Access to specialised social services

Genetic testing/Next Generation Sequencing; Genetic Counselling; neonatal screening, Primary Prevention of rare congenital anomalies
Objective of the Commission Communication underpinning this work: 5.9 Quality management of diagnostic laboratories; and
5.10 Primary prevention

Coordinated approaches to pricing and innovative mechanisms to improve access to rare diseases therapies, including HTA
Comprehensive information systems (Help-lines, information points)
Objective of the Commission Communication underpinning this work:
5.3 Access to Orphan Drugs
5.4 Compassionate use programmes
5.5 Medical devices
5.6 Incentives for Orphan Drug Development

Comprehensive information systems (Help-lines, information points)
Objective of the Commission Communication underpinning this work: Mentioned in 5.2, as above.

E-health
Objective of the Commission Communication underpinning this work: e-health

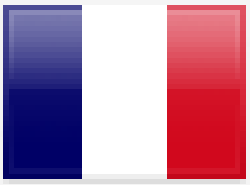
The WP may also explore, as deemed necessary by the Consultative Group, partners and the Expert Group, topics beyond the immediate scope of the

Commission Communication, where these are deemed to meet the changing needs of the field: suggested topics in this category include Best Practices / guidelines on diagnostics, Public Health Indicators and care and Methodology for assessing the Socio-economic Burden of Illness of Rare Diseases.
The precise scope of work in the above areas will form the basis of annual workplanning to reflect current and changing priorities over the course of the JA.

Task 6.1 : Implement a robust policy methodology to support the work of the Expert Group on Rare Diseases
Task Leader (Kate Bushby, UNEW) Contributors (MUW, FPS Health, WIV-ISP, BAPES, MoH CY, MoH Fr, NKCVO, INSERM, EURORDIS, UKF, INERP, OVGU, PTE, SE, OPBG, Veneto, ISS, VULSK, HDIR, Poznan University, DGS, UKCL, FISABIO-Salud P�blica, CIBER, ISCIII, WADOH via the Consultative Group)
Start date: M1 End date: M12
The partners in WP6 together with the Orphanet team and EURORDIS will be constituted to form a Consultative Group to support

COORDINATOR, LEADER CONTACT AND PARTNERS

COORDINATOR



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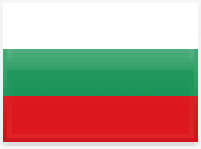

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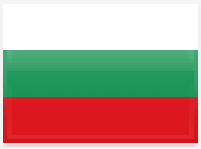

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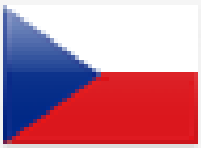

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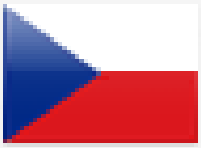

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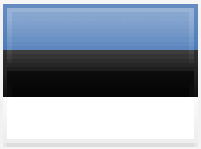

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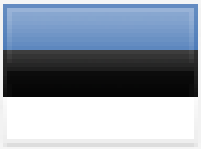

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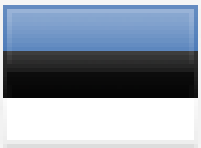

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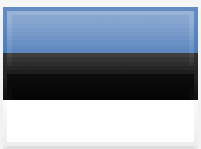

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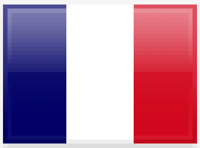

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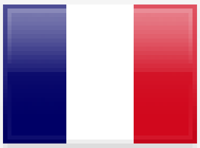

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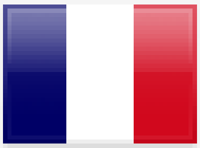

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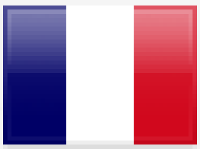

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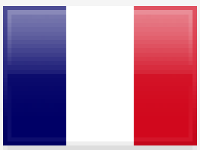

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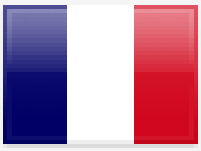

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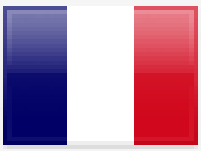

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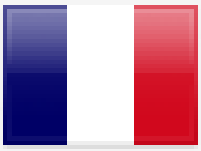

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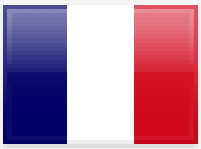

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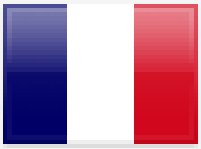

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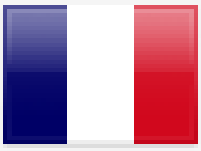

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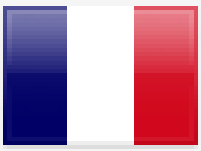

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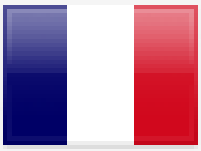

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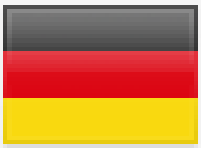

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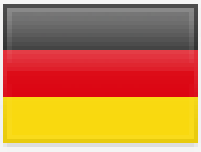

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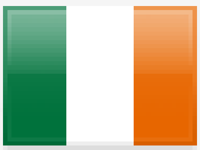

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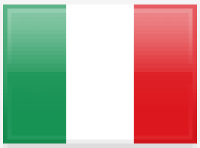

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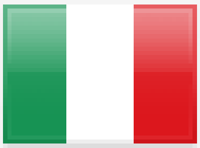

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Country: Norway
Website: http://www.inserm.fr

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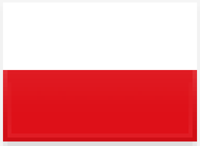

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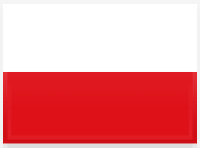

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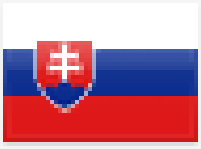

UNIVERZITA KOMENSKEHO V BRATISLAVE
Street: SAFARIKOVO NAM 6
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440
Country: Slovakia
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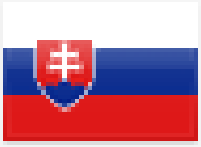

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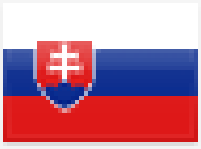

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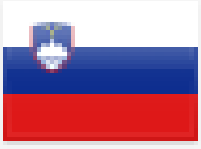

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OUTPUTS

Newsletter Orphanews

INSERM
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 19/06/2018
Produce 60 twice-monthly newsletters of the rare disease community

Layman version of the final report

INSERM
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 31/07/2018
This is a short (e.g. 10 pages) version of the final report, written for the interested public.

Final report

INSERM
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 03/12/2018
This report describes the project implementation and the results achieved. The deliverables are annexed.

Final dissemination report on Sustainable health systems for rare diseases

ISS
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 03/12/2018
Report on the final conference on health systems for rare diseases workshops

Evaluation of the deliverables compared to plans

MUW

Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 03/12/2018
This evaluation is based on the reports of WP leaders to the Steering committee M12,18,24,30,36

2017 Edition of the State of the Art Report

UNEW
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 03/12/2018
Annual Report on key developments in the field, at national and EU levels

Final report on policy delivery and implementation

UNEW
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 03/12/2018
This report will be structured to include specific updates on the thematic areas

Report on Resources to Support the Development and Implementation of Policies within ERNs

UNEW
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 03/12/2018
This report will include all key workshops outputs relative to ERNs

Dissemination of Two editions of the State of the Art Report

UNEW
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 31/07/2018
Dissemination of the Annual Report on key developments in the field, at

national and EU levels Delivered at M18&M30

An European integrated master file

DIMDI
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 31/07/2018
Table fine-tuned according to the test results and containing all information that was provided by member states over the project period

A set of coding helping tools for rare diseases

VR-IIBRD
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 31/07/2018
Electronic sheet and relational DB for the automatic coding of RD (experimentally for a group of RD)

Draft recommendation for routine maintenance

APHP
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 31/07/2018
APHP, VENETO, DIMDI Set of next steps which will help to merge the project results to a long-term maintenance which is stable and available for member states in the draft form of a recommendation.

Orphanet users'survey

INSERM
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 19/07/2018
Perform an online survey intended to website’s users M12,24,36

Orphanet nomenclature with mappings and annotations

INSERM

Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 19/06/2018
Inventory of RD names, classification, mappings to ICD10, OMIM, SNOMED CT, and others, links to genes, annotations with epidemiological data M1 to M36 monthly

Web-based knowledge management platform

INSERM
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 19/06/2018
Data management facility allowing for suggesting updates, to assess demands, to approve/reject, and to insert into the DB To be delivered at M12,24,36.

Orphanet DB versionning and differentials between versions

INSERM
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 19/06/2018
Produce annual versions with differentials between versions To be delivered at M12,24,36.

Annual updates of Orphanet knowledge base of expert resources

INSERM
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 19/06/2018
Mailing to professionals annually To be delivered at M12,24,36.

Orphanet Report Series

INSERM
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 19/06/2018
Thematic reports (frequency depending on each report) To be delivered at

M12,24,36

2016 Edition of the State of the Art Report

UNEW
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 21/08/2017
Annual Report on key developments in the field, at national and EU levels

Interim reports

INSERM
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 20/06/2017
This report describes the activities Carried out, milestones and results achieved in the first half of the project. Deliverables can be attached as annexes. Will be delivered at M12 and M24.

Reports on the external evaluation of Orphanet

MUW
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 19/06/2017
INSERM-DGS (FR) – MUW Reports summarizing the independent evaluations of Orphanet by French institutions and by MS representatives

Standard procedures and guide for the coding with Orpha codes

APHP
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 09/06/2017
To guarantee comparable datasets from MS, a coding procedure should be clearly defined

Annual meeting & meeting reports

INSERM

Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 07/06/2017
Meeting organisation and meeting report. Delivered M12&M24.

Progress dissemination report on health systems equity

ISS
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 07/06/2017
Report on the sustainable health systems for rare diseases preparatory work (workshop and working group discussions)

Sustainability plan for Orphanet core activities

MUW
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 07/06/2017
INSERM- DGS (FR) – MUW Development of a modular representation of Orphanet and elaboration of strategies for a secured legal framework and a sustainable funding of Orphanet in the EU

Progress report on policy delivery and implementation

UNEW
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 16/05/2017
This report will be structured to include specific updates on the thematic areas.

ECRD 2016

EURORDIS
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 12/09/2016
2016 sees the 8th year of the ever-growing European Conference on Rare Diseases & Orphan Products. This biennial conference is a unique opportunity

to come together and exchange: it is the event at which to connect and share with all other members of the rare disease community. The ECRD is the only event which, from its small beginnings, has united all rare disease stakeholders from all European nations- patients and patient representatives, healthcare professionals and researchers, industry, payers, regulators and policy makers alike- in the fight against rare diseases. The ECRD now brings together over 80 speakers and more than 800 participants, covering six themes of content over two days: from the latest research, to developments in new treatments, to innovations in healthcare, social care and support at the European, national and regional levels. New meeting formats to enhance the on-site learning experience will be showcased for the first time in Edinburgh and will include a speed networking session to connect patients, researchers and industry, an open-house “soap box” lunch session, interactive roundtables, audience polling, networking lunches / coffee breaks and dedicated poster sessions. The focus in 2016 will be on Game Changers in Rare Diseases. We will also be holding pre-conference tutorials, which will ensure that you are equipped to learn as much as possible from the conference.

Review document of existing technical implementations for RD coding of MS

APHP

Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)

Published on: 03/06/2016

Analyse at each MS level the situation for RD coding based on previous general work done through precedent JA. This work includes deep analysis of current and future situation. Existing systems in each country might already give satisfactory data from some RDs. Granularity analysis for further coding requirements.

JA Dissemination plan

EURORDIS

Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)

Published on: 18/04/2016

Elaborated dissemination plan indicating for each dissemination action : - what will be disseminated (key message) - to whom (audience) - why (purpose) - how (method) - when (timing

Leaflet

INSERM
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 26/02/2016
A leaflet to promote the project must be produced at the beginning

Website

INSERM
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 16/12/2015
Dedicated website set up and maintenance

Kick off meeting & report

INSERM
Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (RD-ACTION)
Published on: 10/11/2015
Meeting organisation and meeting report