RD ACTION

DATA AND POLICIES FOR RARE DISEASES

TAKING ACTION IN EUROPE TO MEET THE CHALLENGES FACED BY THE RARE DISEASE COMMUNITY

FINAL REPORT

A 3-year
European Joint Action
(June 2015 – June 2018)

34 beneficiaries
30 collaborating partners
40 countries

7 key achievements
32 deliverables

rd-action.eu

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ABOUT

WHAT IS A EUROPEAN JOINT ACTION?

A "Joint Action" is a collaborative action co-funded by the European Commission and Member States of the European Union on priority health issues, that deserve to be tackled at the European level. The objective is to elaborate joint proposals and recommendations aimed at providing a better support at European, national and local levels.

A Joint Action is essentially a large project divided into several sub-projects carried out by the different partners. EU Member States participate on a voluntarily basis and according to their specific interests in the topics covered.

WHAT IS RD-ACTION?

RD-ACTION (www.rd-action.eu) was a three-year Joint Action (JA) co-funded by the EU Health Programme, starting in 2015 and ending in June 2018. The JA was coordinated by Orphanet at the INSERM in France.
The JA aimed at ensuring an integrated European approach to the challenges faced by the Rare Disease (RD) community and at promoting, catalysing and triggering multi-stakeholder debates which are necessary both at European level, to build shared strategies, and at national level, to support the integration of EU policies on rare diseases across Member States.

RD-ACTION brought together 64 partners – academia, Ministries of Health, health institutes, medical universities and patients' organisations - from 40 countries, including all 28 EU Member States.

The strength of RD-ACTION lies in its multi-stakeholder integrated approach and its pan-European and well-structured coverage that included, but that was not limited to, the Orphanet network and EURORDIS (the European patient organisation for rare diseases).

RD-ACTION actively developed partnerships with other relevant RD initiatives and projects beyond RD community possessing a particular value and resonance for rare diseases:

- RD-Connect (integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research)
- JARC (EU Joint Action on Rare Cancers)
- Expand (deploying sustainable cross-border eHealth services in the EU)
- JASeHN (EU Joint Action on to support the eHealth Network)
- INNOVCare
- TO REACH project
- European Reference Networks
- EMA

KEY ACHIEVEMENTS

1

PROVIDING INFORMATION AND DATA ON RARE DISEASES

The Orphanet knowledge base (www.orpha.net; www.orphadata.org) has been funded by the European Commission since early 2000, mainly through Joint Actions, to generate data and knowledge on RD devoted to all the stakeholders, and necessary for policy decision making in a European collaborative manner.
The legacy of Orphanet following RD-Action in the field of rare diseases is threefold:

- **A standard common language for health and research** : the Orphanet nomenclature, (or ORPHA codes) has been improved by making it more easily **exploitable in information systems**. In addition partnerships have been developed, in order to ensure **that sources of data using different medical terminologies can talk to each other when aligned to the Orphanet nomenclature**: this is called "interoperability". Orphanet works with GARD (NIH - NCATS), SNOMED International, and the World Health Organization (ICD 11) to reach this goal. The Orphanet nomenclature exists currently in 9 languages: Czech, Dutch, English, French, German, Italian, Spanish, Polish and Portuguese. **This multilingual approach helps promote interoperability between Member States.**

- **A complete and reliable database for decision-making** (for both Member States and European Reference Networks (ERNs)). Scientific data have been produced for each rare disease (genes are associated to rare diseases, epidemiological and natural history data, and phenotypic traits using the Human Phenotype Ontology (HPO)). Expert resources in each of the 32 countries of the network have been collected and updated in order to provide patients and doctors with a mean to identify resources specific for RD : specialised expert centres and centres of expertise, medical laboratories and diagnostic tests, research projects, clinical trials, patient registries and mutation databases, networks, technological platforms and patient organisations. A cartographic representation of ERNs is also provided to facilitate their identification, data production and data collection procedures and traceability of curation decisions were made available.

- **A more user-friendly website providing information for professionals and patients.**

Behind the scenes, a more collaborative decision-making process, has been developed allowing procedures to be adapted to national/regional situations. An effective partnerships strategy has been developped to avoid duplication of work and potentiate synergies with other relevant projects (including ERNs)). A Quality Management System has been put in place at Orphanet to ensure a controlled continuous improvement cycle and a more user-oriented approach. Finally, concrete steps have been taken to ensure the sustainability of Orphanet: discussions are underway at the "Steering Group for Health Promotion and Disease Prevention", created by the European Commission, as to future financing models.

Task Leader: Orphanet (Inserm Service Unit 14, France)

orphanet
Inserm

A new EU-Project (RD-CODE) will implement ORPHA codes into routine coding systems in different MS, considering resources developed by WP5.

2

IMPROVING IDENTIFICATION OF RDs IN HEALTH INFORMATION SYSTEMS

Rare diseases are difficult to identify in health information systems, because the classifications commonly used for codification include only few RD. An adapted codification, with the correct nomenclature and shared guidelines, has the potential to improve the overall quality of collected data and in turn clinical care and outcomes. RD-ACTION supported the use of ORPHA codes, a stable rare disease nomenclature produced by Orphanet that is multi-lingual and cross-referenced with other medical terminologies with the final aim of improving the visibility and traceability of rare diseases patients' in health information systems, for instance in electronic medical records.

Integrating ORPHA codes into both health and research information systems is crucial for rare disease visibility, system interoperability and data sharing. Cross-referencing is a key step towards making different databases talk to each other.

Based on the European Commission Expert Group on Rare Diseases' (CEGRD) "Recommendation on Ways to Improve Codification for Rare Diseases in Health Information Systems', **RD-ACTION developed a tool set to assist European countries in implementing the ORPHA codes**, including **standard procedures**, and **practical guidance for integration, use and routine maintenance of the ORPHA nomenclature** in health information systems.

A preliminary survey of ORPHA codes use in MS was carried out, showing the value of supporting MS interested in adopting ORPHA codes through the development of common resources and guidelines.

ORPHA codes were tested in real world coding settings, thus proving their consistency and ability to increase RD visibility.

Task Leaders:

- DIMDI (German Institute for Medical Documentation and Information), Germany
- BNDMR (French rare disease registry), AP-HP, France
- Veneto region RD registry, Italy

European Reference Networks

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ERNs SUPPORT

Due to the large number of RDs (over 6,000) as well as the scarce and scattered expertise concerning these diseases, European Reference Networks (ERNs) are seen as the way forward to bring together expert healthcare care professionals, patients, researchers across European countries via a virtual network, with a view to improve timely and accurate access to diagnosis and care. 24 ERNs covering altogether all rare diseases and rare cancers were approved and officially launched on 1 March 2017.

RD-ACTION concentrated many efforts to support the establishment and future development of these ERNs. RD-ACTION partners led by the team of UNEW provided the broad RD field with the following support:

- A workshop to build capacity amongst healthcare professionals and identify possible coordinators of future ERNs, based around the **groups of diseases** defined in the Addendum to the EUCERD Recommendations on ERNs for RDs.

- A **matchmaker tool** was created to allow cross-talk between healthcare centres interested in joining or leading an ERN. The Matchmaker tool received 801 responses in total, and **helped to ensure 24 collaborative and (crucially) non-competitive proposals.**

Meaningful patient involvement in ERNs was promoted. In early 2016, EURORDIS established European Patient Advocacy Groups (**ePAGs**), bringing together patient organisations by groups of diseases, as defined in the Addendum to the EUCERD Recommendations. RD-ACTION ensured that ePAGs were always involved in its activities and workshops. The ePAGs participated in the development of ERNs' applications. Following ERNs' establishment, they are members of the ERNs Board, Steering Committee and Task Forces.

Throughout the duration of RD-ACTION, six major workshops were organised to address a particular policy area or topic in which the ERNs could add value and capitalise on past investments and lessons learned. Many of these workshops were co-organised by DG Santé and involved over 360 multi-stakeholder experts. All these six workshops led to reports, concept papers and Recommendations that are accessible here.

Task Leader: University of Newcastle, UK

Newcastle University

KEY ACHIEVEMENTS

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STATE OF THE ART OF RARE DISEASE POLICIES IN EUROPE & BEYOND

The State of the Art of Rare Diseases provides a broad outline of the history and status quo of rare disease activities across Europe, with an emphasis on topics such as the RD legislative and policy framework, European Commission investments, achievements of the most relevant funded initiatives (e.g. Joint Actions) and research activity. It also provides an overview of the national situation in each EU Member State as well as some other countries.

Within RD-ACTION, the format of the State of the Art was revamped and the consultation process revised for the national section. For each country, three major actors were systematically invited to answer the same online questionnaire to ensure an exhaustive collection of data:

- Ministries of Health;
- Orphanet country partners;
- and National Alliances for rare diseases.

The 2018 edition of the State of the Art on rare diseases can be found here.

Task Leader: University of Newcastle, UK

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SUPPORT TO SUSTAINABLE AND RESILIENT HEALTH SYSTEMS

In line with the Communication of the European Commission and Recommendation from the EU Council on RDs, work was carried out within RD-ACTION to support national authorities to quantify the burden of RDs and identify available resources for sustainable and resilient health systems, taking into account the principles of equity, quality and efficiency, and involving stakeholders, policy makers and civil servants in charge of national RD plans or strategies. Within this work, six Policy Briefs have been released:

1. Resilience
2. Actions on Educational Programmes and Training for Professionals
3. Primary Prevention as an Essential Factor Ensuring Sustainability of Health Systems: the Example of Congenital Anomalies
4. Health Systems for Rare Diseases: Financial Sustainability
5. Integrated Care
6. Patient Empowerment

Task Leader: National Centre for Rare Diseases - Istituto Superiore di Sanità, Italy

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FOSTERING NATIONAL RD PLANS/STRATEGIES

The EUROPLAN National Conferences are the tool designed to promote the adoption and implementation of national plans or strategies for rare diseases in European countries based on the Recommendation of the EU Council on an action in the field of rare diseases of 8 June 2009, as well as to facilitate the integration of EU rare disease policies and recommendations into the national health and social systems (for example on Centres of Expertise and ERNs, use of ORPHA codes, patient registries, research, access to orphan drugs, access to adapted social services for RD patients, etc.).

EUROPLAN National Conferences are jointly organised in each country by a National Alliance (NA) of rare disease patient organisations and EURORDIS. Within RD-ACTION, 19 EUROPLAN National Conferences took place in 15 EU Member States plus Macedonia, Serbia and Ukraine (Belgium organised two conferences).

These conferences had previously taken place in the scope of the EU co-funded project, EUROPLAN (2008 – 2011) and in the EUCERD Joint Action (2012 – 2015). In 2009, only five EU Member States had adopted a national rare disease plan (Bulgaria, France, Greece, Portugal and Spain).

Between 2010 and 2018, altogether 59 EUROPLAN National Conferences took place in 25 EU Member States, as well as in Georgia, Macedonia, Russia, Serbia and Ukraine. Some National Alliances organised several EUROPLAN conferences in their country. As a result, by the end of 2018, **25 EU Member States had put in place a national plan or strategy for rare diseases.**

The outcomes of these conferences have undoubtedly contributed to the adoption of national plans or strategies in many European countries.

The strength of a EUROPLAN national conference lies in its shared philosophy and format. The conferences are patient-led, multi-stakeholder, exhaustive (all strategic areas are covered) and integrated (participants use the conferences as an opportunity to assess the integration of EU regulations, policies and recommendations into national health and social systems).

Task Leader: EURORDIS

EURORDIS
RARE DISEASES EUROPE

26-28 May 2016 Edinburgh
European Conference on Rare Diseases & Orphan Products

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ECRD 2016

The European Conference on Rare Diseases & Orphan Products – ECRD - is a biennal event organised by EURORDIS. Within the framework of RD-ACTION, the 8th edition of ECRD took place in Edinburgh, on 26-28 May 2016. The conference focused on "game changers in rare diseases". It attracted over 800 participants from 48 countries.

One of the key successes of ECRD is to involve the entire rare disease community to share experiences and perspectives: patients' representatives, academia, healthcare professionals, industry, payers, regulators and policy makers. The conference analysed the game changers for rare diseases in the field of research, diagnosis, drug development and access, care provision, social policy and looked at the global dimension of rare diseases.

The conclusions of ECRD 2016 helped inform EU policies impacting on rare diseases and RD-ACTION. Please find the summary report here.

Task Leader: EURORDIS

The ECRD website provides information on previous and future conferences : www.rare-diseases.eu

PARTNERS

RD ACTION
DATA AND POLICIES FOR RARE DISEASES

DESIGNATED PARTNERS

- Medical University of Vienna (MUV)
- Federal Public Service Health (FPS Health) Scientific Institute for Public Health (WIV-ISP)
- Bulgarian Association for Promotion of Education and Science/Rare Diseases Institute
- Croatian Alliance for Rare Diseases
- Coordination Center for Rare Diseases in University Hospital in Motol (NKCVO)
- University of Tartu (UT)
- Rinnekoti Foundation Norio Centre (Rinnekoti)
- Institut National de la Santé et de la Recherche Médicale (INSERM)
  Direction Générale de la Santé, Ministère des Solidarités et de la Santé (DGS)
  Assistance Publique - Hopitaux de Paris (APHP)
- Medizinische Hochschule Hannover (MHH)
  Deutsches Institut für Medizinische Dokumentation und Information (DIMDI)
- Ministry of Health
  Orszagos Tisztifoorvosi Hivatal
  Semmelweis Egyetem (SE)
- Health Service Executive (HSE)
- Istituto Superiore di Sanita (ISS-CNMR)
  Regione del Veneto
  Ospedale Pediatrico Bambino Gesù (OPBG)
- Center For Disease Prevention and control of Latvia
- Vilnius University Hospital Santariskiu Klinikos (VULSK)
- Leids Universitair Medisch Centrum (LUMC)
- The Norwegian Directorate of health (HDIR)
  Norwegian National Advisory Unit For Rare Diseases (NKSD)
- Instytut Pomnik - Centrum Zdrowia Dziecka (IPCZD)
- Directorate-General of Health (DGS)
- University of Medicine and Pharmacy"GR.T.Popa" Iasi Orphanet Center
- Faculty of Medicine in Bratislava (UniBA FoB)
- University Medical Centre Ljubljana (UKC Ljubljana)
- Centro de Investigación Biomédica en red (CIBER)
- Karolinska University Hospital (Karolinska)
- University of Newcastle (UNEW)
  UK Department of Health (UK PHE)
- European Organisation for Rare Diseases (EURORDIS)

DESIGNATED COLLABORATING PARTNERS

- Austrian Health Institute (GÖG)
- Ministry of Health
  Medical University Sofia
- Croatian Institute of Public Health (HZJZ)
- Ministry of Health
- Robert Koch Institut (RKI)
  Universitatsklinikum Frankfurt (UKF)
- Fehlbildungsmonitoring Sachsen-Anhalt an der Medizinischen Fakultät der Otto-von Guericke-Universität Magdeburg
- Institute for Research of Regulatory policies (INERP)
- Pécsi Tudományegyetem (PTE)
- Landspitali University Hospital Ragnar
- Directorate of Health (Ministry of Health)
- Ministry for Energy and Health (MEH)
- Poznan University of Medical Sciences
- Instituto de Salud Carlos III
  Fundación para la Investigación Sanitaria y Biomédica de la Comunidad Valenciana

COLLABORATING PARTNERS

- Ministry of Health
- Center of medical genetics and primary health
- Office Population Health Genomics, Department of Health Government of WA
  Garvan Institute of Medical Research
- Institute of Health Information and Statistics of the Czech Republic
- University Hospital of Aarhus
- Foundation for Genetic and Rare Diseases (GeRaD)
- Institute for Rare Diseases, Institute of Medical Genetics
  The Chaim Sheba Medical center
- Institut National d'Hygiène, Department of Medical Genetics
- Mc Gill University
- Belgrade University
- Geneva University Hospital
- Service de Cytogénétique et de Biologie de la Reproduction, CHU Farhat HACHED, Sousse
- University of Istanbul

Contact us: project.management@rd-action.eu

OUR MESSAGE TO THE GLOBAL RARE DISEASE COMMUNITY

At a time when the achievements of past years of successful rare disease policy are coming to fruition and new challenges and opportunities arise, **we need to accelerate the momentum we have built over the last 20 years to ensure no single person living with a rare disease is left behind.**

We need a **strong drive from EU institutions. RD-Action partners call on the European Commission to reaffirm its leadership in an area where it is unanimously recognised that the EU added value is very high,** by means of:

- A **dedicated policy for rare diseases** that, while creating synergies with other policy areas, effectively addresses the specificities of rare diseases;
- An overarching approach that **integrates rare disease policies within a successful, harmonious and consistent framework;**
- A **multi-stakeholder approach and collaborative strategies** that have proved key to the success of the actions of Europe in the fight for rare diseases;
- A rare disease community equipped with the **most appropriate tools for exchange and policy-making** for the years to come.

For more information about our objectives, achievements and partnerships, consult: www.rd-action.eu

rd-action.eu

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