

How has RD-ACTION supported the conceptualisation and implementation of ERNs?

RD-ACTION (2015-2018) inherited the mandate of the previous Joint Action for Rare Diseases (the EUCERD Joint Action) – i.e. supporting the rare disease (RD) field in adding depth and clarity to the concept of an ERN and preparing the way for implementation of the Networks. Through its Policy and Integration WP (6 - led by Newcastle University) in particular, RD-ACTION has supported the ERNs by organising meetings and workshops and seeking to create policies and guidance *with* the Networks, *for* the Networks.

HANDS-ON SUPPORT TO ENSURE COLLABORATIVE AND NON-COMPETITIVE ERN PROPOSALS

- In preparation for the first Call for ERNs, RD-ACTION organised a major workshop (Summer 2015) to build capacity amongst field leaders and assist the RD community in organising itself around the 21 broad thematic groupings identified by this same team and adopted by the Commission Expert Group on Rare Diseases as the backbone for ERN structure and scope.
- As many experts expressed a desire for a means of identifying Healthcare Providers (HCPs) interested in setting –up/joining an ERN within the same disease area, the RD-ACTION team designed a ‘**Matchmaker tool**’. Launched in December 2015, the Matchmaker ran until May 2016. This tool allowed the specialists to make contact and align intentions. In total, the Matchmaker had received **801** responses across the **21** thematic groups.

ENSURING MEANINGFUL PATIENT INVOLVEMENT IN ERNs

- The European Patient Advocacy Groups (ePAGs) were conceived by EURORDIS, as part of its contribution to RD-ACTION. The purpose is to ensure patients are fully integrated in the development, governance and operations of the ERNs.
- From 2016 onwards, open elections were held to agree formal permanent representatives for each of the 24 ERNs. Over 150 individuals have been appointed as ePAG members, tasked with representing all patients under their broad Thematic Grouping (e.g. rare cardiac), and serving on ERN Boards, committees and task forces.

POLICY SUPPORT FOR ERNs

The ERNs represent a great opportunity to embed good practices and disseminate these into broader healthcare systems. RD-ACTION’s vision was that, as ERNs were established & evolved, dedicated guidance would be important to support but also to ensure a baseline compatibility and interoperability (at many levels) *between* the ERNs. RD-ACTION partners thus developed a workplan for the years 2016-2018, designed to capitalise on the lessons learned in the broader RD field and ‘pilot’ Networks and bring these to the ERN stakeholder community, to agree together how to address shared challenges. By the end of the project, 6 major workshops have been delivered (many co-organised with DG Sante) each addressing a particular policy area in which consensus building was deemed important.

Workshops:

Exchanging data for virtual care in the ERN framework – 27-28 September 2016, Brussels: With over 55 participants, this workshop:

- ✓ Agreed practical advice to enhance the efficiency and utility of virtual consultations;
- ✓ Clarified the legal issues around data protection –especially in view of the new General Data Protection Regulation- and the legal, ethical and social issues relating to consent for the sharing of data in the ERN framework;

- ✓ Brainstormed how patients will enter/ be 'referred' to the ERN for virtual care
- ✓ Shared experiences on the standardisation of data in the RD field, to identify good practices which should be embedded in the ERNs
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Using standards and embedding good practices to promote interoperable data sharing in ERNs – 26-27th April 2017, Brussels: 68 participants (a mixture of ERN Coordinators, EURORDIS and Orphanet partners, data/eHealth specialists, Coding experts, Phenotype ontology experts, data linkage experts, and DG Sante representatives) met to agree how best to capture data collected in the ERNs for care purposes.

- ✓ After previously identifying the value of using the ORDO and the HPO -deemed most sensitive and appropriate ontologies for RD- participants had expressed a desire to learn more about how to use these sorts of tools practically, to optimise the use and re-use of data collection in the ERN context.
- ✓ This was followed by a session on linking data, especially through the concepts of FAIR data and the 'PPRL'.
- ✓ The workshop generated a list of recommended good practices, to enhance the use and reusability of data collected in the operations of ERNs, and created 'tool-kits' on how to practically use the most relevant of these data resources/practices.

Indicators and Outcomes for ERNs - 1-2 June 2017, Newcastle: This workshop united 40 participants to work closely on issues related to the impact and monitoring of ERNs. It allowed the ERN community to:

- ✓ better understand terminology around indicators and become acquainted with the different types of indicators (structure, process, outcome) and their use in health systems
- ✓ to discuss an initial set of indicators, common to all ERNs, for the purposes of monitoring impact, and isolate the definitions needed to advance this selection
- ✓ to identify key issues and challenges on data collection and reporting
- ✓ to elucidate the challenges in selecting clinically-oriented indicators in the rare disease/highly specialised healthcare field

How can ERNs generate, appraise and utilise clinical practice guidelines, to enhance the impact of consensus guidelines in national health systems? 6-7 Dec 2017, Rome: This workshop united 63 participants to discuss for the first time the various ways in which ERNs might add value to a host of activities concerning Clinical Practice Guidelines (CPGs). The group sought to identify good practices which could be shaped into 'recommendations' concerning several aspects of this vast and complex topic, including:

- ✓ methodological approaches to the generation and appraisal of CPGs;
- ✓ strategies for engaging with key stakeholder groups, such as patients and learned societies to partner in all CPG activities
- ✓ highlighting financial issues and time-commitments of guideline activity, proposing strategies to address the need for ethical engagement with stakeholders such as Industry
- ✓ options to engage national authorities, to ensure that CPGs emerging from ERNs can actually be used 'on the ground' in countries
- ✓ outlining concrete future activities needed to support the ERNs in their CPG-related tasks

Creating a Sustainable Environment for Holistic & Innovative Care for Rare Diseases & Complex Conditions - 12-13 April 2018., Frambu, Norway: This workshop, hosted at Frambu Resource Centre, was organised jointly with the INNOVCare initiative. The event united 67 participants from 22 countries, with diverse backgrounds. The workshop addressed many issues, including the following:

- ✓ The state of the art in terms of integrated, holistic approaches to care for people with rare diseases
- ✓ How best to enhance and expand identified good practices to support integrated and holistic care
- ✓ How ERNs -or, sometimes more appropriately, the Healthcare Providers of which they are composed) might add value in this arena

- ✓ The workshop participants proposed practical actions which could be feasible for ERNs and their constituent HCPs (essentially centres of expertise), for instance concerning patient empowerment and involvement; identifying the full medical and societal burden of rare diseases; creating high quality information resources and 'setting the standard' for the types of multidisciplinary, holistic care that patients with a given conditions should receive; etc.

How ERNs can add value to clinical research in rare diseases and highly specialised domains - 29th -30th May 2018, London:

This workshop is being hosted by the European Medicines Agency: the EMA is a co-organiser this time, along with DG SANTE (and RD-ACTION of course). The workshop will unite 64 participants, and has several ambitious goals:

- ✓ To share the state of the art of tools and resources which exist in 2018 to streamline and optimise each 'point' in the clinical research pipeline
- ✓ To better understand the priorities and needs of the ERN community specific to clinical research, and explore case studies
- ✓ To elucidate the services and opportunities offered by the EMA which are of relevance to clinical research in rare and highly specialised domains
- ✓ To identify concretely *how* and *where* ERNs could make a positive difference to each 'timepoint' in the clinical trial pathway, including points of engagement specifically with the EMA, to agree a roadmap to a more strategic and streamlined collaboration in future.

In addition to these large, ERN-focused workshops, RD-ACTION Policy & Integration Work-Package has organised several meetings and workshops in 2015 and 2016, **designed to build synergies between the ERN and rare disease community on the one hand, and the eHealth field on the other** (see <http://www.rd-action.eu/ehealth-and-european-reference-networks/>)

SAMPLE OUTPUTS:

The workshops are a key part of the project's activities in the ERN sphere, but WP6 has been active in uniting and supporting the Networks in other ways, as evidenced from the following sample of outputs:

- [Concept paper on convergence of eHealth and Rare Disease initiatives \(2016\)](#)
- Report on [1st meeting between the ERN Board of MS and the future Coordinators \(Sept '16\)](#)
- [Highlights and conclusions from Workshop on Exchanging data for Virtual Care](#) (Sept '16)
- [Identifying ERN requirements for an IT platform](#) (Aug '16 exploration of IT needs and correlation to published CPMS Tender)
- Canvassing of ERN plans and perspectives re. Registries (Nov '16) and [Analysis of the key issues concerning ERNs & Registries](#)
- [Report on Activities of the JA Task-Force on Interoperable Data-sharing in the framework of the operations of ERNs](#)
- [Canvassing perspectives on ERNs and Research](#) for Maltese Presidency event (March '17)
- Draft Annotated Table of core indicators for ERNs (Summer '17) (subsequently refined by a dedicated ERN Working Group)
- [Tool-Kits on resources for standardising data](#) (Summer '17)
- [Recommended Practices for Data Standardisation in the Context of the operation of ERNs](#) (Sept '17)
- [Recap of Breakout Discussions from the workshop on Integrated and Holistic Care](#) (April '18)

The home page for all these pages/document is <http://www.rd-action.eu/european-reference-networks-erns/>