**RD-ACTION Workshop Co-hosted by DG SANTE: Using standards and embedding good practices to promote interoperable data sharing in ERNs**

**April 26-27th, Brussels**

**Venue: Albert Borschette Conference Centre (CCAB), rue Froissart 36, 1040 Brussels**

**Timing of the Workshop:**

This workshop will immediately follow the Official DG SANTE **meeting of the ERN Coordinators**, which will be held on Wednesday 26th April 10am-4.30pm.

**The workshop, therefore, will commence at 4:30pm on 26th and Day 1 sessions will run until 6pm. The venue for Day 1 will be Room 4C.**

**On Day 2, Thursday 27th, the workshop will run from 9am- 5.45pm. Day 2 sessions will take place in the same building (CCAB, above) but please note that the room will be 0C**

**Aim of the Workshop:** The overall aim of the workshop is to analyse and demonstrate how ERNs and their associated experts can **use standards** to optimise the utility and reusability of clinical data, by sharing the state of the art of rare disease data standardisation practices.

**Ethos of RD-ACTION workshops:** A key objective of the RD-ACTION Policy WP workplan is to continue to provide support to the rare ~~disease~~ community in conceptualising, implementing and evolving robust ERNs capable of meeting the needs and expectations of people living and working with conditions requiring a specific concentration of expertise. As the 1st ERNs are established and evolve, shared consensus guidance is important to support the Networks but also to ensure a baseline compatibility and interoperability (at various levels) between the ERNs.

**Context and Overall Objectives of this workshop:**

ERNs are first and foremost dedicated to care. Once established, and connected by a dedicated SaaS for a clinical patient management platform, the Networks will support the exchange of knowledge and expertise between healthcare providers (HCPs) operating at the top of their game. It is important to emphasise that wherever possible (and appropriate), expertise will travel rather than the patients themselves. Data therefore sits at the centre of this, and as demonstrated in the September 2017 workshop ‘Exchanging data for virtual care within the ERN Framework’ ’ **there are certain standards, ontologies and principles one can apply to rare disease data in particular, to optimise its value by making it interoperable and able to serve a multitude of additional purposes,** besides enabling a virtual consultation for the most complex patients coming to the attention of the ERN.

**ERN communities will be key generators and handlers of rare disease data** -in the everyday functions of HCPs, and not only in relation to the SaaS for a Clinical Patient Management system- **and as such it is important to raise awareness of the potential to embed ‘approved’ and highly recommended rare disease ontologies for the capture and exchange of certain types of data, and to demonstrate on a more practical level how this can be done.** Therefore, in line with the mission of RD-ACTION[[1]](#footnote-1) this is conceived as a follow-up to the September Brussels workshop, to enable experts to  demonstrate how best to use Orphacode and HPO -and other standards deemed important by the ERNs- to optimise the value of ERNs’ (primarily clinical) data. Building upon the discussion from the workshop on ‘Exchanging Data for Virtual Care within the ERN Framework’, this is also a crucial moment at which to update the ERN communities on the good practices highlighted around the pseudonymization of patient data (the concept previously known as a GUID or PUID, which has now finally been agreed at the global level[[2]](#footnote-2) as a PPRL ort Privacy-Protecting Record Linkage); the progress concerning the Informed Consent form activities; and how to make clinical data ‘FAIR’ in line with the data stewardship principles now considered highly relevant and influential in the broader fields of data usage.

An additional, related focus of this workshop will be to clarify how the Networks might, in time, engage with the new Orphanet knowledge management system developed by the Australian partners in RD-ACTION, to:

1. contribute to the updating/future population of the Orphanet database (especially the epidemiological information concerning relevant rare diseases)
2. utilise the resources of the new Orphanet portal to optimise and add value to core ERN operations

**Expected Outputs of the Workshop:**

* **Validated Consensus recommendations on standardising clinical data in the framework of ERNs**
* **Workshop report with PwPs:**
* **Several ‘toolkits’ with relevance to the whole rare disease & highly specialised care community , but with particular emphasis on the ERN community of 900 HCPs:**
* ‘Practical support and steps to use and improve the OrphaCode’
* ‘Practical support and steps to use and improve the Human Phenotype Ontology’
* Recommendations for experts wishing to curate the Orphanet knowledge management platform
* FAIR-ifying clinical data –information and a case study

**Draft Agenda**

**Wednesday 26th 4.30-6pm (Room 4C)**

**Session 1: Sharing data in ERNs – clarifying the status quo and the scope of this workshop**

**16:30-17:00 Welcome and Conclusions of the RD-ACTION workshop on ‘sharing data for virtual care in the framework of ERNs’ re. ‘optimising data’:**

* To begin with a summary of what we know at present regarding the realities of the CDMP (based upon published specifications and official communications only)
* To include presentation of the draft Recommendations (disseminated ahead of time)

[Presentation Here](https://www.dropbox.com/s/apf9qyuvfx04mrn/Sharing%20data%20in%20ERNs%20%E2%80%93%20clarifying%20the%20status%20quo%20and%20the%20scope%20of%20this%20workshop%20%28V%20Hedley%2026.4.17%29.pdf?dl=0)

**17:00-18:00 SoA of data-sharing for care in ERNs: Discussion session**

(Geared around a few key questions, to clarify the different aspects of the debate around sharing clinical data, reusing clinical data, what people plan to do with this data (i.e. what *questions* can it answer for them and what purposes can it serve)).

*Day 1 ends at 18:00*

**Thursday 27th 9am-5:45pm (Room 0C)**

**Session 2: 9-11:30 Using the Orphanet nomenclature and demonstration of the new Orphanet Knowledge Management system (OKMS) (Ana Rath, Annie Olry, Marc Hanaeur, Montserrat Alfaro)**

(Practical interactive session, to include demonstrations and discussions on using the Orphanet nomenclature, how disease areas can improve the codes for their field, how the new OKMS works, and its potential to support ERN work)

* Why the OrphaCode is the most appropriate nomenclature for coding rare diseases (refresher presentation)
* How to use the OrphaCode/ORDO (practical demonstrations)

o To include: how to find codes; how to identify diseases

* How to improve the classification

o Discussion – where are the gaps for your ERN scope/disease area and how can communities work with Orphanet to improve the classification where needed

[Presentation Here](https://www.dropbox.com/s/fp8f29fek83ebl5/Orphanet%20Presentations%20-%20RD-ACTION%20Workshop%2027.4.17.pdf?dl=0)

***Coffee Break: 10:30-10:45***

* Demonstration of the new Orphanet Knowledge Management system (interactive demonstration plus discussion)

o How the new system works and what it can do to support your work

o How might ERN communities take ownership of curating this platform, where desirable, for a particular group of diseases?

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**Session 3: 11:30-12:45- The Clinical Potential and practicalities of using HPO**

Practical interactive session, demonstrating why the HPO has been recommended as the optimal ontology for clinical (phenotypic) descriptions in RD field; how it makes data semantically interoperable; **the different ways to use it, depending on your purpose** (broadly speaking, structured form options for research and free text for clinical summaries and descriptions)

**Why the HPO has been recommended as an optimal ontology for clinical (phenotypic) descriptions** (**Melissa Haendel** and **Sebastian Kohler**) demonstrations and discussions, to include:

* the key things people need to know about HPO and how it works
* the progress to-date in *tailoring* terms to groups of rare diseases/diseases areas
* how to improve the ontology for your ERN’s purposes
* the benefits of using HPO for research (i.e. in a structured dataset)

[Presentation Here](https://www.dropbox.com/s/blxvx5r11cmvlwm/HPO%20Presentations%20%28Haendel%20and%20Kohler%29%20RD-ACTION%20Workshop%2027.4.17.pdf?dl=0)

**12:45-13:45 - Lunch (not provided, but expenses will be reimbursed)**

**(HPO session cont.)**

**13:45- 14:30 How to use HPO-ready tools to generate interoperable free-text clinical summaries** (demonstration by **Tudor Groza**, with space for questions and discussions)

**Session 3: State of the art in linking data**

**14:30 -15:45 FAIR Data concept – what is FAIR data, what does this mean in the ERN context, and why should we aim for this?** (Practical, Interactive demonstration of what ERNs need to do here to make their data FAIR) (**Marco Roos and Claudio** **Carta**) [Presentation Here](https://www.dropbox.com/s/slqfoe2gf0mr5xx/Making%20FAIR%20Data%20in%20the%20ERN%20framework%20%28Roos%20and%20Carta%29%20RD-ACTION%20Workshop%2027.4.17.pdf?dl=0)

***Coffee break 15:45-16:00***

**16:00-16:45 Generating and using Identifiers or ‘Privacy Preserving Record Linkage (PPRL)’ systems, to link patient data**

* **David van Enckevort** - presentation on the PPRL work of the joint IRDiRC & GA4GH Task-Force and the selected demographic data elements. [Presentation Here](https://www.dropbox.com/s/xd8o99bhcl4gq5h/PPRL%20%28David%20van%20Enckevort%29%20RD-ACTION%20Workshop%2027.4.17.pdf?dl=0)
* **Günter Schreier**- Demonstration of the EUropean Patient IDentity (EUPID) Pseudonymisation and privacy preserving record linking service. [Presentation Here](https://www.dropbox.com/s/y4x2kxv3jh7nzsq/EUPID%20Presentation%20%28Gunter%20Schreier%29%20RD-ACTION%20Workshop%2027.4.17.pdf?dl=0)

**Session 4: Identifying further standards for use in the ERN framework**

16:45-17:15 (What else do the ERN experts propose we highlight in the Recommendations, standards-wise, to meet the specific needs of data in their field of highly specialised care/rare diseases) e.g. OMIM for genetics, LOINC for lab reports, systems for quantifying MRI images etc.?)

**17:15 –17:45 Final Discussion and Clarification of practical outputs** (esp. the Recommendations)

***17:45: Workshop Ends***

1. Objective 1. *Support the development and sustainability of the Orphanet database;*

   Objective 2. *Contribute to solutions to ensure the appropriate codification of rare diseases in health information systems across Europe;*

   Objective 3. *Work on priority issues for people living with rare diseases by implementing the actions identified in the ‘EU Council Recommendation on an action in the field of rare diseases’, ensure the sustainability of these actions, and support the work of the European Commission Expert Group on Rare Diseases* [↑](#footnote-ref-1)
2. That is to say, through the dedicated Task-Force of the IRDiRC (International Rare Disease Research Consortium) and Global Alliance 4 Genomics and Health (GA4GH) [↑](#footnote-ref-2)