EUCERD
European Union Committee of Experts on Rare Diseases

EUCERD CORE RECOMMENDATIONS ON RARE DISEASE PATIENT REGISTRATION AND DATA COLLECTION

TO THE EUROPEAN COMMISSION, MEMBER STATES AND ALL STAKEHOLDERS

5 JUNE 2013

BACKGROUND TO THE RECOMMENDATIONS

These recommendations have been based on the outputs of various multi-stakeholder meetings, consultations of the EUCERD and previous publications, including:

- Gliklich RE, Dreyer NA, eds. *Registries for Evaluating Patient Outcomes: A User's Guide. 2nd ed.* (AHRQ, September 2010)
- RDTF: *Patient Registries in the Field of Rare Diseases*, Apr 2009, updated Jun 2011
- EMA/ EUCERD: *Towards a Public-Private Partnership for Registries in the Field of Rare Diseases*, Workshop Report, London, 4 Oct 2011
- EURORDIS/ CORD/ NORD: *Joint Declaration of 10 Key Principles for Rare Disease Patient Registries*, Nov 2012
- EUCERD Joint Action: *Workshop Report on Rare Disease Registration*, Luxembourg, 13 Nov 2012, and drafting group and breakout session discussions (29- 30th January 2013)
- Joint EBE-EuropaBio Task Force on Rare Diseases and Orphan Medicines: *Position Paper for Rare Diseases and Orphan Drugs Registries and Databases*
- EPIRARE Rare Disease Registry survey
- ENCePP E-Register of Studies Guide

It is generated for the review of EUCERD as the basis for producing a set of EUCERD recommendations around governing principles in the field of rare disease (RD) registration and data collection. RD registries should be organised around population health needs and/or RD or groups of RD rather than around a therapeutic intervention. To improve diagnosis, treatment and care of RD, some form of data collection in all MS should be a target and ways to share these data when needed should be in place. Ideally, part of these data should be collected systematically and similarly in all MS. Such collections would together form valuable registries serving many RD-related purposes.

These recommendations were adopted at the 8th Meeting of the EUCERD on 5 June 2013 by consensus.

NOTE FOR CLARIFICATION:

A patient registry is an organised system that uses observational study methods to collect uniform data (clinical and other) to evaluate specified outcomes for a population defined by a particular disease, condition, or exposure, and serves one or more predetermined scientific, clinical, or policy purposes. A registry database is a file (or files) derived from the registry (Gliklich RE, Dreyer NA, eds. *Registries for Evaluating Patient Outcomes: A User's Guide. 2nd ed.* (AHRQ, September 2010)). It is usual to distinguish between population-based registries, which refer to a geographically defined population and aim to register all cases in that population, and non-population-based registries based on clinical centres or other criteria (members of a patient organization, participants registered via an ERN or other disease-specific registry etc.) where the population coverage may not be comprehensive. These types of registry have different uses but both are useful provided they serve identified target aims. Both types of registry are the targets of these recommendations. Multiple RD registries (>600) already exist in Europe (Disease Registries in Europe, Orphanet Report Series, Rare Diseases Collection, January 2013). The key principles proposed apply also to these existing datasets as they adapt to the changing environment for registries in a European and international context. The current recommendations for the basic principles underlying RD registration should take as a starting point generally accepted guidelines for registry development not revisited here (such as "Registries for evaluation of patient outcomes: a user's guide", 2nd edition, AHRQ).

EUCERD CORE RECOMMENDATIONS ON RD PATIENT REGISTRATION AND DATA COLLECTION TO THE EUROPEAN COMMISSION, MEMBER STATES AND ALL STAKEHOLDERS

1. RD patient registries and data collections need to be internationally interoperable as much as possible and the procedures to collect and exchange data need to be harmonised and consistent, to allow pooling of data when it is necessary to reach sufficient statistically significant numbers for clinical research and public health purposes.

1.1 They should use international standards and nomenclature to code the tentative or final RD diagnosis. Either the OMIM code or the Orpha codes are recommended alongside any other coding system in operation in the MS health systems, such as ICD and SNOMED-CT, with a view to establishing a common semantic approach.

1.2 There should be adoption of a minimum common data set across RD that registries should collect, in collaboration with global initiatives, to allow the establishment of national and/or European RD population registries, which have the potential to collect data on all RD patients.

1.3 A minimum common data set should be defined, and supported with a semantic approach and Standard Operating Procedures. Interoperability (via means of mapping) of registry specific data sets towards this common data set should enable comparison across all RD and internationally.

1.4 For disease-specific registries, appropriate core data sets specific to the diseases or disease groups should be adopted. In the future, such disease-specific registries could fall under the remit of RD ERNs. Every effort should be made to incorporate current disease-specific registry initiatives where quality can be assured.

1.5 To avoid duplication and to support Cross-Border Healthcare, the possible benefits of using a global or European RD patient identifier (possibly incorporating the current health identifier) should be investigated to provide a way to link information, samples and research data, and to ensure a quick and secure means of data sharing and protection.

1.6 For countries with regional organisation of healthcare, where multiple registries exist, overlap and duplication between the regional and national registries, should be avoided.

2. All sources of data should be considered as sources of information for RD registries and data collections, to speed up the acquisition of knowledge and the development of clinical research.

2.1 As with all registries, registries for RD should establish clear purposes and objectives of the data collection: the type of data collection should be suited to the need, and the data captured should be appropriate to the proposed use of the data, both in terms of scope and level of detail.

2.2 RD Centres of Expertise, where they exist, should contribute to a registry(ies). Other experts in the field should also contribute to the registry(ies).

2.3 (Electronic) health records from any sector of healthcare delivery are a valuable source for core data collection. Automatic data acquisition from these sources should be envisaged to ease the data collection process.

2.4 Collection of data on RD should be delineated in the National RD plan/strategy.

2.5 A system to allow the collection of data directly reported by patients should be included along with systems for data reported by clinicians.

3. Collected data should be utilised for public health and research purposes.

3.1 RD data collected should be used to support policy development at local, regional, national and international level.

3.2 RD data collected should, where possible, facilitate clinical and epidemiological research and the monitoring of care provision and therapeutic interventions, including off-label use of approved drugs and existing medications.

3.3 RD data collected should, where possible, be used to provide information for multi-centre and multi-national clinical trial feasibility studies.

3.4 Pooling of data across data collections and other resources, including internationally, should be encouraged to reach a critical mass for data analysis. According to the governance/oversight criteria, data should be made accessible to groups with legitimate questions such as researchers and policy/decision makers.

3.5 Access and sharing of data should be defined to control how data is shared and published in the public domain and this should be facilitated through the national RD plan/strategy.

4. Patient registries and data collections should adhere to good practice guidelines in the field.

Specific to the current and future specificities of RD registries:

4.1 Involvement of stakeholders such as patients, policymakers, researchers and clinicians (and industry, where appropriate) in the design, analysis and governance of registries is important to address the complexity and scarcity of knowledge on RD.

4.2 Representatives of all stakeholders should be invited to provide best possible expert support through an advisory board or committee to ensure appropriate information flow and knowledge exchange into and from the registry, and they should define a sustainability

and exit strategy for the registry. Where appropriate, representatives from industry should also provide input.

4.3 This multi-stakeholder model for registry governance should apply not only at a national level but also at the European level and/or pan-European Platform repository of RD registries.

4.4 The process for consenting patients for participation in a RD registry should take into account the wider European and international context to ensure that patients are well informed of this dimension and the consent process is in line with the legal requirements at European and International level.

4.5 Patients already in a RD registry may be required to go through an additional consenting step to ensure compatibility with such systems.

4.6 RD registries should have a system to provide regular feedback to registered patients and their clinical teams, recognising their specific role in the success of registries in this field.

5. Existing and future patient registries and data collections should be adaptable to serve regulatory purposes, where required.

5.1 For the monitoring of therapeutic interventions for RD, a strategy between industry, academia and regulators should be agreed to ensure that data collection is expanded as necessary, and in time embedded in disease-specific registries to serve, for example, the requirements for post-marketing surveillance, and to support development of new therapies. Data access needs to be compliant with agreed guidelines established by the registry.

5.2 As quality assurance is crucial, it is a priority for existing RD registries to explore their capacity to adapt to collect data for regulatory purposes.

5.3 There should be an early dialogue on the type of registry required (and what data is required for regulatory purposes), and/or whether a registry exists for the condition targeted, with all stakeholders, in order to optimise the registration of patients and the generation of knowledge for RD for which a therapeutic intervention is being developed. Collection of data regarding off-label use of approved drugs and existing medications should be encouraged.

6. Patient registries and data collections should be sustainable for the foreseeable timespan of the registries' utility.

6.1 Local, regional, national and European structures contributing to or overseeing data collection should all be supported financially to carry out this role in a sustainable way so that financial responsibility for registries is shared proportionately between stakeholders, MS and the EC and defined in the appropriate funding programmes.

6.2 Public-private partnerships for RD registries should be considered where applicable as a long-term model for optimisation of resources, sustainability and co-creation of knowledge.

6.3 All registries and data collections should have in place an exit strategy in its work plan, including contingency planning for the data in the event that the registry is terminated. There should also be a procedure outlined for succession planning for registry continuation.