Report of the Joint EPIRARE and EJA Workshop on Rare Disease Registries and the European Registry Platform

Paris, 22-23 April 2013

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Overview of workshop

Forty two participants representing member states (MS), European Commission, patient groups, industry and academic experts in registries attended the EPIRARE/EJA workshop held in Paris on 22nd and 23rd April 2013. The draft recommendations for the general principles of RD registries were discussed and minimal changes agreed for circulation to EUCERD for presentation for adoption in June 2013.

To inform the debate on the development of a Rare Disease (RD) registries platform, the current status of the revision of data protection legislation in the EU was presented and the results of the EPIRARE project were reviewed, together with overviews of previous work on registry development in the RD field. All participants contributed actively to discussion, and the views of patient groups, academia, industry and the EC were presented. The European Joint Research Centre (JRC) presented an overview of their position as a part of the research-devoted structures of the European Commission (EC) and their proposed role in the development of a platform for RD registries. In addition to already established RD registries, there is a potential to collect data from designated Centres of Expertise (CoEs) and European Reference Networks (ERNs) when established. This project would provide an unprecedented opportunity within the EU to contribute to public health gains in the field of RD.

The detail of the presentations is provided in the workshop presentations and discussion section of this document.

Executive Summary and Conclusions

The conclusions reached with the meeting highlighted that there are key areas in which a RD registries platform can be seen as having utility and added value and that these should be explored further during the upcoming EUCERD meeting breakouts and plenary session. This presupposes that the issues of data ownership, transfer and governance can be satisfactorily addressed. Details about the proposed RD registry platform are not yet available but it is assumed that plans will satisfy the basic principles laid down in the EUCERD recommendations for RD registration and data collection. The platform will need to be developed taking stock of other on-going related initiatives, such as Parent JA, the Cross-Border Healthcare Directive and other RD initiatives such as the measures of national plans for RD concerning data collection, the designation of RD-CoEs, the establishment of future RD-ERNs and the perspective of the International Rare Disease Research Consortium (IRDiRC) international collaborations. Developments in e-Health and the interoperability of Electronic Health Records (EHR) are likely to contribute significantly to this project in the coming years.

Four main missions for this platform were identified by the workshop participants:

1. Support for new registries.
2. Promotion of the interoperability of existing standalone registries.
3. Act as a hub providing access to all data collection in the field of RD.
4. Provide IT tools to maintain already existing data collection.

1. Support for new registries.

The platform could provide support to new RD registries in areas of unmet need, providing the necessary IT tools, guidelines, consents and standards, education and training.

Ensuring impact: by stipulating that registry initiatives funded via the Directorate-General for Research and Innovation (DG-RTD) or the Public Health programmes are required to operate within this framework, compliance of new disease or project specific registries could be ensured. Standards for national registry developments could be encouraged, evaluated and promoted via national plans, CoEs and ERNs.

2. Promotion of the interoperability of existing standalone registries.

The platform should contribute to the interoperability of existing standalone national registries, as well as fragmented registries dedicated to specific RD or groups of RD. Contribution to interoperability could be achieved via the dissemination of agreed Common Data Elements (CDEs) for RD in general and by disease, and guidelines for the sharing of data. This development of the platform as a source of core agreed CDEs in itself would be an iterative process over the development of the platform. Support to registry holders to develop collaborations and maximise use of their data as well as to better disseminate outcomes should be provided. Information on existing sources of data should be made accessible. Affiliation to the platform would require adherence to standards of quality assurance and active feedback would be required in order to motivate existing registries to contribute to the platform.

Ensuring impact: future ERNs should be required to use the core tools for interoperability as part of their planning process. Registries funded by EU initiatives or under the IRDiRC aimed at data aggregation data should be encouraged to use these standards.

3. Act as a hub providing access to all data collection in the field of RD.

The platform should be a source of information on all existing sources of data collection in the field of RD so as to maximise collaboration and use of data.

4. Provide IT tools to maintain already existing data collection.

Several successful registry initiatives already collect data from different sources, but lack sufficient funding. The platform could provide the sustainable IT tools for data collection and analysis for these projects, so as to decrease the cost mutualising tools and acting as a long-term repository of data. Existing initiatives where CDEs and data sharing have already been agreed could be adopted into this system.

Ensuring impact: sustainability is a major challenge for on-going aggregation of data in many different registry initiatives. The provision of shared IT tools for data collection and analysis for such initiatives (provided the issues of ownership and data access could be resolved), would offer a potentially attractive solution to these challenges.

By fulfilling the above mentioned missions the registries platform could provide support across the board for RD registries and act as an important driver to quality and sustainability. These developments should be elucidated as a series of expected milestones and deliverables.

Workshop Presentations and Discussion

EUCERD Recommendations on RD Patient Registration and Data Collection
(Kate Bushby)

The current draft version of the recommendations were presented and reviewed. Additional comments were incorporated into the draft document ahead of the final review by the EUCERD members.

Update on the EU Regulation on Data Protection
(David Townend)

The EU is currently in the process of revising the Data Protection Directive 95/46/EC. The proposal, now at the legislative stages, is to create a Regulation in place of the Directive – which would have direct effect in Member States' law and thereby address some of the present harmonisation issues. The medical research and rare disease registry communities could receive the draft Regulation presented by the Commission at the start of 2012 positively. The Council's proposed amendments were equally positive. However, the Parliament has produced more than 3000 amendments, many of which arguably cause problems for rare disease registries and for medical research (especially secondary use of data or, for example, biobanking). The process of consent will be strengthened, but for research a separate route to fair and lawful processing, and secondary processing for further possible compatible routes would be available from the EC and Council perspective, but less easily under many of the proposed Parliament amendments. Indeed, many of the amendments, concerned with individual control in safeguarding privacy, do not move further than the current Directive, or even become more restrictive than that position for medical research and rare disease registries.

There remains an obvious acceptance of the need for a balance between individual rights while encouraging health innovation. The aim of lobbying Parliament must be to show that research and registries are needed and this should be weighed against what privacy really means in this context; that this is fully in the public interest. We can demonstrate what has worked as safeguards so far and how approval by local RECs and authorities safeguard privacy while encouraging research. A middle ground could be achieved that both respects the various individuals' sensitivities and the need for a more safe access to data in the public interest through the use of 'dynamic consent model' allowing an on-going dialog with patients and clinicians who are responsive to patients' evolving needs as new information becomes available. This will more likely result in patient empowerment and a strengthening of the therapeutic alliance between patient and clinician.

Two further observations. RD registries are not explicitly covered in the new proposals, in comparison with the Cross-Border Healthcare Directive. As each Member State (MS) has its own supervisory authority that will operate the

Regulation, there remains a question mark as to how harmonisation will be achieved in practice.

EPIRARE surveys on the proposed Registry Platform
(Domenica Taruscio and Luciano Vittozzi)

Several surveys have been conducted by EPIRARE with current registry holders, stakeholders and industry in regard to the definition of a platform model. The first survey (with more than 220 valid respondents) evidenced the main needs of the registry holders and researchers. It also allowed an appraisal of the way of application of ethical and quality guidelines as well as of the expectations from a European Platform. The needs of national authorities and EU institutions were identified on the basis of current national initiatives and EU policies. The needs of industry were communicated by the EPIRARE partner representing industry, on the basis of a consultation and a survey carried out among registries supported by the industry. These results are the main reference for the definition of the platform model proposed by EPIRARE.

Within a scenario of voluntary participation of registries in the platform, the concept of the platform relies on a mix of four main functions: a repository of data referring to a limited number of data elements common to all diseases; a repository of metadata of all data of participating registries; provision of technological tools, advice and services for the start up and operation of registries; a function of information, registration promotion, networking and policy support to ensure that results, activities and actors of data collection are properly brought to the attention of policy makers, patients and the public. The platform mission, served by these functions, is to allow a better use of existing data on rare disease patients and support the registration of patients with rare diseases for which dedicated registries are not sustainable. The platform should receive information from multiple sources to increase the completeness, reliability and allow estimation of the under-reporting and coverage of the platform. The platform would define a minimum set of CDEs, a number of purpose-specific sets of CDEs for pre-defined outputs (information pieces) of the registry platform to the wider public and identified stakeholders. Disease-specific and project-specific CDEs may also be included if and when the relevant experts agree on them.

To test the feasibility of this data structure for the platform, EPIRARE has defined examples of pre-defined outputs and identified the necessary data elements, possible definitions and formats and additional sources of data. Then, another survey (more than 145 valid respondents) was therefore carried out to investigate how many registries are collecting specific data elements regarding the patient, the services used, orphan drug and other treatments used, disability and quality of life, data collected longitudinally, data of donation, participation in clinical trials and on transplantation. The results are still being elaborated to provide a full and solid analysis of the situation. However, it appears that the current registries make up a workable environment where most of the platform outputs are reasonably feasible

using the data collected by at least half of the registries with their current practices; and another fraction of registries declared that their procedures can be adapted to comply with the proposed definitions and formats. Among the least collected data are those regarding quality of life and disability and patient willingness to participate in clinical trials. Two main evidences that may require the attention of policy makers are: 1) patient identifiers are not collected by all registries at central or peripheral level; 2) the RD coding systems used by registries is very fragmented and many registries even use their own coding system or no coding. The need for systematic collection of patient identifiers is a key prerequisite for any attempt to merge data from different collections and to carry out clinical and public health research; our data suggests that the current legal provisions already hinders the collection of personal identifiers; however, the new draft regulation on general data protection will become a killer to any attempt of merging and studying existing data and likely also to merging future data. As to coding, a strong action should be launched to take a policy position on the reference coding system for RD.

Policy Scenarios proposal for a EU RD Registry Platform
(Yann le Cam)

There are many challenges for RD, such as scarcity of patients, costs for maintenance and the need to monitor patients and families where the disease is caused by a genetic mutation. The process of data collection is key and needs to be clarified. Should this process be managed through the future ERNs? The process of data extraction from electronic health records is still unclear and needs to be defined. Another major issue is that stakeholder expectations are varied and how to address them all is challenging.

It was agreed that the platform should provide common tools and serve as a reference for the many challenges ahead. The platform aims to collect data through a framework defined at the EU level using common tools and procedures. The scope of the platform should be for all RD, across all MS (and globally), and provide a long-term storage solution of data with defined quality assurance processes in place. The platform should aim to be a RD-knowledge generation centre and reference for all MS policy makers. Establishing this platform should be defined in the 3rd and 4th Public Health Programmes (2014 to 2020, and beyond).

The added value of the platform is to build a critical mass of data and provide a harbour for ultra-rare RD and those patients with an unclear diagnosis. The platform needs to be safe and sustainable as a public service, and remain neutral from the specificity of RD, MS or other stakeholders. The use of data in the platform should be on a not-for-profit basis. The governance of the platform should involve experts (EUCERD) and stakeholders to help develop the platform strategy and policies – working alongside the Joint Research Centre (JRC) as the executive and operational body of the platform. The structure would also have committees to address issues such as ethics, technical/scientific advice, data requests, patient working group, healthcare and industry working groups. Financial sustainability through EC funding

to host the platform should also involve the RD ERNs, registry initiatives and research projects. MS funding should be targeted to national registries, centres of expertise co-funding in the RD ERNs and the use of registries in research projects. Industry funding can be used to sustain specific activities in clinical research and for regulatory activities. Academic funding through research projects would support the utilisation of current or new registries through the platform. The use of incentives could involve compulsory use of the platform via the CoEs and the RD ERNs, while providing professional credits for entering data into the platform, as well as providing data to industry and distribution of fees to patient groups and/or RD ERNs.

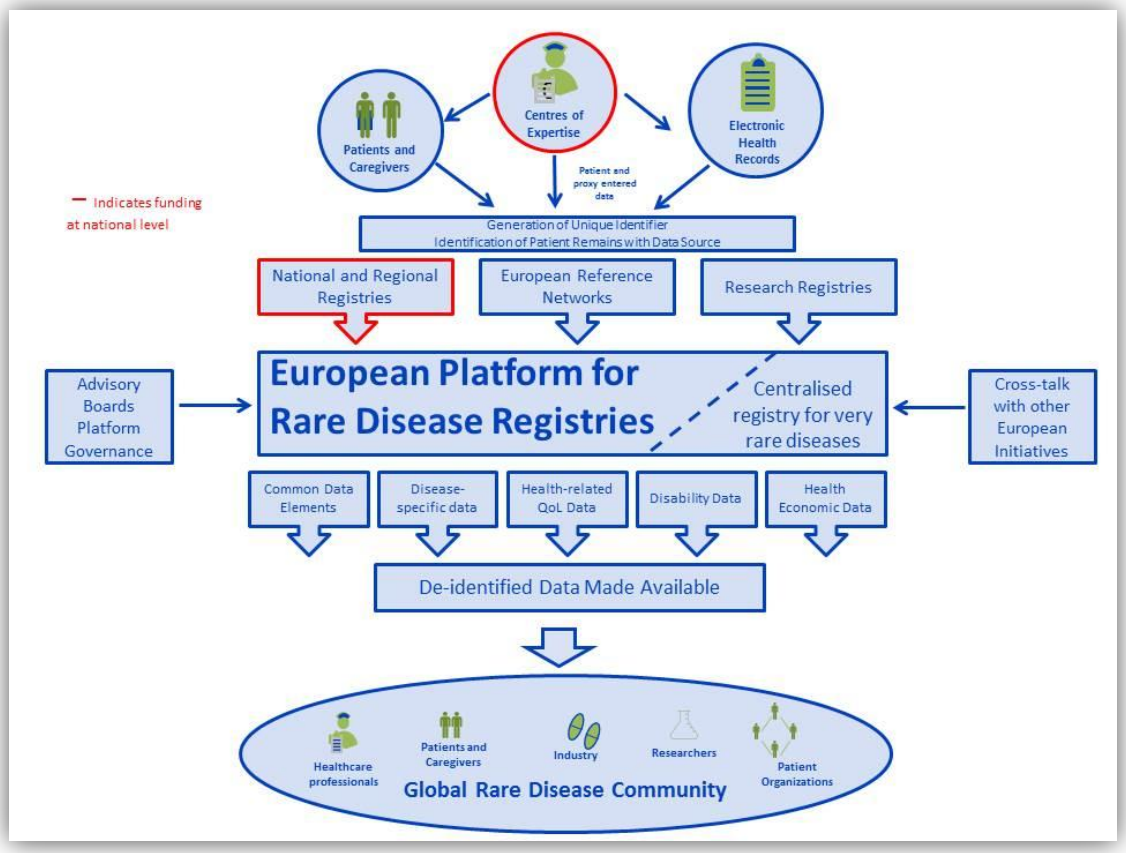

Figure 1. Proposed data flow scenario across the registry platform.

Policy Scenarios proposal for a EU RD Registry Platform
(Ségolène Aymé)

There is a clear need for a flexible and pragmatic approach, built on the long experience of registry leaders, to unite and drive consensus in building the platform. Quality of data is crucial and establishing appropriate quality assurance is difficult and time-consuming. All of this requires substantial investment to ensure this is done properly. Under the French national plan for RD a national committee was established to assess the quality of RD registries and a national platform was established to collect data from multiple sources. However, after three years only

data from centres of expertise are routinely collected, showing the difficulty to set up such a platform. We need a bottom-up approach to the platform, where patient integrity is ensured and has transparent involvement of industry. Therefore, we need to minimise costs and provide support to data producers through training on regulatory, data, ethics and standards, as well as advice on establishing a registry. The platform should be seen as an incentive to collect good quality data with respect to quality standards and monitoring of data, allowing output to be maximised through a critical mass and collaboration. The platform should provide standards on data and access to the data by external groups, and help secure the long-term secure storage of data.

Services need to be developed to ensure uptake of the platform.

- A federated system of national platforms would allow cross-talk across MS initiatives and harmonise their efforts, and would encourage other MS to participate by offering guidance and support of their efforts, while respecting subsidiarity. However, this would not support individual registers.
- Support registration to help with data collection and increase knowledge on as many RD as possible through a technological platform to build disease registers with minimal cost and common data sets. This can be similar to the Patient Crossroads initiative, but this is an expensive model (http://www.patientcrossroads.com/).
- Provide a unique entry point for access to data on RD and document all existing collections. Assess quality and provide an official designation of quality and document the data collected, and provide procedures and rules for access to data.
- Provide guidance, training and support to ease the establishment of local registers. This would need to organise consensus on the appropriate minimal data set for a disease or group of diseases.

It should be noted that the platform will not replace the primary sources of data, except for very rare RD where there is a need to collect primary data. The platform will not decrease the cost of data collection and exploitation, and will not solve the issue of sustainability of the entire data collection, only the data stored through the minimal data set.

The key factors for the success of a platform include selecting clear achievable goals with positive impact for a set of stakeholders, establishing a long-term plan with a credible team, keeping a bottom-up approach at all stages, and not raising expectations among the primary source registers.

Discussion points

- The JRC wants to know what we want to achieve from the platform. The JRC will also look at sustainability of the platform and the data sources, and how to organise the registries.
- We need to know where the data is, and not produce another repository to store the data.

- The MS may not think it is important to have an EU registry platform – so how do we convince them? Need to provide incentives for participation prior to the development of the platform.
- What is really achievable? We need to know what the aim of the platform is and not cover too many expectations of the various stakeholders.
- Achieving the goal will depend on how quickly the incentives can be put in place! Provision of training and support for the primary data collectors/providers to ensure data is collected and utilised for the benefit of the platform and the patients/clinicians is a priority.
- Motivation is important and participation is voluntary.
- The platform needs to provide help and support for collection of data, but the MS need to recognise the benefit of having registries in their national plans. Need to inform at the MS level that this should be in the national plans. Need to incentivise the MS, not only the registry data collectors.
- The EC needs to provide guidance in future funding calls on why future or current registries need to be compliant with the platform.
- The platform should bring the experts to the table and act as a clearinghouse for knowledge and know-how in running the registries and data collection procedures, analysis, retrieval, as well as the funding/sustainability issues.

European Rare Diseases Registration Platform
(Jarosław Waligóra, DG SANCO)

The main challenges to the platform include fragmentation, lack of interoperability, increasing regulatory requirements, and lack of sustainability. The EC is keen to promote cross-talk across the funded projects that involve registries in order to avoid overlaps which they hope will be avoided in the future via the platform. The EC does not aim to sustain all registries, as they need to focus on their aims and what is realistically achievable.

In the 2013 DG SANCO Annual Work Plan there is a call to support RD registries and networks in view of their sustainability. The aim of this action is to set up a sustainable platform to coordinate and maintain registries and networks on RD. The project will be in the form of an administrative agreement with the JRC, with an indicative budget of €2 million over a 2 year period. The JRC and EC are currently negotiating the content of the agreement and want input from the RD community. The platform will need to find its own sustainable funding for the future and so needs a sustainability strategy. The primary aims of the platform include the ability to perform data analysis across many diseases, increase the quality of data, improve patient care, facilitate clinical trials and studies, assess clinical effectiveness and allow comparison of treatments. The support for new registries, IT tools, common standards, training and support in the implementation of new registries will be services provided through the platform. The platform will therefore improve interoperability of existing registries, training in data quality, IT support, data analysis and usage, as well as long-term storage of data.

Eligibility criteria for existing EU registries to participate in the platform should include representation of a RD, willing to share de-identified data with the platform and other databases, agree to adopt CDEs, have an advisory body to assist on issues, such as ethics, privacy, data protection, standards, among others. These registries should also have a good plan to sustain beyond the 2 years of the pilot project.

The data collection through the platform would be limited to the minimal common data sets or just information about data existence in the primary registers. The platform will not collect data per se in the first instance, as this will be the responsibility of the primary registers, but later it may collect and hold data. The platform could also act as a primary source of data by hosting surveillance networks and registries, such as EUROCAT.

The management structure of the platform should ensure stakeholder involvement, but it would be run by the JRC. The EUCERD would provide advice and guidance and ensuring links to other areas of RD policy, but would not be involved in the day-today management of the platform. The stakeholders involved would include MS (health authorities and payers), patient associations, industry, regulators and current and past projects funded by the EC. The platform would need to be compatible with other EU initiatives, such as ERNs, Orphanet, and European Research Infrastructure Consortia (ERICs), such as BBMRI (biobanking). The PARENT Joint Action will also provide advice on the development of the platform within the ERNs and the wider Cross-Border Healthcare initiative.

The platform will also interact with other global initiatives, such as the IRDiRC working group on registries, and the Global Rare Diseases Patient Registry and Data Repository (GRDR) initiative established by the NIH Office of Rare Diseases Research.

The JRC will begin implementation of the platform in 2014 through a step-by-step approach and all future registries supported by the FP programmes will have to adhere to standards agreed if they are to be eligible for funding.

It was concluded that it will be important to involve the various e-Health projects in the development of the platform (maybe via the PARENT Joint Action), as many are involved in the cross-border activities. There are no clear plans for long-term sustainability and security of the data, but sustaining the primary registries and data collectors is also required. The funding for the platform is a separate issue, whereas the funding for the primary registries should come via projects and MS, but the EC can’t guarantee sustainability of these registries or even the platform. We need investment from other sources to sustain both the platform and the registries and a business model needs to be defined. Through the Associated Projects Group within the PARENT Joint Action, these issues will be addressed. One area of interest would be to find synergies between RD and the European Network of Cancer Registries (ENCR) that is also being developed by the JRC. This may help to maximise effort and reduce costs and duplication of work.

Academic vision for the Platform
(Anil Mehta)

RD can be seen as the tip of the iceberg of the more 'common' diseases. RD are seen as explanatory diseases of the more common conditions, so by just being rare should mean they are not ignored. One of the first issues that the RD community faces is that of coding to ensure we are all taking the same language. Therefore, we need to agree a set of interoperable standards to allow data to be entered and inform the knowledge gathered to help cure the disease. By adding genomic data elements gives power to the data and influences how you treat a patient. This requires measuring what is measurable, and make measurable what is currently un-measurable. The Cystic Fibrosis (CF) registry has 30,000 patients and is setting up a de-identification service to allow access by the EC and other organisations.

Understanding the e-Health issues is key to ensure we contain costs and to support data gathering from primary collections and electronic health records in real-time. Maybe we can use EU structural funds to help deliver a solution.

Industry vision for the Platform
(Vinciane Pirard)

Industry is primarily a user of registries data, and not a data provider, and requires an independent source of good quality data.

Industry expectations of the platform would be clearly defined objectives and that it is fit to support the drug development process. A platform would help to increase expertise in RD registries and standards.

Payment for access to data could come through a public-private partnership (PPP) or other agreements. There are PPP models to allow a specific subset of data to be entered into a protocol module for use by industry, and these should be further explored.

A priority for single registries would be to support post-marketing surveillance and pharmacovigilance reporting. It is important for industry to provide the 'right' data to the regulators and for it to be transparent and reported properly. Quality in data collection and reporting is therefore essential.

Industry requires disease registries, to help avoid fragmentation of data and ensure long-term longitudinal follow-up of cohorts. A long-term sustainability plan needs to be in place as longitudinal follow-up of cohorts could last 15-20 years. Industry also wants to know how priorities will be set for data collection and analysis and who is accountable for the outcome?

To become a privileged source of data for industry registries need to establish credibility and performance for example by adopting ENcePP or other organisations standards to ensure quality of observational studies. With observational data a multi-stakeholder approach to design, governance and methods with early input of epidemiologists seems the most appropriate.

Industry wants good partners to help develop and deliver good quality data, but do we really need a PPP for the platform itself? The platform will only hold minimal aggregate data and the use of modules to add on to a disease-specific registry may the best option for collection of regulatory data. Industry should define what it actually needs from a partnership, but this is not easily defined within the concept of the platform and the partnership might ultimately be with the disease-specific registries, not the platform.

Member State vision for the platform
(Helena Kääriäinen)

From the discussions at previous EUCERD meetings, many of the MS representatives were not aware of the EC/JRC call – even though the MS would have approved the call in the current work plan. There are pressures across the EU MS to reduce healthcare budgets, however, collecting data for RD registries is primarily a task of the national centres of expertise (CoEs), who will not receive EU funding, and so they will have to focus on the most necessary tasks, such as improving healthcare for RD patients. The MS recognise the need for collecting RD patient data, but the platform was seen as too ambitious and requiring too many resources. Collecting data from multiple sources, such as electronic health records, registries, mutation databases, etc., would increase the risk of errors in the platform, but collecting a minimal set of data was supported.

Many of the MS felt the plan to have a RD registry platform was too premature as many of the CoEs does not yet exist, but on the other hand any data collection system would need to have common rules and guidance. The issue of data protection was also seen as problematic and the MS recommended that only metadata was collected by the platform.

The plans for a registry of all RD was considered unrealistic by the MS, but disease-specific registries collecting data for a specific purpose was considered useful and would motivate MS to help secure the resources needed to sustain their activities. Collection of specific data sets through clinicians was seen as too laborious and unrealistic, but collection of a minimal data set or data from patients would be more realistic. The least laborious way would be that (all) MS would adopt a coding system that enables identification of RD in the healthcare databases, with electronic health records evolving having more detailed data. Together these would make it possible to collect data nationally from any RD patient group when needed and to share this on an EU or global basis. However, how would a proper consent be ensured? Also, in

reason of data protection issues many MS may not be keen to collect data on EU level.

Patients vision for the platform
(Yann Le Cam and Monica Ensini)

The preliminary data analysis of the EPIRARE patient survey on registries shows that patients have a clear vision of the added value and benefits for them of a comprehensive European approach to Rare Disease registries. It should be emphasised that there is a gap between the purposes of registers in today's reality and the expectations of patients. Patients focus their interests in registries in view of disease knowledge production and healthcare and social planning, treatment evaluation, and natural history of the disease. A very large majority of survey respondents shared the opinion that the legal aspects regarding Rare Disease registries should be regulated at EU level. Moreover, survey respondents are largely in favour of the implementation of a EU registration platform, primarily funded publicly (EU, National, Academic funds) involving patients in all aspects of registry governance (scientific objectives, data to be collected, decision on the use of data, legal and ethical issues). Patients' empowerment and capacity building – information, exchange of experience, networking, and training – are needed for adequate and full involvement of patient representatives in the governance and activities of standalone rare disease registers and the EU registration platform.

The results of the full data analysis will be ready by October 2013 and will be made available to the entire RD community.

The role of the JRC in the platform
(Laura Gribaldo)

The JRC has requested to work in collaboration with the RD community to help develop the platform. They also want to connect with the WHO to ensure global harmonisation. The Institute for Health and Consumer Protection (IHCP) of the JRC will be the host for the platform and provide the scientific and technical support in all aspects of the policy development in regard to the platform. The JRC-IHCP is already in charge of an integrated cancer information system with the secretariat for the European Network of Cancer Registries-ENCR based at the JRC-IHCP. The implementation of this integrated information system could create a model that could be extended to the RD field.

Discussion points

- The JRC-IHCP may want to set up a steering committee of experts to help with the RD registration platform project, but as yet the permanent team at the JRC-IHCP is not yet recruited, they will coordinate the input from experts via meetings, etc.

- However, the JRC-IHCP cannot support specific diseases or organisations.
- The JRC-IHCP will develop a feasibility plan before the next EUCERD meeting (5-6 June 2013) to cover the next two year period. This plan will describe what they need to learn from the RD community. On the other hand, the RD community needs to establish a very clear communication framework with them.
- The JRC is planning a meeting with MS about the creation of an ERIC to help sustain the platform.
- The agreement between the JRC-IHCP and the EC will be signed in the autumn and this will describe how the €2 million will be used.
- The technical annex will be ready soon, and it is important for the RD community to know the timeline for the various milestones and deliverables.
- It is postulated that registries, genomics and coding will be priority areas for funding in the next work plan.

Group discussion session

It was agreed that support for new RD registries should focus on provision of IT tools, standards, training and general support for their implementation to help bring the data together and manage the data accordingly. Official documents should be developed to help guide the new registries, as infrastructure support and guidance has long been needed to ensure they harmonise with current standards. Harmonisation of coding is crucial and this should be one of the roles of the platform.

The IT tools needed can be provided by the infrastructure available at the JRC-IHCP. There is a need to define exactly the tools are needed, whether they are for a repository or for collection, or both. It is important that the JRC-IHCP informs the RD community of what modules they are able to provide. There are many levels on which IT tools can be provided, such as process design tools, software (to collect and manage data, and to analyse data), etc., but these require a lot of resources to implement and maintain. Therefore, we need to define the IT tools before we build the platform and need to keep them simple. Current networks, such as the cystic fibrosis (CF) and neuromuscular diseases have approached the issue from an international perspective and kept the IT tool simple to collect a small amount of key data via an online system. As a lobbying tool the age of death of affected individuals was the key data used to influence policy at a MS level.

Quality criteria will be defined in an EPIRARE deliverable report, and this could become a useful reference in setting up a new registry. It was suggested the JRC-IHCP could establish a new web site with information for the public on the tools available to help new registries, especially those funded under Horizon 2020. These tools will need coded fields to help with harmonisation of data entry, as well as harmonised consent forms, and made available on the web site, updated as policy changes dictate.

Many national health systems will not allow open access onto the web, so how do we get round this firewall issue? An electronic signature, or handshake, could be implemented to allow health systems to talk to the platform. This will be examined by initiatives in the field of e-Health.

There will be an on-going process to inform and disseminate plans concerning the platform to the MS, requiring a step-wise approach to communications. This can be supported by EUCERD and other networks, who can help build a communications strategy to inform and educate allowing the implementation of a future platform through the national plans for rare diseases. The JRC-IHCP can provide support to the communication and education through its meetings with MS and through e-learning facilities.

To increase interoperability we need to agree standards for coding and storing data, and subsequently how to migrate the data to the data fields.

- Need agreement on a suitable dataset model (such as xml) and use this for transferring data to the platform, while the single registry uses its own system for day-to-day functions.
- Need to define the clinical content and then the tools can be made to share the data – so the data to be collected shapes the development of the data model system.
- Need to define the technological model and then the semantic model of what is required to help define interoperability.
- Create a repository of standards to help with data collection and coding into the system.

There are not many standards for electronic health records (EHR) at the moment. The open EHR is a template, but is complicated and not easy to implement presently, but some countries like Sweden have already used it. It would be useful to conduct a review of the best strategies currently available on how to code and collect.

The PARENT Joint Action will carry out some pilot studies in this area and could provide help in this review. They have created a knowledge management platform with guidelines recommendations and methodologies available on the web as part of the framework. This could then provide guidance to the best practices and then provide the correct tools (from the repository) to collect data into a separate registry of registries. This framework concept is currently under testing.

Patient identification is also an issue for interoperability. How can we register patients, especially children, without creating patient identification issues through the use of a system with an identifier and appropriate coding?

Designation of registries into the platform could be via quality management and tools to measure the effectiveness of the registries. At the EU level the platform could help with future designation of data collections to help secure funding and ensure further development of the collection. One role of the platform should be to look at the quality of data, but it needs to give something back to the registries for

providing the data. This assessment would be communicated back to the registries to help them with their plans to improve overall quality. However, assessment should be anonymous as participation in the platform would be voluntary. The EU wants all registries to adhere to the agreed quality standards if they are to receive future EU funding. The MS will need to be informed of the process and agree to it.

The platform could also be seen as a source of data, with EUROCAT as an example. EUROCAT is a repository of data using a common data set, with good practice guidelines in place. The EU has funded EUROCAT for the last 25 years, but this support will not continue. The registries in EUROCAT are anxious about future funding for their collaboration. The JRC is already talking to EUROCAT, whose major issue is sustainability of the networking activities. So the major priority is the existing registry networks for inclusion in the platform instead of building new registries.

The inclusion of federated national platforms needs to be addressed in the platform – using minimum data sets to capture data in certain countries. If it works then it should be considered for inclusion into the platform. The plan would be to organise meetings to inform and discuss and help national registries harmonise their data collections systems through standards and guidelines. The platform intends to federate the national registries and will provide support to help harmonise the data collection.

Finally, it was clear that a key success factor would be to motivate data providers. In the US data providers are rewarded with professional credits and acknowledgements in publications. But how to motivate when no registry exists? The CF community has used a simple model to initiate a data collection system and then build on this, with a financial model that has ring-fenced funds for audit to help train the data providers. Personal incentives such as feedback to the centres on errors and training provided are helpful. Also, feedback on data collected to allow benchmarking by data providers and visibility to the work of the clinician. Again, if their data is used this will also motivate providers.