Exploratory Paper on eHealth Strategies and Roadmaps supporting European Reference Networks and Rare Disease Policies

This document has been drafted through a co-operative process involving members of EXPAND, PARENT Joint Action and the Rare Disease Joint Action (EURORDIS, now RD Action). The document is prepared with the intent to propose a formal process of cooperation between the eHealth and European Reference Network communities via a number of specific actions and activities, which would eventually support a continuing and sustainable process resulting in eHealth strategies encompassing elements to effectively address rare disease (RD) policies and the needs of ERNs and European Patient Registries.

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Executive Summary

Directive 2011/24/EU on the application of patients' rights in cross-border healthcare promotes policy co-ordination and Member States' (MS) cooperation in eHealth through the eHealth Network (eHN), established by Article 14. In May 2014 the eHealth Network adopted four priorities for eHealth (cross border Patient Summary, ePrescription services, patient registries and European Reference Networks). The eHealth Digital Service Infrastructures to be implemented under the CEF 2015 work programme will support the deployment of the cross border Patient Summary and the ePrescription services,[1] including a provision for a central IT Platform supporting European Reference Networks (ERNs).

The initial set of CEF-supported services are expected to put in place the basic infrastructure, governance and interoperability framework needed to enable the exchange of health data based on a clear legal basis, and within a secure and trusted environment. These are important enablers, which lay the foundations for rapid expansion to new health data-sharing services, addressing healthcare challenges beyond emergency care services including, in particular, the priority areas of Directive 2011/24/EU such as ERNs.

Rare disease, patient registries and e-Health policies and strategies have been so far addressed separately and largely without coordination to-date, while interoperability remains an important challenge for sharing data. There is therefore a challenge to create convergence between the eHealth and the public health communities in preparation for extending CEF services to address specific needs of ERNs and rare disease policies.

The document is prepared with the intent to propose a formal process of cooperation between the two communities via a number of specific actions and activities within ongoing and possible future actions, which would eventually support a continuing and sustainable process resulting in eHealth strategies encompassing elements to effectively address RD policies and the needs of ERNs and European patient registries. It is anticipated that this collaboration is necessary in order to ultimately support the work of the Joint Actions on Rare Diseases (RD-Action) and the eHN Joint Action (JAesHN), and eventually the Commission Expert Group on Rare Diseases (CEGRD), the eHN and the HTA Network.

The document summarises the background of policy, strategy and action in the two areas and, based on this, presents the main groups of specific cross-border data sharing challenges faced by the ERNs,

[1] Amended CEF Workplan 2015 https://ec.europa.eu/inea/sites/inea/files/c_2015_7381_f1_annex_en_v3_p1_828057_cef_telecom.pdf

rare disease and registry communities. It then examines how these needs might be addressed as extensions of the Patient Summary and ePrescription services, through specific examples of use case extensions addressing such specific needs. It is not the intention of the document to elaborate a definitive list of such use cases, but rather to elicit the next steps that are needed in order for the stakeholder communities to propose a common strategy and roadmap of use cases to be promoted for deployment of additional cross border eHealth services.

In order to define these steps, it is recommended that the current, project-based expert group and its activity are commissioned as a "Convergence" Task Force that will be tasked to elaborate proposals regarding:

- a stepwise, use-case-based implementation approach and a strategic roadmap of activities supporting convergence between the eHealth domain and the relevant RD ERN and, in time, registry related activities;
- what actions need to be launched immediately in order to support a possible extension of CEF eHealth DSIs within the CEF 2016 work plan; such extensions should represent use cases addressing eHealth Network priorities, which are also priorities of the rare disease community to be implemented through ERNs and other means within the scope of Directive 2011/24/EU;
- the respective immediate activities beyond the scope of the work of EXPAND and the current Joint Actions (for Rare Diseases, Registries and eHealth) that are needed to prepare the assets for these extensions including a unified conformant approach.

The currently running initiatives – projects and existing and new Joint Actions - have supported and are expected to continue to support a certain level of co-operation and activities such as joint workshops, to discuss and validate this Exploratory Paper on eHealth Strategies and Roadmaps supporting European Reference Networks and Rare Disease Policies; however none of the existing projects has a mandate to prepare assets for extended services for CEF. Future eHealth extensions in the CEF annual work programmes will require supplementary activities to this end. On the other hand, a number of projects are selecting and adopting use cases suitable to support work objectives. Likewise, in 2016 the first ERNs will be set up, tasked with carrying out specific objectives, some of which require eHealth support. There is therefore an urgent need for an activity that would reflect on and deliver a proposal for a Roadmap of use cases and associated work elements necessary to address in a stepwise and prioritized approach the full span of cross border eHealth needs implicit in the Directive. This Roadmap could become a valuable policy support instrument in terms of setting common implementation priorities.

Preamble

Directive 2011/24/EU on the application of patients' rights in cross-border healthcare promotes policy co-ordination and Member States' (MS) cooperation in eHealth through the eHealth Network (eHN), established by Article 14. In May 2014 the eHealth Network adopted four priorities for eHealth (cross border Patient Summary, ePrescription services, patient registries and European Reference Networks). The eHealth Digital Service Infrastructures to be implemented under the CEF 2015 work programme will support the deployment of the cross border Patient Summary and the ePrescription services,[2] including a provision for a central IT Platform supporting European Reference Networks (ERNs).

The initial set of CEF-supported services are expected to put in place the basic infrastructure, governance and interoperability framework needed to enable the exchange of health data based on a clear legal basis, and within a secure and trusted environment. These are important enablers which lay the foundations for rapid expansion to new health data-sharing services, addressing healthcare challenges beyond emergency care services including, in particular, the priority areas of Directive 2011/24/EU such as ERNs.

In the rare disease (RD) field, the topics of registries and ERNs have featured prominently on the policy agenda since the 2008 Commission Communication "on rare diseases: Europe's challenge"[3] and the 2009 Council Recommendation on an action in the field of rare diseases[4], which functioned as a roadmap of sorts, outlining necessary national and collaborative measures to alleviate the burden posed by RD to patients, professionals and health and social systems. In January and June 2013 respectively, the European Union Committee of Experts on Rare Diseases (EUCERD) published "Recommendations on RD ERNs" (to which an Addendum was added in June 2015)[5] and "Core Recommendations on RD Patient Registration and Data Collection"[6] based on the outputs of various multi-stakeholder projects, publications and stakeholder meetings. However, there has been limited

[2] Amended CEF Workplan 2015 https://ec.europa.eu/inea/sites/inea/files/c_2015_7381_f1_annex_en_v3_p1_828057_cef_telecom.pdf

[3] Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges COM(2008) 679: http://ec.europa.eu/health/ph_threats/non_com/docs/rare_com_en.pdf

[4] Council Recommendation of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02): http://www.consilium.europa.eu/uedocs/cms_data/docs/pressdata/en/lsa/108383.pdf

[5] http://www.eucerd.eu/?p=2260

[6] http://www.eucerd.eu/wp-content/uploads/2013/06/EUCERD_Recommendations_RDRegistryDataCollection_adopted.pdf

cross-talk between the eHN and its supporting initiatives, on the one hand, and the EUCERD/Commission Expert Group on Rare Diseases (CEGRD)[7] and related RD initiatives, on the other.

This Exploratory Paper represents a joint initiative between –at the time of authoring of this document - the EXPAND Thematic Network, the PARENT Joint Action and the EUCERD Joint Action. It builds on the policies, reflections and assets developed within the two communities of eHealth and rare diseases.

- EXPAND (for more details see Annex I) is a €1M initiative funded by the Competitiveness and Innovation Framework Programme until the end of 2015. It is an eHealth thematic network building upon the achievements of previous projects, notably **epSOS** to ensure sustainable, scaled-up, pan European investments in the field of e-health.
- PARENT Joint Action (Cross-border PAtient REgistries iNiTiative) is based on Article 14b of Directive 2011/24/EU, on methods for enabling the use of medical information for public health and research. PARENT received €3.4M co-funding by the Public Health Programme of the European Commission (EC) and by Member States, to build tools and propose policy actions supporting EU Member States in developing comparable and interoperable patient registries in clinical fields of identified importance (e.g. chronic and rare diseases, medical technology).
- The EUCERD Joint Action: Working for Rare Diseases is a €5.5M policy project co-funded by the Public Health Programme of the European Commission (EC) and by MS, supporting the EUCERD (now the CEGRD) in formulating and implementing healthcare and social policies pertaining to RD across Europe, in order to reduce inequalities in access to quality RD care. It has 5 core WPs and addresses a broad range of RD topics: ERNs; registries; cross-border genetic testing; Centres of Expertise and quality of care; specialised social services for RD; coding, classification and inventorying of RD; and national plans and strategies for RD. **Since September 2015, these policy activities have been carried out under the new Joint Action for RD, RD-Action.**

This document aims to explore means of embedding the eHealth-related needs and activities of ERNs and patient registries into an updated and enriched ***European eHealth strategy and roadmap*** for information and knowledge sharing, describing specific strategies and actions to improve the quality and safety of care for RD patients, including through collaboration in RD research and for promoting inclusion of these elements in national eHealth Strategies and Roadmaps, as well as in Rare Disease National Plans and Strategies . It is not the objective of this document to *elaborate* such a strategy

[7] In July 2013, the EUCERD has been replaced the Commission Expert Group on Rare Diseases.

and roadmap, but rather to signpost a number of activities leading to it, by exploiting a wealth of knowledge, projects, assets and networks currently available and active.

The document is prepared with the intent to propose a formal process of cooperation between the two communities via a number of specific actions and activities within ongoing and possible future actions, which would eventually support a continuing and sustainable process resulting in eHealth strategies encompassing elements to effectively address RD policies and the needs of ERNs and European patient registries. It is anticipated that this collaboration is necessary in order to ultimately support the work of the new Joint Action on Rare Diseases (RD Action) and the eHN Joint Action (JAesHN), and eventually the CEGRD, the eHN and the HTA Network.

1. eHealth addressing the needs of ERNs and European Registries

Delivering high quality, accessible and affordable healthcare is a challenge shared by all countries in Europe. The advent of eHealth offers solutions to these challenges in numerous ways: improving the quality of treatment, health outcomes and quality of life and helping to reduce pressures on public healthcare budgets through facilitating sharing of health data and broadening access to medical care through maximising use of technologies and new services. **These challenges, and the need to coordinate solutions across Europe, are only heightened in the field of RD**: the prevalence of any single RD in any given country will, by definition, be low, resulting in a scarcity of available expertise and budget which can only be redressed though efficient coordination *between* countries. To date, ERNs and patient registries have been identified as fundamental tools in coordinating care for RD patients and facilitating the requisite close interaction between care delivery and research in the RD field. Patient registries, in particular, should be structured and operated in a way that allows for shareable longitudinal patient data across borders, serving multiple purposes incl. re-use of this data for research aiming to improve treatment and care, as well as supporting health policy makers' needs (e.g. through HTA analysis of relevant technologies, surveillance of health services performance and possible socio-economic inequalities).

1.1 Existing Strategies, and their scope

The European eHealth Strategy and Roadmap - as expressed through the EC Recommendation on cross-border interoperability of electronic health record systems[8], the eHealth Action Plan [9]the

[8] Commission Recommendation of 2 July 2008 on cross-border interoperability of electronic health record systems (notified under document number C(2008) 3282) (2008/594/EC), http://eur-lex.europa.eu/legal-content/EN/TXT/PDF/?uri=CELEX:32008H0594&from=EN

[9] eHealth Action Plan 2012-2020: Innovative healthcare for the 21st century, http://ec.europa.eu/digital-agenda/en/news/ehealth-action-plan-2012-2020-innovative-healthcare-21st-century

CALLIOPE Roadmap[10] and the eHealth Network guidelines for Patient Summaries and ePrescriptions[11]- have focused initially on addressing fundamental challenges of interoperability of electronic patient records exemplified through patient summary and ePrescription services; re-using this data for acquiring new knowledge for the improvement of care and care processes is a natural next step. A similar trend is observed at national and regional level.

At the same time, strategies for addressing RD challenges at EU and national levels have also been conceived, as RD are considered a priority area for action at the European level, in view of the potential for European added-value through a collaborative approach. This commitment to RD has been enshrined in a series of key policy documents, including the Commission Communication *Rare Diseases: Europe's Challenge*[12] and the Council Recommendation on an action in the field of Rare Diseases(The Council of the European Union, 2009)[13]. It is also evidenced by the constitution of entities such as the Rare Disease Task Force (RDTF), EUCERD and, most recently, the CEGRD. The EUCERD and CEGRD have issued Recommendations on numerous topics of relevance to RD, to support MS in addressing the challenges posed by RD. To date, Recommendations have been adopted on the following themes: *RD European Reference Networks; RD Patient Registration and Data Collection; the CAVOMP-Information Flow*[14]*; Quality Criteria for Centres of Expertise for RD; Core Indicators for RD National Plans; Ways to Improve Codification for Rare Diseases in Health Information Systems.*[15]

1.2. Implementation Status

Member States have collaborated in the epSOS Large Scale Pilot for exchanging health data on Patient Summaries and ePrescriptions - they have identified challenges for sustainability, and have been addressing these at policy level in the eHealth Network and at the technical and organisational levels through maturation actions within EXPAND. EXPAND will therefore hand over epSOS assets and services for deployment at European and national level through the European Digital Service Infrastructures (DSI) to be developed under CEF.

Likewise, the RD communities across Europe are already at a relatively mature stage in professional and organisational terms, with a long history of cross-border collaborations around sharing evidence, sharing anonymised case studies and pooling knowledge to support research including through using registries to co-ordinate care for patients and to share research data sets, although at present these are largely non-interoperable. With a view to improving patient registry quality and interoperability,

[10] European eHealth Interoperability Roadmap http://www.ehgi.eu/Download/European%20eHealth%20Interoperability%20Roadmap%20[CALLIOPE%20-%20published%20by%20DG%20INFSO].pdf

[11] eHealth Network, Policy, Key Documents http://ec.europa.eu/health/ehealth/key_documents/index_en.htm

[12] http://eur-lex.europa.eu/legal-content/EN/TXT/?uri=URISERV:sp0006

[13] http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF

[14] Clinical Added Value of Orphan Medicinal Products – Information Flow.

[15] These can be accessed via http://ec.europa.eu/health/rare_diseases/publications/index_en.htm#anchor1_more

the eHealth Network has adopted in November 2015 the Guidelines developed for this purpose by the PARENT Joint Action[16].

These policy areas (rare disease, patient registries and eHealth) have been addressed separately and largely without coordination to-date. The e-Health efforts in the registry community, for example, start from the point where data and information has already been gathered at individual registry holder organisations and RD centres and needs to be shared to support patient care and research. The discussion on how has this data/information been collected, by which means, tools, systems and in what format has only recently been considered.

The existing collaborations, and policy and scientific co-ordination in these areas, however, create favourable conditions for a rapid strengthening of the use of interoperable eHealth solutions, for example through new guidelines and decision-supporting solutions to improve the safety and effectiveness of the care of patients with RD (e.g. when presenting at an emergency department or to a GP) and to accelerate the scale-up of clinical and biomedical RD research. ERNs supported through interoperable eHealth services, addressing their specific needs, will also act as an exemplar for European co-operation in ICT-supported management and precision (personalised) medicine of rare and also less rare long-term conditions i.e., where a number of existing relatively common diseases will become sub-profiled into a set of relatively rarer ones.

Action at all levels and a long-term strategy for the convergence of efforts are urgently needed, which should aim to proactively address the needs of people living with rare diseases, scientific communities working on these diseases, Registries and ERNs within European and national/regional level eHealth strategies (e.g. it is necessary to explore how ERN and EU Registry needs may be addressed in the CEF, as extensions of eHealth DSIs). **Likewise, in time eHealth specific considerations should become part of National Strategies and National Plans for Rare Diseases.**[17]

2. Priorities for eHealth in the Rare Disease communities

RDs are defined at EU level as conditions affecting no more than 5 per 10,000 people. By definition, the number of individuals with any single RD is very small; however, the fact that there are 6000-8000 RD means that collectively RD constitute a major public health challenge at national and European level, with approximately 30 million patients across Europe. With a scarcity of patients for any given

[16] PARENT Joint Action D7. Methodological guidelines and recommendations for efficient and rational governance of patient registries. Available at: http://patientregistries.eu/deliverables

[17] MS were asked to elaborate and adopt a national plan or strategy for RD by the end of 2013, to structure RD activities within the framework of the national health and social systems.

disease, and corresponding rarity of experts able to provide diagnostics, treatment and care, collaboration across borders is essential.

Data-Sharing: To realise the vision of cross-border care defined in Directive 2011/24/EU, it is necessary to have **the ability to send data safely and securely between MS, and this should include (anonymised and/or aggregated) clinical research data as well as personal health data,** as the concepts of healthcare and research are very much interlinked in the RD field: digital platforms, such as the platform of RD-Connect, aims to allow clinicians and researchers across Europe to share de-identified genomic and phenotypic data of undiagnosed patients with other professionals, which will support diagnosis and *clinical* practice, as well as research. The eHN priorities of **Registries** and **ERNs** possess unique importance for the RD field, as both are intrinsically linked to concepts of sharing data, which is a major priority in the RD community. Sources of data for RD patients are scarce and, at present, largely fragmented. A priority for any eHealth service should ultimately be to enable the integration of (possibly-disparate) sources of data: this might be data from registries, from EHRs, from biobanks etc.[18]

Linking data for health and research: For this to become possible, unambiguous electronic identification of patients across countries and across databases is key. Appropriate anonymisation services are also necessary to allow for anonymisation of information for research (e.g. Global Unique Identifiers) and diagnostics. It is also becoming increasingly accepted that certain critical information for RD patients is necessary in order to follow them throughout the healthcare pathways (e.g. electronic equivalents of Rare Disease cards).

Interoperability challenges: Ongoing work around ERNs and the services that they should provide suggests that **interoperability between Centres of Expertise** (or healthcare providers as per Directive 2011/24/EU) is recognised as the greatest obstacle that needs to be overcome for full deployment of ERNs. Hence, establishing organisational and legal interoperability as well as common semantic and technical standards is a priority for the RD community, which is directly and intrinsically concerned with ERNs and their previously stated information needs.

Ontological Needs: Key to the cross-border interoperability of data in the RD field is the use of appropriate ontologies. It is recognised that RD are typically far less 'visible' in health information systems compared to more common diseases, as the major systems of disease classification (e.g. ICD 10) are at present unable to capture the complexities and granularity of RD[19]. Developing and promoting use of a shared -and RD appropriate- coding nomenclature is thus a key priority for the field. The Orphanet Rare Disease Ontology (ORDO) is considered to be the most appropriate

[18] "RD patient registries and RD biobanks should aim to be global in geographic scope and practice. **Interoperability and harmonisation between RD patient registries and RD biobanks** should be consistently pursued. Linking and data transfer into existing platforms should be considered "best practice" for RD registries and RD biobanks". (*IRDiRC policies and guidelines, April 2013. IRDiRC is the International Consortium for RD Research, www.irdirc.org*).

[19] ICD10 is able to code only ca. 250 of approximately 8000 RD.

"nomenclature" system for RD.[20] Work has been conducted in the EUCERD Joint Action –and will continue under RD-Action- to cross-reference different systems of coding diseases. When diseases are added to the Orphanet portal[21], they receive an OrphaCode, alongside which one can find the equivalent code in ICD 10, SNOMED-CT, OMIM, UMLS, MeSH and MedDRA.

Harmonization in terms of *phenotypic* ontologies is also essential in sharing RD patient data for health and research purposes: -omics data alone is often not sufficient to support a diagnosis, whereas the ability to link -omics data with a robust phenotypic description of the patient increases the likelihood of 'solving' unsolved and undiagnosed cases. Therefore, ICT tools supporting shared care and research in the area of RD, notably **EHRs and patient registries**, should ideally enable clinical (phenotypic) information to be collected in a computable (semantically interoperable) way, thus enabling the identification and centralisation (or distributed analysis) of information about patients with specific conditions across centres of care, whether Centres of Expertise, local clinics, hospital emergency departments etc. Collecting an adequate threshold of such phenotypic information allows for health improvement, improved research and more empowered patients.

Ensuring RD specificities of broader eHealth tools: To benefit from eHealth advances, it is necessary that RD are adequately integrated both to 'planned' care (or shared expertise and/or care) systems via ERNs, and also to systems dealing with emergency and unplanned care. Given the specificities of RD, it is important that key tools developed outside of the RD 'sphere' per se, to facilitate cross-border care (such as ePrescription and Patient summaries), can adequately convey core information on rare conditions and treatments (e.g. are coded appropriately). There has been little emphasis on this, to date.

Further health potential lies in the exchange of data stemming from improved genome screening techniques and better bioinformatics tools to interpret genome/phenome data.

For all the above, data-sharing should always be supported by an **adequate ethical/legal/social framework** that corresponds not only to ethical standards and legal requirements, but also to **patient preferences**.

[20] See CEGRD Recommendation on *Ways to Improve Codification for Rare Diseases in Health Information Systems*. The importance of a harmonized approach to ontologies for RD is also emphasised in the research sphere: "Ontologies utilized by RD research projects should build upon existing best practice and allow integration and interoperability across different ontologies, including those for model organisms. Ontologies should include a RD classification ontology (nosology), a phenotype ontology with comprehensive coverage of RD manifestations including laboratory values and imaging, as well as ontologies to support biobanking, clinical trials, and research". (*IRDiRC policies and guidelines,* as above).

[21] www.orpha.net. Orphanet is the reference portal for information on rare diseases and orphan drugs.

3. Beyond Cross Border Patient Summary and ePrescription Services

The implementation of Patient Summary and ePrescription services to be deployed under the 2015 CEF work program, will put in place the basic infrastructure, governance and interoperability framework needed to enable the exchange of health data based on a clear legal basis, and within a secure and trusted environment. These are important enablers that set the foundations for rapid expansion to new health data sharing services, addressing health care challenges beyond emergency care services and especially the priority areas of Directive 2011/24/EU.

In addition, the first set of eHealth services to be deployed in CEF will exploit a number of use case-specific semantic and technical interoperability assets developed and validated within the epSOS Large Scale Pilot. There is in addition, a wealth of assets including in the form of disease specific clinical data sets to address priorities of the eHealth Network for patient registries and European Reference Networks (ERNs).

It is therefore sensible to assume that the immediate extensions of eHealth services will need to demonstrate that:

- there is a clear policy priority under the Directive;
- there exists an infrastructural need at a European level that addresses an important integration gap and will deliver real added value to the implementation of this policy if it is met;
- this integration functionality will not be provided through the services that the CEF already plans to deliver; and
- the additional interoperability services can draw on mature assets and specifications and standards that have consensus acceptance within the user community.

The current European patient summary for unscheduled care, provided in a different European country from the patient's usual place of care/place of residence:

- contains generic information that will not be particularly helpful for people seeking to provide continuity of care to patients with rare diseases, nor does it contain the data items that are most needed to select patients that are relevant to rare disease related studies;
- contributes to increased safety in unplanned care situations but less so to continuity of care through e.g. updating of medical records in the patients country of affiliation; it is however noted that this concept has been implemented in the return of the dispensation report in the ePrescription services;
- enables the exchange of codified data elements in electronic clinical documents but does not support the exchange of imaging data;

- makes available to the treating physician in the country of treatment information from a single source i.e. the NCP of the country of affiliation, while shared care requirements through ERNs may require longitudinal care views from multiple sources (members of the ERN);
- does not include tele-consultation components;
- has not been assessed from the perspective of research, quality of care and performance assessment needs.

4. eHealth DSI Extensions - a use case based approach

The following list of use cases is not exhaustive but it can demonstrate how possible use cases addressing ERNs and Registry specificities -in the light of the eHealth Network CEF priorities- may be prioritised as extensions of the patient summary and ePrescription services foreseen for deployment in CEF, as of 2016. The intention is therefore limited to exemplifying how well-defined needs may be addressed through leveraging a foundation of eHealth Digital Service Infrastructures and interoperability assets developed by several communities. The final proposals shall result from extensive consultation within the eHealth and the ERN communities (initially) which should be an integral part of the approximation activities (see section 5).

a. **RD specificities in the European Patient Summary** may be envisaged as an extension of the current data set to include data elements necessary for identifying RD patients and for adequately conveying core information on rare conditions and treatments. This might leverage available RD assets such as the ORDO and -through appropriate links- resources that would help physicians deal with a rare disease encounter.
b. **Supporting planned care through shared care records** may be envisaged as an evolution of the Patient Summary service to address the needs of planned care, starting from selected diseases or groups of diseases. This may be implemented through the Health Care Encounter Report service, extended with the same proposed Patient Summary datasets for RD, which will allow the return of information for incorporation into the patient's health care record.
c. **Exchanging Health Care Encounter Reports (HCER)** i.e., standardised data sets of information generated as a result of a health care encounter and that needs to be shared within other health professionals sharing responsibility for the care of a patient may support ERN needs and set the foundations for services supporting registries by linking EHRs to registries.
    i. these will not be supported under CEF 2015; although the HCER service was analysed in epSOS, only segments were developed in epSOS and Trillium Bridge for demonstration purposes;
    ii. agreed clinical research data sets exist for a number of subsets of rare diseases;

iii. there are mature specifications and standards which have been adopted by the Multistakeholder Platform (MSP).

d. Extending eID services provided for Patient Summary and ePrescription should include the **Global Unique Identifier** being developed by RD-Connect for patients/research participants, which could allow clinicians and researchers to 'link' genome sequencing data with essential phenotypic information gathered in shared care EHRs and registries.

Any extensions of the eHealth DSI to address additional ERN and Registry needs in the light of the priorities of the eHealth Network, are expected to be expressed in the form of additional use cases, which will then be analysed in depth as to the needed interoperability requirements, vis-a-vis existing specs and assets and their further needed preparation and maturation. It is also expected that there will need to be adoption by the eHealth Network of such use cases, as well as actions to prepare additional services for CEF leveraging on existing communities, interoperability agreements, specifications and assets.

The above examples can also help illustrate the kind of additional work which will be needed to implement, test and validate extensions beyond the existing demonstrators, prepare the available semantic assets which might need additional processing, e.g. where appropriate to convert it in the form of a respective Master Value Catalogue, for deployment in CEF and validate the service within a small scale pilot involving a number of centres and countries, members of the respective ERNs.

The prioritisation exercise should also examine gaps that need to be addressed before launching new services on CEF. For example, the epSOS specifications of HCER will need to be tailored to the planned care shared record; this will require introducing extensions to the Patient Summary use case, elaborate specifications, testing, deployment governance and related policies, hence it is unlikely that it could be addressed as such in CEF 2016 and additional work will be needed before deployment. Additional work will be also needed to perform a legal and organisational gap analysis, implement, test and validate these additional services within the small scale pilot involving a number of centres and countries, members of the respective ERNs to assess the organisational, technical and cost implications of accessing data in shared care electronic records; following this to implement possible additional profiles and respective technical extensions, test and validate these additional services within the small scale pilot involving a number of centres and countries, members of the respective ERNs.

5. Bridging the two worlds: an Approximation Strategy

Immediate action is needed to formalise an appropriate strategy and associated preparatory actions for preparing extensions to CEF, capitalising on the experience of EXPAND in preparing and delivering assets produced in project based environments in a form suitable for deployment.

Concurrently it is important to create conditions for bringing the eHealth community and the relevant public health communities together in an appropriate discussion forum. Within the general

framework of expandability versus unmet needs, the stakeholder community may then validate immediate extensions meeting the criteria for CEF for eHealth services.

The existing Joint Actions supporting the eHN and the RD policy may provide the general umbrellas for further work; whether, however, they have the capacity to address the next needed steps to the appropriate depth and span needs to be explored.

The currently running initiatives – projects and existing and new Joint Actions - have supported and are expected to continue to support a certain level of co- operation and activities such as joint workshops, to discuss and validate this Exploratory Paper; however none of the existing projects has a mandate to prepare assets for extended services for CEF. Activities beyond the scope and capacities of currently running projects, to prepare the assets needed for these extensions should also promote a unified approach, taking into account the outcomes of H2020 PHC-34-2014 projects and, in particular:

- Align Medicinal Product description in all relevant cross-border services to the standards identified in OpenMedicine CSA, based both on the EMA Article 57 database and the emerging ISO IDMP standards;
- Duly consider the findings of ASSESS-CT on the assessment of the suitability of adopting a terminology (e.g. like SNOMED-CT);
- Harmonize non-conflicting interoperability standards eStandards, for the revision of the specifications for CEF 2016 programme, to allow coherent call for tenders;
- Consider the business models defined by VALUeHealth, to build the long term sustainability after CEF;
- Pilot and improve further the Guidelines and tools developed by PARENT Joint Action, both in establishing new and in upgrading existing patient registries;
- RD-Connect is an FP7 infrastructure project which is building an integrated platform for RD registries, databases, biobanks and bio-informatics in order to reduce the fragmentation of precious data sources and address some of the obstacles hindering researchers. Healthcare and research are very much interlinked in RD field, and the platform of RD-Connect will allow clinicians and researchers across Europe to share de-identified genomic and phenotypic data with other professionals, which will support diagnosis and research.

If eHealth extensions will be included in the CEF 2016 work programme, ***a supplementary activity to this end will therefore become urgent***.

On the other hand, a number of projects are already selecting and adopting use cases suitable to help them carry out their work objectives. ***There is an urgent need for an activity that would reflect on***

and deliver a proposal for a Roadmap of use cases and associated work elements necessary to address in a stepwise and prioritised approach the full span of cross border eHealth needs implicit in the Directive. This Roadmap could become a valuable policy support instrument in terms of setting common implementation priorities.

An intermediate step is needed to define the action that need to be addressed by any existing or new projects or initiatives. This step could be achieved through commissioning the current, project based expert group and its activity as a ***"Convergence Task Force"*** that will be tasked to elaborate proposals regarding:

- a stepwise, use case based implementation approach and a strategic roadmap of activities supporting convergence between the eHealth domain and the relevant RD, ERN and Registry related activities;
- what actions need to be launched immediately in order to support an extension of CEF eHealth DSIs; such extensions should represent use cases addressing eHealth Network priorities, which are also priorities of the rare disease community to be implemented through ERNs and other means within the scope of Directive 2011/24/EU;
- the strategic elements that need to be incorporated into EU and national and regional strategies for eHealth and for Rare diseases.

The Task Force should also provide support:

- to identify immediate extensions meeting the criteria for CEF for eHealth services; and
- to agree the respective immediate activities beyond the scope of the work of EXPAND and the current Joint Actions (for Rare Diseases, Registries and eHealth) that are needed to prepare the assets for these extensions including a unified conformant approach.