The chair welcomed the Expert Group to their second meeting and announced the adoption of the Commission Implementation report on the Commission Communication: Europe’s Challenges and the Council Recommendation on an Action in the Field of Rare Diseases after the summer. Since the previous meeting the European Conference on Rare Diseases and Orphan Products\(^1\) was held in May in Berlin, attended by many of the members, and the 2014 Health Programme Work Plan\(^2\) was adopted as well as the Delegated\(^3\) and Implementing\(^4\) Decisions concerning the Directive on Patients’ Rights in Cross-Border Healthcare\(^5\).

- **Proposal for the expert group recommendation on rare diseases codification**
  The members of the Expert Group discussed a draft recommendation on Ways to Improve Codification for Rare Diseases in Health Information Systems. The work carried out so far in terms of the inclusion of rare diseases into the next version of the International Classification of Diseases and the cross-referencing of Orphacodes with other internationally recognised terminologies in the field was described. The results of a workshop on Orphacodes in health information systems, held by the EUCERD Joint Action in March 2014 were also presented. The draft recommendation outlines the issues at stake and lays down key points for action, including the organisation of a working group of stakeholders and countries wishing to implement Orphacodes in their health information systems. It was highlighted that these recommendations would support the codification of rare diseases in ICD and SNOMED CT, being in parallel to the ICD process, as the versions of these nomenclatures with fuller representation of rare diseases will not be released for several years. A working party is needed to assess the resources needed and the tools that should be provided to support MS in their endeavours. Support for such work should be sought by the MS with the EC at European level. The draft recommendation, modified according to the discussions, will be circulated to the Expert Group to adopt by written procedure. A workshop on the next steps concerning the implementation of Orphacodes in health information systems will be co-organised by DG JRC and the EUCERD Joint Action on 1-2 October 2014 in Ispra to bring together MS representatives and specialists from coding agencies.

- **Update on Important Activities in Member States**
  Member States representatives informed other Members of important activities in the field in their countries, including progress made concerning the adoption and implementation of national plans/strategies for rare diseases.

- **European Social Insurance Platform (ESIP) – Reimbursement of Orphan Medicinal Products**

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\(^1\) [http://www.rare-diseases.eu/?lang=fr](http://www.rare-diseases.eu/?lang=fr)


An overview of issues concerning the reimbursement of orphan medicinal products in the EU was presented to inform members of problems relative to access to these medicines and possible solutions being sought at the current time, such as adaptive licensing, value-based pricing and the MoCA (Mechanism of Coordinated Access to Orphan Medicinal Products) process. Members of the group welcomed this presentation and the opportunity to discuss this important issue. In the context of EUCERD’s previous work on the Clinical Added Value of Orphan Medicinal Products (CAVOMP) Information Flow, the Expert Group considered work on the issues surrounding access, pricing and reimbursement a logical next step. The Expert Group decided to continue discussions on pricing and reimbursement of therapies for rare diseases and associated regulatory issues in order to review the progress made in other forums. The appropriate EC services will be invited to participate in such discussions. The secretariat will collect suggestions on the topics to be explored in order to prepare discussions in this area at the expert group.

**European Reference Networks for rare diseases**

Members were provided with details concerning the implementation of the Cross-Border Healthcare Directive of 2011, in particular concerning the Delegated⁶ and Implementing⁷ Acts of March 2014 which entered into force in May 2014 relative to the establishment of European Reference Networks in the framework of the Directive. Members cited the need for a strategy and vision in the field of rare diseases within this framework, so as to address the need for ERNs to cover, at term, as many rare diseases as possible. The specific criteria, mentioned in the acts, for rare diseases could also be discussed. The importance of patients in the rare disease community needs to be flagged by the manual to be developed via the call for tender, perhaps simply by providing a link to the EUCERD Recommendations on ERNs for RD. In areas with multiple patient organisations, a scoping exercise will have to be carried out to see how they can federate in the context of the ERN.

**Activities of the EUCERD Joint Action**

An update on the Joint Action which is at Month 29 of its duration was presented. Outcomes were presented and a number of upcoming workshops were announced:

- Evaluation workshop and combined Partner Meeting: 27-28 November 2014 (Lisbon)
- Europlan final workshop: 28-29th October (Rome)
- Guiding Principles for Social Care workshop: 9-10 October 2014 (Norway)

A workshop on centres of expertise and another on genetic testing are foreseen without fixed dates. Another workshop on ERNs on the subject of grouping rare diseases and prioritisation is a possibility and a workshop on rare disease registration could be supported with the collaboration of the JRC if needed.

**Update on Rare Disease Research in Horizon 2020**

An update on the Horizon 2020 programme launched in December 2014 for a total budget of EUR 80 billion was given with specific details given on the support available to the field of rare diseases in the current and upcoming calls. This includes support to the E-Rare ERA-NET, to projects aimed at fulfilling the goals of the IRDiRC in clinical and pre-clinical research, and the INFRADEV1 and INFRAIA1 calls concerning infrastructure for which the Innorare application concerning rare diseases is being elaborated (the details of this application were presented separately). The topic concerning support for ERN modelling has been delayed to 2016 to better fit with the timeline foreseen for the development of ERNs.

**Communications activities**

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The EC presented the website\(^8\) of the Expert Group on Rare Diseases via which information can be found concerning the work of the Expert Group and the previous recommendations of the EUCERD, as well as the section of the EC website concerning rare diseases. The site \(\text{www.eucerd.eu}\) will continue to provide access to previous documents of the EUCERD and information on the activities of the EUCERD Joint Action, and it will be updated for the duration of the Joint Action. The 2014 Edition of the annual Report on the State of the Art of RD Activities\(^9\) was published on 15 July and announced in OrphaNews\(^10\). The five volume report also has individual country reports which should be disseminated widely at national level\(^11\). The OrphaNews editorial board was updated in agreement with the members in line with the new composition of the Expert Group.

- **New Joint Action on rare diseases**

The Joint Actions in the 2014 Call\(^12\) of the 3rd Health Programme 2014-2020 were presented. Amongst the actions cited by the call is the « Support to the implementation of the Council Recommendation and Commission Communication on Rare Diseases », in particular to an EU wide rare diseases information database with a budget of EUR 4.29 million. The RD joint action highlights actions in the area of information provision, codification, European Reference Networks, gathering expertise and provision of support to the Commission Expert Group on Rare Diseases. Participants must be nominated by Member States prior to the invitation to prepare the proposal and nominations should be submitted by 16 September 2014. Members highlighted the importance of support to the implementation of the Council Recommendation, Orphanet and codification of rare diseases.

- **Registries for rare diseases**

An update on the implementation of the European platform for rare diseases registration at the Joint Research Centre in Ispra was given. This initiative aims to address the fragmentation of rare disease data in Europe. The Platform will provide the OSSE\(^13\) open source software for registries and guidelines for sharing data which will allow for decentralised data entry and the creation of a central repository and data hub with a European data search tool. Workshops will be planned on the subject of international standards and nomenclatures for coding. The planned model for the registration platform will be sustainable, and the support given to individual registries will be in terms of tools, guidelines, training and workshops. The implementation phase will require the establishment of an advisory group composed of some members of the Expert Group on Rare Diseases: a call for interest will be launched soon. A presentation of the work of the PARENT Joint Action\(^14\) (Cross-Border Patient Registries Initiative) was also given and ongoing liaison between the PARENT Joint Action and the Expert Group was encouraged.

- **International Rare Disease Research Consortium Activities (IRDiRC)**

The action plan of the IRDiRC has been defined with the following priorities: identification of key resources and of standards, development of a data standards clearinghouse, submission of the roadmaps of scientific committees, and organisation of conferences. The key resources and standards with an international scope directly linked to IRDiRC goals already used by the international community satisfying quality standards and freely accessible, will be given an ‘IRDiRC recommended’ logo. A core set of around 2,300 terms common to all terminologies has been

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\(^8\) [http://ec.europa.eu/health/rare_diseases/expert_group/index_en.htm](http://ec.europa.eu/health/rare_diseases/expert_group/index_en.htm)

\(^9\) [http://www.eucerd.eu/?page_id=163#StateArt](http://www.eucerd.eu/?page_id=163#StateArt)

\(^10\) [www.orphanews.org](http://www.orphanews.org)

\(^11\) [http://www.eucerd.eu/?page_id=154](http://www.eucerd.eu/?page_id=154)


\(^14\) [http://www.patientregistries.eu/](http://www.patientregistries.eu/)
adopted. A data standards clearinghouse will be partially supported by the IRDiRC Scientific Secretariat. The Scientific Committees are finalising or have finalised road maps for their activities. The second IRDiRC conference$^{15}$ will be held in Shenzhen, China on 7-9 November 2015.

- **European Conference on Rare Diseases and Orphan Products, 8-10 May 2014**
The outcomes of the European Conference on Rare Diseases and Orphan Products held in May were presented. The next conference, to be held in Edinburgh in 2016, is being prepared and the programme committee, which will include members of the Expert Group will be formed in the coming months: a call for interested persons will be launched to participate in this process.

- **Rare Disease Day 2015**
The next Rare Disease Day will be held on 28 February 2015 which will again focus on the theme of solidarity.

- **Next meeting of the Expert Group**
The next meeting will be held on 12-13 November 2014.