

## EUROPLAN CONFERENCE SPAIN FINAL REPORT

### I. General Information

<b>Country</b>	<b>SPAIN</b>
<b>Date and Place of the National Conference</b>	<b>November 5 and 6, 2010</b> State Reference Centre for RD (CREER) <b>Burgos</b>
<b>Web Page</b>	<a href="http://www.enfermedades-raras.org">www.enfermedades-raras.org</a>
<b>Organisers</b>	Federación Española de Enfermedades Raras (FEDER), Centro de Referencia Estatal de Enfermedades Raras (CREER)
<b>Steering Committee members</b>	Simona Bellagambi - EURORDIS Advisor Rosa Sánchez de Vega - FEDER Board and vice-president of EURORDIS Miguel Ángel Ruiz - Director of CREER Isabel Calvo - President of FEDER Claudia Delgado - Director of FEDER Concha Colomer - General Director Deputy. Quality and Health Planning Office. Ministry of Health, Social Policy and Equality Ignacio Abaitua - Head of Unit. Research Institute for RD. Instituto Carlos III. Ministry of Science and Innovation Cristina Avendaño - Director of the Spanish Drug Agency Ministry of Health, Social Policy and Equality Miguel García - President of the Spanish General Practitioners Society (Sociedad Española de Medicina Familiar y Comunitaria) Francesc Palau – Scientific Director of the Biomedical Research Network for RD CIBERER Feliciano Ramos - President of the Spanish Society of Human Genetics (Asociación Española de Genética Humana –AEGH-) Pedro Serrano – Member of EUROPLAN Project in Spain. BURQUOL-RD project Co-leader
<b>Names and list of Working Groups</b>	GT1. Methodology and Governance of National Strategy GT2. Definition, codification, inventoring and registries GT2.1. Information and training on RD GT3. Research on RD GT4. Reference Centres and European Networks on RD GT4.1. Standard of care on RD GT4.2. Orphan Drugs GT5.1. Empowerment of Patients GT5.2. Social services and help lines for RD
<b>Chairs and co-chairs of</b>	GT1. Concha Colomer, Claudia Delgado

<b>Working Groups</b>	GT2. Ignacio Abaitua, Mercedes Pastor GT2.1. Miguel García, Salud Jurado GT3. Francisco Palau, Isabel Campos GT4. Rosa Sánchez, Rosa López GT4.1. Feliciano Ramos, Isabel Calvo GT4.2. Josep Torrent, Moisés Abascal GT5.1. Tomás Castillo, Pedro Serrano GT5.2. Begoña Ruiz, Estrella Mayoral
<b>Attached documents (Timing, List of participants, etc.)</b>	<ul style="list-style-type: none"> <li>- Programme</li> <li>- Photo report</li> <li>- Press Clippings</li> </ul>

## II. General Overview: Legal Frame

On June the 3<sup>rd</sup> 2009 the Spanish Ministry of Health and Social Policy announced the establishment of a National Strategy for RD of the National Health System (NHS). The Spanish plan is primarily a coordination tool based upon seven strategic lines that will encompass the domains of information; prevention and early detection; healthcare coordination; therapies (including advanced medicinal products, orphan drugs, and related materials); provision of social and health services; research; education and training. The elements defined in the Spanish strategy follow the recommendations delineated by the European Council Recommendation on an Action in the Field of RD and following the Senate Report, supported by every Political Party.

Given the decentralised health administration (management) of the Autonomous Communities (AC) (Regional Governments), the Strategy will act as a set of recommendations for the different regions which subsequently will be in charge of its implementation. Small funds are allocated through a call for proposals open to the AC in order to facilitate the implementation of the Strategy. Evaluation will take place in two years.

Regarding the Centres of Expertise, there is a **Royal Decree, of November 10, 2006**, for the designation and accreditation of **Reference Centres, Services, Units (RCSU –CSUR in Spanish- ) (RC for short) of the National Health System.**

These RC are designated for diseases that need high technology and high specialization as well as for RD. The Designation Committee is formed by representatives of the AC and of the Ministry of Health, Social Policy and Equality.

### III. Main Report

The EUROPLAN Project has, as the main goal, to develop plans of actions or strategies for RD in every European country before 2013 to address RD with a global approach and face together the health and social problem of these pathologies. The number of people with RD in Europe may reach 36 million, near 3 million in Spain. These figures are remarkable enough to advocate RD as a priority of Public Health in the EU.

We have reached the main goal: identifying and assessing the Spanish situation- at National and Regional -AC- levels, following the European Guidelines at the ***Council Recommendations regarding an European Action on RD. EUROPLAN Conference*** in Spain took place on November 21, 2011 at the State Centre for Rare Diseases (Centro de Referencia Estatal de Enfermedades Raras -CREER-) located in Burgos.

More than 150 experts participated in the event, representing all interested parties: National and Regional -AC- Administrations, Health and Social professionals, scientific community, Industry and patients' organisations

In Spain, different Working Groups were established, appointed by the Steering Committee, in order to have more time to go in depth, as can be seen above. A Moderator and Co-moderator (one of them a patients representative) in every group was appointed, apart from the Secretary/rapporteur.

Then, following EUROPLAN indicators, every Working Group assessed, the suggested actions for every field, pointing out how far, important and feasibility in the **National Strategy of RD**

In short, EUROPLAN Conference in Spain managed to disseminate EU Recommendations successfully, as well as to collect concrete proposals to assure the transferability of these Recommendations to the National Strategy for RD. In addition, it help to identify the existing best practices in Spain and analyse with common indicators (proposed by EUROPLAN) the main priority areas of the National Strategy for RD.

The Steering Committee of the Conference brought together the main representatives of Interest Groups in Spain: Ministry of Health, Social Policy and Equity, Spanish Drug Agency, National Centre for people with RD and their Families, Research Institute for RD, Biomedical Research Network for RD, Spanish Association for Family and Community Medicine (GP), Spanish Society of Human Genetics, BURQOL-RD project researchers, FarmaIndustria and the Spanish Federation for RD. The

presence of Simona Bellagambi, EURORDIS advisor, was key in the whole process of EUROPLAN Conference organization.

There was a lot of work previous to the Conference: two meetings, translation of documents, mailing lists for every working group to share documents and exchange proposals, etc.

Below you can see the main outcomes and proposals, achieved by consensus in every topic.

### **Main outcomes of the Conference:**

- Success in achieving the main goal: **presentation and awareness on the EU Communication Commission** for RD.

- **The quantity and quality of proposals** to improve national and regional policies, in favour of RD, on line with the European recommendations.

- **The balanced participation** of the interested parties in every Working Group.

- **Exchange different points of views**, trying to get common consensus and facilitate the dialogue between all stakeholders. Give the patients the chance to be listened.

- The highly assumed **key role of patients** in their participation in decision making process in RD policies.

- **High impact in terms of audience** through online broadcast during open session, when results were presented.

- **Thorough preparatory work** in every topic/working group

- **The active and lively participation.** Open constructive debate among the different participants

- **The remarkable importance of the Conference**, highlighted in all means of communication.

- The decision for this EUROPLAN Conference to be the seed, to be followed by similar Conferences in the coming years.

- **Common methodology and indicators**, the same for all the Conferences allow comparable outcomes.

- The final report of the **Conference will be "the roadmap"** to improve the extent and implementation of the National Strategy for RD in Spain.

## Main Themes

### **Theme 1 - Methodology and Governance of a National Plan/Strategy (NP/S)**

---

#### Sub-Themes

1. Mapping exercise before developing a National Plan
2. Development and structure of a National Plan / Strategy
3. Governance of a National Plan
4. Monitoring the National Plan
5. Sustainability of the National Plan

### **Theme 2 - Definition, codification and inventorying of RD**

---

#### Sub-Themes

1. Definition of RD
2. Classification and traceability of RD in the National Health System
3. Inventories, registries and lists

#### **2.1. Information and training**

4. How to improve information on available care for RD in general, for different audiences
5. How to improve access to quality information on RD
6. How to ensure adequate training of healthcare professionals on RD

### **Theme 3 - Research on RD**

---

#### Sub-Themes

1. Mapping of existing research resources, infrastructures and programmes for RD
2. Needs and priorities for research in the field of RD
3. Fostering interest and participation of national laboratories and researchers, patients and patient organizations in RD research projects
4. Sustainability of research on RD
5. EU collaboration on research on RD

### **Theme 4 - Standard of care for RD - Centres of Expertise (CoE)/ European Reference Networks (ERN)**

---

#### Sub-Themes

1. Identification of national or regional CoE all through the national territory by 2013
2. Sustainability of CoE
3. Participation in ERN
4. How to shorten the route to diagnosis
5. How to offer suitable care and organize adequate healthcare pathways for patients with RD
6. How to ensure in CoE multidisciplinary approaches and integration between medical and social levels

7. How to evaluate CoE

#### **4.1. Orphan Drugs (OD)**

8. Future of OD
9. Access of Patients with RD to orphan drugs Pricing and Reimbursement
10. Compassionate use and temporary approval of orphan drugs. Off label use

### **Theme 5 - Patient Empowerment and Specialised Services**

---

#### Sub-Themes

1. Involvement of patients and their representatives in decision-making processes in the field of RD
2. Support to the activities performed by patient organizations
3. Specialised social services: Respite Care Services; Therapeutic Recreational Programs; Services aimed at the integration of patients in daily life
4. Help Lines

#### **Horizontal Themes**

"Horizontal Themes" 6 and 7 were addressed in Themes 1 to 5. Therefore, the Steering Committee decided that the sustainability of infrastructures and activities, and the expertise networks at EU level, should be addressed in every working group.

### **Theme 6 – Sustainability**

---

### **Theme 7 - Gathering expertise at the EU level**

---

## **IV. Main conclusions and proposals of the debate**

### **Theme 1 - Methodology and Governance of a National Plan / Strategy (NP)**

---

#### Sub-Themes

6. Mapping exercise before developing a National Plan
  7. Development and structure of a National Plan / Strategy
  8. Governance of a National Plan
  9. Monitoring the National Plan
  10. Sustainability of the National Plan
- 

#### **Workgroup 1**

**Chair:** Concha Colomer (General Director Deputy. Quality and Health Planning Office. Ministry of Health, Social Policy and Equality)

**Co-chair:** Claudia Delgado (Director of FEDER)

**Number of Participants:** 10

#### **1. Mapping exercise before developing a National Plan**

**Is there an awareness of the situation of RD in your country (epidemiologic figures, dimension of the problem)?**

In Spain there is not a comprehensive, accurate, reliable and systematic analysis of the current situation. We have registries and have launched diverse initiatives to know the size of the problem, although they were fragmented (funded by the Autonomous Communities –AC-) and too specific for particular diseases, health centres, scientific societies, etc. giving a heterogeneous and incomplete picture of the situation.

**Conclusion:** It is proposed and agreed that a Workgroup from the RD Strategy will be asked to be in charge of analyzing the situation of the existing RD-related registries in Spain and establish some wide-national recommendations and guidelines in order to unify the existing autonomic registries.

**Is an inventory of the existing resources and actions on RD (from which patients with RD can benefit) being made at the National Healthcare System?**

There is lack of a widespread inventory, although there are specific resources, such as Orphanet, FEDER SIO (Information and Advice System), and a catalogue of centres carrying out genetic diagnosis from the AEGH (Human Genetics Spanish Association), among others.

**Conclusion:** It is proposed and agreed that a Workgroup from the RD Strategy will be in charge of analyzing the situation of the available existing information systems which can be an useful tool to establish quality criteria to validate this type of resources and define updated information mechanisms so that it will be available to all implicated partners (patients, families professionals, etc.).

**Are the unmet needs of patients with RD being evaluated?**

This has been an essential aspect of the RD Strategy. Therefore, the Strategy group included the patient representatives' participation. Moreover, it is necessary to point out that the IMSERSO and the ENSERio reports (research on social and health needs in Spain) have also been considered.

**2. Development and structure of a National Plan / Strategy**

**Is there a legal policy framework in the form of a national plan or strategy created to address the health care and social needs with specific actions?**

Spain has the Strategy on RD, which was approved by the Inter-territorial Board of the National Health System (CISNS) on June 3rd, 2009. The Strategy has been spread out through a printed or electronic document, and has been completely translated into English.

The Strategy document pretends, in base of the information and evidence available, to establish a group of objectives and recommendations that, realistically and according to the available resources and the AC regional frameworks, contribute to upgrade the quality of the actions and subsequent results on RD.

The Strategy identifies the following strategic lines:

**Strategic line 1:** Information on RD

**Strategic line 2:** Prevention and screening

<p><b>Strategic line 3:</b> Healthcare <b>Strategic line 4:</b> Therapies <b>Strategic line 5:</b> Social and healthcare <b>Strategic line 6:</b> Research <b>Strategic line 7:</b> Training</p>
--

Currently, we are working on key-aspects groups to implement the Strategy, as well as on planning its next evaluation.

Patients' representatives considered a priority to avail the EUROPLAN Conference to introduce proposals to improve the current Strategy on RD.

**Is there a timeline for the achievement of priority actions with specific deliverables?**

The Strategy outcomes will be evaluated two years after being started – i.e. on June 2011- and every four years since then.

The source of information for these indicators will be given by the AC within the Strategy Pursuit and Evaluation Committee they take part.

**3. Governance of the National Plan**

**Is there a Steering Committee or Coordination Committee governing the implementation of the plan? Has this Committee the required status for its activities and responsibilities?**

For the Strategy making process, a Technical and an Institutional Committee were appointed:

The **Technical Committee** is formed by patients' representatives and scientific societies, selected on consensus and scientific knowledge- Among the patients' associations we find the Neuromuscular Diseases Spanish Federation, which represents more than 20 pathologies with these characteristics, the Citizens with Chronic Diseases Coalition and the Spanish Federation of RD (FEDER), that shelters over two hundred patients associations.

In the **Institutional Committee** there are representatives designed by the AC Health Services/Departments. One of its main functions has been evaluating the relevance and feasibility of the proposed objectives, since they are the own AC and their health services which are responsible for organizing and giving health care.

Due to the complexity of most RD multiple agents from the Ministry of Health and Social Policy and Equity have also participated:

- GD of Public Health and Abroad Care
- GD of Professional Ordering, Cohesion of the National Health System and High Inspection.
- GD of Pharmacy and Care Products.

- GD of Advanced Therapies and Transplants.
- Spanish Agency on Drugs and Care Products.

Imsero's involvement has been essential to social attention to people affected by rare pathologies, taking also into account the National Reference Centre for People with RD and their Families located in Burgos. The Ministry of Science and Innovation has also participated very actively through the Institute of Research on RD (IIER) and Centre of Biomedical Network Research on RD (CIBERER), both of them belong to the Carlos III Health Institute (ISCIII). Once the Strategy was approved by the CISNS both committees became the Pursuit and Evaluation Committee.

Patients' representatives expressed that the involvement could be enriched by the Senate-reported proposals, such as creating the State RD Organization (OEER).

**Does the Committee meet regularly?**

The Committee has established a minimum of two annual meetings. Moreover, the workgroups developing specific aspects on the Strategy meet periodically to carry out their tasks.

The Committee writes down, through record, its activities-related information in the different meetings.

**4. Monitoring the National Plan**

**Is there a monitoring system for the National Plan? What kind of indicators is used to monitor its implementation?**

A series of indicators related to the achievement of the aims and the carrying out of the recommendations proposed by the Strategy were defined.

**Conclusion:** There is an open process to include in, reformulate and/or enrich the appropriate-considered indicators to evaluate the national Strategy on RD. This process is currently open until June, 2011, when the evaluation process will be started.

During the evaluation process on the Strategy's actions, EUROPLAN indicators will be considered too, being published after the Strategy approval.

**Is the evaluation of the Plan ensured by an external body?**

Exceptionally, in this Strategy, we count in the Chair on Qualitative Research of Barcelona Autonomous University, which is currently working in this process.

**Does the evaluation also include the collection of opinions and satisfaction surveys addressed to patients?**

Some existing indicators will have the patients' associations as a source of information.

**Conclusion:** Patients will have the chance of taking part in this indicators-defining process.

**5. Sustainability of the National Plan**

**Is there a specific budget attached to the National Plan? Does it ensure the long-term sustainability of its actions? What are the main sources of funding of the National Plan?**

There isn't a specific budget for any of the Health Strategies of the National Health System (SNS), since when they are approved in the Inter-territorial Board of the National Health System (CISNS) are included in the SNS budget.

FEDER considers very important to provide the Strategy on RD of the SNS with a specific consignment as is included in the Senate-reported proposal.

**Conclusion:** It is proposed and agreed to create a Workgroup from the National Strategy on RD to analyse and study the issues related to economical funding of all the proposed actions. Following this line, the patients' representatives insisted on the necessity of establishing a specific budget provision for the Strategy as well as a calendar for the actions to be executed.

**Are there specific budget provisions accompanying specific actions?**

There are specific budget provisions for the aims that need them: Centres, Services and Units of Reference (CSUR), research and innovation and others defined by AACC or the Ministry of Health, Social Policy and Equality.

FEDER considers advisable to have information related to budget provisions on RD directly implemented by the AACC.

**Conclusion:** The above-mentioned workgroup will try to identify how much of the cost of the SNS is dedicated to the attention to patients suffering RD.

## **Theme 2 - Definition, codification and inventorying of RD**

---

### Sub-Themes

1. Definition of RD
2. Classification and traceability of RD in the national health system
3. Inventories, registries and lists

---

### **Workgroup 2**

**Chair:** Ignacio Abaitua (Head of Unit. Research Institute for RD. Instituto Carlos III)

**Co-chair:** Mercedes Pastor (Director of Fundación FEDER)

**Number of Participants:** 14

#### **1. - RD definition**

**Is the EU rare disease official definition use in your country? (RDs are those where prevalence is up to 5 in 10 000 inhabitants)**

Spain has adopted the EU official definition for RD

**Are there some alternative or more specific definitions that they are replacing or using in addition to the official definition?**

The official definition has been universally adopted in Spain. However, this definition is not applicable for those diseases which do not have the same difficulties than RD, even though they are low prevalent diseases (i.e.: communicable diseases)

The term "minority diseases" is also used to avoid the stigmatization associated to the meaning of the word "raro" in Spanish. Other synonymous such as "low frequency" or "low prevalence" are also used.

#### **2. RD Classification and presence in the National Health System**

**What type of coding system is used in your country? ICD9, ICD10, SNOMED, OMIM, ORPHAN...**

International Classification of Diseases version 9 (ICD-9) is used for coding the National Discharge Registry in Spain "Conjunto Mínimo Básico de Datos" (CMBD). International Classification of Diseases version 10 (ICD-10) is used for coding causes of death at the National Institute of Statistics (INE).

Both ICD-9 and ICD-10 are sable classifications but they are not valid for the identification of all clinical existing entities. This is why, RDs are usually coded using grouping categories that include not specific codes and generally join a variable number of syndromes and diseases. Hence, RD has a low representation in these classifications and their later tracking is difficult. The IIER has assessed all possible alternatives and finally

adopted the ICD-10 for all purposes. However, the IIER has added two more digits to each one of codes included in the official ICD-10 to provide specificity and a unique code to all RD currently known.

The IIER also considers very important the use of the ORPHANTE list because it is comprehensive for all groups of diseases. This is why; the IIER is working on a new system of relationships between the ICD-10 – with the two added digits - and the Orphan codes provided by the Orphanet list. To facilitate this task, an agreement signed between these two organizations allows to the IIER the possibility of having updated Orphanet lists that are included in the IIER system. However, the lack of stability of the Orphanet list is the main difficult to deal with this list and to apply it in the NHS as the unique coding system for the RD.

One of the main difficulties to gather the needed information for developing the RD follow up is the lack of appropriated IT tools in both primary and specialised outpatient's offices. However, these offices are the clinical settings more frequently visited by the Patients with RD.

The State Reference Centre of Care for People with RD and their Families (Centro de Referencia Estatal de atención a personas con enfermedades raras y sus familias – CREER), IMSERSO, Ministry of Health, Social Protection and Equity, Spain, is planning the creation of the first national observatory of disability and people dependant due to RD. This information will be complemented with profiles of social, health and care data to the RD affected people in our country. CREER is planning to adopt the ICD-10 and the International Classification of Functioning, Disability and Health (ICF) (WHO-2001) for coding the national data of people officially classified and recognised at the moment of the official assessment procedure as disable and/or dependant.

**What are the purposes that lead to use the classification systems (i.e.: follow up, cost analysis, provision of social support ...)?**

The ICD-9 is mainly used for coding the Discharge Registry of the different NHS in Spain. On the other hand, and as it was mentioned before, the ICD-10 is mainly used for the official mortality statistics carried out by the INE as well as for epidemiological studies performed with the information provided by this institute. The ICD-10 is also used for coding diseases at the primary health care sector, in some autonomous regions.

**Is your country ready to adopt the future ICD-11 of the WHO - when it is approved in 2014-, as it was suggested by the EC in the Council Recommendations of RD report?**

Even though the ICD-11 will provide an important achievement for coding RD, it does not seem possible that it can be implemented in the Spanish NHS in due time, nor at the date internationally anticipated (2014).

**What is the level of awareness and knowledge that the health professionals have about the RD classification and coding? What type of actions could be implemented to improve them?**

In general, but particular in the RD field, the level of awareness and knowledge that health professionals have about diseases classification and coding are insufficient. However, thanks to the implementation of IT tools in the health and social areas, important progresses are being achieved.

It is desirable, to promote initiatives as the protocol DICE-APER (<http://dice-aper.semfyec.es>) developed by the Spanish Society of Family and Community Medicine (SEMFYC) and the IIER, that allows to the family doctors to use this new tool for checking if a disease is rare or not as well as criteria for decisions making. Therefore, it is important that these sorts of initiatives could be extended to other medical societies.

It would be also useful to develop specific both undergraduate and postgraduate training courses for improving the RD knowledge among professionals.

### **3. - Inventories, registers and lists**

**Are there official RD lists in your country? Is there a public and official RD registry? And/or RD specific databases (i.e.: conducted by specialised centres)? Are there either follow up projects or programs (i.e.: prevention program, studies...)? What kind of initiatives to the above mentioned issues should be adopted or reinforced in your country?**

There are not RD official lists in Spain, although the IIER is doing great efforts in that way providing the best useful, comprehensive and plausible RD list. It initially considers the Orphanet list and adds, when possible, links to the ICD-9 and ICD-10 codes.

The RD Registry and Biobank were created in 2005 by a Ministerial order. However, this initiative is not started in practical terms until the first months of 2009. Several troubles and obstacles have had to be faced on before this registry was put in place.

There are other autonomous regions registers such as the Extremadura which is on going since 2004, and the Andalucía, Canarias and Castilla-La Mancha which are in their first steps of development. Cataluña is also projected its own registry. There are also specific registers for different pathologies or group of pathologies.

#### **Conclusion:**

- The RD registry of the IIER has to be promoted to the National RD Registry of Spain.
- A legal framework establishing coordination and collaboration mechanisms among public institutions and administrations and/or private centres in order to feed the National registry of the IIER have to be developed.
- The National Registry of the IIER is well prepared and structured as a multiuse platform for allocating every Autonomous Communities registry, if they wish to do it.

**Do the registers or programs receive governmental support? How the long-term registers and databases sustainability is assured by the appropriated financial mechanisms?**

The National Registry of the IIER is placed at the ISCIII; it has been developed by funds allocated by this Institute (ISCIII) and it is managed and administrated by ISCIII staff members.

The National Registry of the IIER is belonging to the Government of Spain and it is currently sustaining by public funds. However, if the registry needs are increased, its resources should be also raised.

It should be established the needed financial mechanisms that guarantee the long-term sustainability of the National Registry of the IIER and the other autonomous registers. These financial mechanisms will imply an adequate provision of funds according with registers needs.

**Is your country participating in the development of the EU RD inventory as it is stated in the Council Recommendations of RD?**

The Spanish registers are being included in the ORPHANET inventory, even though the quality of some registers is not concordant with the criteria requested by the website. In addition, the IIER is contributing to the EPIRARE project that has been just approved by the Executive Agency for Health and Consumers (EAHC). This project aims to harmonize all strategies of patient registers at the European level. On the other hand, The Health Planning and Quality Office as Technical Secretary of the Spanish RD Strategy of the NHS has already made the official request to participate in the next Joint Action of ORPHANET, which it will be started soon. Finally, there are other groups participating in the Joint Action of EUROCAT.

## 2.1. Information and training

7. How to improve information on available care for RD in general, for different audiences
  8. How to improve access to quality information on RD
  9. How to ensure adequate training of healthcare professionals on RD
- 

### Workgroup 2.1

**Chair:** Miguel García (President of the Spanish General Practitioners Society. Sociedad Española de Medicina Familiar y Comunitaria)

**Co-chair:** Salud Jurado (FEDER Board. President of ASTTA, Asociación Andaluza Síndrome de Tourette)

**Number of Participants:** 14

Reflecting on the questions shown above, the workgroup reached to the following recommendations:

#### 1<sup>st</sup> Recommendation

**To introduce the problem of rare diseases (RD) in undergraduate education of the social sciences and health sciences degrees in order to raise the knowledge about them and to promote the awareness with the problem.**

- To propose to the University Coordination Council the curricular inclusion of the body of knowledge of the RD and free elective credits adapting initial training programs in social science and health science studies by raising awareness among undergraduate students in the particularity of the RD and the clinical genetic.
- To perform highly practical workshops, through seminars or a Practicum of last year (supported by professionals and active researchers), in different clinical disciplines that teach skills to undergraduates to enable them suspect the disease, dealing with the patient and his environment and provide them everything needed to alleviate (pharmacological and non pharmacological treatments, rehabilitation, health and social support, etc.).
- To develop national networks of knowledge from university exchange programs for Seneca students in the degrees of Health Sciences and Social Sciences with the aim of promoting positive attitudes to tackling these diseases.

#### 2<sup>nd</sup> Recommendation

**To deepen in the concept and management of RDs in postgraduate training, prioritising the patient knowledge against the disease knowledge.**

- To propose the creation of advanced accreditation diplomas covered by the management law on health professions by each autonomous community, irrespective of the establishment of specific training areas that enable in an agile way the creation of referral units formed by multidisciplinary professionals, covering groups of diseases.

**In postgraduate education, go deep into the concept and management of RD, prioritizing the knowledge about the patient over the knowledge about the disease.**

- Propose the establishment of advanced standing diplomas covered by the basic law on health professions by the Autonomous Communities of Spain irrespective of the establishment of specific training areas. Those diplomas would allow, in a quick way, setting up referral units consisting of a multidiscipline group of professionals that cover groups of diseases.
- Include monographic courses on RD and genetic counselling in the master and PhD degree programs.
- Propose the establishment of an interdisciplinary and interuniversity master on RD.
- Promote, as part of the practicum of postgraduate degree programs, the acquisition of the necessary knowledge and skills to get a professional profile best suited to the needs of individuals affected by RD.
- Promote graduate dissertations on subjects and skills related to the diagnosis and management of individuals affected by RD.
- Promote, in light of the recommendations for the new master and PhD official degrees, the creation of national and international research networks.
- Propose the establishment of grants and awards to projects or PhD dissertations on RD.
- Partially fund these educational activities to facilitate the attendance.

### 3<sup>rd</sup> Recommendation

**Encourage RD-related continuing education initiatives in the Primary Care and Specialised Care. Those must take into account the population, institutional and individual needs and include, as far as possible, the following proposals for the continuing education strategic plans of the Health Services' teaching units of the Autonomous Communities of Spain:**

- Develop continuing education modules aimed at the comprehensive care for those affected by RD, including specific protocols for primary care paediatricians to recognize the relevant signs of a RD.
- Promote the rotation of professionals (physicians, psychologists, nurses ...) among the referral centres for RD, once designated, as well as among primary health centres.
- Promote allocation of RD patients to resident physicians during their outpatient rotation period, getting involved in the patients follow up.
- To encourage the contact, cooperation and information exchange between health professionals and associations of RD patients, thus enhancing the doctor-patient communication.
- To include periods of mandatory rotation in all the aspects of genetic counselling practice for primary care residents and other clinical specialties.
- To propose to all medical scientific societies and other health disciplines the establishment of a "Rare Diseases Section", according to the conclusions of the Senate Presentation in charge of analyzing the particular situation of RD patients.
- To include genetic counselling consultations in all the hospitals and health centres and/or a genetic specialist, as it is legally established.
- To include a section devoted to the RD in the different mechanisms to rate resident physicians (resident's book, portfolio, annual report...) that should be properly completed by the end of the training period.

### 4<sup>th</sup> Recommendation

**To give an official certificate to professionals who work in genetic services or units.**

- To create a National Commission for the Clinical Genetics Specialty, according to the proposal submitted by the Asociación Española de Genética Humana (AEGH) to the Ministry of Health, Social Policy and Equality.

**5<sup>th</sup> Recommendation**

**To promote awareness campaigns about RD and help to generate visibility and information resources. Make the information more accessible to health professionals, those affected, all the sectors involved and, ultimately, the society in general.**

- To update, provide content and enhance the Rare Diseases National Registry, developed and managed by the Rare Disease Research Institute, Carlos III Health Institute (ISCIII-IIER). It must be provided by specific content related to our country as epidemiology maps, associations, experts, reference centres, etc., or any other similar system that would be implanted.
- To make known and promote the Service of Information and Orientation about Rare Diseases (SIO) as a reference service for both affected people and relatives, and professionals and others, by means of collaboration agreements between this service and the central and the regional governmental administrations, with the aim of guarantee its continuity and offer to the administrations an information service about rare diseases.

**6<sup>th</sup> Recommendation**

**Elaboration of "Guides for healthcare of RDs" with the aim of homogenize the actions of the different health care professionals from primary health care and secondary healthcare, and with specific information for affected people, relatives, caregivers and teachers.**

- To request to the Ministry of Health the elaboration of a "General Clinical Practice Guideline of RDs" by Guía Salud.
- To make clinical practice guidelines about diagnosis groups of RDs which include the natural history of the different RDs from each group.
- To develop specific guidelines for those RDs that were prioritised according its magnitude and complexity with the support of the Consejerías de Sanidad (regional healthcare authorities), scientific societies and patient associations. It should be updated every 3-5 years and be available on line in the sites of the Consejerías de Sanidad and Ministry of Health.

**7<sup>th</sup> Recommendation**

**To promote the utilization of the new technologies for a quick access to the specialised information and personalised counseling to the affected person and his/her environment.**

- To use Internet as a mean to promote coping resources among the people affected by a RS and their relatives.
- To facilitate the creation of on line networks to share experiences, information and strategies.

- To promote that the institutional sites include validated links related to the formation-information of the RDs.

### **8<sup>th</sup> Recommendation**

**To favor the creation of a coordination system that includes an accessible directory of formation-information resources that implies the creation of a committee to validate resources and that includes every involved agent.**

### **9<sup>th</sup> Recommendation**

**To make activities of formation specifically designed for affected people and relatives with the aim of acquire theoretical and practical knowledge about different diseases from a biological-psychosocial-societal point of view.**

## **Theme 3 –Research on RD**

---

### **Sub-Topics**

1. Mapping of existing research resources, infrastructures and programs for RD.
2. Needs and priorities for research
3. Fostering and supporting the participation of national researchers and laboratories, patients and patients' organisations in EU-wide projects
4. Sustainability of research on RD
5. EU collaborative research on RD

---

### **Working Group 3**

**Moderator:** Francesc Palau (Scientific Director of the Biomedical Research Network for RD CIBERER)

**Co-chair:** Isabel Campos (FEDER Board. President of Asociación de Ataxias de Castilla La Mancha (ACMA))

**Number of Participants:** 14

#### **1. Mapping of existing research resources, infrastructures and programs for RD**

**Evaluation of RD research resources and infrastructures across different disciplines and sources of funds, both public and private. Considering whether a combination of private and public support is feasible.**

There are many resources and infrastructures for biomedical research in general, so it does not make sense to duplicate or segregate. Existing resources are generalists but usable in all fields, including RD.

The most quantifiable resources are the research groups dedicated to research on RD and the existing programs (see the next section).

Mixed public and private funding for biomedical research is not only feasible but desirable and necessary. In the recent past, Farmaindustria showed an interest in investing in R&D in the RD field, but this initiative did not materialize.

Related to private-public partnership, it is worthy of note, the role played by the Patient Advocacy groups as promoters and collectors of resources.

The centre for Biomedical Network Research on RD (CIBERER) could serve as possible enterprise incubator

#### **Is there a specific programme for funding or recruiting young scientists on RD research?**

- Specific national agenda: Consolider action / CIBER, launched in 2005 by the Ingenio 2010 Programme. CIBERER Budget 2007-2010: 35 M €.

- Regional / autonomic specific program (public non-governmental Foundation): 18th Marathon TV3 (2009).
- Non-specific national program with priority allocation to R&D in RD: Health Strategic Action (AES) of the 2008-2011 National Plan sets research in RD as a priority line.
- The National Program of Fundamental Biology provides funding for research in non-oriented basic research projects, so it funds RD research indirectly.
- Calls for regional R&D projects, which can include research projects in RD.

### **What is the scope of patient-driven research?**

- There is research funded by patients' associations and foundations which allow allocating funds to specific social or scientific research projects.
- Patients' associations should take a more proactive role as researcher "partners". They should also act upon the promotion of specific calls on RD.
- Dialogue and the search for forums where researchers have a chance to explain the results of their research and its real impact should be encouraged.
- The improvement of legislation concerning patronage should be promoted.

### **2. Needs and priorities for research in the field of RD**

#### **Assessing needs and priorities for basic, clinical and translational research, as well as priorities for social research in the field of RD**

Research needs and priorities are defined within the RD CIBER, the National Plan and National Strategy on RD of the NHS. The latter two refer not only to priorities in basic and clinical research but also in the social field, information systems, etc.

### **3. Promoting interest and participation of national laboratories and researchers, patients and patient organizations in research projects on RD**

#### **How to make the link between basic and translational research and Centres of Expertise?**

- Create a dynamic information system regarding research projects in RD with information related to diseases, researchers, institutions, budget, and type of research...
- Establish collaborations in order to capture data from funding agencies: Instituto de Salud Carlos III, Science and Innovation Ministry, Health and Social Affairs Ministry, Autonomous Communities.
- Consider placing the database in the CIBERER.
- Facilitate the intervention of patient associations as mediators to facilitate the contact among their primary care physicians and specialised clinicians and researchers.
- Promotion by the CIBERER of a system of clinical networking by incorporating clinical research groups through an agreement with the institutions where the groups are located.
- Establish mechanisms that allow researchers to fully integrate within clinical services and clinicians to devote time to research without compromising care.

#### **Promoting interdisciplinary approaches to research**

Participation of patients, clinicians, basic and clinical researchers and industry at all meetings and symposiums on RD should be systematically encouraged.

#### **Strengthening the exchanges among patient organisations**

- Patients are real experts in their disease manifestations, its course and its rehabilitation and support needs.
- Their participation may be key to fostering knowledge sharing and cooperation in identifying research topics in need of greater dedication. Their input is also important in order to identify research areas that may have a transverse component of interest to other RD.
- Members of patient associations can also get researchers in contact with patients to participate in research or clinical trials.
- Promote and collaborate in the establishment of patient registries and cohorts.

#### **Develop specific programs for funding or recruiting young scientists on RD research.**

- Specific recruitment takes place through the CIBERER human resources and training programs.
- Non-specific recruitment is channeled through the AES (Health Strategic Action, Chapter II, human resources).
- The lack of a Genetics specialty hinders training opportunities in a field that is fundamental to the approach of RD.
- There is a strong need for the research staff at hospitals to be recognised and integrated into clinical care teams. Clinical research should be considered as part of the activities of medical specialists, at least in university hospitals.

#### **4. Sustainability of research on RD**

##### **How to ensure, through appropriate funding mechanisms, structural and long-term sustainability of research projects and research infrastructures in the field of RD? In particular in respect of public health and social research, as well as transversal infrastructures**

- Maintaining scientific programs over time, including the AES, the CIBERER and other institutions under these programs as well as keeping up transverse infrastructure through adequate financing of CIBERER as a Public Sector State Consortium, which implies and warrants the annual grant in the State's budget.
- Encouraging the creation of new programs in the field of RD. Given the peculiarities of RD, specific programs of research in RD with special emphasis on less competitive areas such as psychosocial, personal autonomy and quality of life should be desirable.
- Maintaining the Strategic Action in Health and RD as one of its priority lines in the 2012-2015 National R & D Plan.

#### **5. EU Collaborative research on ER**

### **How to foster and support the participation of national researchers and laboratories, patients and patients' organisations in EU-wide projects?**

- Increased efforts of patient advocacy groups so that research on RD is maintained as an area of priority concern in Europe and specific calls are promoted.
- Specific provision of funds so that Patients Associations are better informed and trained in order to effectively engage in decision-making.
- Provide mechanisms so that researchers (basic and clinical) have a real institutional support when presenting a European project (release time policies, management support, etc.).
- Develop efficient systems of support to researchers that encourage and facilitate participatory attitudes.
- Create channels of communication with EU Deputy Members.

#### **Conclusions**

The proposal of this working group raises the desirability of having explicit information updated annually regarding the RD research done in Spain. This information could be compiled in a national information system where all financing agencies would send information regarding approved RD projects.

Similarly, it would be desirable to have updated inventories regarding human resources, infrastructure, technology platforms, programs, and budget dedicated to RD.

Research would be greatly enhanced by the accreditation of schools, reference services or specialised units. This would also encourage participation of CSUR (Spanish Centres of Reference) in the European Network of Centres of Expertise.

Promote the concept of translational research to accelerate basic research on RD in the clinical setting as well as to channel to the researcher the needs arising from the interaction between doctor and patient.

### **Theme 4 - Standard of care for RD - Centres of Expertise (CoE)/ European Reference Networks (ERN)**

#### Sub-Themes

1. Identification of national or regional CoE all through the national territory by 2013
2. Sustainability of CoE
3. Participation in ERN
4. How to shorten the route to diagnosis
5. How to offer suitable care and organise adequate healthcare pathways for Patients with RD
6. How to ensure in CoE multidisciplinary approaches and integration between medical and social levels
7. How to evaluate CoE

### **Workgroup 4. Reference Centres**

**Chair:** Rosa Sánchez de la Vega (FEDER Board and vice-president of EURORDIS)

**Co-chair:** Rosa López (Representative of Asociación Española de Cribado Neonatal)

**Participants:** 14

#### **1. Identification of national or regional Reference Centres (RC) all over the national territory towards 2013.**

**How to assure that all the patients affected with ER have access to a RC in their country or abroad? How to support the creation of RC where necessary?**

To achieve this, it is mandatory to launch the following actions:

Before the designation o accreditation of a RC:

a) Localization of specialists

Patients' proposal: To duplicate the process done by France, where a national call for the identification of RC was launched.

b) Creation of RC by groups of diseases

Group the RD by type of involvement o similar clinical characteristics ensuring that (most of) all the RD are covered by the accredited RC.

Patients' proposal: That the "State RC for patients with RD and their families" (CREER), be in charge of the coordination of all the RC.

*The Ministry of Health of Spain has expressed its disagreement to this last proposal, explaining that the RC are part of the National Health System and have nothing to do with the RC of the Social Assistance System, to which the CREER of Burgos belongs. Therefore the CREER cannot coordinate the CSUR of the SNS from the CREER.*

c) Patients' implication in the designation of the RC

Patients' proposal: as there is a Designation Committee of CSUR, the associations of patients with RD claim to participate as experts in the designation of the RC.

Appointment calendar for RC

Develop a chronogram for the designation of the RC in RD.

Subsequent designation/accreditation:

a) Registries and itineraries: Develop a map and a Registry of the socio-sanitary resources (including the RC) all over the national territory.

b) Information and spreading: Make an appropriate spreading of the information about the existing resources.

*The Ministry of Health explained that it has already launched for the RC an information and monitoring system named "SIFCO" designated by the International Council of the National Health Service.*

c) Network: Create some operating and communication network that can establish a direct and effective connection between social and sanitary professionals involved in RD.

**¿Which are the best structures and susceptible solutions to be a Reference Centre in your country?**

**1. Recognition and identification of specialists:**

In the process of designation of RC it should be taken into consideration to identify Specialised Centres/Units that are already working with patients with RD with proven experience and quality.

**2. Organisational structure:**

It should be necessary a transversal coordinated three-level organisation:

- Regional
- National
- European

Local Multidisciplinary Hospital Units within all the Autonomous Communities, where patients with ER are seen and subsequently referred to the corresponding national RC if a suspected diagnosis of a RD has been made. In the national RC specific tests will be carried out for diagnosis, and treatment implemented if available. If a national RC is not available, the patient will be referred to a European RC specialised in his/her RD.

### **3. "Create a Case Manager/Coordinator"**

Create the figure of the "Case Manager/Coordinator" in each RC, to monitor and coordinate all of the different specialists involved in the RD and to maintain communication with the different care levels.

*The Ministry of Health thinks this role must be played by the professional who is closest to the patient and his/her family, such as the family doctor or the primary care nurse, because the figure of the "manager of the case" must be associated with the primary care level that with the RC, which will assist him/her with technical support.*

**¿How can we better apply in our country the criteria proposed in the Report of the EC Workgroup about ER, that possibly will be part of the European Directive about cross border health assistance, art. 15, for the designation of RC?**

- **The elaboration of specific criteria adapted to each RD has been considered essential, as well as the experience approach.**

*For the Ministry of Health the process must be as follows: First, criteria for accreditation of a RC must be defined, and second, once the RC is accredited, it should be officially designated. This is critical to guarantee a minimum quality in the patients and family assistance.*

**Particularly, ¿how to have the certainty that the RC are, as much as possible, experts in the specific ER, including clinical and research expertise?**

**It is necessary to have specific criteria and quantitative measurement tools such as:**

- Accreditation and evaluation standardised procedures
- Quality control procedures
- Intercommunication and collaboration (regional, national and European levels)
- Satisfaction level of professionals and patients
- Innovation, research and good practice guidelines
- Evaluation criteria to assess the experience of the RC in the diagnosis, management, and treatment of the RD of interest.

## **2. Sustainability of the RC**

**¿How to ensure, through the appropriate financial/funding mechanisms, the long-term sustainability of the RC?**

**Organization of information and coordination issues:** The patients are not represented in the Designation Committee of RC and therefore their role in the Monitoring Committee is irrelevant or secondary.

The need of a State-based Agency for the coordination of all the accredited RC in the country. This approach would optimize the costs of RC for our National Health System and a greater satisfaction of the professional and affected groups.

**By the optimization of the available resources:** Design a chronogram for the creation and accreditation of RC in RD. This approach would ensure the initial accreditation, if appropriate, the RC that already exist and have demonstrated experience and results in a specific RD or a group of RD. This network of RC will help to organize the necessary multidisciplinary approach (avoiding unnecessary consults or procedures, erroneous or unchecked information, wrong diagnoses, performing unnecessary laboratory tests and inappropriate treatments, among others).

**Improving the information and formation in RD:** Promote the use of new information and communication technologies (ICT) to spread the knowledge on RD.

**National Funding:** Funds from the "cohesion budget" of the Ministry of Health.

**European Funding:** By the European Social Budget, if there are funds for the National Plans or National Strategies for RD. Funds should be shared with other EU member states to optimize available resources.

### **3. Participation in the ERN**

#### **¿How to encourage the participation of the RC in the European Reference Networks?**

- Improving the information and formation among specialists
- Identifying the specialists and establishing an European map of resources.
- Promoting the coordination between the RC of the EU member states.
- Encouraging and supporting the creation of professional and patients' networks.

#### **¿How to finance patients and/or professionals mobility beyond national borders?**

*Ministry of Health comment: Actually if the health/sanitary need is included in the portfolio of the National Health System (SNS) and if the corresponding AC does not have the necessary resources, a patient transfer is allowed to a specialised centre (RC) of the SNS in another AC if a justified clinical request is filed, and, in case of lack of national resources, to a RC in another EU country.*

Patients' opinion: The problem is that the high specificity of many RD makes difficult to include it in the Hospital Services Portfolio. Prior authorization for derivation and differences in normative among AC pose a serious problem in Spain, especially in cases that are urgent to treat.

### **Workgroup 4.1. Care Model**

**Moderator:** Feliciano Ramos (President of the Spanish Society of Human Genetics (Asociación Española de Genética Humana –AEGH))

**Co-moderator:** Isabel Calvo (President of FEDER)

**Number of participants:** 12

### **4. How to shorten the way to diagnosis**

- Primary care doctor (Family Doctor/Pediatrician): Basic (undergraduate and postgraduate) and continued formation in Rare Diseases (RD). Mandatory. Credits system in continuing education (added value for professional career)
- Actuation protocol (groups of pathologies) in RD
- Immediate access to available resources in RD (professionals and patients associations)
- Diagnostic laboratories accredited with qualified staff (specialists)
- Facilitate the transference of resources among Autonomous Communities (ACs)
- Create the figure of the "Case Manager" in each AC.

Promote the treatment of the patient in their geographic area (mobility of techniques and knowledge).

- Knowledge: ORPHANET, FEDER, AEGH, etc, webs; conferences, courses and meetings
- Techniques: Use of New Information and Communication Technologies (NICT)
- Good practices experiences
- Disclosure through Health/Sanitary Systems of the different administrations. Led by the Ministry of Health and accredited Reference Centres (RC), supported by patients associations of RD.
- Include in the Portfolio (Contract-Programs) of the nation 's major hospitals
- Include this activity among the criteria to recognize a RC
- Adequate and realistic budget

Identify, network and finance diagnostic laboratories working in RD at a national level.

- Directory of accredited RC available to general public (professionals and patients)
- Listings of Centres/Genetic Diagnosis Laboratories (i.e. AEGH)
- Financial support: AC and/or National Health System (Ministry of Health) (include the portfolio –cartera de servicios- of Hospital Services)
- Specific budget

DNA and samples exchange. Reimbursement. E.U/International.

- Contracts with courier companies- Specific shipping codes
- Introduce a flexible and clear process for reimbursement of services between the different administrations to avoid delays in the care of patients with RD
- Reimbursement of S&H expenses and the cost of the genetic studies: Inter-CA Council and National Health System. EU Directory: Across-borders Healthcare System.

Support for European guidelines on newborn general or targeted screening tests.

Mechanisms to develop the implantation of common protocols and recommendations on diagnostic tools, medical care, education and social care of patients and families with RD.

- EU Protocols and Recommendations: Appoint a "Commission of Experts" (which include Spanish and European members)
- Diagnostic tools. EUROAGENTEST, DYSCERNE-DDS, etc.
- Healthcare: Competent professionals: Clinical Genetic Specialists (physicians and non-physicians), basic genetic training for other specialists; Public Health Specialists: Genetic Counselors, etc.
- Education and social care: Information for parents, teachers, educators, speech-therapists, OT (Occupational Therapy) professionals, etc.
- More understandable medical/health reports that include the actual impact of RD
- Involve the "evaluation committees" for impairment / disability / dependence in the follow-up process of patients with RD

## **5. How to offer an appropriate care and organize appropriate ways for health-assistance to patients with RD**

Adoption of clinical itineraries. Inter-disciplinary query funds to cover patients/families transfer costs.

- Clinical itinerary: General derivation scheme (flowchart): from primary care medicine (pediatrician/family doctor) to the hospital (clinical geneticist + necessary specialists). Send the patient back to Primary Care level for monitoring and treatment, coordinated with the social-sanitary and educative services.
- Coordination between AC.
- Financial support: AC and/or National Health System (Ministry of Health)

Link medical knowledge of specialised centres to primary care clinics and social-sanitary/education professionals.

- On-site: Bilateral/multilateral meetings, conferences, etc. Aim: coordination of the comprehensive interdisciplinary approach of patients and families with RD.
- Virtual: Printed documentation (protocols, leaflets, handouts, etc); Internet resources (ORPHANET, FEDER, DICE-APER-SEMFYC); telephone contacts.
- Information returns from the specialised medicine to primary medicine (diverse protocols).

Cooperation between national/international experts. European reference networks. Common sanitary itineraries.

- Cooperation among Spanish experts: Internet (documents, protocols), reunions, meetings (monographic in RD, specialty meetings –sessions, round tables, monographic conferences on RD-), TV conferences.
- Cooperation between European experts: Internet (EUROGENTEST, DYSCERNE-DDS, etc.), congresses, meetings, TV/web conferences (NICTs).

## **6. How to ensure multidisciplinary approaches and coordination among the RC and the health medical and social levels.**

In the Reference Centres (RC):

- Include in Services Portfolio of Hospitals and Primary Care Centres
- RD Committee (Hospitals, Health Districts, AC)
- Conditions or criteria needed to recognize RC

Integration between medical and social care:

- Implant a socio-sanitary integrated system (National/AC)
- Involve the Educational and Occupational Services

### **Conclusions on the Care Model Topic**

The Care Model must be:

- Integral (social, sanitary and educative)
- Interdisciplinary (health and non health professionals)
- Coordinated (AC): Coordination of health, social, educative, and occupational services for individuals with RD and their families.

It is mandatory to create a Directory of Centres/Services/Reference Units accessible to families and professionals (competent and specialised in all the issues related to the care of individuals with RD and their families.

- Establish a compromise by portfolios (contract-programs) to participate in European Networks related with RD. Facilitate professionals' mobility.
- Facilitate the mobility of biological samples and the availability of informatics tools for tele-knowledge. Considerate specific budget (European funding, national funding).
- Organization flow charts and specific budget to speed the creation and guarantee the operational maintenance of the so-called "health highways".
- Supra-autonomic level (national) and supra-national (EU). Specific inclusion of the RD in the "Cohesion Budget" of the Spain's Ministry of Health, which must fulfill visibility and clarity criteria (what, how much, and how).
- Make compatible and harmonize the electronic clinical chart for RD. Use the telemedicine. Consider specific funding for this issue.
- Implant and fulfill protocols and clinical guides that allow the incorporation of new technologies and the network of different care levels to patients with RD. Assign specific budget.
- Create the "Case Manager"/Follow-up of RD. Mechanisms that ensure that care of persons with RD does not pose a financial burden or penalize the centre's budget neither the professionals involved.
- Ensure the multi-interdisciplinary assistance to individuals affected with RD and their families.
- Create the "National Observatory for RD" (*proposed by FEDER*). Specific budget needed.
- Neonatal screening. Homogeneous screening criteria based on the scientific evidence.
- Ensure the accessibility to genetic counseling. Official recognition of the Specialty of Clinical Genetics.

Creation of Units and/or Clinic Genetic Services. Law of Biomedical Investigation (14/2007) that regulates diagnostic genetic testing. Accessibility from all the healthcare levels.

- Genetic testing quality. Fulfillment of strict quality control evaluations. Implement external quality control programs.
- Directory of clinical diagnostic laboratories (genetic and non-genetic). Mandatory. Public accessibility to professionals, affected individuals and associations of RD.
- Codification system to facilitate recognition and reimbursement of services, rehabilitation treatments and drug therapies. Validate their utility.

## **4.2. Orphan Drugs (OD)**

8. Future of OD
  9. Access of Patients with RD to orphan drugs Pricing and Reimbursement
  10. Compassionate use and temporary approval of orphan drugs. Off label use
- 

### **Workgroup 4.2.**

**Chair:** Josep Torrent (Director of Fundació Dr. Robert, Associate Professor of Pharmacology, Autonomous University of Barcelona)

**Co-chair:** Moisés Abascal (FEDER Board. President of Fundación FEDER. Secretary of ADAC, Asociación de Deficiencias de Crecimiento y Desarrollo)

**Participants:** 13

## **8. Future of OD**

### **Number of marketed OD and treated patients.**

At the moment 48 OD are funded by Spanish National Health Service (NHS).

MSPSI (Spanish Ministry of Health, Social Policy and Equality) website should provide updated and accessible information to the list of definitive prices of funded OD.

Implication of all agents involved in RD (RD) in order to provide information about number of patients treated with OD.

**Obstacles to successfully have access to OD approved by UE within timeframe set in legislation (180 days). Please see comments below.**

## **9. Accessibility of OD patients to process of price regulations and reimbursement of OD.**

### **How to improve and speed up national procedures for pricing and reimbursement of OD?**

- Fast- track assessment system for evaluation of pricing and reimbursement conditions within NHS system.
- Updated transparency of decision- making procedures (e.g. website) about pricing/ reimbursement and terms of use of OD.
- OD marketing authorizations are mostly given under special circumstances or conditionally. Those are annually revised. Pricing procedures should be revised taking into account system economic sustainability in the short and long terms.

**In particular, which mechanisms should be implemented to use UE report on added clinical values of OD in order to minimize the delay in access to OD during the national decision-making process about OD pricing/ reimbursement?**

- Evaluation on added value of OD at European level is done by the EMEA with the involvement of national experts and based on public reports.
- Reports mentioned above are used by Spanish State Office for Pharmacy Services and Medical Devices (AEMPS) to set the prices of OD. However, these decisions are re-evaluated by Spanish Autonomous Communities, not following the same criteria when evaluated. It would be advisable for the regional health administrations to work together in a coherent and coordinated way in order to avoid delays and minimize the lack of equity in the access to OD treatments

**How to promote a national policy on pricing/ reimbursement of OD based on recommendations concerning the UE High Level Pharmaceutical Forum "Improvement of access to OD"?**

- Promotion of early dialog (with approval from EMEA) between industry and administrations, including HTA (Health technology assessment) in order to use common criteria to assess efficacy and added value provided by OD.
- Creation of a cohesion fund at national level by the Spanish Health Service is recommended, in order to guarantee equal and fair access to OD in all Spanish Autonomous Communities.
- Creation of specific budgets at regional level that would avoid negative impact on local hospital budgets, especially in RD centres/ services/ units.

**Access to OD through Reference Centres.**

It is considered essential to provide priority access to OD through Reference Centres.

**Participation at EU level in the assessment of added clinical value of OD at the EMEA.**

Spanish Government is involved at the moment along with others EU members at EMEA and UE working groups and committees established with regard to OD.

**10. Compassionate use and temporary approval of OD. Off label use.**

**How to promote access to OD through compassionate use?**

- Compassionate use (case- by- case or cohort use) is a critical issue in the development and early access to OD under investigation.
- Spanish Agency for medicine and medical devices (AEMPS) should be responsible of transmitting information and professional formation regarding procedures and requirements needed for applying for compassionate use according to Spanish regulations currently in force (Spanish Royal Decree).
- All decisions regarding issue mentioned above should be made public in the AEMPS website.
- Harmonization of accessibility criteria in the different Spanish Autonomous Community Regions.

- Participation and collaboration of patient associations in the decision- making process is recommended.

**¿Could off- label medicines be prescript and reimbursed if there is evidence of a benefit to patients?**

This question is of relevant importance for OD, since its use should be promoted by AEMPS in collaboration with scientific and professional organizations, along with patients according to legal regulations currently in force.

**Which measures could be studied and implemented to provide another treatment, besides medicines, when there is evidence of a benefit to patients?**

Encouraging patient associations participations in analyzing, identification, evaluation and prioritisation of the need for adjuvant drug treatment, including among others, cosmetics, special nourishment, medical devices, such as the working group created by the National Strategy in Rare Disease.

## **Theme 5 – Patient Empowerment**

### Sub-Themes

---

1. Involvement of patients and their representatives in decision-making processes in the field of RD
  2. Support to the activities performed by patient organisations
  3. Specialised social services: Respite Care Services; Therapeutic Recreational Programmes; Services aimed at the integration of patients in daily life
  4. Help Lines
- 

### **Workshop 5.1. Participation of Patients**

**Chair:** Tomás Castillo (FEDER Board. President of Federación Española de Fibrosis Quística)

**Co-Chair:** Pedro Serrano (Member of EUROPLAN Project in Spain. BURQUOL-RD project Co-leader)

**Number of participants:** 11

#### **1. Involvement of patients and their representatives in decision-making processes in the field of RD**

**Are involved both, patients and their representatives in decision making processes in the field of RD?**

Both, patients and their representatives (when needed) should participate in national and regional plans or strategies, in information provision, in establishing and management of reference centres / European Reference Networks, in defining Health Policy Research on RD.

Increasing participation of patients and their representatives should occur in other areas such as the development and management of disease registries, clinical trials, assessment of the clinical added value of drugs, educational and therapeutic programmes, professional training of physicians, nurses, social workers and paramedics as some of the most relevant health care professionals.

**It is fundamental:**

**1.1. To improve the perceived value of patient associations of RD by:**

- The patient associations themselves, taking on their corresponding leadership.
- Governmental organisations, health care professionals and researchers.

**1.2. To develop a more active role, moving from Consultation to Collaboration in all processes related to RD: Health planning, policy development, research design and implementation and assessment of health care outcomes.**

- Patient empowerment through leadership training.
- To establish POLICY GUIDELINES (Circular, Order, Decree, etc.) for institutionalizing the participation (collaboration) of the Associations of People with RD in the design, goal setting, monitoring, etc., of policies.

**For this, is important to consider the basic Principle of encourage** the transition of people with RD from people who is CONSULTED, to people who is ACTIVELY COLLABORATING in:

- The development and evaluation of national and regional strategies for RD.
- The development of Information Systems and information provision.
- The establishment and Management of the Reference Centres.
- The definition of policy and research objectives in RD.

**How to ensure, through appropriate funding mechanisms, the representativeness of patients in decisions about the processes to be performed related to the RD?**

- Provide funding for participation: Participation costs money as well as time. This is not to remunerate the time, but to offset the costs involved participation: telephone, transportation costs...
- Mechanisms for public funding from income tax, in which citizens choose which organization wants to allocate part of their taxes (PIT 0.7)
- To promote the practice of corporate social responsibility of enterprises.
- To establish partnerships with the pharmaceutical industry within a transparent and ethical framework.
- To provide to RD Associations of tools to facilitate their work and improve the quality of their management: RD registers database, instruments to assess the social-economic impact of RD developed by BURQOL-RD project.

**2. Support to the activities performed by patient organisations**

**How to support the activities carried out by the patients' organizations?**

- **Awareness:** Looking for more free space in the media. Linking leaders or celebrities who can make the RD more visible.
- **Training and Education:** Developing training programs for the leaders of patients associations, to improve the leadership, organization and coordination skills. This objective should be in line with the Strategic Plan of FEDER. Co-financing by regional governments.
- **Exchange of information and good practice:** Promoting of meetings, training sessions and exchange of best practices to improve the results of the patients' associations' management, to share knowledge and ideas.
- **Co-financing** by the national and regional governments to maintain the infrastructures developed in the field of information.
- **Important potential role of CREER** in promoting this exchange. Annual Training School with these exchanges.

**How to promote networking?**

- Promoting the call of events that create models of participation, encouraging those projects that use networking and posing it as a condition for granting aid.
- Outreach of the most isolated patients
- Support from the existing social service networks at the local level (social workers at the city halls)
- Information system for professionals, who are in touch with the needs of the population, can be very useful for the detection and monitoring of many situations.
- Primary Care and the role it can play in identifying isolated cases of persons with RD.

**What mechanisms can be implemented to support the activities of empowering patients and their representatives in all EU bodies?**

- The European Forum of People with Disabilities is a good example of how one platform influences the decisions of the European institutions.
- EURORDIS could have that role as a forum for ongoing initiatives to the European Council, the European Parliament and the European Commission. A strong alliance Eurordis - European Forum of People with Disabilities should be also created, as many aspirations are common.

To promote joined activities (nationally and regionally) on Members of European Parliament to ensure their involvement in the social and health policies of RD.

## **Workshop 5.2. Social specialised services**

**Chair:** Begoña Ruiz (Responsible of the Technical Area, CREER)

**Co-Chair:** Estrella Mayoral (Director of SIO, Servicio de Información y Orientación- FEDER)

**Number of participants:** 12

### **3. Social specialised services: Respite Care Services; Therapeutic Recreational Programmes; Services aimed at the integration of patients in daily life**

#### **What kind of programmes supporting patients with RD and/or, in general, disabled people and their families are there in your country?**

It is necessary to say that, in Spain, aids and benefits depend directly on disability recognition or on dependency valuation, those valuations show some difficulties for the affected by RD, since the disability degree of these disorders is extremely complicated to be determined, due to factors such as the lack of knowledge, they are chronic, the acute processes, among others.

#### **Relevant to Social Services:**

- Community Social Services (Information, Valuation, Guidance and Advice / Home Help
- Help Line Service
- Social Specialised Services: (Centres for Valuation and Guidance of disabled people, Resident Centres, Day Centres and Respite Care Programme)
- Tutelage Institutions
- Promotion of Personal Autonomy and Attention to Dependency

#### **Relevant to National References Centres: National Reference Centre for People with RD and their Families:**

The National Reference Centre for People with RD and their Families (CREER) in Burgos must promote the social and health coordination because of its own functions and the privilege of being run by the current Ministry of Health, Social Policy and Equality.

Its main role consists of addressing the National Health System with the Social Services System and integrating in an only strategy the benefits dealing with the equality of opportunity system for disabled people.

#### **Relevant to Patients' Organizations: Federations, Foundations and Associations of RD:**

The reference association is RD Spanish Federation (FEDER). Its umbrella covers over 200 associations. FEDER is the voice of 3 million people suffering RD in Spain and dedicates its efforts to promoting their rights and supporting their interests.

### **Proposals for addressing the disability valuation of people suffering RD:**

- Modifying the disability schedules to include: chronic character, degenerative processes and outbreaks (so that other factors apart from functional character should be considered).
- One-criterion application on the valuation order in all the Autonomous Communities in order to avoid inequalities.
- Working out a model report necessary for professionals (doctors, social workers, psychologists, therapists) attending to patients to provide the specific data needed by the team working on disability and / or dependency evaluation.
- More specific approach of the evaluation team to the patient's situation, with the Associations' information help.
- Updating training for the evaluation teams.
- Guidebooks for a better valuation of the different collectives' needs.

### **Proposals relevant to the accessibility of the Services:**

- National schemes allowing families and affected accessing to them can have paid licence.
- Promoting wide-conciliatory schemes.
- Training for primary attention professionals at Health Centres.
- Spreading the services through a primary attention guideline.
- Promoting patients suffering RD to join the IIER Registry
- Developing a specific guideline for educative centres and universities.
- AACC schemes assuring Total Integration.
- Including patients-attention itineraries in AACC schemes with the access to these services.

### **What can be done to improve their availability and accessibility of such services, including public funding?**

#### **Proposals for upgrading the availability and accessibility:**

- Information about the resources and services from Primary Attention to Base Social Services.
- Promoting Itinerant Primary Attention, mainly in rural areas.
- Extending the motherhood and/or fatherhood licence per dependant-cared child.

### **How are specialised social services financed? By government institutions and budget? By private initiative or patient associations?**

There is public funding, but the services management can be public or private.

#### **Proposals for better funding:**

- Users co-payment
- Taxes reduction for private entities.
- Social charity involvement in the access-related-to-services projects.

#### **Proposals for assuring the sustainability of the social services:**

- Carrying out and evaluation and real monitoring of the centres and specialised services.
- Implementing quality systems.
- Professionalizing the attention staff and evaluation of their attitudes and skills before signing them up.

- EU financial support.
- Using the current resources, increasing and reinforcing them.
- Unification of the access criterion to resources by AACC.

#### **4. Help-lines**

##### **What kind of help lines do they exist in Spain to assist Patients with RD and healthcare professionals?**

At present, the help lines in Spain are:

- FEDER Information and Advice Service
- CREER Information Service
- IIER RD Registry
- Associations Information Services

##### **How to develop or consolidate existing patient-run help lines?**

###### **Proposals for consolidating the help lines services:**

- Interaction of the different help lines.
- Service arrangement with Public Administrations (Administration funding, Entity running) and with professionals' colleges or private institutions.
- Increasing the number of full-time professionals attending this service.
- Optimizing the actual resources: support on other help lines.
- Spreading help lines through Primary Attention Guideline.
- Spreading them out in the media (radio, newspapers, television).
- Questionnaires for users.
- Carrying out researches on help lines as a whole to optimize resources.
- AACC funding through the RD National Strategy.

##### **National measures to establish the 116 European number.**

FEDER SIO and the help lines European project have signed a supporting-European 116 agreement. In Spain, it is necessary the whole AACC consensus. Other numbers steps (e.g.: 112, 016) could be followed.

##### **How are help lines financed? By private initiative or patient associations? Is there any government funding?**

- FEDER SIO is mainly public-funded.
- CREER Information Service is public-funded.
- The ISCIII Diseases Registry is funded by the own Carlos III Health Institute, shared by CIBERER.
- Associations Information Services are public and private funded.

##### **How to ensure their long-term sustainability?**

To ensure RD help lines sustainability is essential:

- Service benefit recognition (10-year experience) by Administrations supporting it as an agreed service.
- Supporting commitment in order to guarantee its upgrading and long life.
- Proper technology.

## I. Document history

Status (Draft/Reviewed/Final)	<b>Final</b>
Version n°	
Author(s)	<b>Federación Española de Enfermedades Raras (FEDER)</b>
Reviewer	
File name	<b>2011_01_EUROPLAN _ INFORME FINAL</b>



De izquierda a derecha, Miguel Ángel Ruiz Carabias (director general de CREER), Isabel Calvo García (presidenta de FEDER) y Jesús Norberto Fernández Muñoz (subdirector general de Ordenación, Planificación y Evaluación del IMSERSO).



La italiana Simona Bellagambi (representante de EURORDIS) destacó que se trata de un proceso en marcha para la Unión Europea.



Ignacio Díaz Azcárraga, gerente de Servicios Sociales de la Junta de Castilla y León.



Durante dos días intensos, los participantes enriquecieron el debate con sus opiniones.



Los asistentes reclamaron el reconocimiento de nuevas especialidades (genética, neuropediatría,...) y alertaron sobre la autoconplacencia.



Las sesiones tuvieron traducción simultánea en lenguaje de signos.



Moisés Abascal, presidente de la Fundación FEDER, explicó que de los 68 medicamentos aprobados por la EMEA en España sólo hay 45.



Francesc Palau, director general del CIBERER, insistió en que, para avanzar, conviene saber lo que tenemos.



Jesús Norberto Fernández Muñoz, subdirector general de Ordenación, Planificación y Evaluación del IMSERSO.

