

# SPAIN

## EUROPLAN NATIONAL CONFERENCE

### FINAL REPORT

20-21 November 2014, Madrid

# FOREWORD

**The EUROPLAN National conferences are aimed at fostering the development of a comprehensive National Plan or Strategy for Rare Diseases addressing the unmet needs of patients living with a rare disease in Europe.**

These national plans and strategies are intended to implement concrete national measures in key areas from research to codification of rare diseases, diagnosis, care and treatments as well as adapted social services for rare disease patients while integrating EU policies.

The EUROPLAN National conferences are jointly organised in each country by a National Alliance of rare disease patients' organisations and EURORDIS – the European Organisation for Rare Diseases. For this purpose, EURORDIS nominated 10 EURORDIS-EUROPLAN Advisors - all being from a National Alliance - specifically in charge of advising two to three National Alliances.

**EUROPLAN National conferences share the same philosophy, objectives, format and content guidelines.** They involve all stakeholders relevant for developing a plan/strategy for rare diseases. According to the national situation of each country and its most pressing needs, the content can be adjusted.

During the period 2008-2011, a first set of 15 EUROPLAN National Conferences were organised within the European project EUROPLAN. Following the success of these conferences, a second round of up to 24 EUROPLAN National Conferences is taking place in the broader context of the Joint Action of the European Committee of Experts on Rare Diseases (EUCERD) over the period March 2012 until August 2015.

The EUROPLAN National Conferences present the European rare disease policies as well as the EUCERD Recommendations adopted between 2010 and 2013. They are organised around common themes based on the Recommendation of the Council of the European Union on an action in the field of rare diseases:

1. Methodology and Governance of a National Plan;
2. Definition, codification and inventorying of RD; Information and Training;
3. Research on RD;
4. Care - Centres of Expertise / European Reference Networks/Cross Border Health Care;
5. Orphan Drugs;
6. Social Services for RD.

The themes “Patient Empowerment”, “Gathering expertise at the European level” and “Sustainability” are transversal along the conference.

## I. GENERAL INFORMATION

<b>Country</b>	Spain
<b>Date &amp; place of the National Conference</b>	20-21 November 2014 Ministry of Health, Social Services and Equality (MSSSI) Madrid
<b>Website</b>	<a href="http://www.enfermedades-raras.org">www.enfermedades-raras.org</a>
<b>Organisers</b>	Federación Española de Enfermedades Raras (FEDER)
<b>Members of the Steering Committee</b>	<p><b>Juan Carrión</b>, President of FEDER</p> <p><b>Simona Bellagambi</b> EURORDIS Advisor for the EUROPLAN project</p> <p><b>Paloma Casado</b> Deputy Director General of Quality and Cohesion Directorate General of Public Health, Quality and Innovation Ministry of Health, Social Policy and Equality (MSSSI)</p> <p><b>Carmen Pérez Mateos</b> Directorate General of Basic Services of the National Health System and Pharmacy Ministry of Health, Social Policy and Equality (MSSSI)</p> <p><b>Aitor Aparicio</b> Director of the National Reference Care Centre for People with Rare Diseases and their Families (CREER) Directorate General of Social Services and Equality Ministry of Health, Social Policy and Equality (MSSSI)</p> <p><b>Manuel Posada de la Paz</b> Director of IIER Institute of Health Carlos III</p> <p><b>Ángel Abad Revilla</b> Head of Patient Information Area Directorate General of Patient Health care Department of Health Autonomous Community of Madrid</p> <p><b>María Luisa Arteagoitia González</b> Director of Health Planning, Management and Assessment Department of Health</p>

	<p>Autonomous Community of the Basque Country  <b>Arancha Gordo</b>  National Centre for Educational Innovation and Research (CNIIE)  Ministry of Education, Culture and Sport  <b>Roser Vallès</b>  Director General of Health Management and Regulation  Department of Health  Autonomous Community of Catalunya  <b>Pilar Magrinyà Rull</b>  Directorate of Health Care  Catalan Health Service  <b>Luis Cruz</b>  Spanish Association of Laboratories of Orphan and Ultra-Orphan Drugs (AELMHU)  <b>Teresa Pampols</b>  Coordinator Committee of Experts of FEDER  <b>Justo Herranz Arandilla</b>  Member of Executive Board of FEDER  <b>Begoña Martín Laucirica</b>  Member of Executive Board of FEDER  <b>Alba Ancochea Díaz</b>  Director of FEDER</p>
<p><b>Names and list of Workshops</b></p>	<ol style="list-style-type: none"> <li>1. Methodology, Governance and Monitoring of the National Plan</li> <li>2. Definition, codification and inventorying of RD <ol style="list-style-type: none"> <li>2.1. Definition, codification and inventorying</li> <li>2.2. Information and Training</li> </ol> </li> <li>3. Research on RD</li> <li>4. Care for RD - Centres of Expertise and European Reference Networks for Rare Diseases <ol style="list-style-type: none"> <li>4.1. Centres of Expertise</li> <li>4.2. Care Model</li> </ol> </li> <li>5. Orphan Medicinal Products</li> <li>6. Social Services for RD</li> </ol> <p><b>Additional Workshops:</b></p> <ol style="list-style-type: none"> <li>7. Labour Market Inclusion</li> <li>8. Educational Inclusion</li> </ol>
<p><b>Workshop Chairs (and Rapporteurs, where applicable)</b></p>	<p>Workshop 1: Juana M<sup>a</sup> Saénz, Julián Isla  Workshop 2:  <ol style="list-style-type: none"> <li>2.1. Mónica Rodríguez, Gema Chicano</li> <li>2.2. Isabel Motero, Vanesa Pizarro</li> </ol> Workshop 3: Teresa Pampols, Francesc Palau</p>

	<p>Workshop 4:            4.1. Begoña Martín, Ignacio Blanco            4.2. Enrique Galán, Justo Herranz            Workshop 5: Juan Manuel Fontanet, Luis Cruz            Workshop 6: Miriam Torregrosa, Tomás del Castillo  <b>Additional Workshops:</b>            Workshop 7: Fernando Torquemada, Modesto Díez            Workshop 8: Natividad López, M<sup>a</sup> Carmen Murillo</p>
<p><b>Annexes :</b></p>	<p><b>I. Programme in English</b>  <b>II. List of Participants</b>  <b>III. Photographs</b></p>

## II. MAIN REPORT

### Plenary Report – Opening Session

EUROPLAN National Conferences aim to encourage the development of National Plans or Strategies for Rare Diseases addressing the unmet needs of patients living with a rare disease in Europe.

On 3 June 2009 the Spanish Ministry of Health and Social Policy announced the establishment of a National Strategy for RD within the National Health System (NHS). An update of the Strategy was adopted in June 2014.

In 2010, the First Spanish EUROPLAN National Conference was held with the aim of getting to know and analysing the situation of Spanish policies on RD, taking as reference the European Guidelines for the Council Recommendation on a European Action in the field of Rare Diseases.

Four years later, the Spanish Federation of Rare Diseases (FEDER) organised the Second Spanish EUROPLAN Conference using the same methodology, to assess RD issues, the progress made in rare diseases and the challenges and needs that must be met.

The EUROPLAN II Conference in Spain was held at the Ministry of Health, Social Services and Equality (MSSSI) on 20 and 21 November 2014.

Paloma Casado, Deputy Director General of Quality and Cohesion from the MSSSI, opened the Conference. She stressed the importance of participation and implication of all parts involved to improve the National Strategy for Rare Diseases launched by the Ministry of Health in 2009. She also declared that the lines of work proposed at the I EUROPLAN Conference, held in 2010, had served as a guide for further work. The Ministry representative said, "EUROPLAN is a good starting point for further progress along these lines seeking at all times to improve."

Juan Carrion, the President of FEDER, declared that "EUROPLAN transmits the urgent and imperative need for Rare Diseases to become a social and health priority in Spain." He further stressed the importance of networking as "the true key to learning more about these diseases and the key to improving the quality of the family life".

Simona Bellagambi, EURORDIS Advisor for EUROPLAN, spoke of RD as a challenge in Europe, describing the framework of EU policies on TD and highlighting the most important pillars of the Council Recommendation.

She also spoke of the EUROPLAN project, giving a vision from EUROPLAN I to this second stage EUROPLAN 2012–2015, which is part of the "EUCERD Joint Action: Working for Rare Diseases". The aim is to support and coordinate 24 National Conferences to promote the National Plans/Strategies on RD organised by National Alliances in Europe.

She concluded saying that the purpose of these conferences was to search for results to work on the national strategies in each country, and the conclusions of each country would help with other conferences at European level and serve as an example of good practice.

## Workshop Reports

### Theme 1 - METHODOLOGY, GOVERNANCE AND MONITORING OF THE NATIONAL PLAN

#### Sub-themes:

- Policies and resources mapping
- Development and structure of a national plan/strategy
- Structure of the national plan/strategy
- Steering committee and other governance mechanisms
- Transparency of the governance structure
- Dissemination and communication of the national plan
- Monitoring and evaluation of the national plan
- Sustainability of the national plan
- National Plan financial support

#### Background and current situation:

The Strategy is a well-structured document, but more like a statement of intentions than a concrete workable plan.

The aim of the Strategy is to help improve the services provided throughout the national territory for patient with rare diseases and their families, based on the principles of quality, fairness and coherence.

Coordination structures include a Technical Committee and an Institutional Committee responsible for management of the Strategy:

- The Technical Committee is made up of representatives of patient's associations and scientific societies, selected by consensus on the basis of their scientific knowledge.
- The Institutional Committee consists of representatives appointed by the regional departments of health and the Ministry of Economy and Competitiveness (MSSSI) through Institute of Health Carlos III. There is a scientific coordinator and a technical secretary who report to the Quality and Cohesion Section of the MSSSI.

Patient participation is contemplated in certain phases of the Strategy but not all stakeholders are represented.

The Strategy includes specific actions under the umbrella of 24 general goals framed in 7 strategic lines, but no specific goals or measurable results are established, only recommendations.

- The Strategy is assessed every 4-5 years. There is a checkpoint every 2 years.
- Proposals are incorporated through the Institutional Committee but only superficially, without assigning responsibilities.
- The set of indicators established for assessment of the 2009 Strategy are like a number of questions focused on just checking that progress is being made.
- The Technical Committee has so far been efficiently updating the Strategy.
- The Institutional Committee has checked the relevance and suitability of the proposed goals, but the regional governments and their health services are responsible for managing and providing the final health care service.
- The indicators that will influence future developments have been checked, the goals and recommendations have been updated and were tested by the CISNS (“Consejo Interterritorial de Salud”, Inter-territorial Health Council) in June 2014. The indicators for the 2014 Strategy have not yet been published.
- The participation by representatives of patients and scientific societies, along with other stakeholders, was taken into account when drafting the Strategy, but no full gap analysis of patient’s unmet needs has been made.
- Although the needs of the different regions are expressed through the Institutional Committee, they do not appear to be specifically contemplated in the Strategy.

Awareness of rare diseases has grown in Spain:

- The Strategy is published on the MSSSI website, but this does not mean that the general population or patients’ families are familiar with it.
- There is an inventory of the CSUR (Centres of Reference) but not a common inventory of all existing resources.

There is no specific budget for the implementation of the Strategy objectives.

- MSSSI budget provisions are distributed among the regional governments (with the exception of Navarra and the Basque Country) to fund specific actions implementing the national plan. We consider the budget assignment insufficient and clearly symbolic.
- There is no cost estimation of the Strategy goals or associated activities.

### **Proposals and Conclusions:**

#### **Policies and resources mapping**

1. Request the regional governments to create specific plans for rare diseases, assigning the necessary structures and budget and following national plan recommendations.

2. Include patient organisations in the executive committees of the regional plans.
3. Find resources for the training of health professionals (focusing on primary health care and paediatric physicians) to raise awareness of the need for early diagnosis (public-private coalitions, scientific societies...).
4. Conduct a survey to measure and assess the level of knowledge on rare diseases among the population at large. Organise training for patients based on the results of this survey.
5. Develop performance indicators in respect of patients' unmet needs.

#### **National plan/strategy development**

6. Develop an implementation plan for the new 2014 Strategy:
  - Clear assignment of task and responsibilities
  - Measurable goals: development and adoption of performance indicators
  - Effective coordination between agents
  - Tasks execution monitoring
  - Clear goals, results and milestones
7. Provide sufficient resources to enable implementation of the national plan/strategy.
8. Create a monitoring and assessment committee to issue a status report tracking implementation of the goals of the new Strategy.
9. Create an organisation within the MSSSI to coordinate and combine the implementation and development of the proposals of the new national plan.

#### **Strategy governance**

10. Engage all agents involved in rare diseases in the Technical and Institutional Committees (geneticists, industry, scientific associations, information technology, academic bodies, etc.).
11. Publish on the MSSSI website and inform on the responsibilities, activities and conclusions of committee meetings in order to create an impact for the RD population.

#### **Dissemination and communication of the Strategy**

12. The Strategy must ensure that information on regional plans is distributed in order to guarantee an effective impact.
13. Increase awareness in the social networks and specific task forces or working parties in different areas of the development of the Strategy.
14. The national plan must be publicized by key opinion leaders.

#### **Monitoring and evaluation of the National Plan**

15. Embrace new information technologies for the implementation of regional Plans.
16. Create a scorecard to monitor indicators.

### **National Plan sustainability and Sources of funding**

17. Provide sufficient financial resources for achievement of the goals of the Strategy.
18. Request regional governments to explore the possibility of obtaining grants from EU structural funds.
19. Analyse the cost of proposals for implementing the Strategy.
20. Consider developing sustainability mechanisms for the development of new drugs and access by patients.
21. Consider the connection with the pharmaceutical industry. Explore public-private partnerships models.

## **Theme 2 - DEFINITION, CODIFICATION AND INVENTORYING OF RD**

### **2.1 DEFINITION, CODIFICATION AND INVENTORYING**

#### **Sub-themes:**

- Definition of RDs
- Codification of RD
- Registries and Databases

#### **Background and current situation:**

##### **Definition**

Spain has adopted the EU RD definition.

##### **Codification**

The Spanish National Health System (NHS) uses de International Classification of Diseases versions 9 and 10 (ICD-9 and ICD-10). Both these classifications have a stable structure but cannot embrace all existing diseases, so most RD are classified under residual categories with little representation in this classification system. ICD-9-CM is used mainly in the codification of hospital discharges throughout the NHS. ICD-10 is used primarily for mortality statistics and primary health care codification in some Autonomous Communities. The NHS will introduce soon the ICD-10- CM in the Hospital Discharge Registry. Despite the major breakthrough for RD codification, ICD-11 is not expected to be published in the EU before 2017.

##### **Registries and Databases**

In our country there is a national policy on data collection through the National Rare Diseases Registry of Institute of Health Carlos III (ISCIII), controlled by the Institute of Rare Diseases Research (IIER). This national policy is set out in both the National Rare Disease Strategy of the National Health System (NHS) which was approved by the Interregional Council in June 2009,

and in the strategy update approved by the Interregional Council in June 2014.

The National Registry has a dual purpose as a population registry and a specific patient registry for specific rare diseases or groups of RD.

The National Rare Diseases Registry is financed partly with IIER funds and partly through the budgets of regional health departments. The population registry was financed through the project “Spanish Rare Diseases Registries Research Network (SpainRDR)” project, with human resources obtained for both the IIER and the Autonomous Communities. This funding will end in a few months’ time, making it impossible to continue with the project. Regional provisions have been adopted on this matter in most Autonomous Communities.

The population registry has been boosted through the “Spanish Rare Diseases Registries Research Network (SpainRDR)” project, with the support of the International Rare Diseases Research Consortium (IRDiRC). This project strengthens the Registry development, as it has agreements with all Autonomous Communities whereby cases detected in the different regions can be included in the National Registry.

A Procedures Manual with a common methodology for all Regional Registries and the National Registry was prepared within the framework of the “Spanish Rare Diseases Registries Research Network (SpainRDR)” project. This Manual contains Common Data Elements and Minimum Data Set. Other aspects of the methodology used to ensure harmonisation of data collection are specified, such as data sources, codification and classification of rare diseases, municipal codification, etc.

Data sources used in population registries are highly varied. The data sources that provide the largest number of cases are the Minimum Data Set (CMBD), the Medical Record of Primary and Specialized Health care and the Mortality Registry. However, congenital defects registries, orphan drugs registries, metabolopathies registries, chronic kidney diseases, among others, are also used.

With regard to Patient Registries, specific records of specific diseases or groups of diseases, embracing hundreds of different diseases, were recently added to the National Registry. These Patient Registries are developed in collaboration with specialists in specific areas with considerable experience and a great interest in them.

All Patient Registries in Spain, both those developed outside the National Registry of Rare Diseases and those developed with the support of the National Registry, follow international data models whenever there are specific international registries. The ultimate aim is to establish agreements with international registries, so it is essential to follow the standards already established.

Patients can participate in the National Registry of RD by requesting their inclusion in the Registry. Once included, they are provided with a password, which allows them to obtain information about their disease; they can also complete online questionnaires and donate samples for the National Biobank of Rare Diseases, which is linked to the Registry.

The Patient Registries integrated in the National Registry are supported with IIER funds. Some of them have also received support from Scientific Societies. Patients Registries outside the National Registry are financed by Scientific Societies or with grants from the Pharmaceutical Industry.

### **Recommendations and Conclusions:**

#### **Definition**

1. Keep the European Union definition for RD, which is based on both quantitative criteria of prevalence (fewer than 5 cases per 10,000 inhabitants in the EU) and criteria regarding the severity, chronicity or generation of disability.

#### **Codification**

2. It is important to prioritize suitable codification, and although this is very complicated, Orphanet codes should be included in the national health care information system, as it is probably the most complete European inventory of RD.
3. Furthermore, development of the Electronic Medical Record is expected to be completed in Spain within the short or medium term, enabling the use of SNOMED-CT and interoperability in Spain, among other advantages.
4. The existing coding systems should be interoperable at European level, based on Orpha codes, with a single codification using the specific codifications.

#### **Registries and Databases**

5. Support the NORD, EURORDIS and CORD Joint Declaration of 10 key principles for Rare Disease Patient Registries.
6. ECONOMIC SUSTAINABILITY of the Spanish Rare Diseases Registries Research Network (SpainRDR). International interoperability of Registries and Databases is a recommendation of the EU Committee of Experts on RD to facilitate the pooling and sharing of data knowledge. We propose providing national and European budget, with priority on a national level, for the Population Registry and for the Registries Network, in order to consolidate this Registries Network.
7. Urge the Spanish political authorities to support the amendments tabled to the new European Regulation on Data Protection, which includes a high level of protection of personal data which is generally considered detrimental to research.

#### Population Registry

8. LEGAL FRAMEWORK: The basic legal framework regulating and consolidating the Spanish Rare Diseases Registries Research Network must be urgently adopted, ensuring its sustainability over time. This sustainability currently has an expiry date in the coming months, so we urge all authorities to keep their commitment to ensure the sustainability of the Population Registries.

9. It is urgent and essential to have a National Registry, guaranteeing its sustainability and an undertaking by all Autonomous Communities to participate.

#### Patient Registry

10. The involvement of all interested parties, including patients, is required in the design, support and governance of registries, for example, by providing data or sitting on Ethics Committees.

11. A greater link between the Patient Registries and the National Registry must be fostered.

#### Database interoperability for Patient Registries

12. The interoperability of the existing overlapping databases and control of duplicated entries should be encouraged and improved. Data source limitations should be improved to adapt them to the new requirements or needs as appropriate.

13. Networking will be fundamental; generosity and willingness to collaborate in the sharing of information across all the sectors involved (social, health, educational and labour, in both public and private sectors) should be praised and supported. This is the only way to improve the epidemiology information available.

## **2.2 INFORMATION AND TRAINING**

### **Sub-themes:**

- Information on RD, for different audiences
- Helplines
- Training of health care professionals

### **Background and current situation:**

#### **Information on RD, for different audiences**

#### What are the existing information sources in Spain? Are they of good quality?

- Internet. The participation of committees of experts is considered vital for assessing and deciding on what information is to be published.
- Orphanet
- Scientific societies (working parties or task forces).
- **PubMed** is a search engine providing free access to the MEDLINE database of citations and abstracts of articles on biomedical research, offered by the U.S. National Library of Medicine. MEDLINE has around 4800 journals published in the United States and over

70 countries around the world, from 1966 to the present.

- Medical consultations.
- Patient associations. It is necessary to promote the committees' work in translating not only the English but also the strictly scientific language which most patients do not understand into plain, easily comprehensible language.
- Research Centres and Institutes.
- Specialist journals. These sources of information must also be addressed to patients and their families.
- Manuals and guides.
- Universities.
- Institutional sources. It is necessary to improve institutional sources, mainly linked to the information issued on RDs by the regional departments of health.
- Others. Problems are encountered for access to quality information and resources for certain diseases, owing to the idiosyncrasies of the disease.

Proposals for improvement in respect of sources of information should be geared to:

- ✗ Publishing information in good quality networks, starting at the hospitals where the patients are diagnosed.
- ✗ Agreements between information source access services and central and regional governments to ensure widespread access through all relevant media.
- ✗ Promoting institutional websites with validated links to training and information on RD.

#### How are specific disease (or group of diseases) information networks organised?

Associations are one source of information: there are about 2,512 patient organisations in Europe, according to Orphanet. Some are organised into national alliances for rare diseases and others belong to a European organisation for a particular disease. Most are included in a European organisation such as EURORDIS, which brings together organisations from 61 countries and enables direct communication between the European Commission, players involved and patients with rare diseases. The proposal for improvement is geared to establishing:

- ✗ The grouping of networks for specific diseases or groups of diseases under the umbrella of national, European, Latin-American and global federations.

#### How are their European/international activities promoted and supported?

- The measures adopted within the European Union policies and the cooperation between Member States, are very important. In the European context, patient organisations play an active and practical role in promoting and supporting activities.

- Promotion and support of activities as a result of the national and international network structure of organisations. This structure supports activities such as: facilitating contact between people from different countries suffering with the same disease; collecting and publishing information on a specific disease at an international level; developing networks with medical professionals and cooperating with them, and patient networks; developing common projects (such as youth exchanges, summer camps, workshops, meetings and conferences); representing common political interests on a European level (or international level); promoting research, knowledge transfer, equipment and easy access to treatment; helping people with rare diseases in under-developed countries; helping small local groups to create an organisation; exchanging "examples of best practices" and guidelines for certain procedures (how to do public relations, fundraising, organise conferences, etc.); working with the European Medicines Agency, etc.
- The FEDER Information and Guidance Service, as member of the European Network of Helplines, also works with the sources it identifies for answering consultations with the help of the FEDER Advisory Committee, patients, other professionals, etc. with the following sources proposed by the Network:

- The Cochrane Library (<http://www.cochrane.org> )

Many international centres have documents on "evidence-based medicine", including a Spanish centre:

- "Iberoamerican Cochrane Centre"

Jordi Pardo Pardo, Iberoamerican Cochrane Centre, Hospital de la Santa Creu i Sant Pau, Barcelona

Abstracts are free and have been translated into Spanish. They can be consulted in alphabetical order at: <http://www.update-software.com/abstractsES/mainindex.htm> or just the most recent abstract, at:

<http://www.update-software.com/abstractsES/newreviews.htm>

Payment is required to view the full text of the documents. The Spanish Ministry of Health and Consumption has reached an agreement to provide "free universal access to the /BCP/ throughout Spanish territory".

<http://www.bibliotecacochrane.net/Clibplus/ClibPlus.asp>

- National Guideline Clearinghouse ([www.guideline.gov](http://www.guideline.gov))

Made in the United States, the largest database is, unfortunately, only available in English, but all the texts are free. Articles can be found by category or treatment.

The Information and Guidance Service (SIO) of FEDER has a protocol in which it has classified its information sources as follows:

## 12. SOURCES

### 12.1 BIBLIOGRAPHIC INFORMATION SOURCES

- DATABASES
- PORTALS
- BOOKS / GUIDES / MANUALS

- ARTICLES

## 12.2 OTHER INFORMATION SOURCES : CENTRES / REFERENCE UNITS

- Spain
- International

## 12.3 OTHER CENTRES

- Spain
- International

## 12.4 ASSOCIATIONS

- Spain
- International

## 12.5 RESOURCES ON GENETICS

### **Helplines**

What kind of helplines (all diseases) exist in Spain to assist RD patients and health care professionals?

- FEDER's Information and Guidance Service. [www.enfermedades-raras.org](http://www.enfermedades-raras.org)
- CREER's Information Service <http://www.creenfermedadesraras.es/>
- IIER's Registry of RD. <https://registoraras.isciii.es>
- Association Information Services
- DICEAPER Protocol (Primary Health care Protocol) <http://dice-aper.semfycc.es/>  
Publicising of this protocol to disability evaluators throughout Spain
- Links on recognised websites
- Telecare Services. For example, Deusto University in the Basque Country: telecare service for neuromuscular diseases since 2004

Proposals for improvement in access to helplines:

- ✗ Helplines interaction
- ✗ Arrangement with the authorities (funding by the government; service provided by the organisation), and with professional bodies or private organisations
- ✗ More professionals and full-time dedication of these professionals to the service
- ✗ Optimise existing resources: support in other helplines (e.g. "Salud Responde")

How to develop or consolidate existing patient-run helpline services for RD?

- Interaction between different helplines
- Arrangement with the authorities (funding by the government; service provided by the organisation), and with professional bodies or private organisations
- More professionals and full-time dedication of these professionals to the service

- Optimize existing resources: support in other helplines (e.g. “Salud Responde”)
- Work common topics online (uncertainty, managing emotions, isolation, helplessness, etc.), but also taking into account the difficulties that arise when dealing with heterogeneous groups
- Support Associations with grants for helplines based on fiscal or social criteria, taking account of the cost of long-term care
- Promote funding of initiatives such as Rare Connect, <https://www.rareconnect.org/es> which connect rare disease patients globally, with a view to increasing patient networks and encouraging their development.
- Promote the funding of initiatives such as the FEDER Information and Guidance Service, which puts patients in touch with one another, not only in Spain but also in other countries through the European helpline network, as well as in Latin America; boosts training in associations; provides guidance in cases that are difficult to diagnose; provides information for patients, relatives and professionals in all areas; has 12 years of experience, 7 professionals; has never worked with volunteers; has a protocol for action with identified sources of information, an advisory committee, several agreements with entities and a database containing information on over 40,000 consultations; specific resources for RD; works with an open list established by consensus with the IIER, containing more than 1,800 diseases, many of which have not yet been coded. It is the service with most experience, which covers most information.

How to improve the service offered and improve their visibility, especially for patients?

- Publicise helplines through primary health care protocol.
- Publicise helplines through the health portals of the different Autonomous Communities and the Ministry of Health, Social Services and Equality.
- Publicity in the mass media (radio, press, television)
- Spread key messages that identify people with rare diseases: loneliness, isolation, time taken to obtain diagnosis, etc.
- Promotion of initiatives such as WOP project <http://www.walkonproject.org/>, with a massive impact, in which tens of thousands of people participate. Nowadays few people can say they do not know the X child's case of Leukodystrophy.
- High impact viral campaigns: Parallel Lives (FEDER), Shaking hands (ASEM), Bucket of cold water (ADELA) or "Get wet" (Multiple Sclerosis) campaigns, among others.
- National publicity campaigns on TV, radio, press, posters in strategic locations (hospitals, health centres, etc.), give visibility through government/civil service intranet, etc.
- Advertising campaigns with periodic reminders on key dates such as Rare Disease Day and the Days of specific diseases.

- Institutional support by celebrities and distinguished persons.

How are helplines financed? By private initiative or patient associations? Is there any government funding?

The following is proposed:

- To conduct a study on existing helplines in Spain in order to optimise resources (ensure that services are not underfunded or underutilized).
- Regional governments may request Health Strategy grants to maintain Patient Information Services.

How can their long-term sustainability be ensured?

- Interaction among the different helplines.
- Arrangement with the authorities (funding by the government; service provided by the organisation), and with professional bodies or private organisations
- More professionals and full-time dedication of these professionals to the service

What national measures can promote to reserve a 116 phone number for European RD Helplines?

- Actions taken by FEDER to promote the European helpline number 116:

Sending a letter of support for the project presented on 13 May 2013 at the meeting of the Ad Hoc Group on 116 helpline numbers within the Communication Committee (COCOM) of the European Commission, in which the Directorate General of Telecommunications represents Spain. Ángel León Alcalde, Coordinator of the Telecommunication Operators Area, Head of the Spanish delegation on the Communications Committee, contacted the Spanish Help Line (SIO) of FEDER to coordinate actions regarding Spain's position on the assignment of the number. We held several meetings and are about to inform the new Minister of Health on the project.

Status report on 116 numbers

The number 116 000 (missing children hotline) is only established in 22 Member

Request to reserve number for information on rare diseases

The proposal to reserve a European number for information on rare diseases, received by the Commission in July 2013, had letters of support and undertakings to put the corresponding line into operation from 5 countries (France, Spain, Romania, Italy and Portugal), so did not meet the requirement established in 2011 of having at least 14 supports to proceed to reservation of the number.

Spain offered to collaborate with the Commission to prove the solvency of the Spanish entity that had seconded the proposal (FEDER – Spanish Rare Diseases Federation).

The Commission agreed to contact the applicant organisation (established in France), suggesting that it could set up a network of contacts with equivalent organisations in other member states and thereby obtain the 14 supports and undertakings to establish the service as required to reserve the number.

EURORDIS and its partners continue the negotiations with the European Commission regarding the possibility of requesting the reservation of a 116 common number for rare diseases. It is, therefore, a Community initiative and should have European funding.

### **Training social and health care professionals and patients**

#### How are health care professionals trained in your country?

- With undergraduate and postgraduate degree programmes; with the inclusion in university course syllabi.
- Within the Psychology degree at the University of Deusto, knowledge on Rare Diseases is included in the subjects Neuropsychology and Clinical Evaluation. At the Universities of Seville and Zaragoza, two optional subjects are available.
- Knowledge on RD should be incorporated in PhD programmes.
- With specialised training, providing economic investment and resources to foster the exchanging of professional experiences:
  - Official Masters degree in knowledge of Rare Diseases - UNIA and University of Pablo de Olvide, Seville.
  - I Masters Degree in Integral Psychological Intervention in Rare Diseases – University of Seville
  - Courses available at the *Escuela Valenciana de Estudios en Salud (EVES)* and the *Fundación Universitat-Empresa (ADEIT)*. Implementation of a specific Masters degree in “Rare Diseases”
  - I University Masters Degree in Rare Diseases in Murcia.
  - Rare diseases awareness programme in schools and training for teachers.
  - I National Conference on Rare Diseases which has become a benchmark. Social and health professionals and families spend three days together exchanging information and experiences.
  - I Course in Social Work and Rare Diseases "A New Challenge for Social Work" organised by the School of Social Practice, within Murcia University’s School of Social Work.

- Training in Primary Health care and Hospital Health care. Greater emphasis is perhaps needed in Primary Health care, since as mentioned throughout the document, this is the main access to health care for patients with RD. Training should be addressed at professionals with different profiles who form part of multidisciplinary teams (nursing, general medicine, paediatrics, etc.), dealing with adults, children and young people.
- Training through the School of Social Sciences of Extremadura and the School of Public Administration of Extremadura. An expert from FEDER is a trainer offering an annual course for workers in the Extremadura Health Service and others on “Integral Handling of Rare Diseases”.
- Training courses in the Autonomous Communities. For example in Galicia, in the past three years, two annual training courses have been run in the past three years leading to official qualification in the prevention, detection and monitoring of rare diseases for primary health care staff of the *Servizo Galega de Saude (SERGAS)* in the *Fundación Escola Galega das Administracions Sanitaria (FEGAS)*.
- Training within scientific societies such as SEMFyC, SEMERGEN, SEMI or SEN to increase awareness and training of health care professionals. The activities of these groups so far include organising and held courses on "Train the Trainers" and PhD courses related with RD, workshops such as "Rare patients or uncomfortable doctors?", round tables on the issue of RD within their National Congresses and publication of articles on RD in the journals of different scientific societies.
- On 27 June 2012 the Interterritorial Council of the NHS approved the creation of a Network of Health Schools for Citizens, offering training courses for patients with rare diseases and their carers.

It was also proposed to:

- Collaborate and support knowledge by providing these groups with public resources (as in the case of CREER) to help raise the level of training of our health care professionals.
- Create reference groups engaged in the publication of texts (textbooks, e-books, etc.) for use in teaching. The publications should be interdisciplinary, specialising in specific pathologies and include the main bibliography.

## Recommendations and Conclusions:

### Information sources

#### PROPOSAL National Level:

1. Establishment of a coordinating body to bring together and validate existing information sources.

2. Financial support and dissemination of the information helplines, such as:
  - DICE-APER protocol.
  - Information and guidance services on rare diseases, such as the SIO of FEDER.
3. Support the request for reservation of a number in the 116 numbering range in countries involved.
4. Design a protocol for information management for professionals, patients and students by a working group made up of the Ministry of Health, IMSERSO-BELIEVE, IIER RD Registry, CIBERER, universities, FEDER and Associations. This working group would analyse existing information sources and confirm their reliability.
5. Consolidate the RD Observatory as a benchmark for data analysis.
6. Designate people to direct or coordinate follow-up of the information initiatives proposed in this regard.
7. On a general level, begin to include information on RD in school textbooks.

PROPOSAL Autonomous Communities level:

8. Obligatory declaration of RD to the Registry (or to the central regional node)
9. Support the population-based National Registry of RD ensuring continuity and sustainability of the collaboration between Autonomous Communities and IIER-ISCIII.
10. Provide health care professionals with information and access to specialist and general consultations in order to expedite special need issues in RD. This can be done through fold-outs in their clinical protocols.
11. Use patient workshops to promote and encourage information on RD to professionals in any field.
12. Autonomous Communities should provide health care professionals with access to new technologies so that they can access protocols and validated information.

**Training sources**

PROPOSAL National Level:

13. Patient empowerment through training activities in social and health policies by FEDER-CREER.
14. Guidelines for RD Care developed by working groups formed by professionals and patients, which would be endorsed by the government.
15. Training through Scientific Societies for professionals who are treating RD patients.
16. Training within the MIR [houseman] programme.
17. Training by Spain RDR

18. Promote the training of trainers through expert groups.
19. Recover the bio-psycho-social approach to RD care from all areas of intervention (humanistic dimension).
20. Agreement with the professionals who are to receive training on the development and content of the training.

**PROPOSAL Autonomous Communities level:**

21. All Autonomous Communities should facilitate access by professionals to training in tools, protocols, RD registries in the Autonomous Communities, through concrete actions such as contract programmes.
22. Inter-university and interdisciplinary postgraduate training: include information on RD in existing masters degrees on epidemiology and genetics, primary health care, general health psychology and education. Specific specialist training in bio-psycho-social care in RD.
23. Encourage the inclusion of compulsory and optional subjects providing clinical, social and educational knowledge on rare diseases in the catalogue of university degrees and non-university qualifications of Health Sciences, Social Sciences, Communication, etc.
24. Provide more training courses through health science schools corresponding to the regional departments of health and make that training and attendance of the courses compulsory for professionals in the health services.

**Theme 3 - RESEARCH ON RD**

**Background and current situation:**

**Knowledge, classification/codification, Registries and Research**

**Indicator 10. Existence of a national policy on rare disease clinical practice guideline development and implementation**

AREA OF COUNCIL REC.:		2
INDICATOR DESCRIPTION:		
TYPE OF INDICATOR:		Process
SHORT ANSWER	DETAILED ANSWER	INITIATIVES IN PROGRESS

YES	<p>YES, there is a policy on CPG development</p> <p>YES, there is a policy on adaptation of CPG</p> <p>YES, there is a policy on CPG implementation</p>	<p><b>GuíaSalud.</b> body of the National Health System (NHS) in which the 17 Autonomous Communities created in 2002 participate. Adopted by the Interterritorial Council in 2003 as an instrument for enhancing the quality of health care in the NHA, begun in Aragon. The goals of GuíaSalud are: promote the development (preparation, adaptation, updating) of Clinical Practice Guidelines (CPG) and other products based on scientific evidence (OPBE) following a common methodology; facilitate access by the CPG and OPBE to the National Health System (NHS); and foster implementation and utilisation of the CPG and OPBE, among others.</p>
-----	---	---

**Indicator 11: Type of classification/coding used by the health care system**

AREA OF COUNCIL REC.:		2
INDICATOR DESCRIPTION:		
TYPE OF INDICATOR:		Process
SHORT ANSWER	DETAILED ANSWER	INITIATIVES IN PROGRESS
<i>Type of coding used by the system</i>	ICD-9	<b>NB:</b> The Codification and Classification working group may provide detailed information on this aspect.
	ICD-10	
	OMIM	
	SNOMED	
	MESH	
	ICD-O	
	OTHERS	
The ORPHA Code is used in addition to the national code	YES	The ORPHA number is used in addition to the national code, although such use is voluntary.
	NO	In the future Joint Action on Rare Diseases (2015-2017) there will be a specific working package for implementation of the ORPHA codes in national health systems.

**Indicator 12: Existence of a national policy on registries or data collection on RD**

AREA OF COUNCIL REC.:		2&3
INDICATOR DESCRIPTION:		
TYPE OF INDICATOR:		Process
SHORT ANSWER	DETAILED ANSWER	INITIATIVES IN PROGRESS
YES	YES, for national/centralised registry and data collection	<b>NB:</b> The Registries working group may provide detailed information on this aspect.
	YES, for regional registries and data collection	
NO		

**Indicator 13: Existence of RD research programmes and/or projects in the country**

AREA OF COUNCIL REC.:		3
INDICATOR DESCRIPTION:		
TYPE OF INDICATOR:		Process
SHORT ANSWER	DETAILED ANSWER	INITIATIVES IN PROGRESS
YES	YES, Specific research programme	<p><u>Specific national programme:</u> Acción Consolider/CIBER launched in 2006 within INGENIO 2010. With the existence of CIBER-ER, which exclusively addresses RD research.</p> <p><u>Specific national programme::</u> Telemarathon “<i>We are all strange, we are all unique</i>” organised by the Spanish Rare Diseases Federation (FEDER), the Spanish Neuromuscular Diseases Federation (ASEM) and the Isabel Gemio Foundation. which uses the funds raised for RD biomedical research.</p> <p><u>Specific regional programme:</u> Marató TV3 (Non-governmental public foundation), which applied the funds raised in 2013 to neurodegenerative diseases (including a large number of RD).</p> <p><u>Unspecific national programmes:</u> With priority allocation to R&amp;D projects on RD: the Strategic Action in Health (AES) of the State Scientific and Technical Research and Innovation Plan 2013-2016, which includes RD as one of the fundamental aspects of Research, Development and Innovation in Spain.</p> <p>National Non-Specific Fundamental Research Programme, which finances basic RD research projects.</p>

		Open R&D regional calls Online Cooperative Health Research Networks (RETICS). Not specifically RD.
	<b>YES</b> , specific RD research projects within the research programme	Between 2008 and 2013, 260 specific RD projects out of a total of 4,150 (6.27%) were financed through the Institute of Health Carlos III/FIS.  With regard to the Non-Specific Fundamental Research Programme: The State T+D+I Plan of the Ministry of Economy and Competitiveness, within its Non-Specific Fundamental Research Programme, also finances research projects on rare diseases. However, we have no reliable information as to how many projects there are or what percentage of total projects they represent.  Between 2008 and 2013, CIBERER financed 100 RD Intramural Projects.
NO		

**Indicator 14: Participation in European and international research initiatives**

AREA OF COUNCIL REC.:		3
INDICATOR DESCRIPTION:		
TYPE OF INDICATOR:		Process
SHORT ANSWER	DETAILED ANSWER	INITIATIVES IN PROGRESS
YES	<b>YES</b> , E-RARE and IRDiRC	<p><u>European initiatives:</u></p> <p>ERA-Net for Research Programmes on Rare Diseases (ERARE) Call in which ISCIII has participated as financing institution since it began (2009) and in which projects in which Spanish groups participate are financed, in some cases as coordinators.</p> <p>H2020 Calls: which, in line with the IRDiRC goals, promote research on RD (mainly focusing on diagnosis of the vast majority of RD and the development of 200 new RD therapies in 2020).</p> <p>Participation in EUCERD Joint Action (2012-2015): CIBERER as coordinator of a work package. Quality of Health care and Centres of Experts.</p> <p>Participation in Orphanet Europe Joint Action (2011-2014): CIBERER as Spanish partner.</p> <p><u>International Initiatives:</u></p> <p>Call for collaboration projects promoted by ISCIII within the framework of the International Rare Diseases Research Consortium (IRDiRC) in 2011, financing 3</p>

		<p>RD research projects.</p> <p>Participation in the International Rare Diseases Research Consortium (IRDiRC): ISCIII and CIBERER participate in this international consortium and its different working groups.</p>
--	--	--

**Financial support**

**Indicator 15: Public funds specifically allocated for RD research actions/projects in the country**

AREA OF COUNCIL REC.:		3
INDICATOR DESCRIPTION:		
TYPE OF INDICATOR:		Process
SHORT ANSWER	DETAILED ANSWER	INITIATIVES IN PROGRESS
YES		<p>Between 2008 and 2013, 260 specific RD projects were financed through the Institute of Health Carlos III/FIS in a total sum of 31,796,907 €, representing 7.73% of the total supported.</p> <p>With regard to the Non-Specific Fundamental Research Programme: The State R+D+I Plan of the Ministry of Economy and Competitiveness, within its Non-Specific Fundamental Research Programme, also finances research projects on rare diseases. However, we have no reliable information as to how many projects there are or what percentage of total projects they represent.</p> <p>Financing of ISCIII for the IRDiRC: 10 M\$ for the period 2012-2016 at the rate of 2M\$/year.</p> <p>Financing of ERARE: -500,000 €/year to ISCIII for 3 years.</p> <p>The financing of Intramural Projects by CIBERER in the last two calls, corresponding to 2012 and 2014 was 1.1 M€.</p>
Under development		
NO		

**Indicator 17: Public funds specifically allocated for RD research actions/projects per year since the national plan started**

AREA OF COUNCIL REC.:		3
INDICATOR DESCRIPTION:		

TYPE OF INDICATOR:		Outcomes
SHORT ANSWER	DETAILED ANSWER	INITIATIVES IN PROGRESS
<i>Number</i>	Value	The annual financing from CIBERER received by ISCIII between 2008 and 2013 was 36.5 M€.
	Value just for funds allocated by the NP	
	N/A: incorporated in the total value of funds	

### Recommendations and Conclusions:

#### **B1-Map of existing research resources, infrastructure and programmes for RD**

##### Public Sources of Financing

##### HUMAN RESOURCES AND INFRASTRUCTURES

##### TECHNOLOGICAL PLATFORMS

1. PRB2 IIER

2. CENAG

3. IIER

4. CIBER

##### ACTIONS ON NATIONAL LEVEL:

- R+D+I AGENCY, Secretary of State, MINISTRY OF FINANCE (MINECO)

- ISCIII STATE AGENCY

- HR Programme: MINECO has pre-doctoral researcher training programmes and post-doctoral contracts for personnel in general, not specifically to research on RD, although many of these researchers work in the area of RD.
- Infrastructure programmes.

ISCIII is the main public research organisation, which manages and carries out biomedical research in Spain. In management terms, it is allocated to MINECO and reports functionally to MINECO and MSSSI. It does not work specifically for RD, but includes its own RD research centre in the IIER (Institute of Rare Diseases Research), it set up CIBERER and RD play an important role in the FIS (Medical Research Fund) programme.

- Strategic Action in respect of Health: Research Projects, Human Resources Programmes (Post-doctoral researchers, post-houseman researchers, intensification and aids for pre-doctoral and post-doctoral for innovation, Independent Clinical Research Programmes and Cooperative Research Networks. The Advanced Therapies network is worth mentioning.

CIBERER of the ISCIII finances staff contracts, scholarships launching researchers who are starting out and some intramural projects and support programmes for RD conferences and meetings. With 62 research groups and related 10 clinical groups over its 8 years of existence it has become firmly established as a benchmark RD research network centre on a national and international level.

- Cofinancing of International Programmes: E-rare ERA-NET (which must be cofinanced by ISCIII if awarded). B.3. Intramural Financing of the IIER. B.4. Financing of 3 IRDIRC Programme Projects (CIBERER, IIER Registries, Drug Discovery Platform at the Scientific Park in Barcelona).

Private Sources of Financing: Foundations such as Ramón Areces, with a budget allocation for RD. Since 2007 it has allocated 5 million euros to 53 rare disease projects. Merck Serono, who offers an aid of 25,000 euros for 1 rare disease research project every year. Foundations belonging to banks, savings banks, companies (Iberdrola) and several private foundations donate solidarity funds for RD research. RD patient and family associations, having to cope with a reduction of funds due to the crisis, have been turned into enterprises to bear the cost of research. Some Autonomous Communities have specific programmes, such as the Department of Equality, Health and Social Policies of the Andalusian regional government, which has signed an agreement with Cellgene to research on RD. It has also set up the Andalusian Laboratory of Cellular Reprogramming (under an agreement with the Californian Institute for Regenerative Medicine). The Prince Felipe Research Centre in Valencia has set up an “RD Biomedical Research and Technological Innovation Institute (IBIT)” within its strategic plan 2012-2015.

#### Research promoted or financed by patients or patient associations

What scope must be given to the research promoted by patients or patient associations?

In view of the shortage of research on RD, many patient associations have been converted into enterprises raising solidarity funds in order to hire researchers, pay for research material and even bear the cost of therapy. There are patients’ enterprises of various types: some act in response to competitive calls and others provide direct funding for laboratories and clinics that are experts in the matter, since patients know which clinics know most about their disease and also which groups conduct the research they wish to finance. It is important to continue encouraging dialogue between patient associations and researchers.

Inventory of national RD research working groups: There are none as yet. This is an aspect yet to be solved. The CIBERER brings together many groups that work on RD, but there are other individual groups and groups in networks which also work on rare diseases. ISCIII is making an

inventory to be published on the Orphanet portal, which currently contains information on Spanish research projects, but it is not possible to make a list; projects must be sought for each particular disease separately.

There is a map on the CIBERER website of the research done at CIBERER.

IRDIRC publishes the entire ISCIII list of RD projects participating in IRDIRC, not just those included in Orphanet.

Technological platforms: Within the inventory of resources, it is important to highlight technological platforms. The CIBERER biobank, the SEFALER for the phenotypes of animal models, the BIER or bioinformatics platform, the CEGEN or genotype centre, the CIBERER exome server and the SPANEX project of databases on exomes and genomic information on the Spanish population, are all very important for making progress in the knowledge of RD.

### **B.2 RD Research programmes with specific funds**

There is a CIBER that focuses specifically on RD (CIBERER), as well as other networking structures also financed by ISCIII which conduct research on RD. The calls of the State Scientific, Technical and Innovation Research Plan 2013-2016, including the aids for Strategic Action on Health (AES) managed by ISCIII, always includes research on RD as a preferential line.

- Where are the funds located?

In ISCIII, nominated subsidies earmarked in the programme budget, allocated through HR programmes, CIBER; there is also the IIER, plus hospitals, universities, CIEMAT, CSIC...

What is their governance model? There is a governing council and a consortium of research groups.

- Is a research and coordination model like the Fondation Maladies Rares feasible? What activities could be centralised?

“La Fondation Maladies Rares” is not a rare diseases research centre, but a financing agency and driving force for RD research in France. Part of the duties of the Fondation Maladies Rares are covered by the ISCIII through its own centres, the networking structures (especially CIBERER) and research projects. CREER, the National Reference Care Centre for People with Rare Diseases and their Families, set up in Burgos in 2008, could be similar in certain social aspects, since it provides information and guidance, psychological support and support in physiotherapy and early stimulation, family rest, it holds seminars, meetings with RD associations, etc. Activities depending on technological platforms (evolving alongside technical knowledge).

### **B.3 Sustainability of RD research programmes**

- How can the long-term structural sustainability of research projects and infrastructures in the field of RD be guaranteed through appropriate financing mechanisms?

By maintaining the National Agency, following R+D+I budgets increased over the State Plan 2012-2015.

- Are there specific programmes to finance research on RD? Do they make long-term research possible, ensuring the sustainability of the projects they finance?

There are no calls for specific projects. The sustainability of projects is guaranteed at 3 years.

- Are there any measures to inform on the national RD projects that are financed?

There are no measures in place at present, although they are being developed.

- How are the research programmes established? What mechanisms enable the continuity of successful initiatives and projects?

The RD research lines follow the R+D+I plan of focused calls.

EX-Post Assessment by the National Agency for Assessment and Forecasting (ANEP) and peer review.

- Is there any public-private collaboration? Is any model proposed?

We need a type of model like that of the Fondation de las Maladies Rares, which is based on donations. Another model of proactive action would be through partnerships (public/private).

A final recommendation would be a Sponsorship Act.

Public-private collaboration needs to be improved. Very few specific collaborations are made, mainly in European projects in which companies need to be involved. There are some initiatives, which are not sufficiently utilised, such as ALINNSA or ASEBIO. Some pharmaceutical companies are beginning to invest in orphan medicines, delivery technologies and other actions related with advanced therapies, but this is totally insufficient. The tax allowance for scientific sponsorship has yet to be addressed in Spain.

#### **B.4 Needs and priorities for research in the field of RD**

- Have priorities been set in Spain for both basic translational clinical research and social research?

Within the field of basic, clinical, translational research, Spain has followed the guidelines of Horizon 2020 and IRDiRC. It is essential to return at least to the R+D+I funding levels of 2009. We have lost 41%, between human resources and research in general. There is still insufficient awareness in society of the importance of research. It must be preserved in education. And the first people that need to be made aware are politicians, who allocate public financial support. The shortages are even greater in the field of research on living conditions and quality and social research. Social research lags further behind. Less priority is given to ENSERIO/CREER.

- How can awareness in research on living conditions and quality and social research on RD be improved? How can funds be secured for this type of research?

If we had the map of RD and resources, we would know what RD needs exist in Spain and what resources we have. In any case, it would be necessary to: establish coordination among working groups; promote the value of research in health and society; publicise the economic cost of RD.

The recommendation would be to join forces in the line of social, non-hospital research, such as Crónic-Care, research in primary health care, diagnosis in other areas such as chronic diseases whose tools (technological assessment agencies) are useful, creating research groups in sociology.

### **B.5 Promotion of interest and participation of national laboratories/researchers in RD projects**

- What measures need to be adopted for multi-centre studies? What national networks are needed?

Encourage clinical trials in general, diagnose where clinical research on RD is performed, foster collaboration between Reference Centres.

- How can bonds be established between basic and translational research and Reference Centres?

What solution can be devised in Reference Centres to enable researchers to participate in clinical services and clinics to spend time on research without jeopardising their activity?

Encourage consortiums with the participation of public/private agents, clinics, patients and researchers.

- What mechanisms could be put in place to facilitate clinical trials on small groups of people in Reference Centres?

Generate patient registers in the Reference Centres, services and units (CSUR) connected with other medical centres in respect of a given disease. Institutional engagement is necessary.

- Are there any specific programmes for attracting young scientists to RD research?

Specific programmes need to be promoted to attract young scientists to participate in research on RD. CIBERER awards 10 scholarships a year with a duration of 1 year for young researchers who want to begin their careers in CIBERER Groups. This is an excellent initiative, but it is not enough. In order to institutionalise patient participation in research, especially at Centres of Reference, a model of collaboration could be proposed seeking intrinsic unity among clinics, patient associations and researchers. In this way, work would be done in rare disease units, where the pillars supporting the unit are doctors, patients and researchers.

- How can patient participation in research be institutionalised, especially in Centres of Reference? What national collaboration models can be proposed?

Patient empowerment through their associations participating in the CSUR Advisory Board.

### **B.6 Research infrastructures and registries in Spain**

If there is a policy of centralised registries in the RD system, there must be two types of register:

1. Population registry, which must be a single register with epidemiological value.
2. Registries for each disease must be centralised in the different CSUR, in order to

have the information required for clinical trials.

### **B.7 International and EU collaboration in RD research**

Spain participates in ERA-NET and RD Connect (coordinated from the UK). Parc Científic de Barcelona participates in the creation of an integrated platform connecting registries, genetic biobanks and bioinformatics clinics for RD research and there are Spanish groups in EUROFANCOLEN, headed by CIEMAT, with the participation of ICS, and in LEUKOTREAT. CIBERER is a partner and coordinator of EUCERD. But RD research collaboration still needs to be strengthened more and better through the European Reference Networks.

## **Theme 4 – CARE FOR RD - CENTRES OF EXPERTISE AND EUROPEAN REFERENCE NETWORKS FOR RARE DISEASES**

### **4.1. REFERENCE CENTRES, SERVICES AND UNITS**

#### **Sub-themes:**

- Scope and functionality of Reference Centres
- Criteria for the designation and evaluation of Reference Centres
- Sustainability of Reference Centres
- Referral procedures/routes. Effectiveness in patient care
- Patient (case) coordinator/manager
- Collaboration of Centres of Reference with European Reference Networks
- Patient and patient-organisation involvement in Centres of Reference

#### **Background and current situation:**

##### **Scope and functionality of Centres of Reference\***

##### **\*CSUR (Reference Centre, Services and Units)**

#### Analysis of the Spanish definition for a Centre of Reference and comparison with the EUCERD recommendation

The Autonomous Communities are responsible for the provision of health care. However, in order to guarantee equal access to a safe, efficient and good-quality level of service for people with rare diseases (who need a greater concentration of cases to receive appropriate care), the NHS has established the procedures for the designation of Centres of Reference. This does not contemplate a continued provision of care at the Reference Centre, but instead the Reference Centre would act as the leading centre for the confirmation of diagnosis, definition of therapeutic strategies and continued treatment, as well as a reference point for those health care centres which provide the day-to-day care.

The NHS tries to follow the EUCERD Recommendations on “Quality Criteria for Centres of

Expertise for Rare Diseases In Member States”, adopted on 24 October 2011, following several consultations with relevant stakeholders. The first recommendations are as follows:

“1. CEs [Centres of Expertise] tackle diseases or conditions requiring specific care due to the difficulty in establishing a diagnosis, to prevent complications and/or to set-up treatments.

2. CEs are expert structures for the management and care of RD patients in a defined catchment area, preferably national, and at international level if necessary.”

4. CEs bring together, or coordinate, within the specialised healthcare sector **multidisciplinary** competences/skills, including paramedical skills and social services, in order to serve the specific medical, rehabilitation and palliative needs of rare diseases patients.”

The EUCERD provides a set of quality criteria to ensure that the RD Centres of Reference / Centres of Expertise provide high-quality and specialised healthcare, whilst recognising a degree of flexibility on the scope of centres, as stipulated in the Recommendation No. 14:

“14. The scope of diseases covered by each CE, or by a CE at national level, will vary depending on the size of the country and the structure of the national health care system.”

It is also important to mention that EUCERD, among others, recommends the use of ICT (information and communication technology) whenever necessary to assist patients who are geographically remote from the centre of reference; and to favour the dissemination and mobility of knowledge and experience to ensure the correct care of patients in proximity.

#### Scope of Centres of Reference in Spain

Centres of Reference in Spain must have a nationwide scope and access to those centres must be guaranteed on equal terms to all patients of the National Health System, no matter where they live, improving the effectiveness and efficiency of health care.

Centres of Reference of the Spanish National Health System must provide care through multidisciplinary teams which will provide care as well as confirmation of diagnosis, definition of therapeutic/monitoring strategies and as a reference point for those health care centres which provide the day-to-day care. Centres of Reference must also guarantee continuity in health care and treatment between the different stages of a patient’s life (e.g. from paediatric services to adult care) and the different levels of care and assess the results.

The purpose of the Reference Centre project is to guarantee equal access to safe, efficient and good-quality health care for people with diseases which, due to their characteristics, need professionals with a high degree of specialisation and high technology, concentrating the cases in a small number of centres.

#### Degree of coverage of rare disease patient needs

The approach in Spain to designating Centres of Reference for rare diseases is, just as in Europe, being directed towards creating groups of diseases with important aspects in common. Although progress has been made in recent years, the designation process is very slow. It is very important to continue with the identification of groups of diseases which could

need a centre of reference.

How can we make sure that rare disease patients have access to the appropriate centre of reference?

Within the Spanish National Health System, the autonomous communities are responsible for the provision of health care and if they lack the necessary services and resources they can refer patients to another community or outside the NHS. These referrals require authorisation by the patient's community of residence and are currently financed by the "*Fondo de Cohesion sanitaria por compensación entre comunidades autónomas*" [Health care Cohesion Fund through offsetting between autonomous communities]. The community which recommends the referral (the patient's community of residence) will usually cover the related expenses (travel, board & lodgings and costs of the carer, if necessary) and each community regulates what expenses it will cover.

National and regional health care powers and responsibilities

The autonomous communities are responsible for providing health care to people with rare diseases. However, equal access to care for those living with rare diseases must be guaranteed throughout the national territory, so Centres of Reference must be better identified and well-known in order to facilitate such equal access.

The central government is responsible for ensuring that the basic rights of all citizens are met on equal terms throughout the country and must take appropriate action if access to medical services and health care through the different Reference Centres are not provided on equal conditions for all citizens throughout the country.

The procedure for designation of Reference Centres is led and coordinated by the "Reference Centre Designation Committee" which is overseen by the Interterritorial Council of the National Health System. This Committee is chaired and managed by the Directorate General of Basic NHS Services and Pharmacy and made up of representatives from all the autonomous communities, *INGEST*, the National Transplant Organisation, the Carlos III Agency for the Evaluation of Health care Technologies and the Subdirector General for Quality and Cohesion.

Royal Decree 1302/2006 sets out the broad outlines of the designation and accreditation procedure for Reference Centres of the National Health System.

<https://www.msssi.gob.es/profesionales/CentrosDeReferencia/ProcedimientoCSUR.htm>

Once the Reference Centre has been designated, the autonomous communities must inform the hospital that is has been designated as a reference centre and inform the primary health care centres and hospitals of the following: the diseases and procedures which have a reference centre; the Reference Centres designated; the procedure for referring patients to them.

Networking and Telemedicine

To date, telemedicine is not being actively incorporated in the healthcare system and networking amongst professionals should be encouraged. This would avoid patients having to travel and promote communication between health care professionals (in both primary health care and hospitals , including social health care) and the Centres of Reference.

**Criteria for the designation and evaluation of Centres of Reference**Adaptation of the designation criteria to the characteristics of the disease or group of diseases covered by each reference centre

The criteria and requirements that must be met by a reference centre are developed ad hoc by the responsible team, taking account of the characteristics of the disease or group of diseases to be handled at the centre. The working team includes scientific societies, experts and specialists in the disease(s), appointed by the regional governments and the Ministry of Health. The involvement of patient associations in the expert groups of the designation committees could ensure that Reference Centres are better adapted to patient needs. However, in most cases, this does not happen yet and would need to be implemented.

Comparison between designation criteria in Spain and those recommended by EUCERD

The Spanish designation criteria for Reference Centres largely follow those recommended by EUCERD on a European level.

Involvement of patient organisations in the designation of Reference Centres

Since 2011 the Spanish Federation for Rare Diseases (FEDER) has held regular meetings with the team in charge of the designation of Reference Centres, which it attends in an advisory capacity. Moreover, the Ministry has included some of the experts in RD suggested by FEDER in the working groups of experts. However, this participation is insufficient and other ways of increasing the involvement of patient organisations in the designation process must be explored.

Assessment procedure for Reference Centres

Reference Centres are assessed before their official designation, which is made for a maximum period of five years. The designation must be renewed before the end of that time, subject to reassessment by the National Health System Quality Agency to make sure that the designation criteria are still met. If any of the criteria are no longer met, the Ministry of Health will, by agreement with the CISNS, revoke the designation.

One of the problems encountered in the validation of Reference Centres is the shortage of human resources to make those assessments, slowing down the process considerably.

In 2009, the Designation Committee put in place an Information System (IS) for annual

monitoring of the activity, indicators and results of referrals and inter-community travel by patients.

What quality indicators have been adopted and what measurements of results are considered?

The designation criteria to be met by the Reference Centres are specified for each disease or group of diseases, including experience, human resources, equipment and training, with adequate indicators and information systems.

Need to include patient satisfaction as a prerequisite

Assessment of the satisfaction of patients and their relatives, and of the health care professionals treating the patient needs to be introduced. This has not yet been possible due to lack of resources. This assessment should be included within the indicators of the Information System monitoring Reference Centres.

At present, neither patients nor patient organisations are directly involved in the assessment of Reference Centres or in the designation processes.

**Sustainability of the Centres of Reference**

Appropriate funding mechanisms to ensure the long term sustainability of Reference Centres

RD 1207/2006 regulated management of the Health Cohesion Fund (FCS) and included as an activity for compensation by this FCS, "health care for patients resident in Spain referred between autonomous communities for care in NHS Centres, Services and Units of Reference (CSUR)". Until 31 December 2012 the FCS has paid compensation for health care in CSUR. As from 1 January 2013 "the application of Article 2.1(a), (c) and (d) of Royal Decree 1207/2006 of 20 October regulating the Health Cohesion Fund is suspended" and no allocation is made in the MSSSI expenditure budget for compensation of health care provided in Reference Centres for patients referred between Autonomous Communities, Ceuta and Melilla. After deducting any negative balances from the payments to be made to the autonomous communities by the National Social Security Institute (INSS), the positive balances resulting from the FCS settlements will be paid to the autonomous communities from extra-budgetary funds as the net positive balance of the overall amount for health care coverage.

Impact on the functioning of Reference Centres caused by the change in compensation for care provided at the Centres of Reference

Significant decrease in the number of patient referrals (autonomous communities are more reluctant to refer patients); increased rejection of patient referrals by the receiving autonomous communities; concerns in hospitals that are Reference Centres over the lack of resources to treat patients referred.

It is essential to include a specific budget item in the General State Budget to finance health care provided for patients referred between autonomous communities, Ceuta and Melilla.

## **Referral procedures/routes. Effectiveness in patient care**

### Procedure for referring a patient to a Centre of Reference

There is a defined procedure for referral agreed by the Interterritorial Council of the National Health System in 2008 whereby each designated Reference Centre undertakes to treat all patients from other autonomous communities when the latter request assistance through SIFCO (the Health Cohesion Fund Information System) and may only reject such a request in exceptional cases, which must be duly evidenced.

The care provided at Reference Centres to patients referred from other autonomous communities is provided on the same conditions and with the same guarantees as that given to the residents of the autonomous community in which the reference centre is situated (Article 2.4 Royal Decree 1302/2006).

### Efficiency of the current patient referral procedure

In practice, patients face many problems in getting referrals. People are generally unsure of how to proceed and do not know about SIFCO or the cover and rights included in the health care provided at Reference Centres. There are many cases in which this right to referral is denied in the patients' autonomous community and cases are also refused by the receiving autonomous communities. This may be due, among other causes, to financing and sustainability problems of the Reference Centres and difficulties encountered in obtaining payment of compensation for the health care provided. The problem is also often due to the fact that although requested by the patients, the clinics at which they receive health care do not issue the referral report so the referral many times is not processed or it takes long.

## **Patient Case Manager**

### Role of the Patient Case Manager at a Centre of Reference

This person should act as a liaison between specialists and the patient and his/her family, to ensure proper coordination between services and professionals (identifying the best referral route) and offer emotional support and assisting the patients in anything they might need throughout the care process. This person may be a nurse or doctor, depending on the situation, and must be familiar with access to the different resources available enable multidisciplinary work.

### Incorporation of case managers at existing Centres of Reference

The figure of the case manager is not usually specified as such in the Reference Centres. It is required within the designation criteria for Reference Centres, which must have a care coordinator to guarantee coordination of patient care by the medical team of the multidisciplinary unit and any other units collaborating in the care of these patients. Only certain kinds of Reference Centre, such as the neuromuscular rare diseases centre, have been explicitly required to have a case manager.

## **Participation by Centres of Reference in European Reference Networks**

According to European legislation, centres wishing to participate in the European Reference Networks may do so in three ways, provided they meet the pre-established criteria. The three forms of participation are: a) network members (designated Reference Centres); b) associated national centres; and c) collaborating national centres.

Two pilot experiences are currently being conducted under the EU Directive 2011/24/EU on “the application of patients’ rights in cross-border healthcare and NHS centres are participating in one of them. As European networks are set up, all the designated NHS Reference Centres that meet the criteria established for those networks will be able to join them.

### **Patient and patient-organisation involvement in Centres of Reference**

Patients are not normally involved in areas to which they could make a valuable contribution. The involvement of patient associations depends largely on the professionals in the Reference Centre and their interest in encouraging that participation.

It is considered important to promote collaboration and active participation by patients and their associations in order to improve the results of Reference Centres.

### **Recommendations and Conclusions:**

1. Strengthen, pursue and accelerate the procedure for designating Reference Centres for all rare diseases based on previously identified needs of patients and their families.
2. Step up the allocation of resources to the Reference Centre project in order to speed up the designation process.
3. Claim a specific allocation in the General State Budget for 2015 and following years, a specific budget for health care provided in Reference Centres.
4. Include the figure of Case Manager in the Reference Centres.
5. Publicise the Reference Centres designated in the NHS and the necessary referral procedures.
6. Facilitate patient referral to Reference Centres, ensuring that patient welfare prevails at all times over all other considerations.
7. Improve the financing of the costs incurred by the transfer of those patients (related expenses such as travel, board & lodgings and costs of the carer, if necessary) and advance part of the outlay required to cover transfers.
8. Incorporate patient participation in the Reference Centre designation process, through the designation committee expert groups, and in the Reference Centre evaluation system.
9. Create and implement the Reference Centre evaluation system, quality controls and their corresponding improvement plans, in collaboration with patient associations. The satisfaction of patients and their families must be a key parameter in the assessment.
10. Urge the health authorities in the autonomous communities to recognise the

professionals of Reference Centres through their professional careers.

## 4.2. CARE MODEL

### Sub-themes:

- Early diagnosis
- Appropriate care and patient referral procedure
- Multidisciplinary approach
- Guarantee of treatment

### Background and current situation:

RD are often characterised by their clinical variability and by multisystem, severe, chronic and highly disabling dysfunction. It is known that around 80% of rare diseases are genetic and mostly appear in childhood. This makes them one of the current challenges for public health and of multisectoral holistic care.

There are also other factors that make them unique: invisibility and social exclusion, medical ignorance, difficulties in the diagnostic process and in obtaining access to adequate treatment, difficulties in monitoring the disease and providing information and guidance to patients and their families and the historical lack of institutional commitment.

For the above reasons, it is necessary to consider improving care for RD right from the appearance of the first symptoms, setting the protocol in motion to tackle the real priorities of RD far more decisively, these priorities being:

- × Early diagnosis
- × Appropriate care and patient referral procedure
- × Guarantee of treatment
- × Multidisciplinary approach

In other words, the mere fact of being born with (or acquiring) a rare disease must not put the patient at a disadvantage compared with other patients, or be an emotional or financial burden on top of their disease per se, as is the case at present.

Both in Europe and in Spain, thanks to the work of patient associations and numerous professionals, RD have gradually become institutionally recognised and a public commitment has been adopted to improve patient organisation care. This is why we want patients to be present, through their associations, in the relevant decision-making bodies and procedures, because they often stand in for the authorities in the services the latter ought to provide, and also because nobody knows better than they do the day-to-day implications of these diseases.

### **Recommendations and Conclusions:**

Different aspects must be taken into account for the development of the care model for rare diseases. These aspects (below, proposals from 1 to 25) are interconnected and make up a comprehensive model of care:

#### **Primary Health care**

1. Consider the possible inclusion of warning signs and symptoms of RD in the healthy child care programmes (regional).
2. Multidisciplinary work, as most of these diseases are complex, requiring monitoring with interdisciplinary coordination. The diseases are multisystemic and the person is much more than just a disease: he or she has a social life, family, studies, works...
3. Academic training, further training, medical guidelines and implementation of the DICE APER protocol in all national primary health care. All healthcare professionals must be given training and they in turn must provide training for patients and their families.
4. Step up neonatal screening and guarantee comprehensive care, including access to all treatment.
5. Address the existing gap in the care and monitoring of undiagnosed patients.

#### **Creation of information units, monitoring and coordination of care**

6. Information units, monitoring and control, non-clinical interdisciplinary units. These are administrative units within the regional health services, which should have comprehensive information and be at the disposal of professionals in primary and specialist care and professionals who cooperate with regional, national and foreign experts.
7. Proposal:
  - a) Whenever any suspicion, symptom or sign is detected in primary health care, the information units should be contacted and those units would help refer the patient to the most appropriate hospital and specialist.
  - b) If this specialist or hospital is able to make a diagnosis, they would establish the treatment and monitoring with Primary Health care through the Case Manager. If they do not feel qualified, they would refer the patient to the appropriate Reference Centre, even a Reference Centre in a different EU member state if necessary, through the Case Manager.
  - c) A protocol must be established for informing patients of the diagnosis: who? how? where? Psychological support and genetic counselling must be provided.
8. Share the monitoring between primary and specialist health care.
9. There must be an assigned budget provision.

10. Guarantee access to a second medical opinion nationwide.

#### **Reference Centres and case managers**

11. Reference Centres are required due to the complexity, ignorance and large number of rare diseases. Not all medical professionals have the necessary knowledge and experience to treat them and patients accept that information and response should be concentrated at those centres, while demanding a systematic referral free from bureaucratic obstacles so that all patients, wherever they live, have access to them.

12. These centres are in contact with the referring hospitals and primary health care. Neither the patient nor the specialist need always be present. The use of telemedicine is becoming increasingly popular. They are specialised centres with experience and with exchange and research capacity. They are interdisciplinary centres where teamwork is a basic premise. They have a holistic concept of the person. Finally they need state budget allocation to guarantee their basic premises: they are state centres.

13. Case manager duties include ensuring continuity or conclusion of health care and guaranteeing support and information.

Location: hospital, dealing with primary care manager.

Profile: as considered most suitable by the service.

Function: Concentrate complexity.

#### **Comprehensive concept of the Care Model**

15. Ensure comprehensive treatment of the person.

16. Protocol between Health, Social Services and Education. Educational inclusion.

17. At an employment level, support enterprises in providing employment for people with RD, facilitating adaptation of the jobs.

18. Recommendation for medical reports to contain information on the disease and the basic activities and skills in everyday life.

19. Relationship between disability evaluators, health professionals and patients or their representatives.

#### **Access to drugs, benefits, Directorate General of Basic Services**

20. Resume inter-territorial cohesion funds for orphan drugs and Reference Centres

21. State budget allowance for orphan drugs and compassionate use.

22. Grant for medical devices not recognized as drugs.

23. The service portfolio should include psychological support, physiotherapy and rehabilitation.

24. The psychologist should be involved throughout the process, right from diagnosis.
25. There should be equal access and treatment time in all Autonomous Communities and there must be a professional to coordinate at national level to supervise (with the intervention of FEDER).

### CONCLUSIONS:

“The care model must be defined taking into consideration the opinions of patients, health care and management professionals and different government agencies, which is also recognition of the professionals’ and centres’ track record and historical contribution.”

It is considered necessary for the model to be based on three pillars: **diagnostic systems and early diagnosis, comprehensive care plan and support and assistance for patients and their families.**

The model should prioritize a multidisciplinary care system, benefiting from the pooled knowledge and experience, quality and outcomes of care and its improvement, in an integrated care network regulated by the principles of equal access and outcomes.

It should also be borne in mind that the low or very low prevalence of RD makes it necessary to concentrate resources and knowledge, so that the quality improvement criteria can be met, obtaining optimal results, promoting systematic evaluation and training of professionals, improving efficiency and opening up more powerful lines of research and innovation.

RD health care must be provided as a multidisciplinary, integrated and coordinated combination of different services and care levels and across other areas as education, social welfare, family, business and employment.

The low or very low prevalence of RD also requires a particular concentration of resources and knowledge, as a result of which the level of expertise is concentrated in a Unit of Clinical Experience (UCE) or a UCE network for a rare disease or group of RD.

This RD care model aims to optimise available resources and adequacy of care, establish care levels and circuits that ensure quality and efficiency, the continuity of care and support for patients and their families in the different stages of the disease.

The following features are particularly important:

- 2 levels of care:
  - ✘ A more specialist, expert care level, concentrated in a Unit of Clinical Experience or in a UCE network for a rare disease or group of RD.
  - ✘ A territorial or community level, near the RD patient’s residence, including primary care, regional specialist reference care, other regional health services (social and health care, mental health care, rehabilitation, etc.) and also social, educational and labour resources available in the region.
- An integrated network of the different resources involved in the care of the patient,

based on active professional participation according to the responsibility of each one in the integrated care.

- Requires collaboration and the use of ICT to foster the coordination of resources and connectivity to share information and knowledge.
- Multidisciplinary care team, both at the level of Units of Clinical Experience and at regional or community level, made up of all the professionals participating in the process of diagnosis, therapy and monitoring.
- Both the more specialised level and the territorial level must guarantee patients and their families the organisation and coordination of their care processes as well as information and support.
- Guarantee a definitive diagnosis from the UCE and a care plan including therapeutic strategy and shared monitoring for anyone with or suspected of having a rare disease.
- A care protocol established by consensus among experts in the RD must be available and contain scientific evidence of clinical practice guidelines.

## **Theme 5 – ORPHAN MEDICINAL PRODUCTS AND MEDICAL DEVICES**

### **Sub-themes:**

- Support for Orphan Drug (OD) development
- Access to treatment
- Compassionate use programmes
- Off-label use of medicinal products
- Pharmacovigilance
- Other therapies

### **Background and current situation:**

In 2010, the first recommendations on the use of drugs and medicinal products were made in EUROPLAN I. There is still considerable room for improvement in the implementation of those measures and further work is required to cover the needs of RD patients and their families.

The following recommendations are intended to define actions to be taken, which should be monitored to measure their degree of implementation.

**Recommendations and Conclusions:**

**Support to Orphan Drug (OD) development**

1. Prioritize resources from all current support programmes (PROFARMA plan, FIS –the Medical Research Fund-, independent research of the pharmaceutical industry, competitive projects or European projects, among others). Contemplate specific measures to stimulate the research and development of medicines destined for the treatment of rare diseases. When possible (for example PROFARMA plan, FIS, etc.), award additional points to research and development projects related with orphan medicines.
2. Adequately publicise the list of Reference Centres and units designated for different rare diseases in order to facilitate research on these diseases.
3. Speed up the process of creating Reference Centres on rare diseases and prioritize coordination between them to encourage the recruitment of patients and to promote the conducting of clinical trials, safeguarding at all times the patients’ personal data.

**Access to treatment**

4. National coordination of early information on new products with the Health Technology Assessment agencies (HTA), price makers, payers and regulatory agencies, to be able to represent Spain in European level decision-making.
5. Prioritize and expedite the preparation and publication of Therapeutic Positioning Reports (TPRs), pricing decisions and reimbursement conditions for orphan drugs for diseases without any alternative therapy, to minimise the time between authorisation and putting the product on the market.
6. Encourage the relevant public institutions to participate in European initiatives to avoid inequalities, favour equal access to orphan drugs and medical devices throughout the EU and ensure greater transparency in decision-making.
7. The TPRs directed by the Spanish Agency for Medicinal Products and Medical Devices (AEMPS) and developed by the different agents of the health system (AEMPS, the Ministry of Health, Social Services and Equality (MSSSI), regional governments, medical experts, different medical and healthcare professionals and patient associations), which make a single assessment of the value added of medicinal products for the entire Spanish territory, are important in securing equal access to these medicines in all regions of Spain.

In this respect, and provided that the reports have been prepared by consensus, the role of TPRs should be consolidated and its conclusions used to favour equal access to the therapies throughout Spain. It should be borne in mind that once the TPR on a medicine has been published, and is thus binding on all Autonomous Communities, the information it contains will have to be updated from time to time in the light of new scientific evidence and the results on health obtained through use of that drug.

8. Enable the appropriate budgetary systems to prevent discrepancies on purely

economic grounds in access to medicinal products and medical devices by patients with similar clinical situations and stimulate coordination in this respect between the Ministries involved (MSSSI and Ministry of Economy and Finance) and Autonomous Communities.

9. Promote the creation of rare disease Reference Centres to improve access to therapy (medicinal products and medical devices for rare diseases) and generate greater scientific knowledge on therapies, recognising the special nature of these centres (rarity, complexity and adequate budgetary support).

#### **Compassionate use programmes**

10. There is specific legislation on the compassionate use of medicines in Spain. Adequate publicity should be given to the existence of this access to treatment.

11. Under AEMPS leadership, coordinate conditions for access to medicines authorised for compassionate use and foreign medicines among the different autonomous communities and hospitals.

12. Take into consideration the criteria and opinions of relevant patient associations when making decisions on the authorisation of medicinal products for compassionate use.

#### **Off-Label prescription**

13. There are specific laws on the off-label use of medicines in Spain. Adequate publicity should be given to the existence of this access to treatment.

14. Encourage exploration of the possibility of reimbursement for medicines used off-label when such use is justified by scientific evidence and there are no authorised medicines for the purpose.

#### **Pharmacovigilance**

15. Encourage the reporting by all and any parties involved, using the mechanisms provided, of any adverse side effects associated with pharmacological treatment.

16. Step up the information and training provided to patients and professionals on the importance of reporting adverse effects.

#### **Other Therapies**

17. Conduct studies on the need for medicinal products and other medical devices (cosmetics, dermatological, dietetic, dressings or orthopaedic and prosthetic material, among others) financed or otherwise by National Health System (NHS), for specific groups of patients with rare diseases, when they are considered essential in view of the clinical properties of the disease suffered. Recognise the therapeutic value of these products and include them within the exceptions contemplated in the supplementary portfolio of NHS (RD 16/2012 of 20 April) for specific groups.

18. Select medicines and medical devices for their properties, characteristics and clinical

evidence, rather than on economic grounds.

## **Theme 6 –SOCIAL SERVICES FOR RARE DISEASES / AID LINES / DISABILITY / DEPENDENCE**

### **Sub-themes:**

- Social Services structure
- Spanish Social Services system

### **Background and current situation:**

The SAAD (System for the Autonomy and Attention to Dependency) service catalogue sets out a compendium of benefits, resources and services to promote personal autonomy and dependency care. These services are not RD specific.

There are no specific services or benefits for people with rare diseases. The only public recourse within the social services is the State Reference Centre for Rare Diseases (CREER) in Burgos.

The information is scattered.

Associations make up for this lack of information and, very often, resources for RD patients. They propose initiatives which sometimes receive government support (especially at regional level: examples can be found in Andalusia, the Basque Country and Madrid).

At present there is no social benefit model or any coordination between Health Services and Social Services in Spain, although some progress has been made in this direction (The management of social services has been transferred to the autonomous communities (regional differences)).

- There is a lack of coordination between different levels (primary care - specialist services) and between different systems (education, health, social, labour, etc.).
- Types of access to resources (through primary care):
  - a) Access to benefits and aids for disability and dependence.
  - b) Access to health care resources: early treatment centres, rehabilitation, speech therapy, etc.

The economic crisis had an impact at both public/government and individual level (families and affected people):

- Cutbacks and reduction of available services.
- Difficulty in meeting the cost of treatment required for RD or services such as personal assistance or home help.

- The main sources of information for families are: specific associations, school, doctors, etc. (resources such as the dependency benefit, disability certificate, early treatment, special education, orthopaedic and prosthetic aid, allowances for the care of children with cancer or other serious diseases).

### **Recommendations and Conclusions:**

1. Spain needs a Social Services Act laying down the criteria for coordination among health, employment and educational services, guaranteeing equality for people living in different regions of the country and regulating people's right to receive support and services to enable personal autonomy and participation in the community. The lower the prevalence of the disease, the greater the need to reinforce the coordination of resources.

#### Indicator

1.1 A national law by consensus within the coming 4 years

2. A common platform is needed for sharing information between health, social services, education and employment professionals, enabling access to the necessary data of each person required for different benefits, without having to repeat administrative procedures and thus expediting the response. This computerized system would include, among other features, a directory of the existing resources to guide each person rapidly to the most appropriate support for their particular situation regarding health, disability or dependence. Information would be accessible to the person concerned.

#### Indicators

2.1 Have the initiative approved by the Ministry of Health by 2015, creating a multidisciplinary group for its design, with participation by patient associations.

2.2 Get 50% of the autonomous communities to share their information over the next two years and all of them within four years.

3. Urgent approval is required of the disability percentage tables following ICD criteria.

#### Indicator

3.1 Publication of the tables before the end of the current term of government.

4. A map of the social resources available and a directory easily accessible by people are needed.

#### Indicator

4.1 Publication of the map next year (2015).

5. It is necessary to unify the processes for assessing disability and dependency to avoid contradictory reports, save having to repeat formalities and make the evaluation process more efficient, since dependence usually goes hand in hand with a significant degree of disability.

Indicator

5.1 Regulate the coordination between professional teams in autonomous communities to enable joint assessment within two years.

6. Urgently enable access to patients' medical records by the professionals responsible for evaluating disability and introduce ICD-10 codes.

Indicator

6.1 Access to this information should be enabled. The ICD-10-CM should be introduced in the Hospital Discharge Registry by the NHS in 2015.

7. Cooperation agreements should be made with third sector organisations to fast track the paperwork for people who urgently need their dependency assessment and access to the resources they require.

Indicator

7.1 Over the next two years, spread to all autonomous communities the experience of Madrid, where there is a collaboration agreement in place between FEDER and the Department of Coordination of Dependency.

8. The use of existing resources should be encouraged for disabled or dependent people, coordinating existing programmes, sharing available services between organisations and creating resource banks. The government should adequately fund joint projects between organisations prioritizing this practice.

Indicators

8.1 Number of programmes shared between two or more organisations per year.

8.2 Number of services shared.

8.3 Have a resource bank in each autonomous community and a national resource bank within three years' time.

**ADDITIONAL WORKSHOPS**

**Theme 7 –LABOUR INCLUSION**

**Sub-themes:**

- Labour market inclusion of people with rare diseases
- Labour market inclusion of carers of people with rare diseases

**Background and current situation:****Labour market inclusion of people with rare diseases**

Labour inclusion must be addressed, including not only the problems of "access to employment" but also and above all, of performance once a job has been found.

Inclusion can only be achieved through employment. Only through employment will it be possible to secure the integration, participation, autonomy and independence of people with disabilities; in short, true equality.

People with rare diseases are a particularly vulnerable group when talking about labour inclusion. 80% of the people with rare diseases have a disability rating of at least 33%.

There is no access to normal jobs in competitive businesses on equal conditions with other workers. There is a low impact of the labour integration model of "Supported Employment". This alternative model can be considered as the most integrating as it actually leads to "normal" employment (where most of the employees do not have any disability), exactly the same job (albeit with the necessary adaptations) and on the same conditions as any other worker in terms of work, wages and working hours.

In defence of the "supported employment" labour integration model, some studies have shown that it is beneficial in terms of value for money when analysed from the different points of view, namely of the worker, taxpayers and society, as it implies more income for the individuals and a smaller outlay for taxpayers, generating a positive result for the whole society. The same study also noted that this model obtained better results in a cost-effectiveness analysis using quality of life criteria; the benefits for people in supported employment were also higher than those of workers in sheltered employment.

One of the main downsides is the high, unavoidable rate of absenteeism among people with rare diseases. People with RD often find themselves - through no fault of their own (because of "ultra-dependence" on health care) and completely unavoidably - affected by a number of circumstances which can cause conflicts of different kinds at work. Unjustly and without any reasonable cause, their productivity is questioned, especially in a highly competitive context in which the only valid measurement criterion is productivity.

Most RD are debilitating, degenerative illnesses involving physical, mental and sensory disabilities forcing patients to stay at home or in hospital, sometimes for long periods of time. In short, RD patients suffer a high dependence, mainly on health care, which causes serious difficulties for them to do their work normally. It should also be remembered that the vast majority of rare diseases are multisystemic and multi-pathological, requiring patients to see a number of different specialists. This causes increases dependence y on healthcare.

Moreover, people with rare diseases generally have no legal protection. Labour laws do not protect this kind of specific situation, which is unfortunately the one faced by people with RD in their everyday life. Moreover, these people often have to cope with "mobbing", or workplace harassment, which is closely related with the above-mentioned problems of

absenteeism.

Unfortunately, numerous cases of mobbing have been detected among people with rare diseases, where the employer tries to undermine them so that they will just leave their jobs. Through this practice, the employer seeks to spare himself the cost of what would otherwise (had the employee not left of his/her own accord) have been declared unfair dismissal, in which case he would have to pay the corresponding severance pay to the worker.

Another problem frequently encountered by people with RD is the battle to enforce their right to have their job adapted and/or changed.

We must be very aware, that people with rare diseases, just like anybody else, are very keen on keeping their jobs. For them, incapacity to work is never a solution. They will always prefer to first see whether it is possible to adapt their workplace, and only if this is not possible, change their job, maintaining their professional profile and qualification.

But in actual fact, people who are diagnosed with rare diseases while working in a given job often have to face the inevitable situation of not being able to do their job because of their particular health situation. In this regard, this situation mostly affects workers diagnosed with some rare disease whose employment relationships are protected by the Workers' Statute or the specific collective agreements of the sector they work in. For this large group of workers, who do not fall within the scope of the Basic Statute for Civil Servants, not even the procedures have been established, so it is impossible to exercise the workers' right to be moved to a different job and/or have the workplace adapted because there is no procedure established for requesting this.

It is nevertheless true and should be stated clearly that the problems of access to employment, absenteeism and adaptation and/or change of job can often not be blamed entirely on the employers. They cannot ignore productivity and often do not have the necessary tools to deal with these situations. The employer is by no means always responsible. Workers with rare diseases have no legal backing to cope with their diseases within their employment, nor do employers have sufficient aids (tax allowances, full coverage by the National Health Service of the cost of time off work) to be able to cope with the difficult situations that sometimes arise.

### **Labour market inclusion of carers of people with RD**

Most of the problems mentioned above and suffered by people with rare diseases at work are equally applicable to their carers. Adequate health care and the state of health of a person with RD depend on his or her carer. Therefore, all the problems related with absenteeism (in this case due to having to accompany the sick person to all kinds of appointments, check-ups, tests, treatment and medical and complementary interventions) and, consequently, the mobbing or workplace harassment put down to those absences from work (even when such absence is duly justified) are also suffered by carers. It is, therefore, doubly important to solve these issues because they affect both RD patients and their carers.

Taking care of a dependent person is a long-term commitment, since with rare diseases, most of which are chronic, taking care of a sick relative spreads over a long period of time.

The family usually does most of the care for these people (72% of aid). In most families, only one person bears most of the responsibility for care. Most of these carers are women, so much that eight out of ten people who are looking after a sick relative in this country are women. Moreover, these women are mostly aged between 45 and 65, so are in the working age group.

At this point, we would like to try to find an answer to the following question: How can they solve certain problematic situations that arise when they try to combine looking after the relative and doing their work?

We are all aware of the specific problems entailed in the constant care required by a child or relative with a rare disease, which is often chronic, multisystemic, needs complex treatment and supervision by different medical specialists and with unpredictable ups and downs.

It is not easy for carers to organise themselves in these situations or respond as required by companies. Furthermore, working hours are not compatible with the timetables of RD patients (often in school age) and labour laws offer no protection for the specific situations in which carers of RD patients find themselves.

In this sense, the following difficulties are encountered:

1. No paid leave to look after sick relatives with rare diseases.
2. The reduction of working hours with entitlement to a compensatory allowance to look after a child with severe disease has serious shortcomings, such as the automatic withdrawal of the benefit when the child turns 18 (with no possibility of extension or review) and failure to contemplate the right to benefits if the child goes to school (establishing benefits only when the child is hospitalized or confined to home).
3. Since 13 July 2013:
  - a) The benefits for carers of dependent people with RD have been suspended for a period of two years.
  - b) Social Security coverage of carers of dependent people with RD, which was previously compulsory, has been made voluntary so carers have to pay their contribution costs.

#### **Documentation and relevant legislation:**

- Action Plan for the Spanish Strategy on Disability 2014 – 2020.  
This Action Plan has been established by the Directorate General of Disability Support Policies, the Secretary of State for Social Services and Equality and the Ministry of Health, Social Services and Equality. It was approved by a Resolution adopted by the Council of Ministers on 12 September 2014.

- Support for carers of dependent relatives at home: IMSERSO Award for "Infanta Cristina" Social Research and Studies 2006
- Report on Rare Diseases and Employment. Fundación Adecco. 2014
- Report on labour inclusion of people with RD. Fundación Fernando Pombo and International University of La Rioja, UNIR. 2014
- Report on the hiring of people with disabilities. FEACEM, Spanish Business Federation of Associations of Special Employment Centres. April 2014

## **Recommendations and Conclusions:**

### **Labour market inclusion of people with Rare Diseases**

#### Inclusion and access to employment

1. Demand a six-monthly report by the appropriate agency of the Ministry of Employment indicating the degree of effective implementation of Article 17 of the Rights of the Disabled Act 1/2013 on the following points:
  - Right to keep employment and return to work;
  - Professional guidance taking account of the real capacity of the person with RD.
2. Remove from Royal Decree 1851/2009, establishing the requirements for disabled workers to apply for early retirement, the requirement of having a 45% disability and the list of rated diseases set out in Article 2.
3. Enforce the obligation to inform applicants of the recognition of their disability and their right to obtain a report stating their abilities or skills with a view to labour inclusion, in the procedure set out in the Order of 2 November 2000 on evaluation of disability.

#### Absenteeism from work

4. The Statute of Workers' Rights should specifically contemplate:
  - A paid leave per year to participate in activities with a direct effect on improving the quality of life of the affected person.
  - Absences caused by serious illness cannot be ground for dismissal.
5. The Labour Procedure Law should include a provision whereby:
  - Employers of people with RD may compute sick leave on the same terms as maternity leave.

#### "Mobbing" or workplace harassment

6. Labour laws should recognise the psychological effects of mobbing and consider them occupational accidents.

7. Labour inspection should be able to require employers to prevent these situations in their staff policies and, should they occur, to prove that the company has taken sufficient measures to remedy and analyse the situation in order to prevent it from happening again in the future. Accordingly, following measures must be taken:

- Companies must have procedures in place to deal with any problems of psychological harassment, failures inherent in the organisation of the work or problems of cooperation and collaboration among workers.
- Companies must have a protocol binding on all workers contemplating effective measures to be applied immediately on detection of mobbing and must make an analysis to see whether the mobbing is due to an inadequate organisation of the work.
- The workers submitted to mobbing or harassment must receive immediate help or support.

Adaptation and/or change of job

8. Companies with more than 50 employees should be required by law to include in their internal procedures channels for their employees to request and enforce their right to adaptation and/or a change of job.

**Labour market inclusion of carers of people with RD**

9. Amend Art. 37.3(b) of the Workers' Statute to extend paid leave for as long as the sick relative remains in convalescence, displacement or in hospital.

10. Royal Decree 1148/2011 of July 29 should be amended as follows:

- a. Article 2 should expressly include in the protected status:
  - i. That the child with a serious illness should be allowed to go to school if it is proved that that he/she is receiving the necessary care at the school.
  - ii. The care required should not be exclusively for medical or health care.
- b. Article 7 should remove the automatic cancellation of the right when sick person reaches the age of 18. Instead, it should expressly contemplate the possibility of extension if the protected status continues.

11. Royal Decree Law 20/2012 of 13 July should be amended as follows:

- a. Supplementary Provision Seven establishing the two-year suspension of the economic benefits for care in the home environment and support for non-professional carers should be annulled.
- b. Supplementary Provision Eight making it voluntary to sign the special agreement with the Social Security for non-professional carers of people in situations of dependency should be annulled.

12. Recognition and tax allowance for employers who hire carers of people affected by RD with the possibility of working flexible hours or from home (teleworking).

13. Express recognition in the Workers' Statute of the possibility of part-time work for carers without incurring in a strictly proportional reduction of wages, powers or responsibilities.

## **Theme 8 –EDUCATIONAL INCLUSION**

### **Background and current situation:**

Organic Law 8/2013 of 9 December 2013 for improvement of the quality of education (LOMCE) guarantees equity, ensuring equal opportunities for the full development of personality through education, inclusive education, equal rights and opportunities to help overcome discrimination and ensure universal access to education, and offset any economic, social, cultural or personal inequalities, paying special attention to those resulting from any disability. *(Chapter 1 PRINCIPLES AND AIMS OF EDUCATION. Article 1. Principles, b) Equity, ensuring equal opportunities for the full development of personality through education, inclusive education, equal rights and opportunities to help overcome discrimination and universal access to education, and offsetting economic, social, cultural and personal inequalities, paying special attention to those resulting from any kind of disability.)*

80% of rare diseases are congenital and have an early start in life (two-thirds appear before the age of two), affecting children and adolescents.

65% of these diseases are serious and highly disabling.

Some characteristics, such as the lack of information, lack of specialist knowledge by the professionals in different care systems and/or poor development of adequate support systems have psychological, social, family and school level implications that hinder access to health, educational or social services, and limit the exercise of rights such as THE RIGHT TO EDUCATION, with implications for education, training and access to employment of people with rare diseases. This affects their personal development and hinders their social integration and participation.

While insufficient intervention in early stages deprives this group of their right to higher education, equal opportunities for access to and continued participation in quality EDUCATION must be defended, not only at the initial stages but throughout their life cycle.

Children with rare diseases are particularly vulnerable so the rights of children and persons with disabilities must be recognised and guaranteed, such rights being tolerance, respect, non-discrimination, equal opportunities and valorisation of differences.

The "Educational Report on Rare Diseases" (FEDER, March 2014) defines problems associated with suffering from a rare disease within the school context.

Such problems arise at different levels in the educational environment, causing a domino

effect. They begin with ignorance and lack of social understanding, which in turn implies indifference from the authorities and becomes apparent through the absence of appropriate resources for students. Both levels can be detrimental to the involvement of teachers and families and in turn result in discrimination and rejection in the classroom. All this affects the child and his/her personal development.

**Current situation:**

- ✘ Shortage of organisational, material and human resources (healthcare professionals, physiotherapists, counsellors, teachers of therapeutic pedagogy and hearing and language) in schools, especially in nursery, up to 6 years of age, and after the age of 16, when education is no longer compulsory.
- ✘ The requirement of access to forms of "home care" schooling does not suit the situation of students with rare diseases (RD) for several reasons: number of convalescence days above 30.
- ✘ Insufficient training of teachers and other members of staff at schools to meet the needs of students with RD.
- ✘ The educational psychology assessment conducted among students with RD does not reflect their needs.
- ✘ Students with RD are sometimes forced to go to special education centres, since they are the only centres that have the qualified professionals required to ensure maximum development.
- ✘ Existence of RD students enrolled in schools without support, especially in rural areas.
- ✘ Syllabus adaptations do not meet the comprehensive needs of students with RD.
- ✘ Only the 33.96% of public infant schools and the 27% of private infant schools are prepared for the care requirements of children with RD.
- ✘ Difficulties in access to the educational system and in changes between different stages of schooling.
- ✘ There is lack of communication between professionals of different care systems for children with RD.

**Recommendations and Conclusions:**

**1. OBJECTIVES**

**1.1. Overall Objective:**

Encourage educational inclusion for students with RD, offering an educational response adjusted to the needs of each student.

### **1.2. Specific Objectives:**

- Raise awareness in the society and government that inclusive education is a right of all children and young people with rare diseases.
- Promote awareness campaigns and actions in schools aimed at pupils, families, teachers and non-teaching staff.
- Ensure organisational and curricular measures, providing the necessary means and resources, to promote the comprehensive development of students with RD, responding to each of their needs.
- Promote and adapt the inclusion of students with RD in the form of education that best suits their needs.
- Open schools and the education community to patient associations, establishing channels for coordination and effective collaboration, since associations can provide schools with specialised information.
- Promote the training of teachers and all professionals involved in providing education for children and young people with rare diseases.
- Promote the creation of networks for knowledge management, educational innovation and the identification, recognition and generalisation of good practices.
- Promote coordination protocols and inter-institutional collaboration.
- Promote care and support schemes for families of students with RD. Ensure that adequate information is provided on difficulties/educational needs, educational goals and the support plan proposed by the centre.
- Develop a census of pupils with rare diseases attending school.

## **2. RECOMMENDATIONS**

### **2.1. Inclusive culture**

1. Establish agreements between the education authorities, FEDER and other national or regional associations for annual awareness campaigns on RD aimed at the entire school community (students, parent-teacher-student associations, teaching and non-teaching staff).

#### Indicators

1.1 Agreements signed by each autonomous community.

1.2 Number of schools which have implemented awareness campaigns on RD in each autonomous community.

2. Support and monitor access and transition between stages of schooling and create educational pathways to ensure educational continuity and success.

Indicators

2.1 Increased numbers of students with RD who complete compulsory secondary education and continue in the education system.

2.2 Number of schools that offer Home Plans for students with RD.

2.3 Number of schools with Plans to identify and eradicate situations of physical and / or psychological bullying on the grounds of RD.

2.4 Is there is a figure of reference for RD students at school?

**2.2 Inclusive Policies**

3. Establish channels for coordination between social, health and educational services.

Indicator

3.1 Number of coordination protocols, agreements, etc. developed between social, health and educational services.

4. Commitment of educational authorities (national or regional) to publish guidelines for the development of protocols for students with rare diseases. These protocols must be drawn up with experts recommended by associations dealing with the disease in question and other expert groups.

Indicators

4.1 Number of Educational Authorities that have developed guidelines for the preparation of protocols on students with rare diseases.

4.2 Number of students with RD benefiting from these protocols versus all enrolled students with RD.

5. Incorporate training on intervention with RD in the basic and further training of teachers and school counsellors.

Indicators

5.1 Presence of academic credits linked to specific training on RD in university education.

5.2 Calls for further training on RD for teachers and school counsellors.

**2.3 Inclusive Practices**

6. Regulation and implementation of a permanent forum for educational professionals to compile and share experiences and good practices for inclusive education of students with RD. This forum would be coordinated by FEDER and representatives of the educational authorities.

Indicators

6.1 Creation of the forum in 2015

6.2 Number of experiences and good practices identified and shared

6.3 Number of beneficiaries

7. Let students with RD and their families have a say in the building and assessment of the educational inclusion process.

#### Indicators

7.1 Number of regular family-school meetings

7.2 Family satisfaction with the academic and personal development of their children

7.3 RD Student satisfaction

### **3. CONCLUSIONS:**

In their globally renowned publication "Index for Inclusion", Booth and Aisncow (2002) argue that to achieve genuine inclusion it is necessary to act in three dimensions: developing inclusive policies, cultures and practices.

In this line and to achieve educational inclusion of children and people with rare diseases, a process must be set underway with changes generated in these three dimensions.

#### **A change of culture**

To achieve real inclusion of students, we have to promote the perception in society that people with rare diseases have more in common with others than differences; that they can develop their skills and abilities and deserve the same respect as any other human being.

This requires a change in attitude from very young ages and the involvement of everyone in the educational community: students, parents, teachers and administrative and service staff.

For rare diseases it is essential to establish adequate coordination among all the services involved in childcare (health, social and educational), who must collaborate closely to ensure a comprehensive intervention, which is only possible through a change of culture.

A decisive step for achieving this goal would be to open up the educational community to patient associations, which can offer specialised information and resources.

#### **New educational policies**

Laws and regulations contain a number of rights which must be reflected in the statutory instruments and their implementation.

Make schooling more flexible, including home and hospital care, adapted to suit the needs of these students.

Allocate educational assistants and specialist professionals (occupational therapist, speech therapist, nurse, etc.) regardless of geographic location and educational level, especially at non-compulsory education levels where these resources are currently not available.

Students with RD require genuine accessibility, beyond the demolition of architectural barriers. Some examples include administering medications, health care, special menus, proper integration of rehabilitation and educational services, adapting furniture, care of air conditioning conditions or addressing specific risks of these students.

Guarantee the availability of technological resources and technical aids that respond to the needs of these students.

And regulate and ensure early intervention through an integrated model that ensures the prevention, early detection, diagnosis and treatment of children, from a very early stage.

### **Inclusive Practices**

Attending students with rare diseases requires a balance between the perceptual-cognitive, motor and emotional development, which also involves care for their health needs. It is also necessary to establish collaboration between them. It is the educational establishment which must adapt to the student, not the other way round, at all levels, including pre-compulsory, compulsory and post-compulsory levels.

A change in culture and attitude towards recognising the value of each and every member of a society and the development of legislative frameworks for inclusive policies is not the sole guarantee for progress in the process towards inclusive education. All stakeholders have to get involved in order to develop new ways of doing things, new approaches, new methods, new practices, new ways of organising technical and human resources to transform the school into a SCHOOL FOR EVERYONE that guarantees access, participation and the success of everyone in quality education given in conditions of equality and equity.

### **Report of the Closing Session – Conclusions**

Terkel Andersen, President of EURORDIS, and Justo Herranz, Representative of the Steering Committee of the Spanish EUROPLAN Conference II and member of the Executive Board of FEDER, spoke at the closing session. They both highlighted the most important conclusions of the working groups.

One of the principal conclusions highlighted by Terkel Andersen was the need to strengthen, continue and expedite the procedure for designating Reference Centres in order to encompass the largest possible number of rare diseases based on the needs of patients and their families. He also stressed the need to include a figure of vital importance that of the “case manager” to coordinate all the professionals involved in health care for RD patients.

Justo Herranz underlined the importance of the proposals submitted by each working group through consensus and the participation of all the stakeholders involved.

He stressed the importance of having a body to coordinate the entire National Strategy, combining information and actions and the importance of the Centres of Reference as a key to the entire process.

He concluded by underscoring the need for a legal framework to guarantee protection of the

health and social rights of people with rare diseases. This demand corresponded to the right to equity, the need for specific treatment not covered by the Social Security and the proposal for holistic treatment of people with rare diseases.

The Spanish EUROPLAN Conference II thoroughly analysed the current situation of RD in Spain and provided an opportunity to identify the aspects requiring greater effort. The achievement of a consensus document making that analysis and detection, with the participation of representatives of different stakeholders, gives the content unmeasurable value.

The situation of the political organisation of Spain (a central government and 17 autonomous regions), the concern for sustainability of the initiatives already in place and the proposals launched, together with the recognition of patients as another important stakeholder to be taken into account, were present practically throughout the entire process.

At the end of the conference, there was a Debriefing Session to inform the competent authorities for rare diseases of the recommendations stemming from the EUROPLAN conference II.

### III. ANNEXES

#### ANNEX 1: PROGRAMME

Thursday, 20 November 2014

9:00	<b>REGISTRATION</b>
9:30	<b>WELCOME</b> <i>Juan Carrión, FEDER President</i> <i>Alba Ancochea, FEDER Director</i>
	<b>PLENARY SESSION</b>
9:40	<b>"European context on Rare Diseases and EUROPLAN Project"</b> <i>Simona Bellagambi, EUROPLAN EURORDIS Advisor</i> <i>Yann Le Cam, EURORDIS Director</i>
10:30	<b>Discussion</b>
11:00	Coffee Break
	<b>WORKSHOPS</b>
11:30	<b>Sharing results of workshops</b>
14:00	Lunch Break
15:30	<b>Final Report of workshops</b>
17:30	End of session
18:00 – 19:30	<b>GROUP REPRESENTATIVES SESSION</b>

Friday, 21 November 2014

8:30	<b>REGISTRATION</b>
9:00	<b>OPENING CEREMONY</b> <i>Juan Carrión, FEDER President</i> <i>Alba Ancochea, FEDER Director</i> <i>Paloma Casado, Deputy Director General of Quality and Cohesion</i> <i>Directorate General of Public Health, Quality and Innovation</i> <i>Ministry of Health, Social Policy and Equality (MSSSI)</i>
9:20	<b>PLENARY SESSION</b> <b>"National Plan on Rare Diseases: From Europe to Spain and back"</b> <i>Simona Bellagambi, Europlan Eurordis Advisor</i>
9:40	<b>WORKSHOP CONCLUSIONS</b>
9:40 - 9:55	Group 1 "Governance and Monitoring of the NP"
9:55 - 10:10	Group 2 "Definition, codification and inventorying of RD"

10:10 - 10:25	Group 3 "Information y training on RD"
10:25 - 10:40	Group 4 "Research on RD"
10:40 - 10:55	Group 5 "Centres of Expertise"
11:00	Coffee Break
11:30	<b>WORKSHOP CONCLUSIONS</b>
11:30 - 11:45	Group 6 "Care model"
11:45 - 12:00	Group 7 "Orphan Medical Products"
12:00 - 12:15	Group 8 "Social Services on RD"
12:15 - 12:30	Group 9 "Work Inclusion"
12:30 - 12:45	Group 10 "Educational Inclusion"
12:45	<b>CLOSING SESSION</b> Terkel Andersen, EURORDIS' President Steering Committee Representative
13:30 - 14:30	<b>DEBRIEFING SESSION</b>

**ANNEX 2: LIST OF PARTICIPANTS**

Name	Organisation	Role	Stakeholder group
Aitor Aparicio García	CREER	Managing Director	Public administration (MSSSI)
Alba Ancochea	FEDER	Director	FEDER
Ana Mingorance_Le Meur	Dravet Syndrome Foundation	Scientific Director	Patient representative
Ángel Abad Revilla	Directorate General of Patient Health care Department of Health, Autonomous Community of Madrid	Head of Patient Information Area	Public administration (MSSSI)
Ángela Almansa	Rare Diseases Registry, IIER, ISCIII		Public administration (MSSSI)
Anna Ripoll Navarro	FEDER	Representative of FEDER Catalunya	FEDER
Antonio Blázquez Pérez	Spanish Agency for Medicinal Products and Medical Devices (AEMPS)	Head of Medicines for Human Use Service	Public administration (MSSSI)
Antonio Liras	Committee of Experts of FEDER /Comisión Científica de la Real Fundación Victoria Eugenia	President	Academic/Researcher
Antonio Molares	Researc in Rare Diseases Institute (IIER).	Head of Clinical Projects Service	Public administration (MSSSI)
Antonio Pérez Aytes	Committee of Experts of FEDER	Neonatology Service. La Fe University and Polytechnic Hospital	Academic/Researcher
Arantxa Gordo Santos	National Centre for Educational Innovation and Research (CNIIE)	Head of Section, Coexistence and Human Rights Area	Public administration (MECD)
Begoña Martín Laucirica	FEDER	Board member	FEDER
Begoña Ruíz García	CREER	Head of Technical Area I	Public administration (MSSSI)
Belén Pérez	AEGh		Academic/Researcher
Belén Ruiz de Miguel	Spanish Ictiosis Association	Board member	Patient representative
Blanca Segurola Lázaro	Corporate Pharmacy Programme, Osakidetza	Coordinator	Public administration (Autonomous Community of the Basque Country)
Carmen Ayuso	Fundación Jiménez Díaz Hospital	Head of Research Area, Chief of Clinical Genetics Department	Academic/Researcher
Carmen González Vargas	FEDER	Administrative Assistant FEDER Andalucía	FEDER
Carmen López Rodríguez	FEDER	Coordinator of Galicia FEDER	FEDER
Carmen María García Quiñonero	Rare Disease Association D,Genes	Speech therapist	Patient representative
Carmen Pérez Mateos	Technical Department, Secretariat of Basic NHS Services and Cohesion Fund. D.G. Basic NHS Services and Pharmacy		Public administration (MSSSI)

## SPAIN– EUROPLAN National Conference Final Report

Caterina Aragón	FEDER Catalunya	Manager of FEDER Catalunya	FEDER
Clotilde de la Higuera		APERTCras	Patient representative
Cristina Pérez Vélez	CREER	Psychologist	Public administration (MSSSI)
Elvira Bel	Committee of Experts of FEDER / Main Lecturer of Barcelona University		Academic/Researcher
Encarnación Guillén	Committee of Experts of FEDER		Academic/Researcher
Enrique Galán Gómez	Committee of Experts of FEDER		Academic/Researcher
Enrique López	Spanish Narcolepsia Association	President	Patient representative
Enrique Peiró Callizo	Osakidetza	Coordinator of Public Health and Patient Safety programs	Public administration (Autonomous Community of País Vasco)
Esther Cámara	AEPPEVA	President	Patient representative
Esther Vicente Cemborain	Rare Diseases Poblational Registry of Navarra. Public Health and Labour Institute. Autonomous Community of Navarra		Public administration (Autonomous Community of Navarra)
Estrella Guerrero Solana	DEBRA España	Responsible of Social Area	Patient representative
Estrella Mayoral Rivero	FEDER	Responsible of Social Area of FEDER	FEDER
Eva Bermejo	Research in Rare Diseases Institute (IIER). Institute of Health Carlos III (ISCIII)	Main Researcher; Coordinator of UIAC_IIER	Public administration (MSSSI)
Evanina Morcillo Makow	DEBRA	Director	Patient representative
Federico Eduar Arribas Monzón	Representative of the Autonomous Community of Aragón in the RD Strategy of the NHS		Public administration (Autonomous Community of Aragón)
Félix Casado Soto	5p- Syndrome Foundation	Secretary	Patient representative
Fernando Torquemada	FEDER	Legal Adviser	FEDER
Francesc Palau Martínez	CIBERER. ISCIII. MINECO Príncipe Felipe Research Centre, Valencia	Director	Public administration (MINECO)
Gema Chicano Saura	FEDER	Representative of FEDER in EURORDIS; AADE	FEDER
Gema Esteban Bueno	FEDER	Board member	FEDER
Graciela Susana Bosca	Costello y CFC Spanish Association	Social Worker	Patient representative
Guadalupe Álvarez-Rodríguez Desch	Rare Disease Association D,Genes	Physiotherapist	Patient representative
Guiomar Pérez de Nanclares	AEGH		Academic/Researcher
Ignacio Blanco Guillermo	Germans Trias Hospital   Gerència Metropolitana Nord   Institut Català de la Salut	Coordinator – Programa d'Assessorament i Genètica Clínica	Academic/Researcher
Ignacio Abaitua	Research in Rare Diseases Institute (IIER). Institute of Health Carlos III (ISCIII)		Public administration (MSSSI)

## SPAIN– EUROPLAN National Conference Final Report

Imanol Amayra	Deusto University	Director of the researcher group on Neuromuscular and Neurodevelopment Disorders	Academic/Researcher(university)
Inés Elizaga González	Spanish Rubinstein-Taybi Syndrome Association		Patient representative
Inmaculada Arroyo Manzanal	CREER	Nurse	Public administration (MSSSI)
Isabel Fernández	FEDER Murcia	Manager of FEDER Murcia	FEDER
Isabel Moreno Portela	Technical Department, Secretariat of Basic NHS Services and Cohesion Fund. D.G. Basic NHS Services and Pharmacy		Public administration (MSSSI)
Isabel Motero Vázquez	FEDER Andalucía	Manager of FEDER Andalucía	FEDER
Javier Alonso	Research in Rare Diseases Institute (IIER). Institute of Health Carlos III (ISCIII)	Head of Human Genetic Division	Public administration (MSSSI)
Javier López Pisón	Representative of the Autonomous Community of Aragón in the RD Strategy of the NHS		Public administration (Autonomous Community of Aragón)
Jesús Sueiro Justel	SEMFYC		Academic/Researcher
Jordi Cruz	FEDER	Board member	FEDER
José Antonio de la Rica Giménez	EAEren Koordinatzaile Soziosanitarioa	Social and Health coordinator	Public administration (Autonomous Community of País Vasco)
José Antonio Sánchez Alcázar	CABD-CSIC University - Pablo de Olavide University (Sevilla)	University Lecturer	Academic/Researcher(university)
José Luis González Cerezo	Spanish Miastenia Associaton (AMES)		Patient representative
José M <sup>a</sup> Espinalt	AELMHU	Vice President	Industry
José María Casado	FEDER	Navarra's coordinator	FEDER
José María Millán	Genetic Unit. La Fe University Hospital	Scientific Coordinator of the RD Strategy of the NHS	Academic/Researcher
José María Soria de Francisco	Spanish Ictiosis Association	Board member	Patient representative
Jose Vicente Sorli Guerola	SEMFyC		Academic/Researcher
Juan Cruz Cigudosa	AEGh	President	Academic/Researcher
Juan Manuel Fontanet	Committee of Experts of FEDER /Clinic Pharmacologic Service, Santa Creu i Sant Pau Hospital		Academic/Researcher
Juana M <sup>a</sup> Sáenz	FEDER Basque Country	Representative FEDER Basque Country	FEDER
Julián Isla	Dravet Syndrome Foundation	President	Patient representative
Justo Herranz Arandilla	FEDER	Board member	FEDER

## SPAIN– EUROPLAN National Conference Final Report

Laura Ranz Carazo	Foundation Against Pulmonary Hypertension	Department Manager	Patient representative
Luis Cruz	AELMHU	President	Industry
Luis Miguel Ruíz Ceballos	D. G. Health care and Regulation. Department of Health and Social Services, Cantabria	Head of Health care Service	Public administration (Autonomous Community of Cantabria)
Luisa María Botella Cubells	Spanish HHT Association		Patient representative
M <sup>a</sup> Carmen Murillo	FEDER	Technical of Promotion of a Positive Image Area	FEDER
M <sup>a</sup> Dolores Camero Melero	Andalucian Haemophiliacs Association	President	Patient representative
M <sup>a</sup> Dolores Montero	Spanish Agency for Medicinal Products and Medical Devices (AEMPS)	Head of Farmacovigilance Division	Public administration (MSSSI)
M <sup>a</sup> Elena Escalante	FEDER	Representative of FEDER Madrid	FEDER
M <sup>a</sup> Jesús García Barcina	Osi Bilbao-Basurto	Responsible of Genetics Laboratory	Public administration (Autonomous Community of Basque Country)
M <sup>a</sup> Jesús Muñoz Huerta	Rare Diseases Association of Guadalajara		Patient representative
M <sup>a</sup> Patrocinio Verde González	Spanish HHT Association		Patient representative
M <sup>a</sup> Victoria Benavides Sierra	Sub-Directorate of Information & Care for Patients. DG Patient Services. SERMAS Madrid	Assistant Technician	Public administration (Autonomous Community of Madrid)
Mamen Almazán Peña	FEDER Madrid	Psychologist	FEDER
Manuel Errezola Saizar	Department of Health; Autonomous Community of the Basque Country	Head of Registries and Health Information Service.	Public administration (Autonomous Community of the Basque Country)
Manuel Hens Pérez	Research in Rare Diseases Institute (IIER). Institute of Health Carlos III (ISCIII)	Head of Service	Public administration (MSSSI)
Manuel Pérez Fernández	Association of Pharmacists of Seville (RICOFS)	President	Academic/Researcher
Manuel Posada de la Paz	Research in Rare Diseases Institute (IIER). Institute of Health Carlos III (ISCIII)	Director IIER	Public administration (MSSSI)
Marga Iniesta	AELMHU	Executive Director	Industry
María Ángeles Baños Baños	CREER	GP	Public administration (MSSSI)
María del Prado Capilla	Phelan McDerimid Syndrome Association		Patient representative
Mario Margolles	Representative of the Autonomous Community of Asturias in the RD Strategy of the NHS		Public administration (Autonomous Community of Principado de Asturias)
Marta Fonfría Solabarrieta	CREER	Social Educator	Public administration (MSSSI)
Marta Moreno	CELGENE	Director of Market Access and Regulation	Industry

## SPAIN– EUROPLAN National Conference Final Report

Mercedes Jaraba Sánchez	Secretariat for Participation & Supervised Entities. D.G. Support for Policies for the Disabled	Deputy Director General	Public administration (MSSSI)
Mercedes Martínez Vallejo	Secretariat for Quality of Drugs and Medicinal Products. D.G. Basic NHS Services and Pharmacy	Technical Adviser	Public administration (MSSSI)
Miguel García Ribes	SEMFYC		Academic/Researcher
Miriam Torregrosa Granado	FEDER Madrid	Manager of FEDER Madrid	FEDER
M <sup>o</sup> de Luján Echandi García Herrera	FEDER	Head of Associations Management	FEDER
Modesto Díez Solís	FEDER	Representative of FEDER Extremadura	FEDER
Mónica Blanco Marengo	CRECER Madrid		Patient representative
Mónica Rodríguez	FEDER	Board member	FEDER
Montserrat Cabrejas del Campo	CREER	Pre-primary education	Public administration (MSSSI)
Natividad López Langa	Madrid Association of Nursing in Education Centres. AMECE	President	Patient representative
Norma Alhambra Jiménez	Phelan McDermid Syndrome Association		Patient representative
Nuria María Garrido Cuenca	Spanish Mastocytosis Association		Patient representative
Óscar Zurriaga	Committee of Experts of FEDER /Directorate Gen. of Public Health. Department of Health. Valencian Regional Government	Chief of Service of Epidemiological Studies and Health Statistics	Academic/Researcher
Pablo Rodríguez Martínez	Representative of the Autonomous Community of Valencia in the RD Strategy of the NHS		Public administration (Autonomous Community of Valencia)
Paloma Casado Durandez	Directorate General of Public Health, Quality and Innovation (MSSSI)	Deputy Director General of Quality and Cohesion	Public administration (MSSSI)
Patricia Carrillo Ojeda	Canaries Health Service Management	Head of Information Systems Service	Public administration (Autonomous Community of Canarias)
Patricia Carrillo Ojeda	Canaries Health Service Management	Head of Information Systems Service	Public administration (Autonomous Community of Canarias)
Patricia García Luna	Andalusian Association of ELA - CRECER Association		Patient representative
Patricia García Primo	Rare Diseases Registry (IIER)		Public administration (MSSSI)
Pilar Magrinyà Rull	Directorate of Health Care Department of Health Autonomous Community of Catalunya		Public administration (Autonomous Community of Catalunya)
Pilar Sánchez-Porro	Centro Base nº 7, Social Services Department. Autonomous Community of Madrid	Research doctor	Public administration (Autonomous Community of Madrid)

## SPAIN– EUROPLAN National Conference Final Report

Pilar Soler Crespo	Technical Secretariat of the Rare Disease Strategy of the SNS, Quality and Cohesion Section. D.G. Public Health, Quality and Innovation	Head of Service	Public administration (MSSSI)
Roberto Zarrabeitia Puente	HHT Unit. Internal Medicine Service. Sierrallana Hospital of Cantabria		Patient representative
Rocío García Yuste	Phelan McDermid Syndrome Association		Patient representative
Rufino Álamo Sanz	Directorate General of Public Health. Department of Health. Autonomous Community of Castilla y León	Head of Public Health Information Service	Public administration (Autonomous Community of Castilla y León)
Ruth Rabaneda	APERTCras	President	Patient representative
Santiago Culí	Boehringer Ingelheim	Director of Communication and Public Affairs	Industry
Santiago de la Riva	FEDER	Board member	FEDER
Sara Pérez	APERTCras	Neuropsychologist	Patient representative
Sofía González	Centro Base nº 5 , Department of Social Services, Autonomous Community of Madrid	Psychologist	Public administration (Autonomous Community of Madrid)
Sonia Grandes Velasco	Williams Syndrome Association Madrid		Patient representative
Teresa Pampols	Committee of Experts of FEDER	Coordinator	Academic/Researcher
Thais Pousada García	Committee of Experts of FEDER		Academic/Researcher
Tomás del Castillo	FEDER	Board member	FEDER
Vanesa Pizarro Ortiz	FEDER	Project Manager	FEDER
Verónica Alonso Ferreria	Research in Rare Diseases Institute (IIER). Institute of Health Carlos III (ISCIII)	Main Researcher, RD Epidemiology Area	Public administration (MSSSI)
Victor Volpini	AEGh	Vice President	Academic/Researcher
Yolanda Ahedo	CREER	Pedagogue	Public administration (MSSSI)
Yolanda Barrios García	CREER	Social Worker	Public administration (MSSSI)

## 2nd Spanish EUROPLAN National Conference, 20-21 November 2014



**Workshop: Methodology, Governance and Monitoring of the National Plan**



**Workshop: Research on Rare Diseases**



**Workshop: Care Model**



**Workshop: Definition, codification and inventorying**



**Workshop: Centres of Reference**



**Workshop: Orphan Medicinal Products**



**Workshop: Information and Training**



**Workshop: Social Services for Rare Diseases**



**Workshop: Educational Inclusion**



**Workshop: Labour Inclusion**

