

# PORTUGAL

## EUROPLAN NATIONAL CONFERENCE

### FINAL REPORT

27- 28 February 2015, Lisbon

# 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 Commission RD Expert Group (former EUCERD) over the period March 2012 until August 2015.

The EUROPLAN National Conferences present the European rare disease policies as well as the European Commission RD Expert Group 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 Medicinal Products;
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>	Portugal
<b>Date &amp; place of the National Conference</b>	February 27 and 28 2015, National Parliament, Lisbon
<b>Website</b>	<a href="http://www.aliancadoencasraras.org">www.aliancadoencasraras.org</a> <b>Conference website:</b> <a href="http://ddr2015.aliancadoencasraras.org/">http://ddr2015.aliancadoencasraras.org/</a>
<b>Organisers</b>	Aliança Portuguesa de Associações das Doenças Raras (Portuguese Alliance of Rare Disease Associations) - Aliança
<b>Members of the Steering Committee</b>	Filipe Assoreira   P-Bio, Portuguese Association of Bioindustry Francisco George   Director-General of Health Inês Leal de Faria   Aliança Jorge Sequeiros   Orphanet-Portugal Luís Brito Avô   Rare Diseases Working Group, SPMI Maria de Belém Roseira   Member of the National Parliament Marta Jacinto   Aliança Lene Jensen   EUROPLAN Advisor
<b>Names and list of Workshops</b>	<b>Workshop theme 1:</b> Methodology, Governance and Monitoring of the National Plan <b>Workshop theme 2:</b> Definition, Codification and Inventorying of Rare Diseases (includes information and training) <b>Workshop theme 3:</b> Research on Rare Diseases <b>Workshop theme 4:</b> Centres of Expertise (CE) / European Reference Networks / Cross-Border Health Care <b>Workshop theme 5:</b> Orphan Medicinal Products and Rare Disease Treatments <b>Workshop theme 6:</b> Social Services for Rare Diseases
<b>Workshop Chairs (and Rapporteurs, where applicable)</b>	<b>Workshop 1:</b> <b>Moderator:</b> Rui Gonçalves   Hospital D. Estefânia (D.

	<p>Estefânia Hospital), Lisbon  <b>Rapporteur:</b> Teresa Coelho   Unidade Clínica de Paramiloidose Corino de Andrade, CHP (Paramyloidosis Clinic of Oporto Medical Centre), Oporto</p> <p><b>Workshop 2:</b>  <b>Moderator:</b> Margarida Reis Lima   Hospital Lusíadas (Lusíadas Hospital), Oporto  <b>Rapporteur:</b> Lina Ramos   Hospital Pediátrico, CHUC (Pediatric Hospital of Coimbra), Coimbra</p> <p><b>Workshop 3:</b>  <b>Moderator:</b> Patrícia Maciel   ICVS, Universidade do Minho (ICVS, University of Minho), Braga  <b>Rapporteur:</b> Isabel Marques Carreira   Faculdade de Medicina, Universidade de Coimbra (School of Medicine, University of Coimbra), Coimbra</p> <p><b>Workshop 4:</b>  <b>Moderator:</b> Eduardo Silva   Faculdade de Medicina, Universidade de Coimbra (School of Medicine, University of Coimbra), Coimbra; and CHLN, Lisbon  <b>Rapporteur:</b> Luísa Diogo   President, SPDM (Portuguese Society of Metabolic Diseases), Coimbra</p> <p><b>Workshop 5:</b>  <b>Moderator:</b> Fátima Vaz   IPO (Portuguese Oncology Institute), Lisbon; and INFARMED (National Authority of Medicines and Health Products)  <b>Rapporteur:</b> Carolino Monteiro   Faculdade de Farmácia da Universidade de Lisboa (School of Pharmacy, University of Lisbon), Lisbon</p> <p><b>Workshop 6:</b>  <b>Moderator:</b> Paula Silva   Faculdade de Medicina da Universidade do Porto (School of Medicine, University of Oporto), Oporto; and IPATIMUP, Oporto  <b>Rapporteur:</b> Helena Machado   CES, Universidade de Coimbra (CES, University of Coimbra), Coimbra</p>
<p><b>Annexes:</b></p>	<p><b>I. Programme in English</b>  <b>II. List of Participants</b></p>

## II. MAIN REPORT

### Plenary Report – Opening Session

The plenary session was chaired by Professor Constantino Sakelarides, scientific ambassador of APADR – Aliança Portuguesa de Doenças Raras (Aliança) for the Rare Disease Day.

The national situation was presented by João Lavinha, from INSA (National Institute of Health Dr. Ricardo Jorge); and the European situation was presented by Lene Jensen, EURORDIS representative, EUROPLAN Advisor and Chief Executive Officer of the Danish National Alliance for Rare Diseases.

#### Country status

João Lavinha presented the Portuguese situation in the field of Rare Diseases, listing the adverse characteristics of these diseases and their inherent problems.

Then, he described chronologically the development of policies for Rare Diseases in Portugal (legislation):

- 2008 – Approval of the National Plan for Rare Diseases (NPRD), involving many contributions from all stakeholders: doctors, patients, pharmaceutical industry, researchers and caregivers
- 2011 – Approval of the National Network of Centres of Expertise for Rare Diseases (RD)
- 2013 – Three Centres of Expertise for lysosomal diseases were established (but no criteria for choosing these centres were ever defined)
- 2014 – RD Card launched by the DGH (Directorate-General of Health); 513 requests and 175 cards issued by November 2014
- 2014 – Regulation for National Centres of Expertise (“Centros de Referência”, CE) not specific for Rare Diseases, but with specific reference to them. The concept of CE was laid down and a national committee to set them up appointed. This national committee has annually to submit the centres of expertise to be established each year to the Ministry of Health
- 2015 – January: a list of centres of expertise to create in 2015 was issued and approved, which included as per RD, the hereditary metabolic disorders and paramyloidosis

In this sense, João Lavinha emphasized that this policy has been erratic, causing the stakeholders to be 7 years behind.

In fact, he informed that the National Strategy for Rare Diseases<sup>1</sup> will be announced and repeal the 2008 NPRD. This Strategy shall create a macro structure, an inter-ministerial committee

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<sup>1</sup> Note from Aliança: This strategy was publically announced on 28 February, after the end of the conference

with representatives from the Ministry of Health, Social Security, Education and Science, which will constitute its governing body. The inter-ministerial coordination may also be extended to other areas of governance, such as the line ministry for spatial planning which can regulate accessibility for transportation, urbanisation, infrastructures and leisure facilities, etc.

In fact, nothing guarantees that this Strategy will be implemented, either because of lack of funding, which is never mentioned, or because of the type of coordination foreseen. Regardless of the number of members, the presidency will be under the Directorate-General of Health (DGS), which does not seem a good idea, once that it is one of the partners. This body should be independent and executive, in order to facilitate the operationalization of decisions among stakeholders. The governing body for this new Strategy does not include associations or patient representatives, nor health professionals specialized on RDs.

In addition to the legislation, and moving on to the operational field, other problems were noticed, such as the existence of multiple centres treating only a small number of patients, the absence of systematic national registries (and the few that exist are not consistent), and the existence of two aggregating structures of RD patients associations, which can weaken the intervention of rare disease patients and their families in the defence of their interests.

On the positive side, the universal screening of newborns for congenital hypothyroidism and a group of inherited metabolic disorders, working well for many years, free of charge and has had very good results, covering 98-99% of the national population.

Also, the research conducted is of high-level, despite the financial support being primarily centered on the Foundation for Science and Technology (FCT). There are also small groups of researchers with little national competitive ability, but which take part in several international consortia.

### **European policy and guidelines**

Lene Jensen presented the European situation in the field of Rare Diseases.

To begin with, she raised the following question: Why an European dimension? Well, she continued, for rare diseases there are few patients, less doctors, nurses, scientists, etc. and getting efforts together all over Europe is the most useful attitude to help patients and families.

Organizing patients and making their voice heard is of the uttermost importance, for they are experts on their diseases, and have the most gain and are the ultimate users of RD policy.

EURORDIS, a pan-European organisation that fights for patients' rights in Europe, supporting people living with rare diseases in building a strong community, was also presented to the audience. This organisation has over 600 Patient Associations, which represent over 4.000 rare

diseases in more than 60 countries.

Next, Lene Jensen presented some of the EU measures to support the RD community:

- the strengthening cooperation and coordination among European countries;
- the creation of European Reference Networks, linking centres of expertise and professionals in several countries, to share knowledge and experience across borders;
- encouraging research
- evaluation of current screening population practices;
- support to RD registries and project for an European platform for RD registration.

Various actions have been taken since the early 1990s, in order to attain these objectives, and public health programmes have been adopted in the different EU countries, more efficiently in some than in others.

She highlighted the importance of the creation of ORPHANET. Afterwards, she highly stressed that Orphanet-Portugal is one of the best organised in Europe, with very reliable information and a highly specialised team.

Afterwards, a few key documents issued between 2000 and 2011 were presented, with links to the original sources, namely:

- Regulation (EC) number 141/2000 of the European Parliament and of the Council of 16th December 1999<sup>2</sup> on Orphan Medicinal Products provides rules for designation as an «OMP», when intended for the diagnosis, prevention or treatment of a life threatening or chronically debilitating condition affecting no more than 5 in 10.000 persons
- Commission Communication on rare diseases: Europe's challenge of 11 November 2008<sup>3</sup> offers perspectives on the elaboration of some of the common instruments to tackle rare diseases, especially on diagnostics and medical care and European guidelines on population screening
- Council Recommendation on an action in the field of rare diseases of 8th June 2009<sup>4</sup>
- Directive of the European Parliament and of the Council of 9 March 2011 on patients' rights in cross-border healthcare<sup>5</sup>

The establishment of the EUROPLAN project<sup>6</sup>, with both EUROPLAN I (2008-2011) and EUROPLAN II (2012-2015) was also highlighted:

- The EUROPLAN recommendations provide tools for Member States to develop a plan or strategy, linking it with a common framework at the European level

<sup>2</sup> [http://ec.europa.eu/health/files/eudralex/vol1/reg\\_2000\\_847/reg\\_2000\\_847\\_pt.pdf](http://ec.europa.eu/health/files/eudralex/vol1/reg_2000_847/reg_2000_847_pt.pdf)

<sup>3</sup> <http://ec.europa.eu/transparency/regdoc/rep/1/2008/PT/1-2008-679-PT-F1-1.Pdf>

<sup>4</sup> <http://eurlex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:pt:PDF>

<sup>5</sup> <http://eurlex.europa.eu/legalcontent/PT/TXT/PDF/?uri=CELEX:32011L0024&from=EN>

<sup>6</sup> <http://www.europlanproject.eu/Documentation?idDocumentationType=2&idDocumentationTypeChild=3>

- This "double level" approach is respectful of national decisions, but is expected to ensure a coherent and consistent progress in EU care for rare diseases
- The project includes 15 EUROPLAN National Conferences in 2010-2011 and 25 EUROPLAN National Conferences in 2012-2015; these conferences share the same format and content guidelines
- Over 50 indicators were defined to evaluate the achievements of rare diseases initiatives, to capture relevant data and information on the process of planning, implementing and monitoring of plans and strategies
- 22 key indicators have been selected by the European Commission Expert Group on Rare Diseases, to be collected once a year from all EU Countries<sup>7</sup>

Finally, she mentioned that a Commission Expert Group on RD has been created<sup>8</sup>. The members of this group are National Authorities from Member States, Third Countries and Candidate Countries, Medical professions, Corporate International organisations and, of course, patient representatives!

### **Debate**

Considering the realistic and disturbing picture presented on the Portuguese situation, the debate ended up focusing on four major themes: the tendency to seek inter-sectoral solutions for problems involving one sector; the report with 22 indicators on rare diseases; small, multiple organizations and their interaction; the state of regulation in Portugal.

When something does not work well in a sector, there is a tendency to seek an inter-sectoral solution. This is not a good idea, because complex and difficult inter-sectoral solutions always require a strong focus on strategic coordination in each sector. Therefore, an inter-sectoral organization should be concerned about the several aspects of rare diseases beyond the health sector, which will work if the health sector is organized in such a way that it allows it to do so. The one sector's organization failures should not be transferred to the inter-sectoral level.

An inter-sectoral strategy only makes sense if the pillars on which it is based are well structured, which is far from happening. In the Ministry of Health's present situation, it is risky to repeal the work already structured, which needed only to be put in place.

Moving on to a more complex situation as the one proposed, with communication difficulties among the different sectors involved without the consolidation of each pillar, can be a disaster waiting to happen. Nevertheless, those heard by the DGH regarding the new Strategy hope that once the policy-makers were alerted, there has been sensitivity from lawmakers and

<sup>7</sup> [http://www.eucerd.eu/wpcontent/uploads/2013/06/EUCERD\\_Recommendations\\_Indicators\\_adopted.pdf](http://www.eucerd.eu/wpcontent/uploads/2013/06/EUCERD_Recommendations_Indicators_adopted.pdf)

<sup>8</sup> [http://ec.europa.eu/health/rare\\_diseases/expert\\_group/index\\_en.htm](http://ec.europa.eu/health/rare_diseases/expert_group/index_en.htm) and [http://www.eucerd.eu/?page\\_id=15](http://www.eucerd.eu/?page_id=15)



policy-makers to pave the way and incorporate some of recommendations made.

Regardless of the national plans or strategies, the most important for patients and their families is to get a diagnosis and treatment, if it exists. Given that for most cases there is no treatment, these patients need support in their daily lives, and it is imperative to see everything as a whole. The first concern is if health care system works well.

Currently, and despite the various existing reports, and opinions from the several stakeholders, in the end what remains is the report from the Ministry of Health, which responds to the 22 indicators for RD requested from the Portuguese government. In this report, the conclusion always shows that in Portugal all runs beautifully well, which often does not correspond to reality.

It is important to have two reports: one from the government and the other from patients, and that the European Commission (EC) accepts these two documents. The 22 indicators can be the shared structure to report. It is important for them to contain what happens and promote few actions, ones that can make a difference.

Therefore, it is very useful for patient organizations to be heard in an EU organization, even if informally. It is of the utmost importance to coordinate the various skills, so that there is a good response for people with RD and this should be the role of a programme for rare diseases.

Let our voice be heard in what concerns the European Commission RD Expert Group reports about the state of the art in each country.

The existence of small and local organizations is a good thing in itself, because they will be able to work better in the interest of patients. The problem is that they do not communicate with each other, and no convergence exists in terms of making demands at governmental level.

On the other hand, the absence of appropriate strategies eventually leads to the existence of many organizations which do not meet the needs of the community as a whole.

The National Plan for Rare Diseases (NPRD) is a document involving knowledgeable people on Rare Diseases – doctors, scientists, associations, patients and others, and it was a good basis for it. At a moment when there is already an awareness and extensive involvement in the subject, to create an inter-sectoral organization which in our country has never been put into practice, corresponds to throwing it all away. There are already good initiatives with good results. One should not waste more time on organization than with practical actions, and everything should be done for the patient and not for the government.

Unfortunately, things have been done and redone. For example, the Committee on RD did a good job based on the European practices and the existing situation in Portugal, but this work has been completely modified in the next phase. You cannot spend your life changing things, in

particular the work done by technical experts which cannot be modified every time a government changes: one thing is politics and another thing is the work done by technical experts.

When we started we were one of the most advanced countries regarding some standards, at this moment we are very far away from them.

It is important to stress that the European recommendations remain in place, whichever government, so it even makes less sense to change the health policies every time there is a change in government. An option is to work with the permanent officials, since they remain in office and know the situation better than we do.

These leverage points can change the situation for Portugal to reach a different level.

### Official Opening Session

Given that the EUROPLAN National Conference was held in National Parliament, the Steering Committee decided to include as additional session, an official opening session.

Present at the session<sup>9</sup>:

- Maria de Belém Roseira (that presided) – Member of National Parliament, representing the President of the Parliament Health Commission
- Jorge Sequeiros – Coordinator of Orphanet-Portugal
- Constantino Sakelarides – Scientific ambassador of Aliança for the Rare Disease Day
- Ana Bacalhau – Aliança’s Ambassador
- Marta Jacinto – Aliança’s President

All stakeholders were represented in the audience: academia/researchers, doctors, nurses, therapists, other health care professionals, industry, social services, educators, patients, caregivers/relatives, politicians, public administration (local, regional or national).

Maria de Belém Roseira opened the session and thanked Aliança for organizing the meeting and gave the floor to each member.

Jorge Sequeiros started by mentioning that several documents had been elaborated by experts, scientists and Patient Associations since 2008, and that none of the proposed projects were taken forward. He defended that there was a fear that the specificities of rare diseases would be lost with the new National Strategy to be announced soon, though these specificities are emphasised by the EU, the European Expert Group on Rare Diseases (former EUCERD) and DG SANCO.

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<sup>9</sup> Mr Alexandre Diniz was appointed by Mr Francisco George, Director-General of Health, to represent the DGHealth. Nevertheless Mr Diniz did not participate in the conference

Marta Jacinto thanked the National Parliament on behalf of the Aliança's Board for providing the space and the setting, as well as the Steering Committee's participation, and mentioned that organizing this conference was quite a challenge. Aliança decided to take up such challenge because of the importance to implement a plan for RD and alert the decision-makers on the needs of rare disease patients.

Then, she asked everyone, especially the National Parliament, to support 2019 as the European Year for Rare Diseases. Finally, she expressed the Aliança's readiness to hold information sessions for the members of Parliament, highlighting the difficulties affecting rare diseases every day.

Constantino Sakellarides began by saying that the issue of RD is a matter of democracy. Democracy is only achieved when it includes minorities, such as people with rare diseases, and provides specific answers to very particular needs.

To avoid the eventual idyllic descriptions made annually by the national health authorities on the 22 indicators set out by the European Commission, it was proposed that the Patient Associations together with the professionals who work with them, elaborate an annual description of the country's situation: this report elaborated by the patients and professionals who work with them would have the advantage of dialoguing with the National Report, therefore improving it.

On the other hand, it is known that such problems require an inter-sectoral response. But each sector must be strong in its strategy, because otherwise it will take its weaknesses, and not its strengths, into the inter-sectoral level.

With scientific and knowledge breakthroughs in various disciplines, we are all paving the way to being rare persons. Therefore, by being concerned about people with rare diseases today, we are anticipating the future.

Ana Bacalhau thanked for the invitation, which she accepted with great pleasure.

Maria de Belém Roseira welcomed the other member of the Parliament who attended the session and underlined the importance of holding this conference at the National Parliament - home of democracy, where a lot can be heard, discussed and prepared, so as to be applied in the future. It is an important stage for having the attention of the media and giving visibility to the existence of rare diseases.

She suggested to engage the National Parliament, in particular the members of the Parliament's Health Commission, to have internal competence to legislate and supervise properly, ensuring that things are well done and well legislated.

Finally, she thanked the presence of all Parliament Officials that supported this conference, in the person of the President and Secretary-General of the NP.

## Report of Workshops

### Theme 1 - Methodology, Governance and Monitoring of the National Plan

#### Sub-Themes:

- 1.1 Mapping policies and resources
- 1.2 Development of a National Plan / Strategy
- 1.3 Structure of a National Plan / Strategy
- 1.4 Governance of a National Plan
- 1.5 Dissemination and communication on the National Plan
- 1.6 Monitoring and evaluation of the National Plan
- 1.7 Sustainability of the National Plan

#### Workshop:

- 1.1 Mapping policies and resources

#### Debate:

From the discussion, it was said that there was no palliative or continuous care for rare diseases.

#### Proposals:

- Mapping patients' needs, and those of caregivers and of associations.
- Seek collaboration from academia, for example, research grants for this work.

- 1.2 Development of a National Plan/Strategy

#### Debate:

In Portugal, there is a National Programme for Rare diseases (NPRD<sup>10</sup>) since 2008, based on a participatory approach by several RD stakeholders.

- 1.3 Structure of a National Plan/Strategy

#### Debate:

The NPRD is based on 3 main pillars, namely: intervention strategies, training, collection and data analysis. A cross-cutting approach to RD should be ensured.

The general objectives<sup>11</sup> and specific objectives<sup>12</sup> are defined, as well as a time frame (which

<sup>10</sup> Note from Alliance: The NPRD may be consulted on <http://www.portaldasauade.pt/NR/rdonlyres/555DD3B3-45F0-4F74-B633-28889E721BF1/0/i010420.pdf>

was not respected). Clear measurable results associated with each action were not defined. Most of the objectives of the NPRD that were actually achieved were the result of the work of the national team of Orphanet, and from single actions carried out by proactive persons.

#### 1.4 Governance of a National Plan

##### **Debate:**

The debate ended up by focusing on the identification of what did not go well in the NPRD's implementation, which was not fulfilled.

Possibly the methodology and the type of governance, as well as weak monitoring, prevented the implementation of almost all of the NPRD. It was necessary to establish plans in order to facilitate their implementation, governance, and monitoring. The latter would fulfil a timeframe for priority actions and reports. It happens that no time frame (other than that the included in the NPRD which was not implemented) was ever elaborated.

There is still a need for sustainability, in addition to structures, continuity and implementation, which depends on the money allocated. There was no allocation of economic resources for the implementation of the NPRD.

Additionally, there are no statistics and measures taken from a global point of view. A mention was made that the smallest progress has been in terms of social support.

##### **Proposals:**

- It is essential to have a real NPRD and not just a virtual one
- Create a Steering and Monitoring Committee, chosen in a transparent manner, including patients and their representatives and professionals specialised in RD
- Actions should be taken from a global point of view: from the patient, caregiver and social resources

#### 1.5 Dissemination and communication of the National Plan

##### **Debate:**

It was considered that the assessment should take into account the number of actions carried

<sup>11</sup> *Note from Alliance:* a) Improve the national responses to the unmet health needs of RD patients and their families; b) Improve the quality and equity of health care provided to RD patients.

<sup>12</sup> *Note from Alliance:* a) Creating a national network of centres of expertise for rare diseases; b) Improve the access to appropriate care for people with rare diseases; c) Improve the mechanisms for integrated management of RD; d) Improve the responses to the needs of patients and families; e) Increase the knowledge and national research on rare diseases; f) Promote therapeutic innovation and accessibility to Orphan Medicinal Products ; g) Ensure cross-border cooperation within the EU and Community of Portuguese Speaking Countries – CPLP.

out and mapping the knowledge on rare diseases.

According to the participants, knowledge on RD and the NPRD is scarce. In effect, even if there is broad knowledge on biotechnology, in health centres, public contact services and hospital emergency rooms, there is often insufficient or no knowledge at all.

With regard to associations, there was a lack of communication between the Committee for the implementation of the National Programme for Rare Diseases (CNPRD) and patient organizations, as well as between the CNPRD and other partners.

There is a lack of transparency from the DGH on the NPRD, which prevents patient organisations to consult an entity because of being unaware of its existence.

The Committee for the announced new National Strategy for RD, has already been designated and will not contemplate health professionals linked to RD or Patient Associations, etc. The workshop participants were unaware of its action.

**Proposals:**

- Conduct training sessions aimed at health professionals, namely from General Practice and Family Medicine.
- Conduct training sessions for high-school and university students, for example with the support of the Portuguese Society of Human Genetics.
- Propose changes in the curricula to include more information about genetics and RD.
- Create a template roadmap to give each family when a new diagnosis is made, accompanying the diagnosis, follow-up, treatment, etc.
- Propose a trans-disciplinary discussion within the Ministries of Education, Social Security and Health, etc., on all problems faced by RD patients.
- Follow the strategy adopted by other countries where there was the same difficulty of dissemination.
- Include all stakeholders in governance, and monitoring of the implementation of the NPRD or whatever replaces it.
- Implement a strategy to ensure support on the specificities of RD and its related difficulties, for which diagnosis is difficult and quite often much delayed, and there is no treatment or preventive measures.
- Integrate what is possible in treatment protocols for other diseases. Only focus on what is different from other diseases in RD. What can be integrated should be integrated.
- Take advantage of the work developed so far and maintain continuity.

## 1.6 Monitoring and evaluation of the National Plan

### **Debate:**

The participants tried to map the existing multidisciplinary consultations.

It was concluded that there is a lack of knowledge on the patients' real situation, namely from a social and psychological point of view. There is also a lack of management of patient situations by the health authorities. Finally, there is lack of coordination between hospital structures and the ministries involved.

On the other hand, supervision of paediatric patients until adulthood only exists in some hospitals and the participants only identified one in those conditions. Still, in this case, as for all others, there is no mapping of existing cases, patients or consultations.

### **Proposals:**

- Establish and promote the link between structures (health, social, etc.) and information within hospitals.
- Define a practical structure in the hospitals for referral of a RD patient to the right consultation.
- Prepare the transfer of paediatric patients with chronic disease or RD between 16 and 18 years old to a closer structure where they will be supervised as adults.
- Maintain, during that period, mid-term consultations to ensure transition.
- Prevent fragmentation of the various specialties in the care for RD.
- Mandatory appointment of a person in charge/manager/partner to centralize the care to be provided to any given patient when there is a need for multidisciplinary care.
- Each patient should be entitled to have a RD manager.

## 1.7 Sustainability of the National Plan

### **Debate:**

More than giving answers, this debate raised several questions.

Is there a methodology to achieve the objective? Are there any funds? Where and how should they be applied? Equitably by all *stakeholders*?

Where should the existing budget allocation focus, in order to maintain long term sustainability?

Will there be a budget dedicated to RD, or should one assume that this budget will not exist?

How to pay for treatments that are always expensive and how to equally treat all patients?

How to transport patients to treatment?

**Proposals:**

- Establish a fund to train family doctors with regard to RD.

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

**Sub-Themes:**

- 2.1 Definition of RD
- 2.2 Codification of RD and traceability in national health system
- 2.3 Registries and databases
- 2.4 Information on available care for RDs in general, for different audiences
- 2.5 Help lines
- 2.6 Training healthcare professionals to recognise and code RD
- 2.7 Training healthcare professionals

**Workshop:**

- 2.1 Definition of RD

This sub-theme did not raise any questions, since Portugal adopted the European Definition of Rare Diseases: a disease affecting no more than 5 in 10.000 individuals (less than 1 in 2.000 individuals).

- 2.2 Codification of RD and traceability in national health system

**Debate:**

*“Without codification there is no disease”* was used as the motto for the discussion on this sub-theme.

In the Directorate-General of Health (DGS), there is a Unit dedicated to codification for the Ministry of Health (MH). The ICD-10 statistical classification is officially used by all health units, and the codes are registered by computer in all clinical files. To this end, there are Coding Doctors paid by the Ministry of Health, which review the files and code them according to ICD-10. Codification is used for the purpose of identifying the disease (registries, disability tables, insurance tables, etc.), as well as statistics and invoicing. Therefore, Portugal follows, in this field, the general EU and WHO indications.

Certain Medical Services adopt internally, in addition to the mentioned ICD-10, other complementary codification systems. For example, some genetics departments and laboratories also use MIM and Orphacode. Pathology departments also use SNOMED and



ICDO.

So, the Orphacode is not frequently used, and it is expected to integrate ICD-11, but is officially used in the Identification Card of People with Rare Diseases. Orphanet-Portugal has suggested to DGHealth that Orpha codes are used in the NHS (as it is already done in other countries), but this has not yet been implemented.

No special strategies are required to introduce ICD-11, because Portugal follows Europe and the WHO in this field. This classification already includes Orphacodes that will allow the effective recognition of RD. When available (2017?), it will certainly be adopted, after the respective specific training, translation and IT adaptation.

It was acknowledged that as long as all persons affected by RD obtain their Rare Disease ID Card, the Directorate-General of Health will have an automatic registry of patients with RD in Portugal. It should also be emphasized that this RD Card requires the informed consent of the patient for their privacy and protection of their personal and clinical data (and this should be approved by the CNPD, the National Data Protection Authority).

**Proposals:**

- While the ICD-11 is still not applied, implement the use of Orphacode as a complement of ICD-10 in the codification of RD.

2.3 Registries and databases

**Debate:**

Currently, there is no entity responsible for RD registries.

There are various official RD registries, exclusive for RD or that include them, for instance: RENAC - National Registry of Congenital Anomalies, at INSA; the registry of Neuromuscular Patients from the Portuguese Association of Neuromuscular Patients (APN); the National Registry of Amyloidosis (RPP), at INSA; the registry of patients with diseases screened by the National Programme for Early diagnosis (Neonatal screening), at INSA.

There are also "hidden" registries in medical departments, medical laboratories, genetic departments, Patient Associations, social security (for example: patients with disability, disability benefits, support of a third person), etc.

Therefore, there is a growing awareness that a personal effort is being made to register RD and that there are several databases and registries, but they are not standardized or unified, and even if, in general, they try to follow European standards, they do not follow uniform or official rules.

To ensure sustainability, the solution will be to allocate the registries to Institutions within the

National Health Service (NHS), central or peripheral. The participants did not reject the contribution of private entities and companies, provided that the rules are perfectly transparent. The role of patients and associations as enablers of registries should always be considered, ensuring their right to be part of monitoring and even coordination committees/registry curators.

Other options raised were the possibility of associating the registry of rare disease patients with a biorepository of biological samples (biobank); and to include in the Census a question about the existence of a member with a Rare Disease in the household.

It is important to have concrete data on Portugal and to define who is entitled to do so, but also the internationalization and networking of related registries as in the case of the pulmonary hypertension network.

It is also important to emphasize the European work already developed on the registries and publication of good practices of the European Platform for RD Registries (EpiRare), as well as the work carried by the IRDiRC - International Rare Diseases Research Consortium and working groups (which does not yet include Portugal).

The protection and security of personal data and the role of clear, informed and comprehensible consent by the interested parties were also highlighted, since delicate ethical issues are raised<sup>13</sup>.

**Proposals:**

- Encourage Patient Associations to make an effective dissemination of the RD Card to their members and public in general – *DGHealth Regulatory decree nº8, 21-07-2014*, as a starting point for a National Automatic Registry of Rare Diseases.
- Define the host institution for the National Registry of Patients with Rare Disease: INSA, which already hosts other national registries; some other NHS institution; DGH; Infarmed; etc.
- Establish a National Commission / Observatory to monitor and improve the codification and the RD registries after the survey of the current national situation. This committee should be small, with power of decision, national scope and encompass all interested partners.

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<sup>13</sup> *Note from Aliança:* The national legislation requires that any databases or registries that contain genetic information and may enable the identification of at-risk relatives must be kept or supervised by a physician that, whenever possible, should be a medical geneticist. National legislation also stipulates that any biological samples that are identified or identifiable should be coded and the respective codes kept separately, but always at a public institution.

## 2.4 Information on available care for RDs in general, for different audiences

### **Debate:**

It was consensual that enough, reliable, quality and validated channels exist to convey information on RD, although almost all are international initiatives, such as Orphanet, EURORDIS, NORD (the US patient organisation for rare diseases). Orphanet, in particular, has played a vital role in Portugal and for other Portuguese-speaking countries since 2003.

It was agreed that there should be more information translated into Portuguese, as well as more involvement of Patient Associations.

It was agreed that more visibility should exist in the media on Rare Disease Day.

### **Proposals:**

- The Patient Associations will have to play a more relevant role in raising awareness about RD among citizens, in particular in schools and at all education levels.
- Implement the translation into Portuguese of RD information and recommendations and use ORPHANET and EURORDIS for dissemination.

## 2.5 Help Lines

There was not much discussion on the topic, as there are two Help Lines: an effective and general Health Line 24 (808 24 24 24), and a specific Rare Line for RD (300 505 700) run by the “Associação Raríssimas” with state support.

The national team of Orphanet has always been providing information about RD and answering many requests and inquiries, usually by email, coming through Orphanet international, from Aliança or specific Patient Associations, from Portuguese or Portuguese speaking people from many different countries, thus also acting as an informal help line.

It was not deemed useful to have more Help Lines or to elaborate on the subject, since there are no proposals to submit.

## 2.6 Training healthcare professionals to recognise and code RD

Since in Portugal there are coding physicians with specific training, it is expected that ICD-11 will make a difference in codification.

The recognition of RD training is intertwined with academic training in general and was discussed under the following sub-theme.

## 2.7 Training healthcare professionals

### **Debate:**

This topic sparked a lot of interest, since it is widely known that there is not enough educational training, regardless of its level.

The introduction of this theme in the curricula was considered important, since the need for specific training is felt at all levels: from primary school to university, in the specialization of health, education and social service professionals. For example, this theme can be addressed together with the theme on disability/difference, which is already covered since primary school, and adapted to several age groups. This will contribute to learning tolerance, respect and solidarity.

For the younger population, the approach to this theme involves simulated experiences like blindfolding or immobilizing a limb for some time, so as to better understand blindness or motor limitations.

Introduce the theme in secondary education and university, by using European and International initiatives such as the DNA day or the Rare Diseases Day.

The use of IT, such as e-learning and videoconferences could be an important asset.

Greater involvement of Patient Associations is deemed essential in terms of training at all levels, by going directly to schools, as already happens with some dissemination and awareness-raising initiatives, like “Rare: Inform without dramatizing” from the Aliança or other similar initiatives from other associations, or even with the study of life stories, brochures or other. Here, the Learned Societies have a great opportunity to become involved.

The close link with the theme on the Prevention and Genetic Counselling component was considered vital.

Finally, it was emphasized the need for specific training of psychological, ethics and human components, as well as communication techniques, such as learning how to deliver bad news.

### **Proposals:**

- Introduction or allocation of more time and greater relevance of the theme on RD in university and post-graduate education of Health Professionals in general.
- Strengthen the role of Scientific Societies in the overall training of Health Professionals in Rare Diseases.
- Strengthen the role of Patient Associations in the overall training of rare diseases in schools and secondary education and increase the time for the discussion of this theme.
- Provide a greater importance to components on Psychology, Ethics and Communication

Techniques in the education and training of Health Professionals.

**Theme 3 - Research on RD**

**Sub-Themes:**

- 3.1 Mapping of existing research resources, infrastructures and programmes for RDs
- 3.2 Dedicated RD research programmes and governance of RD research funds
- 3.3 Sustainability of research programmes on RD
- 3.4 Needs and priorities for research in the field of RDs
- 3.5 Fostering interest and participation of national laboratories and researchers, patients and patient organisations in RD research projects
- 3.6 RD research infrastructures and registries
- 3.7 EU and international collaboration on research on RD

**Workshop:**

After the opening of the session, participants began to refer to the most important extracts in the terms of Council Recommendations, as well as the situation in Portugal on the implementation of these recommendations.

The discussion focused on questions for which the answers were different from a definite no. These questions raised a lively debate.

There was a clear notion that the lack of implementation of NPRD in Portugal contributed significantly to the less positive aspects of the current situation.

- 3.1 Mapping of existing research resources, infrastructures and programmes for RDs

**Debate:**

With regard to the actions taken or under way, a successful example was given for Huntington Disease. Its American and European Associations created a network where there are computerised registries, shared with other research networks, in order to submit valid projects, thanks to which the research on this disease has progressed quite a lot in the recent years. It is an example that can be followed by other associations and for other diseases.

As a rule, the link between research and the “Centres of Expertise” is centralized by the doctor because of lack of human resources. It is very hard for only one person to deal with all the aspects concerning the patient’s situation and this will lead to a lack of articulation between “Centres of Expertise” and Research.

**Proposals:**

- Assign the intermediary role to Associations, in order to organize the contact between the patient and researchers, eventually with database registry of all disease occurrences of the studied disease.
- Promote the mapping of existing resources by Patient Associations. For example, a group at the Catholic University informs families on home therapies to improve the symptoms in order to disseminate good results.
- Apply what has been established in the 2008 NPRD.

3.2 Dedicated RD research programmes and governance of RD research funds

**Debate:**

With regard to funding, and according to the information obtained from the Foundation for Science and Technology (FCT), 7% of the biomedical research projects funded by this entity are dedicated to RD.

Presently, there are no exclusive research funds for RD. The existing funding cannot support but a minimal portion of all candidates for research projects.

**Proposals:**

- Assign a research quota for RD, rather than an application to the general biomedical research grants from the Foundation for Science and Technology.
- Promote the interconnection between several entities, namely Ministries of Health and Education to enable the joint work of researchers and Centres of Expertise.

3.3 Sustainability of research programmes on RD

**Debate:**

The participants debated if it would be feasible to create a Centre of Expertise for RD in Portugal and which type of activities should it have. It was proposed, for example, the scenario of an incubator for small and medium enterprises.

In particular, in Portugal, there is a lack of technological infrastructures, such as those existent in other countries, with experts to help researchers and associations to apply and be granted, as well as selling projects. Essentially, there is the need of having European projects led by Portuguese researchers.

In the teaching hospitals in Portugal there is clinical research and remarkable Research

Centres, but there is an absence of coordination and centralization in terms of research on RD.

Funding is scarce, there is a lack of long-term sustainability for existing specific research programmes and there are no mechanisms guaranteeing the continuity of research projects on RD.

Another problem identified was the fact that these programmes are only assessed at the end. And still, the participants underlined that there is no difference between monitoring a successful or an unsuccessful initiative. Even if research is successful, sustainability and long-term funding are not ensured in the current model of the Foundation for Science and Technology.

When a clinician submits a project to a scientific entity applying for research funds on RD's, he does not have the possibility of simultaneously requesting time to conduct this research. Thus, there is great difficulty in funding and in conducting research by clinicians and their departments.

There is no research in private hospitals and universities.

**Proposals:**

- Implement the clinical research programme under preparation, where it is intended that the clinician can be freed of clinical duties up to 75% for research.
- Establish a commitment for an objective with defined timeframe. For example, funding for studies proposing to constitute large series of patients at national level, supporting the pluriannual period considered adequate for the work in question.
- For very rare diseases, it is important to properly set up the coordination with other countries where research on these diseases is carried out.
- Constitute INFARMED also as an institution coordinating research, because it has more freedom and versatility for this issue.
- It is necessary to be included in Europe and well-fitted in a centralising structure, not allowing, in principle, the existence of more than one centre conducting the same research. When considering the number of RD in Portugal and its fragmentation, the number of existing cases is always small.

3.4 Needs and priorities for RD research

**Proposals:**

- Consider which are the intervention priorities: Registries? Research? Epidemiology? Clinical Trials? Organisation of services? Health care coverage?

### 3.5 Fostering interest and participation of national laboratories and researchers, patients and patient organisations in RD research projects

#### **Debate:**

With regard to Associations, the difficulties in participation/involvement in research projects are related to several factors, such as:

- Lack of structural and financial capacity and logistic resources to provide support to research.
- Lack of proximity to Research Centres.
- Insufficient information on the progress of research.
- Difficulty communicating results to the families.
- How to involve the families and patients in research.
- Difficulty knowing which patient to recommend.
- Difficulty knowing what information should be given.

#### **Proposals:**

- Promote participatory research, where patients and their families take part in its various stages, suggesting that they play a positive role in contributing and building research agendas that can support researchers.
- Create grants to support students on RD during a shorter or longer period.
- Assign a quota for RD research, rather than applying to the global amount from the Foundation for Science and Technology.
- Put pressure on the awarded grants so that they have substantial funds, in order to allow joint work with international teams.
- Constitute teams, namely from specialized secretariats that work with doctors.
- Create a map or web of what exists in Portugal, so that there is an effective sharing of reciprocity.
- Always ensure the existence of national representation: North, Centre, South, and Autonomous Regions.

### 3.6 RD research infrastructures and registries

#### **Debate:**

There was much talk on Associations promoting registries. But, there was no consensus on whether the registries elaborated by Patient Associations would be the best model.



**Proposals:**

- Map the working groups at national level and keep this data updated.
- Also, map the ongoing research, classifications, etc.
- Create a map or web of what exists in Portugal, so that there is an effective sharing and reciprocity.

3.7 EU and international collaboration on research on RD

**Debate:**

There is presently difficulty in accessing research projects at national and international level.

**Proposals:**

- Check the European priorities and the possibility of integrating these priorities in Portugal.

**Theme 4 – Care for RDs - Centres of Expertise and European Reference Networks for Rare Diseases**

**Sub-Themes:**

- 4.1 Designation and evaluation of CE
- 4.2 Scope and functioning of CEs
- 4.3 Multidisciplinarity, healthcare pathways & continuity of care
- 4.4 Access to information
- 4.5 Research in CEs – How to integrate research on RDs and provision of care
- 4.6 Good practice guidelines
- 4.7 Diagnostic and genetic testing
- 4.8 Screening policies
- 4.9 European and international collaboration – Cross-border healthcare and ERNs (European Reference Networks)
- 4.10 Sustainability of CEs

**Workshop:**

The discussion on these sub-themes was divided into different questions, listed below, followed by a summary and concrete proposals to solve them.

- 4.1 Designation and evaluation of CE

**Question 1:** *What is the reality in Portugal on CE? What is the perception of that reality by different actors: 1. Medical community; 2. Scientific community; 3. Pharmaceutical industry; 4. Patient Associations; 5. Decision making/political power*

**Debate:**

In Portugal, there are no Centres of Expertise (CE) for rare diseases, although there are some centres recognised by the health authorities as “highly differentiated” or which follow specific pathologies, like the Treatment Centres of the National Newborn Screening Programme.

The current centres emerged from the need felt by patients and doctors, with differentiation driven by proactivity, resulting in very asymmetrical conditions for providing health services. Furthermore there are not reference networks for the vast majority of patients.

The foregoing is not due to the lack of legislation, which exists in Portugal, namely: the 2008 National Plan for Rare Diseases; the approval of a document for a National Network of CE for RD by the Ministry of Health in 2011; the appointment of members of the National Committee for Centres of Excellence (order n.º 13163-C/2014 – Official Journal n.º 209/2014, 2<sup>nd</sup> Supplement, 2nd series of 2014-10-29 Ministry of Health – Minister’s Cabinet); the concept, establishment, identification process, approval and recognition of National Centres of Excellence for health care provision, namely for diagnosis and treatment of RD (n.º 194/2014 – Official Journal, (Diário da República) n.º 188/2014, 1 series of 30-09-2014, Ministry of Health)<sup>14</sup>.

It is agreed that there are (from the medical and scientific community to Patient Associations) too many bodies legislating, decree-laws which are not implemented and lack of coordinated action in the field. The implementation of “Centres of Excellence” for Fabry’s disease that did not consider the installed capacity and just before the establishment of a commission for the creation of CE (including the CE for RD), is an example of this.

There is no mapping/registry of rare diseases in Portugal. There was an attempt to elaborate a RD national registry in the past, which was unsuccessful. There is the need for a RD national registry, at the CE and controlled centrally, taking into account the fact there can be a multiplication of patient registries for the need of a second opinion. The question on data protection cannot be overlooked.

The RD card for patients with rare diseases, proposed by the Aliança and claimed by the Patients’s Associations, if properly implemented, can be used as a census, though it does not replace a national registry.

<sup>14</sup> rapporteur’s note: and on the very day of the debate, Order n.º 2129-B/2015, Official Journal, (*Diário da República*), 2<sup>nd</sup> serie—N.º 41— of 27 February 2015, approves the Integrated Strategy for Rare Diseases 2015-2020.

Finally, the major difficulty experienced by the patients was mentioned to be the transition from paediatrics to adult care.

**Proposals:**

- It is necessary to conduct a survey on the number and types of rare disease patients which exist in Portugal, i.e. a national patient registry.
- The designation of pathologies to be included in the Centres of Expertise (CE) should consider the Portuguese reality, namely the number of patients with diagnosis already being followed at Hospital Services and the inherent pathology (for example, familial amyloid polyneuropathy or paramyloidosis)
- Taking into account the installed and proven capacity and the experience “in the field”, the Scientific Societies, Patient Associations and Pharmaceutical Industry can make an important contribution to recognising these groups.
- The CE should have multidisciplinary teams, including paediatricians and adult specialists, in order to make the age transition properly within a given CE.
- The CE can concentrate several poles, clinical and laboratorial research – these would be “affiliated centres”, but not disregarding the implementation of National CE networks for each pathology group.
- The Pharmaceutical Industry should support the CE as partners, namely for the conduction of Clinical Trials.
- The CE should function adequately, based on well-defined objectives and their implementation monitored.

**Question 2: *National Network recognised and with mandatory circuits?***

**Debate:**

There is no national network. It was the unanimous opinion of the participants that a network of CE should be officially recognized by the decision-makers, but also and especially identified as such by the professionals providing the service and by its beneficiaries.

The possible mandatory circuits in the future were not considered a positive point by all patient representatives. In fact, some defended a free choice of the Centres of Expertise based on confidence, and the freedom to obtain a second opinion.

Finally, it was emphasised that some recent decree-laws show lack of adequate technical opinions, apparently because the caregivers and patients were not consulted.

**Proposals:**

- The CE network should be based on the NHS, with possible partnerships with the health services and diagnostic laboratories, public or private, and defined on a case-by-case

basis. The Patient Associations must be partners in the decision process

- The CE need to be implemented according to the quality criteria already well defined by European Commission RD Expert Group, with specialised technical (not honorific) committees.

**Question 3:** *What is the real link with the European networks? Reality or myth?*

**Debate:**

The link with European Centres of Expertise was conducted upon the individual initiative of clinicians and researchers, and it can be said that it formally does not exist.

As for Patient Associations, few have close links with their European and even North-American Counterparts. Successful examples are the Association of Pulmonary Hypertension and the European Huntington Disease Network.

**Proposals:**

- In Portugal, the CE should follow the criteria defined by the European Commission RD Expert Group (previously named EUCERD), so that in the future they can integrate the European Networks.
- Patient Associations should play a more active role, namely, through contacts and partnerships with their European Counterparts.

**Question 4:** *Would it make sense in the Portuguese reality to create/implement these High Differentiation Centres, by sacrificing quantity over quality and linking them to Treatment Centres, in a greater medical proximity? And, simultaneously linking this network with European ones? What kind of role do the Patient Associations have or should have in the designation and creation of CE? How to promote an effective dissemination of a National Directory of CE?*

**Debate:**

That would make sense, but not forgetting that the Treatment Centres (TC) should be articulated with the Centres of Expertise and should not be mere centres for administering medicines or other treatments. Additionally, we cannot privilege proximity over quality.

The Patient Associations have a notion of the vital work carried out by the specialised health professionals, but there are also social integration determinants which go beyond clinical care. On the other hand, it is important for clinicians to recognise the role of the Associations and to be their close partners.

A National Directory of Centres of Expertise, when they exist formerly, will have to be disseminated. And the way to do so could be a web page, inspired from the way Orphanet displays information. However, Orphanet has a different role, namely, self-referencing and

sources with diverse information.

**Proposals:**

- The CE should have multidisciplinary teams to carry out the evaluation and link with the social and educational supports which patients with rare disease require.
- The CE should have the ability of providing telemedicine or remote support.
- The CE should send professionals and portable means to other centres, whenever feasible, to support patients and local professionals.
- The CE should contribute to informing about of Associations of patients.
- The Associations should be represented in the CE related to the diseases they represent, contributing to the evaluation of results, namely through satisfaction surveys.
- We could use the Ophanet structure and adaptations to elaborate a national directory of CE.
- The dissemination of the national directory of CE, as well as RD in general, should be done beyond the Health Services, on the internet, mass media, Social Security Services and in schools. Associations should also contribute to the dissemination of the directory with their members and the civil society

**Question 5:** *In defining the CE, is there an absolute need for the concept of multidisciplinary or to link resources/specialties. Does it make sense in our reality, to have CE with a pathology or group of pathologies? Does it make sense for these units to be autonomous? Is there a real evaluation of CE in Portugal? Which are the quality indicators used?*

**Debate:**

The CE should incorporate pathologies or related group of pathologies in view of the estimated number of patients and in coordination with the European Networks. Given the limited number of European Networks, even though there are 6000 to 8000 rare diseases, it makes no sense fragmenting the CE.

As for autonomous funding, it was not possible to reach a consensus.

Considering the absence of formally recognised CE, no evaluation exists either.

**Proposals:**

- Funding, autonomous or not, has to be adjusted to the objectives and their earmarking and follow transparent rules. These objectives of the CE should be well-defined.
- The official nomination of CE should follow closely the centres specialised in RD that are already recognised (seen as such) by other healthcare professionals, Patient Associations, the industry and other stakeholders in the field of a RD or group of RDs.
- The official establishment and evaluation process of the CE should be strictly based on the Recommendations of the European Commission RD Expert Groups (formerly

EUCERD) as quality criteria for CEs.

#### 4.2 Scope and functioning of CEs

**Question 1:** *The definition of CEs in Portugal is in accordance with the European Commission RD Expert Group recommendations relating with the Criteria of Quality<sup>15</sup>?*

**Debate:**

As widely highlighted, there are no CEs officially recognised in Portugal.

**Proposals:**

Already mentioned above.

**Question 2:** *What kind of collaboration with patients and respective associations is established in CE? These agents play an active role in the management and decision-making process of these CE? How is the level of competence of each CE measured? Is there a real sharing of human resources and competences at National and International level? How can this aspect be approved?*

**Debate:**

Since there are no officially recognised CE, no formal collaboration exists with patients, which therefore have no active role in management. In the same way, there is no evaluation.

The sharing which exists is occasional and results from individual initiative, as was previously identified.

**Proposals:**

- The establishment of a network of national CE, integrated in the respective European Networks, and according to the European Commission RD Expert Group standards would answer these questions.

#### 4.3 Multidisciplinarity, healthcare pathways & continuity of care

**Question:** *Are the CE in Portugal based on a multidisciplinary approach? Do they have the ability to congregate or coordinate, “in the field of specialized health care, competences/multidisciplinary skills, including *paramedical and social service skills that meet medical and rehabilitation needs and specific palliative measures of patients with rare diseases*”? (European Commission RD Expert Groups Recommendations). Are the CE linked to*

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<sup>15</sup> The audience was reminded that the “CE are expert structures for the management and care of patients with RD, with determined coverage, national preference, and if necessary, international. The CE address pathologies, sometimes very difficult to diagnose, and which require specific care, planning of treatment/intervention and prevention of complication plans.”

*specialized laboratories and other units?*

**Debate:**

In general, the existing (non-official) centres comply with that list of criteria, but there is usually no national networking.

**Proposals:**

The ones already mentioned.

4.4 Access to information

The establishment of CR should include these issues.

4.5 Research in CEs – How to integrate research on RDs and provision of care (How to integrate research in RD and the provision of treatment)

The establishment of CR should include these issues.

4.6 Good practice guidelines

This sub-theme was not discussed.

4.7 Diagnostic and genetic testing

**Question 1:** *In parallel and in close partnership with the CE, there is the need to compile an inventory of clinical laboratories to confirm the RD diagnosis? Which decisions should be taken in terms of best clinical-laboratorial link? In terms of diagnosis, it is equally important to know which tests are legal/allowed? Who defines these criteria?*

**Debate:**

A list of accredited diagnostic laboratories is indispensable. There are only two medical genetics laboratories accredited in Portugal by IPAC, following ISO 15189 (at INSA, Lisbon, and CGPP-IBMC, Porto). The recognition by accreditation of diagnostic labs and its registry was done jointly by EuroGentest (their quality database now taken by the ESHG Quality Committee) and Orphanet. That will avoid the duplication of specific laboratorial and senseless waste of resources.

The implementation of CE will automatically resolve the clinical-laboratorial link, which will determine the needs of the centres, and should have in view who and what already works well in the field.

The European Reference Network of CE will have an underlying diagnostic laboratory network.

Only confirmed quality tests should be requested and according to clinical indication, following the recommendations of good practices.

**Proposals:**

- The list of clinical laboratories to confirm the diagnosis of Rare Diseases should be included in the National Directory of RD or be attached to it.
- Diagnostic tests should have confirmed quality and ideally be accredited
- The CE should include or be articulated with clinical laboratories that are accredited and/or are recognised as the reference in the field.
- It is necessary to take into account accessibility and access equity.

**Question 2:** *What arrangements do exist to enable the travelling of biological samples, radiological images as well as other diagnostic materials? At the national and European level? What sort of reimbursement agreements and policies do support these exchanges? How to best integrate the EuroGenTest recommendations on genetic counselling into national practices?*

**Debate:**

There are no specific measures. The mentioned travelling occurs only from individual initiative. Application is made to DGH, who decides if a sample for diagnosis or a patient for examination or treatment should travel to another country, but only if those services are not available at the national level. The Orphanet national directory of specialised centres and diagnostic labs is supposedly used by DGH to make that decision.

The Portuguese legislation highly recommends and entitles all persons with RD of genetic origin (the vast majority) to genetic counselling. In Portugal, there are already officially recognised clinical geneticists and genetic counselling professionals.

**Proposals:**

- The exchanges with other CE abroad will be reinforced in the national and European Networks.
- Guidelines for that are provided in the quality criteria of European Commission RD Expert Group.
- Medical geneticists should always be part of the multidisciplinary teams of CE.

4.8 Screening policies

**Question:** *The population screening programme especially the new-born screening (NBS = Newborn Screening) – is it adequate / enough in the present state of science? What measures*



*could be put into place to improve its performance and effective coverage of the population? Should or can the diagnosis/genetic screening replace biochemical evaluation?*

**Debate:**

Neonatal Screening (NS) has a close to 100% coverage. It is currently based on biochemical parameters only. The diseases included/to include were the subject of a debate but without reaching a conclusion.

The Patient Associations present had no opinion on neonatal screening (NS).

The clinical screening was deemed insufficient, decades have elapsed until obtaining a diagnosis.

The genetic screening should not replace biochemical evaluation when it is guiding. However, it is lawful to perform genetic screening in the presence of clinical manifestations of unknown cause.

**Proposals:**

- The neonatal screening (NS) should be monitored.
- It is imperative to implement the CE, allowing to strengthen knowledge and provision of laboratory diagnostic tests, which may be biochemical and/or genetic in view of the state of the art of each situation.

4.9 European and international collaboration – Cross-border healthcare and ERNs (European Reference Networks)

**Debate:**

The measures needed to comply with the European Directive on cross-border healthcare should not be implemented before establishing the national network of CE.

No specific measures are being taken in preparing for the participation in European Reference Networks (ERNs) or to exchange information between CE and healthcare givers. Nor is there a structure to promote and support the common designation of CE that may be associated to the future ERNs.

According to Patient Associations, it is important to have arrangements (both at the national and at the European level) that facilitate the establishment of and integration in ERN. Healthcare should be European, so that the patient can be transferred to a European clinic in accordance with established agreements.

Patient Associations should be partners of their European (and American) counterparts, so as to achieve synergies.

**Proposals:**

- The need for a European Health Care Network will be resolved with the implementation of a national network of CE in conjunction with the European networks of CE.

4.10 Sustainability of CEs

- **Question 1:** *What mechanisms do ensure that CEs are established and operate at national level with a sustainable plan? How are activities performed by the CEs, but not strictly related to patient treatment (e.g clinical research, production of guidelines for diagnosis and care, in-depth clinical and biological investigations, coordination of international networks, etc)? How to make the best use of Structural Funds in the forthcoming period 2014-2020? Will they be a mirage? Is there scope for investments in rare disease CEs in the national strategic reference frameworks for Structural Funds?*

**Debate:**

There are no officially recognized CE, as mentioned above.

Activities, in addition to financial assistance from the State, could be funded by clinical trials, patronage from Patient Associations or others.

There was a national sense of European Funds being underused, perhaps partly because of lack of structure to support applications for funding.

**Proposals:**

- The CE sustainability will involve its creation in the NHS, according to the quality criteria of European Commission RD Expert Group.
- A structure to define, certify and supervise the CE (in terms of assistance, training and research) needs to be established.
- This structure should be public, centralized and have well-defined composition and transparent operating rules.
- It should incorporate representatives of healthcare professionals, researchers, Patient Associations, pharmaceutical industry, in addition to a core group of collaborators, designated by the MH/DGH, based on technical competence.
- It should include also a professional office to support community funding.

## Theme 5 – Orphan Medicinal Products

### Sub-Themes:

- 5.1 Support to Orphan Medicinal Products (OMP) development
- 5.2 Access to treatments
- 5.3 Compassionate use programmes
- 5.4 Off label use of medicinal products
- 5.5 Pharmacovigilance

### Workshop:

The discussion began before the systematic approach to the sub-themes, during the presentation of the Council Recommendations on RD.

#### **COUNCIL RECOMMENDATION V (e) ON RD:**

“The sharing Member States’ assessment reports on the therapeutic or clinical added value of Orphan Medicinal Products at Community level where the relevant knowledge and expertise is gathered, in order to minimise delays in access to Orphan Medicinal Products for rare disease patients (Clinical Value and Therapeutic Added Value).”

#### **Debate:**

Since 2006, the medical products that have medical use must undergo a prior approval and then a contract is signed between Infarmed and the industry’s representatives. This contract is reassessed every 2 years.

In Portugal, the added value of a new drug is highly regarded (valued) in relation to another one that already exists with the same application and economic value.

#### **COUNCIL RECOMMENDATION Nº15:**

“Include, in their plans or strategies, the necessary conditions for the diffusion and mobility of expertise and knowledge in order to facilitate the treatment of patients in their proximity.”

#### **Debate:**

As basic indicator, use the number of Orphan Medicinal Products (OMP’s) with an EU marketing authorisation, and the number of existing OMPs available in the country, priced and reimbursed.

Comparing the reimbursement of OMPs in Portugal and in other EU countries, in Portugal,

amongst all approved medicines, there are those for hospital use, and among them OMPs.

**COUNCIL RECOMMENDATION Nº16:**

“Encourage centres of expertise to be based on a multidisciplinary approach to care when addressing rare diseases.”

**Debate:**

As for the compassionate use of orphan medicinal products, in Portugal, is there a system that provides medicines to patients whose distribution hasn't yet been authorised?

In Portugal, compassionate use requires the doctor to ask for a special use authorisation usually granted by Infarmed. There are 2 types of authorisations: one scientific and the other economic. It doesn't seem difficult provided there is a medical recommendation.

An example was mentioned about an orphan drug which did not exist in Portugal, whose formula was obtained in France, and later prepared in Portugal. It represents the case of a described, recommended drug, ready to be used but not available in Portugal. Infarmed authorised its preparation and use. The entire process took 15 days.

**Proposals:**

- As in some countries, like France and Italy, establish laws for compassionate use of Orphan Medicinal Products which allow patients to access new medicines before formal approval, as this delays the use of OMP.

5.1 Support to Orphan Medicinal Products (OMP) development

**Question 1:** *What kind of support is given to small and medium sized enterprises for the development of Orphan Medicinal Products (OMP)?*

**Debate:**

In Portugal, the authorisation of a wide range of benefits related to patent exemption, tax exemption, etc., so as to foster the development of OMP, is centralised through the European Medicines Agency (EMA).

The EMA has an important support programme. We should start considering the existence of any type of specific support in Portugal, because there are already small enterprises which help developing new OMP.

**Proposals:**

- To have a scientific body which provides support in addition to funding, such as IPAMEI or COTEC.

- There should be scientific counselling in terms of the Informed support that already existed in the past. It is up to the companies to put pressure to continue with this support.

**Question 2:** *Are there specific programmes that support the development of ODs?*

**Debate:**

There are specific programmes, in accordance to EMA that should exist in any EU country. If a *start-up* wants to obtain national incentives for an orphan drug, it has to submit an application to the EMA. There is no specific national programme supporting companies dedicated to the development of Orphan Medicinal Products .

**Proposals:**

- Create a specific programme to promote the development of OMPs.
- Promote the inclusion of OMPs in the innovation programme for which there is plenty of money in the EU.
- Not to give up from submitting new development projects.

## 5.2 Access to treatments

**Debate:**

It is essential to understand how to improve and accelerate the procedures and definition of reimbursement, in order to minimise the delays and speed up the support to Orphan Medicinal Products.

One of the difficulties is the economic decision to use drugs with authorization (not only for OMPs). This decision was once centralised but, a few years ago, it became the hospital's responsibility, which caused a problem of equity, namely between central and local hospitals.

Therefore, the problem of patient access to OMPs is an economic one and not Informed's authorisation. It should also be noted that the value attributed to Orphan Medicinal Products is not coordinated nationally.

The question was raised on the existence of a committee to decide on the access to Orphan Medicinal Products, debating whether it would make more sense to have a committee for each pathology, or a central body. Indeed, they are very specific diseases. A pool of experts could be created, supported by a small administrative organisation, with a special function to address special and rare issues. Time can be crucial and it is essential to streamline the access to Orphan Medicinal Products. Thus, the management of the patient expectations would be contemplated, in addition to scientific ones.

Eventually, availability and access could be managed from the Centre of Expertise.

Nevertheless, the budget should always be at a national level to guarantee equity.

The question was raised about Portugal being aware or not of the European Recommendations from European Commission RD Expert Group on the development of Orphan Medicinal Products, in particular on the clinical added value of orphan medicinal products (CAVOMP – Clinic Added Value of Orphan Medicinal Products)? And if it was willing to exchange information with other member states and European Medicine Authorities? How would Portugal participate in these efforts?

This appears to be an early discussion. It is necessary to compile evidence and obtain a study plan for an assessment of relative effectiveness.

Besides, the participants did not know whether Portugal had participated in the MoCA (Mechanism of Coordinated Access to orphan medicinal products). MoCA seeks collaborative ways to identify and assess the added value of orphan medicinal products: Under the "Access to Medicines in Europe" platform, EU Member States, European Economic Area countries, and relevant stakeholders - including patient organisations - were invited to become involved in a process for the concept of a coordinated access to orphan medicinal products based on the creation of programmes between companies and groups of competent authorities, and on a mechanism for the assessment of clinical added value of orphan medicinal products. Stakeholders joined this MoCA process on a voluntary basis.

**Proposals:**

- There should be a centralised fund for authorisation of medicinal products that require it, a centralised economic decision for these cases, in order to avoid inequalities between hospitals and regions.
- The medical teams should put pressure so the patients can have access to medicines.
- The value attributed to the OMPs should be coordinated at national level.
- A time limit should be established for approval of OMPs.
- Greater involvement of the Patient Associations is needed.
- There is plenty of information on RD, but we need to deepen the reliable and competent sites and improve them with the doctors and Umbrella Patient Association.
- Improve the dynamic flow of information between doctors, patients and associations.
- Information on the treatment of RD should be disseminated more effectively.

5.3 Compassionate use programmes

**Debate:**

It was discussed how to promote access to compassionate use programmes, what is the best way to inform professionals, patients and Associations of the opportunities which exist at this level and how to adopt compassionate use.

Compassionate use is a treatment option for a patient suffering from a disease for which no satisfactory authorized alternative therapy exists and/or who cannot enter a clinical trial. There are specific compassionate use programmes in some countries of Europe.

In Portugal, the framework is the following: when a disease is diagnosed, a medical centre conducting the treatment is contacted. Then, the family is informed that there is a clinical trial for the patient's pathology and is given the medical team and centre's contact information. From there, the patient's family, and not the doctor, contacts the team. The doctor's role is just to inform the patient that he/she can be integrated in a clinical trial.

In Portugal, compassionate use is free of charge for patients that are included in the clinical trial and, when they leave the trial, they can continue treatment until there is access to medicinal products in the country. In practice, compassionate use is the only free of charge treatment using a drug that has not yet been approved in Portugal.

So, compassionate use is post-clinical trial, implicit and only for patients registered at a national centre.

#### 5.4 Off label use of medicinal products

##### **Debate:**

The *off-label* use consists of a drug being used for a given disease, though it has been authorised for another use/indication.

In Portugal, the off-label use of medicinal products from the start cannot be reimbursed, even if they can benefit patients.

Improving and simplifying the use of medicinal products for reimbursement in an off-label situation is essential.

If there is a medical report concerning a specific situation, Infarmed will assess it with the hospital and there may be a positive decision in accordance with the Portuguese legislation.

Currently, this use depends on the Hospital's financial condition to approve or refuse the delivery of the medical product. Therefore, it has to be a doctor from the hospital where the patient is treated to make the request and give the approval for the financing of a particular product, since the hospital is paying and therefore it has always to approve, regardless of an expert's second opinion. For Infarmed to assess the application, it is essential to have the Hospital's approval, if not the process is considered incomplete and therefore not assessed.

##### **Proposals:**

- Create very solid concepts in Infarmed, rather than creating specific groups, which stimulate the industry and facilitate the off-label use of medicinal products.

## 5.5 Pharmacovigilance

### Debate:

Often, the risk of these medicinal products is superior to other drugs, because they are sometimes used without having been adequately tested and the risk can be great.

Regarding the compassionate use and *off-label* use, Portugal is doing well because the pharmacovigilance system is centred in Infarmed with offices in the north, centre and south of the country. Any person (either clinician, patient, relative or caregiver) can report an adverse reaction of any drug, in particular an *off-label* medicinal product or compassionate use.

An orphan drug leads to a commitment by the pharmaceutical company to implement an organisational study to further study the disease, by taking notes of all events during treatment.

When you are in a situation where off-label use is not occasional, you can request a special authorisation, but to go beyond and create a specific committee might not be effective.

### Proposals:

- In terms of authorisations, it is more logical to be aligned with Europe, as the European structures are already in place, thought and experienced. If the request is made directly at European level, the authorisation will be faster.

## Theme 6 –Social Services for Rare Diseases

### Sub-Themes

- 6.1 Social resources for people with disabilities
- 6.2 Specialised social services for rare diseases
- 6.3 Policies to integrate people living with rare diseases into daily life
- 6.4 International–supranational dimension

### Workshop:

The discussion on these sub-themes was divided into different questions, listed below, followed by a summary and concrete proposals to solve them.

- 6.1 Social resources for people with disabilities

**Question 1:** *Considering the national schemes supporting people with disabilities, the way disability is detected and assessed so to trigger the entitlement to measures of public support;*



*how do rare diseases “perform”? How visible are they?*

**Debate:**

Rare Diseases are not sufficiently represented in the context of national mechanisms to support people with disabilities and their visibility is thus limited. For example, the framework that RD may have within the concept of disability is not clearly defined.

The reduced visibility of RD is due to several factors: Lack of specific legislation on social support for RD; lack or incomplete information on RD and social support; patients, health professionals and decision makers not properly informed, leading to increased difficulties in the access to medical diagnosis; reduced ability of association and collective mobilisation; shortage of biomedical references.

The reduced representation and visibility of RD can be mitigated by strengthening the role of Associations and creating measures to raise awareness among patients of the need to join them.

An increase in the number of members could mean an improvement in the representativeness of Associations. However, the Patient Association representatives recognised the enormous difficulty in having access to patients, since there is no way of knowing who is diagnosed.

Additionally, Patient Associations are also confronted with difficulties in attracting patients to increase the number of their members. Sometimes, patients seek compensations/offsets, which cannot be guaranteed by Patient Associations.

It is necessary to find strategies that ensure the appropriate visibility of caregivers in order to raise awareness of policy makers, employers, and the society in general, of their crucial role in caring for patients on a daily basis.

It is essential to improve the access to information on RD. It is also necessary to define a clear and adequate legislative framework for RD.

**Proposals:**

- Mobilisation of Patient Associations in order to have greater representativeness and, therefore, to be heard and gain more visibility in the public sphere and with policy makers.
- Investment in training health professionals on RD, in order to enable equal access to an effective, fast and affordable medical diagnosis.
- Creation of multidisciplinary teams, by involving health professionals, social workers and psychologists that together can define short, medium and long term strategies, allowing access to social supports, health care and the proper conditions of social, educational and professional integration.
- Creation of the figure of “case manager(s)” for complex cases, who in practice may be

a single individual, but preferably a multidisciplinary body that should/could provide support to patients and their families.

**Question 2:** *What mechanisms effectively support the allocation of social security benefits to people living with rare diseases?*

**Debate:**

The patients feel little support in social terms, most of them having to rely exclusively on family members.

Participants recognised the caregivers' heavy workload, and that there are no specific, social benefits for them, as well as no official recognition of their role.

Patients have difficulty in accessing the information, noticing also a lack of knowledge among social workers, health professionals and educators, with regard to social security benefits for people living with rare diseases.

In addition, many of the existing benefits were suppressed or diminished in recent years, as a result of the financial crisis in the country. These benefit cuts affected in particular the school system, today witnessing, namely, a huge decrease in human resources supporting Special Education.

For these reasons, there are specific mechanisms allocating social benefits to people with Rare Diseases. The general support for people with disabilities is not always adjusted to particular and specific situations of people with rare diseases. The social benefits that exist for people with disabilities have been decreasing in recent years.

**Proposals:**

- Create mechanisms for integration and greater interaction of social workers with Patient Associations in order to facilitate the access to social benefits.
- Recognition of the “case manager(s)” (single individual or multidisciplinary body), which should support the patient and family, and/or a personal assistant. These case managers should receive specific training, aimed at the needs of people with rare diseases.
- Patients with Rare Diseases should have an easier and faster access to the RD Cards (the RD card is already available, but most patients and physicians are unaware of its existence and how they can obtain/deliver it, and its distribution has been residual).

**Question 3:** *Which national security social schemes exist to support families and patients with disabilities? How are the social resources distributed at national level distributed? Is there an official directory of social resources for people with disability?*

**Debate:**

There are difficulties in locating/identifying the information on social resources for families and people with disabilities, as well as to the effective access to these resources.

Patients who live in more isolated and distant locations from large urban centres experience greater difficulty in accessing information as well as obstacles to integrate Patient Associations.

Indeed, large geographic zones of the country and smaller towns indicate a lack of knowledge on the social support mechanisms for people with rare diseases.

Therefore, we are looking at an unequal geographic distribution of access to existing social resources due to a greater lack of information and access to health care and public services, mainly in interior and rural areas. There is official information on these resources, but no specific information aimed at RD.

**Proposals:**

- Creation of an updated portal providing direct and specific information about RD.
- Reduce the inequalities in access to information. More than accessing information, it is important to know how the information reaches the patients and families. Social workers and other specialised professionals (for example, family doctors) should be sensitised to meet the needs of the patients and their families, in order to facilitate the access to information on social support mechanisms.

6.2 Specialised social services for rare diseases

**Question 1:** *What actions of the national plan or strategy on RDs are or will be aimed at “guiding and structuring relevant actions in the field of rare diseases within the framework of ...the social systems”? (Council Recommendation)*

**Debate:**

The existing actions for RD are limited in scope, and characterised by the fragmentation and unequal distribution of resources and access in geographic terms.

It is essential to make information on RD accessible to all patients and their families, by creating specific support mechanisms for people with rare diseases, investing in training several stakeholders (in the clinical, therapeutic, educational and psychological support to patients and their families in the field of social services).

**Question 2/5:** *Is there any specific information path that RD patients could use to find their way through existing legislation and schemes?*

The topics discussed and suggestions included those already mentioned for the first sub-

theme, item 3.

In addition, participants were unanimous to the fact that there were no specific information channels on legislation and social benefits for people with rare diseases. The existing information is dispersed and there is no specific legislation for RD.

The difficulties in accessing information were also mentioned by health professionals, social workers and other specialised professionals.

**Proposals:**

- Disseminate Orphanet as the portal for RD, with credible and scientifically validated information about RD, specialised centres, diagnostic laboratories, tests, patients associations, research projects and clinical trials, OMPS.
- Creation of a portal with organised and updated information, written in a clear and simple language, in other words, user friendly and adjusted to a lay public.

**Question 3:** *How are specialised social services for rare diseases funded? Is there a specific fund to support the long-term sustainability of such measures?*

**Debate:**

The participants noted the lack of specific funding in the field of RD, and of funds ensuring sustainability of any measures in this field, the discussion on the topic did not last.

**Proposals:**

- Creation of specific funding in the field of social services for people with rare diseases and definition of strategies allowing long-term sustainability.

**Question 4:** *How are specialised social services for people living with RDs evaluated? What quality systems are adopted or guidelines followed to ensure an adequate level of service provision?*

**Debate:**

The participants unanimously agreed that there is a high fragmentation of assessments on the quality of social services provided to people with rare diseases. Such quality assessments are fragmented, occasional, dispersed and not comprehensive. There is a need to define general guidelines adjusted to the specificity of RD.

There is no link between structures, for example, between the Ministry of Health and the Ministry of Education. In general, even within the same hospital, there is no interaction between the various services and specialists.

There is no centralized database circulating patient information between different specialists and structures. After the transfer from paediatrics to adult care, patients are told they have to go to another hospital (often districtal, not central), where many times doctors have never heard of RD.

**Proposals:**

- Introduce survey questionnaires on patient needs and treatment evaluation. The same can be used by Patient Associations.
- Define measures allowing transfer and transition from paediatrics to adult care, in order to ensure permanence and quality care.
- Avoid fragmentation, by creating a manager of various specialties, such a collegial body that integrates the perspective of several people.

**Question 6:** *What of the following measures do exist and which ones need to be fostered? (examples from EUROPLAN Recos): educational support for patients, relatives and caregivers; individual support at school at different schooling levels, for both pupils with RD and teachers, including disease-specific good practices; promotional activities to foster higher education for RD patients; support mechanisms to enter and stay in school and participate in work life for people with disabilities.*

**Debate:**

The EUROPLAN recommendations are not implemented.

The EuroGentest patient leaflets have all been translated into Portuguese and are easily available (both in html and printable pdf files) from the EuroGentest, ESHG and Orphanet websites. In addition, some Patient Associations and some genetics services have developed their own educational materials, sometimes very specific to a given disease or group of diseases, that also need to be centralised and better diffused.

It was unanimously said that the existing resources are scarce in terms of educational support to patients, relatives and caregivers, stressing several difficulties in terms of staying in school and integration in work life.

Several participants stressed that the rights of people with RD, namely children and young adults in schools, are regressing (for example, access to speech or physical therapy).

**Proposals:**

- The implementation of the EUROPLAN Recommendations should be accompanied by strengthening the training of clinicians, caregivers, educators and teachers and human resources in schools supporting Special Education
- It is necessary to empower the people with RD towards social integration.

**Question 7:** *What role do Centres of Expertise have in developing or facilitating specialised social services aimed to improve the quality of life of people living with a rare disease?*

**Debate:**

All participants acknowledged the potential importance of Centres of Expertise for that role.

It is important to create Centres of Expertise with multidisciplinary teams, not only aimed at the medical and therapeutic aspects, but also to establish the link between the social, clinical and psychological needs of patients.

The Centres of Expertise could perform early diagnosis and predict better the prognosis and outcome for the future of the patients (for example through annual screening).

**Proposals:**

- The creation of multidisciplinary teams, aggregated to the Centres of Expertise, where there should be case managers.
- The organisation of Centres of Expertise, taking into consideration the geographic areas may create more comprehensive areas of expertise than the Centres of Expertise foreseen in the National Plan.

**Question 8:** *How are existing social resources for people living with RD mapped at national level? Is there an official directory of social resources specifically for people with RDs?*

The topics and suggestions overlap with the first subtheme, question 3, thus are not being repeated here.

**Question 9:** *What national schemes do exist that promote access of people living with RD and their families to: Respite Care Services; Therapeutic Recreational Programmes; Adapted housing; Resource Centres*

**Debate:**

No such types of supports were identified.

The participants highlighted the fact that accessibility to public buildings, like schools and hospitals or health centres, were often not adjusted to patient needs.

The participants working in the field of social and therapeutical support, as well as patients and their families, stressed the absence of life-long and continuous care, emphasising that this is a particularly serious problem, especially when the patient has elderly parents.

Also, the delay in accessing basic supporting products was underlined. For example, there are

patients waiting one year for a wheelchair.

**Proposals:**

- Complete removal of architectural barriers in public spaces.
- Creation of continuous care centres.
- The State should be accountable for access to social resources, breaking the dominant trend of relegating that responsibility solely on the family or channelling it to the individual sphere

**Question 10:** *What level and sources of information do RD patients have on existing social resources?*

The topics and suggestions discussed overlap with the first sub-theme, question 3, thus are not repeated here.

6.3 Policies to integrate people living with rare diseases into daily life

**Question 1:** *What of the following measures do exist and which ones need to be fostered: educational support for patients, relatives and caregivers; individual support at school at different schooling level, for both pupils with RD and teachers, including disease-specific good practices; promotional activities to foster higher education for RD patients; supporting mechanisms to enter and stay in school and participate in work life for people with disabilities*

**Debate:**

The EUROPLAN recommendations are not implemented and should be so quickly.

**Proposals:**

- The implementation of the EUROPLAN recommendations should be accompanied by strengthening the training of clinicians, caregivers, educators, and teachers and human resources in schools supporting Special Education.
- We must give voice to the patient, also in training.

**Question 2:** *Please discuss about patient-centred measures on individual intervention plans or “Complex Case Managers.”*

**Debate:**

The participants considered the creation of individual intervention plans to be very important, since they are currently non-existent and to associate the figure of “complex case manager”.

The complex case manager may be a collegial body congregating different specialties in the

clinical, therapeutics, psychological, social and family pathway of the patient.

**Proposals:**

- The complex case manager could emerge from setting up multidisciplinary teams related to the creation of Centres of Expertise.

**Question 3:** *“Complex Case Managers”, in the French National Plan are defined as the element that can ensure a better coordination in the care pathway of RD patients, functioning as a link between the medical and the social needs of the patient. What is the profile/competences? What is the institutional framework?*

The topics and suggestions discussed overlap with the first sub-theme, question 3, therefore are not repeated here.

**Question 4:** *What national schemes do exist that promote the educational support for patients, relatives and caregivers?*

**Debate:**

The participants are unaware of the existence of schemes at national level aimed at promoting educational support for patients, relatives and caregivers, so it is assumed that they do not exist.

**Proposals:**

- Create schemes at national level to promote educational support for patients, relatives and caregivers.
- Create the position of personal assistant to remove the physical and psychological burden of the caregiver.

**Question 5:** *In addition to support in school and work life, what support is provided to improve accessibility, in particular to public services?*

**Debate:**

The participants mentioned the lack or absence of support in school and work life, and they also referred to the absence of other support to improve the access to public services.

**Proposals:**

- Create the mechanisms of support to patients at school and in work life.
- Strive towards support to improving the access to public services.



#### 6.4 International–supranational dimension

**Question 1:** *What social guidelines can be shared based on the experiences in some European countries?*

This question was not discussed, partly due to lack of knowledge on the experience of other countries.

**Question 2:** *How existing tools can be best disseminated, validated?*

The topics and suggestions overlap with the first sub-theme, question 3, thus are not repeated here.

**Question3:** *How to raise awareness of the existence of such tools?*

##### **Debate:**

The participants considered that, currently, there is usually access to social benefits (or information about them) only when the patient is institutionalised.

The clinicians, social workers, teachers and schools, as well as Patient Associations, can play a key role in raising awareness of the existing resources and tools. But the social resource mechanisms are virtually non-existent. The solution would be to create biomedical references and also collect information and make it available, as well as standardise, update and centralize it.

The Associations can only apply to project funds if they are Private Social Solidarity Institutions (IPSS). The constitution of an Association as an IPSS is a demanding procedure in bureaucratic terms and incompatible with the fragile structure of most Patient Associations.

##### **Proposals:**

- Strengthen the training of clinicians, not only in terms of biomedical, but also the social dimension.
- Aliança can serve to foster information and experience-sharing among Patient Associations.

##### ***Transversal to the theme***

Emphasizing the analysis and assessment of social services and RD:

##### **Debate:**

Absence of specific resources in this field: The social mechanisms supporting people with RD

are practically non-existent.

**Difficulties to access information:** The RD invisibility means that there are deep gaps in terms of biomedical references, and that the access to information is difficult. The information which exists is dispersed and its dissemination more difficult in regions far away from the large urban centres.

**Persistence of social inequalities:** The responsibility of providing health care and supervision to a person with a rare disease relies almost exclusively on the family. Still with regard to inequalities, there has been a worrying situation of shrinking social resources in schools, namely Special education. The participants mentioned the experiences of children and young people with special education needs who have been diagnosed but do not have support (it is denied), or, simply do not go to school. Many are apparently integrated, since they do not have a Special Education teacher or teaching assistant. The economic crisis reduced the rights of children and young people with RD at school, despite disseminating, in official terms, the idea of an inclusive School.

**Proposals:**

- The solution, in general terms, involved creating specialized collective entities that can intercede before decision bodies, to legislate and implement specific social resources for RD.
- The State should collect specific data on social resources for people with RD, standardise, update and centralise it.
- The State should assume its social responsibilities to the welfare of patients and families with RD.
- The social benefits for these patients and families with RD should be assumed as a public mission, in favour of equity.

**Additional Workshops**

No additional Workshops were held.

**Report of the Closing Session Conclusions**

The closing session was chaired by Glória Isidro, from INSA.

The moderators/rapporteur of each theme referred to the main conclusions and practical proposals, emphasizing the more relevant ones for each theme:

**Theme 1:**

- There has to be a committee on RD selected in a transparent manner, including all stakeholders, namely doctors, caregivers, patients and Associations.
- Mapping patient needs should be carried out. Associations can conduct this with the

support of research grants and people from the academia in order to direct the Associations in a valid way.

- It is essential to disseminate data on RD, in particular, to health professionals (mainly general practitioners and family doctors).
- We need to ensure curricular changes in the various school subjects on health, including information on genetic and rare diseases. It is also important to conduct training sessions within Higher Education Institutes of Health and secondary schools, namely for students who will take health courses.
- Implement multidisciplinary consultations, defining the care manager for each patient; mapping what exists in this field at national level; ensure the transition from paediatrics to adult healthcare; organise all RD in a network of care, so that patients do not have to travel several times to be seen by several doctors and other health professionals at the same CE or other institution.
- Prepare written information to give to the patients, namely, a road map at the very beginning of the process.
- Mapping the existing patients – how many and where? The card for rare diseases may be an option, but there is a need to develop a more concrete proposal.
- Mapping epidemiology. There are projects like EPIRARE, whose purpose is to constitute registries, where such mapping could be integrated.
- Create a real NPRD (and not a virtual one), to facilitate the performance of all healthcare and other actions.

**Theme 2:**

- In Portugal, there is a trend to centralize the responsibilities in the field of RD, which can be useful in a small country like ours, but there is a risk of removing democracy from the process and of eliminating the participatory role of all interested partners.
- The existence of two aggregating structures of Patient Associations does nothing to help the patients with RD to have greater visibility and power of lobbying.
- There is a clear lack of concerted actions by several actors that should have a platform for meeting, agreement and coordination. This platform could be this aggregating organisation of Patient Associations.
- There is still a lack of effective information with regard to general data and European opportunities, and a fragmentation of information at the national level. It was proposed that the DGH might have on its website a section with specific information (both national and international) in that field.

**Theme 3:**

- At European level, contact the EU Commissioner for Science, who is Portuguese, to find out what are the methodologies, to be better prepared when applying to the existing funds for this purpose.
- At National level, create a national observatory for RD, possibly within Infarmed, with a

scientific committee and a committee for Patient Associations. This observatory should be a small and strong structure, among others, identifying research opportunities.

**Theme 4:**

- The creation of a national network of Centres of Expertise in cooperation with the European ones, in its support, training and research pathways according to the European Commission RD Expert Group criteria will enable, in coordination with the educational and social resources, effective support of patients with RD.
- The national observatory for RD could constitute an integrating tool of all resources, provided that it is composed by competent people, part of which full-time, working with well-defined rules and objectives, and receiving adequate funding, being transparent, less bureaucratic and proactive, and having a clear representation of all stakeholders in the field of RD and OMP.

**Theme 5:**

- The most important point discussed in this workshop was equity of access. Given the national situation, social inequalities and unequal budgets in hospitals, we need a centralized and dedicated budget for RD, which covers all patients equitably, regardless of their geographical area and place of residence.
- It is necessary to have some structuring support framework, in order to obtain data and guarantee access to European programmes, where it is possible, for example, access European funds (like Horizon 2020).
- The Informed information should be friendlier in some fields. It is also important to have greater quantity and quality of the information provided by bodies like Informed and Patient Associations, in order to provide health professionals and all patients with insightful information, by managing scientific quality and expectations that might exist.
- On the other hand, there is interest in knowing the expectations of the implementation of the measures mentioned by the MoCA group.
- Timing is essential to implement all these procedures.

**Theme 6:**

- In the debate and measures regarding RD, there is a predominant clinical pathway (including treatment). It is necessary to include the educational and social support pathways, as well.
- Through Patient Associations, there should a mapping of unmet needs of patients with RD and ensure their continuous care throughout life, at all ages.
- The links among the Ministry of Health, Ministry of Education and Science and Ministry of Solidarity, Employment and Social Security is essential.
- There is a need to consolidate the training of family doctors in the field of RD and genetic risks. For example, establish continuous training with specialised courses for

general practitioners and family doctors.

- Have a National Centre of Expertise and then regional centres with expertise consultations to help the social service perspective.
- It is very important to cross-check clinical research with research in social sciences, including psychology (to assess and understand how patients and their families live, and also the social role of Patient Associations).

The main conclusions that are cross-cutting to the 6 themes, from 2 days of discussion and participation of various *stakeholders*, are:

- Patients must be heard and represented in all situations that concern them, such as committees, working groups or other organizations on RD.
- It is essential to create centres of expertise for rare diseases, both to improve the patients' diagnosis and to allow the monitoring, as well as research in rare diseases and the inclusion of Portugal in the European network.
- The allocation of funds for the implementation of a plan/strategy for rare diseases is essential for it to work.

### III. ANNEXES

#### ANNEX 1: PROGRAMME

##### Day 27 February, Friday

##### 11h00: Reception and Welcome

##### 11h30 - 13h00: First Plenary Session

Moderator: **Constantino Sakellarides** | Scientific Ambassador of Aliança Portuguesa de Associações das Doenças Raras Aliança (Portuguese Alliance of Rare Disease Associations) for the Rare Disease Day

National Situation: **João Lavinha** | INSA

European Situation: **Lene Jensen** | Chief Executive Officer of Alliance for Rare Diseases Denmark and EURORDIS representative

##### 13h00 - 14h00: Lunch

##### 14h00 - 14h30 : Official Opening Session

**Maria Antónia Almeida Santos** | President of the Health Commission

**Francisco George** | Director General of Health (to be confirmed)

**Jorge Sequeiros** | Orphanet Portugal

**Marta Jacinto** | Portuguese Alliance of Rare Disease Associations

**Constantino Sakellarides** | Aliança's Scientific Ambassador for the Rare Disease Day

**Ana Bacalhau** | Aliança's Ambassador

**14h45 - 16h45**

**Workshop theme 3: Research on Rare Diseases**

Moderator: **Patrícia Maciel** |  
Universidade do Minho (University of Minho)  
Rapporteur: **Isabel Marques Carreira** |  
Faculdade de Medicina, Universidade de Coimbra (School of Medicine, University of Coimbra)

**Workshop theme 6: Social Services for Rare Diseases**

Moderator: **Paula Silva** | Faculdade de Medicina da Universidade do Porto (School of Medicine, University of Oporto) and IPATIMUP  
Rapporteur: **Helena Machado** |  
Universidade de Coimbra (University of Coimbra)

**16h45 - 17h15: Coffee Break**

**17h15 - 19h15**

**Workshop theme 2: Definition, Codification and Inventorying Rare Disease**

**(including information and training)**  
Moderator: **Margarida Reis Lima** |  
Hospital Lusíadas Porto (Lusíadas Hospital Oporto)  
Rapporteur: **Lina Ramos** | Hospital Pediátrico, CHUC (Pediatric Hospital of Coimbra)

**Workshop theme 4: Centres of Expertise (CE) / European Reference Networks/ Cross-border Health Care**

Moderator: **Eduardo Silva** | FMUC e CHLN (FMUC and CHLN)  
Rapporteur: **Luísa Diogo** | Presidente da SPDM (President of the Portuguese Society of Metabolic Diseases)

**Day 28 February, Saturday**

**9h00 - 11h00**

**Workshop theme 5: Orphan drugs and Rare Disease Treatment**

Moderator: **Fátima Vaz** | IPO (Portuguese Oncology Institute) and INFARMED (National Authority of Medicines and Health Products)  
Rapporteur: **Carolino Monteiro** |  
Faculdade de Farmácia da Universidade de Lisboa (School of Pharmacy, University of Lisbon)

**Workshop theme 1: Methodology, Governance and Monitoring of the National Plan**

Moderator: **Rui Gonçalves** | Rui Gonçalves | Hospital D. Estefânia (D. Estefânia Hospital)  
Rapporteur: **Teresa Coelho** | Clínica de Paramiloidose do Centro Hospitalar do Porto (Paramyloidosis Clinic of oPorto Medical Centre)

**11h00 - 11h30: Coffee Break**

**11h30: Closing Plenary Session**

Moderator: **Glória Isidro** | INSA

**ANNEX 2: LIST OF PARTICIPANTS**
**Stakeholder Groups:**

- Academic/Researcher
- Clinician/GP
- Healthcare Professional (other than clinician or GP)
- Industry
- Insurer
- Medical /Learned society
- Patient representative
- Politician
- Public administration (local, regional or national)
- Social worker
- *Media*
- Other

**Roles:**

- C:** Chair
- F:** Facilitator
- KS:** Speaker in plenary
- M:** Moderator
- P:** Participant
- R:** Rapporteur
- S:** Speaker
- V:** Venue/ComAgency

Name	Surname	Stakeholder group	C	R	KS	S	M	P	F	V
Alice	Margaça	Patient representative						x		
Ana	Afonso	Industry						x		
Ana	Bacalhau	Other				x				
Ana	Branquinho	Other								x
Ana	Correia	Academic/Researcher						x		
Ana	Gonçalves	Patient representative						x		
Ana	Martins	Academic/Researcher						x		
Ana	Romão	Academic/Researcher						x		
Ana	Santos	Academic/Researcher						x		
Ana	Tavares	Healthcare Professional (other than clinician or GP)						x		
André	Santos	Clinician/GP						x		
António	Silva	Patient representative						x		
António	Veiga	Healthcare Professional (other than clinician or GP)						x		
Baldomero	Teixeira	Patient representative						x		
Bárbara	Marques	Healthcare Professional (other than clinician or GP)						x	x	
Bruno	Colaço	<i>Media</i>								
Carina	Brandão	Other						x		
Carlos	Matos	Academic/Researcher						x		
Carolino	Monteiro	Academic/Researcher		x				x		
Catarina	Carvalho	Industry						x		
Catarina	Marcelino	Industry						x		
Catarina	Pyrrait	Patient representative						x		

Name	Surname	Stakeholder group	C	R	KS	S	M	P	F	V
Cláudio	da Cruz	Patient representative						x		
Clévio	Nóbrega	Academic/Researcher						x	x	
Conceição	Queiós	<i>Media</i>								
Constantino	Sakellarides	Other	x			x				
Corália	Loureiro	Other						x		
Cristina	Bastos	Patient representative						x		
Cristina	Dias	Patient representative						x		
Cristina	Serra	<i>Media</i>								
David	Silva	Patient representative						x		
Diogo	Lopes	Patient representative						x		
Diogo	Martins	Patient representative						x		
Eduardo	Silva	Clinician/GP					x	x		
Elisa	Leão Teles	Clinician/GP						x	x	
Elsa	Mateus	Patient representative						x		
Elvira	Dias	Patient representative						x	x	
Emerência	Teixeira	Clinician/GP						x	x	
Fátima	Godinho	Patient representative						x		
Fátima	Vaz	Clinician/GP					x	x		
Fernando	Bastos	Patient representative						x		
Fernando	Canhão	Healthcare Professional (other than clinician or GP)						x		
Filomena	Lobo	Public administration (local, regional or national)						x		
Francisco	Nunes	Industry						x		
Glória	Isidro	Healthcare Professional (other than clinician or GP)	x					x	x	
Gonçalo	Rijo	Patient representative						x		
Helena	Machado	Social worker		x				x		
Helena	Rodrigues	Other								x
Heloísa	Santos	Clinician/GP						x		
Hugo	Canhão	Industry						x		
Inês	de Faria	Patient representative	x					x	x	
Isabel	Carreira	Healthcare Professional (other than clinician or GP)		x				x		
Isabel	Martins	Patient representative						x		
Jesuíno	Jacinto	Patient representative						x		
Joana	de Melo	Healthcare Professional (other than clinician or GP)						x	x	
Joana	Martins	Healthcare Professional (other than clinician or GP)						x		
Joana	Santos	Patient representative						x		
João	Cabrita	Healthcare Professional (other than clinician or GP)						x		
João	Fonseca	Industry						x		
João	Gabriel	<i>Media</i>								
João	Lavinha	Academic/Researcher			x			x		
João	Silva	Clinician/GP						x	x	



Name	Surname	Stakeholder group	C	R	KS	S	M	P	F	V
Joaquim	Brites	Patient representative						x		
Jorge	Marques	Clinician/GP						x		
Jorge	Saraiva	Clinician/GP						x	x	
Jorge	Sequeiros	Academic/Researcher	x			x		x	x	
José	Bento	Patient representative						x		
José	Robalo	Public administration (local, regional or national)						x		
Jytte	Puls	Other								
Lene	Jensen	Patient representative			x			x		
Leonor	Guedes	Clinician/GP						x		
Lina	Ramos	Clinician/GP		x				x		
Luís	Brito Avô	Medical /Learned society	x					x	x	
Luísa	Diogo	Clinician/GP		x				x		
M <sup>a</sup> de Belém	Roseira	Politician	x			x		x		
Manuela	Azoia	Other								x
Marco	Padrão	Patient representative						x		
Margarida	Reis Lima	Clinician/GP					x	x		
Maria	Barreiros	Patient representative						x		
Maria	Bento	Patient representative						x		
Maria	Brandao	Patient representative						x		
Maria	de Abreu	Industry						x		
Maria	Ferreira	Patient representative						x		
Maria	Gomes	Other								x
Maria	Marques	Academic/Researcher						x		
Maria	Namora	Social worker						x		
Maria	Pereira	Patient representative						x		
Maria	Rodrigues	Patient representative						x		
Maria	Saraiva	Patient representative						x		
Marina	Magalhães	Clinician/GP						x	x	
Marta	Beirão	Patient representative						x	x	
Marta	Jacinto	Patient representative	x			x		x	x	
Michelly	Abreu	Patient representative						x		
Miguel	Leão	Clinician/GP						x	x	
Milena	Paneque	Academic/Researcher						x		
Miriam	Silva	Academic/Researcher						x	x	
Nuno	Lages	Industry						x		
Nuno	Machado	Healthcare Professional (other than clinician or GP)						x		
Patrícia	Arinto	Academic/Researcher						x		
Patrícia	Maciel	Academic/Researcher					x	x		
Patrícia	Rama	Academic/Researcher						x		
Paula	Finuras	Patient representative						x		
Paula	Marques	Healthcare Professional (other than clinician or GP)						x		
Paula	Pinto	Academic/Researcher						x	x	
Paula	Silva	Social worker					x	x		

Name	Surname	Stakeholder group	C	R	KS	S	M	P	F	V
Paulo	Bacalhau	Patient representative						x		
Paulo	da Cruz	Patient representative						x		
Paulo	Sá Santos	Patient representative						x		
Raquel	Palha	Industry						x		
Rita	Vassal	<i>Media</i>								
Rogério	Lima	Social worker						x	x	
Rui	Bernardino	Patient representative						x		
Rui	Gonçalves	Clinician/GP					x	x		
Rute	Verdade	Academic/Researcher						x	x	
Sandra	de Jesus	Patient representative						x		
Sandra	Fernandes	Healthcare Professional (other than clinician or GP)						x		
Sandra	Santos	Industry						x		
Sílvia	Silva	Patient representative						x		
Sofia	Dias Gaspar	Patient representative						x		
Sofia	Loução	Social worker						x		
Sofia	Machaqueiro	Other								x
Sónia	Albuquerque	Industry						x		
Sónia	da Costa	Patient representative						x		
Susana	Moura	Patient representative						x		
Teresa	Caeiro	Politician						x		
Teresa	Carvalho	Academic/Researcher						x		
Teresa	Coelho	Clinician/GP		x				x		
Teresa	Montalvão	Other								x
Teresa	O'Neill	Industry						x		
Tiago	Barros	Industry	x					x	x	
Verónica	Gómez	Academic/Researcher						x		
Victoria	Hedley	Other						x		