

EUROPLAN National Conferences

CONFERENCE FINAL REPORT

I. General information

Country	GREECE
Date & place of the National Conference	26-27 NOV.2010 EUGENIDES FOUNDATION, ATHENS
Website	www.pespa.gr
Organisers	Greek Alliance for Rare Diseases: Mrs. M Lambrou, Dr. K. Yannoukakos, Dr. J Traeger-Synodinos; Mr. D. Synodinos, Dr. G. Voutsinas, Mrs. P. Traka, Mrs. A. Alexandropoulou, Mrs. D. Delga
Members of the Steering Committee	<p>Mr. A. Dimoploulos, General Secretary of the Ministry of Health and Social Solidarity;</p> <p>Mrs. A. Dretta, General Secretary of the Ministry of Labor and Social Security,</p> <p>Dr. M. Skourouliakou, Vice-President of the National Organisation of Pharmaceuticals;</p> <p>Professor E. Kanavakis, Professor of Genetics, Athens University;</p> <p>Mrs. M. Lambrou, President of the Greek Alliance of Rare Diseases (PESPA) and the Tuberculosis Association of Greece;</p> <p>Mrs. V. Tsala, Head of the Education Section of the Hellenic Centre of Infectious Disease Control & Prevention (KEELPNO);</p> <p>Mr K. Frouzis, Vice President of Novartis Hellas and Secretary General of the Greek Association of Pharmaceutical Companies;</p> <p>Mr A. Kypraios, General Director of Genzyme Hellas and Vice-President of the Greek Association of Biotechnology;</p> <p>Mrs E. Gavriil, Production Manager of Pharmaceutical Institute for Research & Technology;</p> <p>Dr. M Petersen, Head of Genetics Dept, Institute of Childhealth and Representative of Orphanet in Greece.</p>
Names and list of Workshops	<p>Workshop 1: Methodology and administration for the National Plan- Strategy</p> <p>Workshop 2: Definition codification and inventorying of Rare Diseases; Information and</p>

	<p>training</p> <p>Workshop 3: Research on Rare Diseases; Orphan Drugs</p> <p>Workshop 4: Evaluation of level of care for Rare Diseases: Centres of Expertise, European Reference Networks for Rare Diseases</p> <p>Workshop 5: Empowerment of Patients organisation, Specialised services</p> <p>Horizontal themes: Sustainability and Gathering Expertise on Rare Diseases at European Level</p>
Chairs and Rapporteurs of Workshops	See each section
Attachments (programmeme, list of participants, etc.)	>350 attendees

II. Overview

The Greek EUROPLAN National Conference for Rare Diseases and Orphan Drugs was organised in the framework of the EUROPLAN Project, funded by the European Commission.

The conference was held over 2 days on the 26th & 27th November 2010, at the Eugenides Foundation, Athens, under the **Hospice of Chairman of Greek Democracy Karolos Papoulias** and was attended by >350 people, from all the main stakeholders in Greece, such as representatives of the Ministry of Health and Social Solidarity, Ministry of Labor and Social Security, Health Insurance Departments, the Hellenic Centre of Infectious Disease Control and Prevention, the General Secretariat for Research & Technology of the Ministry of Education and Life Long Learning, Secretary General of the Greek Association of Pharmaceutical Companies, Vice-President of the Greek Association of Biotechnology, the National Organisation of Pharmaceuticals, Academic Professors, Healthcare Professionals (doctors, nurses, psychologists etc), the representatives of Orphanet in Greece, Researchers in the field of RD, Students, Representatives of patient associations, Patients and their families and the general public.

In Greece the level of awareness on the issue of Rare Diseases (RD), both of the general public and also of the health professionals and the scientific world is relatively low. This includes information related to approaches and methods to prevent or diagnose RD, and also on how to manage patients with RD, including optimal treatments, new treatments and psychosocial support.

The current Greek National Plan of Action for Rare Diseases 2008-2012 was compiled and published in Athens in 2008, based on recommendations by PESPA (acronym for the Greek Alliance for RD), which were in turn based on the French National Plan, but modified by the Greek Ministry of Health of that time (2008). It has not yet been applied.

In the continued absence of effective public services for patients with RD and their families in Greece, it is important to lay the foundations on which these services can be built through promoting the steps towards applying the National Plan.

In this context, the Greek Europlan Conference aimed to:

- present and publicise the specific strategies and aims of the Greek National Plan for rare diseases to all the relevant stake-holders, including the Greek Ministries and Health Authorities, Health Professionals, Scientists and Researchers, Patients, as well as the wider society ,
- make the Greek rare diseases stakeholders familiar with the EU Council Recommendation on RD and EUROPLAN recommendations for actions, to present and gather proposals on them, to discuss their transferability and application in Greece,
- evaluate the status of RD awareness and patient care currently in Greece,
- draft proposals for actions to improve prevention, diagnosis, treatment and social integration of patients with RD,
- promote specialised training of healthcare professionals; promote research and availability of orphan drugs in the field of RD,

In preparation for the conference, a Steering Committee was formed, all stakeholders were invited, a press release was made and a TV Spot and Radio Spot were played regularly in the month preceding the

conference. Mrs Simona Bellagambi came to Athens twice to support the conference organisation, and she also attended the Greek conference itself.

The conference was characterised by extremely fruitful discussions throughout, by all participants present. Following the initial plenary session, there were 5 concurrent workshops to cover the 7 main themes and the 2 horizontal themes. Members of the Board of Directors of PESPA acted as Facilitators (Chairs) for the presentations and discussions of each workshop. Following completion of the workshops midday on the second day, a second plenary took place in which the conclusions and recommendations from each workshop were presented by the relevant facilitator and were opened to discussion by all delegates. The summary of all workshops and themes follows below.

III. Main Report

WORKSHOP 1: METHODOLOGY AND GOVERNANCE OF A NATIONAL PLAN

Facilitator (Chair): Mr. D. Synodinos

Present at the workshop with active participation were:

- Ms Pantazopoulou representing the Ministry of Health and Social Solidarity.
- Ms Matsika representing the Ministry for Labor and Social Affairs.
- Ms Tzala representing Hellenic Centre of Infectious Disease Control & Prevention (KEELPNO).
- Members of patients associations as well patients, their families and students.

Having taken into account the European Council recommendations, the EUROPLAN recommendations and indicators, the workshop first answered the questions posed in slides, and then proceeded to discuss the current situation in Greece with RD and set out proposals appropriate and relevant for Greece.

With reference to the Europlan Indicators

Slide 13 answers

ACTIONS	INDICATORS	TYPE	ANSWERS (GREECE)
Development of Regulations/ Laws	Existence of Regulations/laws that support the creation and development of a RD plan	Process	“Not existing not implemented”
	National / regional (percentage of regions)	Process	“Does not exist”
Establishment of Coordination mechanisms	Existence of a coordination mechanism	Process	“Not existing not clearly stated”
	Existence of an expert advisory committee	Process	“Does not exist”

Slide 14 answers

ACTIONS	INDICATORS	TYPE	ANSWERS (GREECE)
Establishment of an external evaluation of the plan/strategy Procedure	Existence of an external evaluation body / procedure	Process	"Does not exist"
Degree of comprehensiveness	Number of priority areas included	Process	"7" (on a scale of zero-10)
Establishing of a budget for developing the plan/strategy	Budget of plan/strategy	Process	"Does not exist"

The current **Greek National Plan (NP) of Action for Rare Diseases 2008-2012** was compiled and published in Athens in 2008, based on recommendations by PESPA (in turn based on the French National Plan), but modified by the Greek Ministry of Health of that time (2008). The evaluation of this plan by all present at the EUROLAN meeting concluded that it is generally acceptable.

The strategic priorities in the Greek NP are:

- 1) The recognition of the specificity of RD
- 2) The systematic monitoring of the epidemiology of RD and a registry of RD and patients with RD
- 3) To develop the information for patients, health professionals and the general public concerning RD.
- 4) To improve the quality of all services involved in timely diagnosis, early intervention, treatment and rehabilitation of patients with RD.
- 5) To increase access of citizens screening
- 6) To promote research and innovation on RD notably for treatments.
- 7) To respond to the needs of people suffering from RD especially for new and more effective treatment.
- 8) To develop a common platform of action on a national level in the sector of RD and participation in the equivalent European Network.

Modification to the current strategies of the Greek NP during the workshop included:

- To strategic priority: No.4 the term "early intervention" was added
- To strategic priority: No.7 the word "specific" before needs was deleted.

To achieve strategies 1-8, the aims are shared between **6 work packages** as follows:

- 1) Timely diagnosis
- 2) Therapy
- 3) Welfare
- 4) Research
- 5) Education
- 6) Development of strategies dependant upon EU membership co-operation

Modification to the current work packages (axis and actions) of the Greek NP during the workshop included:

- Under “Therapy” the action to create “Centres of Treatment” was changed to “Centres of Reference” (Centres of Expertise) [to be preceded by “Centres of Co-ordination for RD- see recommendations of Workshop 4]
- The term “Welfare” (Providence) was changed to “Social Care and Rehabilitation” and the word “specific” was deleted before “needs” and “early intervention” was added.
- To “Education” the terms “Awareness and Sensitivity” were added, as well as “develop awareness of public opinion and centres of decision”.
- In WP6 following the term “National Platforms” the words “with extension to local and district level” were added.

The WG1 on the Greek National Plan of Action for Rare Diseases at the EUROPLAN meeting also made the following proposals and serious observations:

- At the moment there is no kind official body (committee or other authority) which is monitoring or evaluating the NP.
- No publicity is given to the NP or its content.
- As things stand, the legal/policy framework in this National Plan of Action for RD 2008-2012 is in the form of a written document with a fairly clear structure but it has no legal binding up to date, which precludes a monitoring and evaluating system.
- It was also decided at this workshop that an external committee under the KESY (Central Council for Health which is under the auspices of the Ministry of Health) should be established. The committee will include major Stakeholders in RD.
- This committee will monitor, evaluate and deliver opinion on the progress of the Greek NP for RD.
- Currently in our country the awareness of the situation of RD – epidemiologic figures, inventory of existing resources – is poor and the unmet needs of RD patients are not being evaluated.

Finally it was decided that the **priorities of action** should be:

- 1) Registries for RD and RD patients.
- 2) Timely diagnosis and
- 3) The development of awareness, information and education both for health professionals and the general public.

On the crucial subject of **sustainability** of the NP, the answer is that currently there is NO budget and NO allocated source of funding.

After the discussions within the workshop the results were presented at the plenary session, but no comments were made altering or adding to what was decided and proposed in our WG1.

WORKSHOP 2 - DEFINITION, CODIFICATION AND INVENTORING OF RD

Facilitator (Chair): Dr. G.E. Voutsinas

Present at WG with active participation were

- Dr. E. Kanavakis, Professor of Medical Genetics, University of Athens, Greece
- Dr. Sofia Douzgou, Clinical Geneticist, representing the Dept of Genetics, Institute of Child's Health, Athens, Greece, formal representative of Greece in the international network ORPHANET
- Ms P. Traka, representative of the Greek Alliance for Rare Diseases
- Members of patients associations as well patients or relatives and students.

EUROPLAN Indicators for Definition, Codification and Inventoring of RD

EUROPLAN Indicators part 1

ACTIONS	INDICATORS	TYPE	ANSWERS
To officially adopt the EC RD definition (no more than 5 cases/10,000 inhabitants)	Adoption of the EC RD definition	Process	• No
To include the best Rare Diseases classification currently existing into the public health care related services	Type of classification used by the health care system	Process	• ICD-10

EUROPLAN Indicators part 2

ACTIONS	INDICATORS	TYPE	ANSWERS
To include the best Rare Diseases classification currently existing into the public health care related services	Developing policies for recognising RD by the care information systems	Process	• Not existing, not clearly stated
Defining a surveillance system based on a patient outcomes registry	Registering activity	Process	• No registry at all
	Number of diseases included	Outcomes	•

1. Definition of RD

Question 1

▪ Is the EU official definition (RDs are those affecting up to 5 out of 10 000 person) used in your country?

Answer

▪ Officially no. Used informally by the people who already know the percentage 5/10.000

Suggestion

▪ Be legislated by the Greek Parliament with law of state and they commit for his functional adoption relative Ministries: (1) Health and Social Solidarity, (2) Employment and Social Insurance and (3) Economy, Competitiveness and Shipping

Question 2

▪ Are there alternative or more specific definitions used instead or in addition?

Answer

▪ No

2. Classification and traceability of RDs in the national health system

Question 1

▪ What classification system is used in your country? ICD9, ICD10, SNOMED, OMIM, ORPHAN...

Answer

▪ Recently the ICD10 classification was officially adopted.

Question 2

▪ For which purpose is (are) the classification system(s) used, e.g. surveillance, reimbursement, provision of social support, etc.

Answer

▪ With the exception of a handful of clinics currently making their first steps in adopting ICD10, no registry of patients is being done and thus used in daily practice.

Suggestion

▪ Involved Ministries should promptly exercise pressure on state and private supervised entities for adopting ICD10 in practice at all levels of National Health and Welfare system.

Question 3

▪ Is your country prepared to adopt the WHO-led system, the ICD-11, recommended by the EU in the Council Recommendation on RD, when ready (2014)?

Answer

▪ No.

Suggestion

▪ Adoption of ICD10 in practice must proceed, coupled with a change in the attitude vis-à-vis disease inventorying that will be the foundation for ICD11 acceptance.

Question 4

▪ What level of awareness and knowledge do healthcare professionals have of the RD classification and codification? What can be done to improve it?

Answer

▪ They are not well informed.

Suggestions

▪ Implementation of ICD10 in daily clinical practice will help in systemizing the inventorying process and in turn will promote the level of knowledge and awareness on RD of health professionals. Moreover, it is also recommended that Ministry and Patient Associations functionaries would take initiatives, during medical congresses, by means of relevant printed leaflets, by setting up seminars in Medical Schools and/or hospitals, by providing information via the Athens and Thessaloniki Medical Societies, as well their respective printed publications.

3. Inventories, registries and lists

Question 1

▪ Are there official lists of RD in your country? Is there an official governmental RD registry? And/or specific RD databases e.g. held by Centres of Expertise? Are there RD surveillance projects or programmes (e.g. sentinel programmes surveys)?

Answer

▪ Official lists of RD do not exist in Greece.

Question 2

▪ What kind of initiatives should be taken or reinforced in your country?

Answer

▪ Doctors concerned with specific RDs and Patient Associations have in their possession such directories. PESPA holds already a large directory of patients, either belonging in associations-members of PESPA, or individuals. In order to have single and sustainable inventorying of RD patients and RDs in Greece, a large meeting with representatives from all concerned parties (related Ministries, health professionals, patient associations) must be organised, for acknowledging this effort and establishing the registry's location. This meeting may be setup by initiative of PESPA and under the aegis of the Ministry of Health and Social Solidarity. Last, to ensure sustainability of inventorying, a certain amount of state funding must be provided for, a thing fairly feasible since the sum needed is not big.

Question 3

▪ Do these registries and programmes receive government support?

Answer

▪ Existing non-state, individual or collective, inventories are not being supported by the state.

Question 4

▪ How to ensure, through appropriate funding mechanisms, the long-term sustainability of registries and databases?

Answer

▪ As mentioned above, funding must derive from state sources, since no exuberant amounts of money are needed. If this is not feasible, the option of financing registries and/or databases by private sources must be examined, once eventual requirements by individuals are considered.

Question 5

▪ Does your country participate to the development of an EU inventory of RD as recommended in the Council Recommendations on RD?

Answer

Our country does not participate in the EU-driven RD patient inventorying. However, the Children Health Institute's Division of Genetics, as formal representative of Greece in the ORPHANET world network, collects data on Greece that are forwarded to update the above directory. The ORPHANET directory

contains information on European services on RD with respect to clinics, biochemical and molecular laboratories, research activities and active patient associations.

1.1. Information and training

1. How to improve information on available care for RDs in general, for different audiences

Question 1

▪ What are the existing information sources in the country? Are they of good quality? Do they receive public funding or Patients Org. funding?

Answer

▪ Scarce official and unofficial Centres of Expertise (state-funded but not to inform the public on RD treatment) and Patient Associations (non state-funded). Also, as mentioned above, the Children Health Institute's Division of Genetics, as formal representative of Greece in the ORPHANET network, collects data on Greece that are forwarded to update the above directory. Information provided is usually of high quality, although there is always room for improvement.

▪ Suggestion

To be translated the Orphanet network in the Greek language.

• Question 1.1

○ Is there a national official website for RD in the country?

• Answer

○ No.

• Suggestion

• To establish an official national website on RD in our country funded by the Ministry of Health and Social Solidarity and potentially by private entities. PESPA may undertake this effort on behalf of the RD patients.

• Question 1.2

○ Are there help lines for both patients and healthcare professionals? Are they known to the public?

• Answer

○ In terms of patient help lines, the telephone numbers of patient associations are the ones used for that purpose, and PESPA plays an important role in informing and supporting RD patients. There are no help lines for health professionals. Patient associations are not particularly known to the wider public despite their efforts to gain exposure.

• Suggestion

○ Establishment of a telephone help line, bearing a phone number compatible with operating principles of such lines in other European countries. To do so, subject to detailed feedback by colleagues from other EU countries, with relevant experience, a task force should be formed which all interested parties should take part in (patient groups, clinical doctors, psychologists, etc), to determine such help line's operating principles and rules of partnerships among interested parties, and to also appoint persons in charge of each specific action (psychological support, medical information).

• Question 1.3

○ Are there initiatives of centres of expertise and/or patient organisations or programmeme to stimulate the development of information and educational material for patients or specific publics (teacher, social workers, etc.)?

• **Answer**

○ There are. They consist in initiatives taken by certain hospital clinics, pharmaceutical companies, PESPA, and other patient associations. They are addressed mainly to patients and general public. Due to scant or even zero financing. These efforts are sporadic. Unfortunately, a major problem lies in the observed lack of volunteerism by patient families and, in general, of will to take part in common demands.

• **Question 1.4**

○ Are existing resources at EU level, Orphanet and Eurordis used: (information on diseases, specialised centres and patient groups, ongoing research projects, clinical trials)?

• **Answer**

○ At a limited degree, mainly by those having access to internet and speaking foreign languages (mainly, English).

• **Suggestion**

○ Orphanet be translated in the Greek

• **Question 1.5**

○ Are there initiatives to raise awareness on RD such as a RD Day?

• **Answer**

○ Yes. Greece, by virtue of PESPA, was one of the countries to have adopted and established the Rare Disease Day (February 28th 2008) and participates ever since hosting various events. Initiatives for raising patient and general public awareness on RD are being taken mainly by PESPA, as well as other patient associations.

2. How to improve access to quality information on RDs

Question 1

▪ How are these information sources and initiatives publicised?

Answer

▪ These initiatives take various forms. For instance, PESPA hosts, apart from RD Day, a number of events all year around aiming at informing patients and the general public on RD, e.g. happenings during events in downtown Athens, TV and radio advertising spots, articles published in daily newspapers and periodicals, hosting speeches, seminars and congresses, visits to hospitals, attendance of members of the scientific committee to conferences, TV shows on RD issues, etc.

Question 2

▪ How to make sure that they reach out to the target audiences? And to the wider public?

Answer - Suggestions

▪ In order for these initiatives to reach targeted audiences, as well as the general public, activation and more massive participation of volunteers from patient families is needed, in order for these awareness campaigns to become more efficient. Actions that may currently be taken to this direction, are the following:

- Utilization of Health Visitor staff, who are graduates of the related technological Educational Institution of Athens.
- Suggestions to Pedagogical Schools to organise seminars in Special Education Departments, and to municipalities for speeches in primary and secondary education schools.
- Positioning of printed informational material in pharmacies.

3. How to ensure adequate training of healthcare professionals on RDs

- Training for healthcare professionals may include:

Question 1

- Training to make them aware of the existence of RDs and of resources available for their care

Answer

Suggestions

- To provide post-training to doctors in Medical Societies by means of appropriate seminars.
- To publish articles in the Medical Association and the Pharmaceutical Association Journals.
- To prepare and dispatch printed information material to all primary care centres in the countryside, and a large number of pharmacies.
- To raise awareness on services provided by the Orphanet network and translation of its website in Greek.

Question 2

- Medical training in fields relevant to diagnosis of RDs (e.g. genetics, oncology, immunology, neurology, paediatrics)

Answer

- There is such training, but limited. There is no specialisation on Clinical and Laboratory Genetics.

Suggestions

- To establish, in Greece, the specialization on Clinical and Laboratory Genetics, upgrading thus the field of diagnosis to clinical and laboratory standards.
- To hold a conference on medical, laboratory and legal aspects of diagnosis.

Question 3

- Medical training of young doctors and scientists in the field of RD

Answer

- As mentioned before, there is no specialisation on Clinical and Laboratory Genetics. If established, there will be increased interest in RD treatment at clinical and laboratory level.

Suggestions

- To provide post-training to doctors in Medical Societies by means of appropriate seminars.
- To publish articles in the Medical Association and the Pharmaceutical Association Journals.
- To prepare and dispatch printed information material to all primary care centres in the countryside, and a large number of pharmacies.

- To raise awareness on services provided by the Orphanet network and translation of its website in Greek.

Question 4

- Exchange and sharing of expertise between centres within the country

Answer

- It is so, but to a limited extent.

Suggestions

- To establish a workshop, open to experts of various specialisations, dealing with RD.
- To set up a committee, made up of experts from Centres of Expertise, aiming at coordinating efforts vis-à-vis common problems. Patient associations should also be represented in this committee.

Question 5

- How to ensure that existing and validated international guidelines are used at national level to guide diagnosis and treatment of RD?

Answer

- To establish a health professionals committee in charge of defining principles of best practice in clinical and laboratory diagnosis, as well as therapeutic protocols for each disease. This committee may be supported by other doctors and scientists, experts on specific diseases, and RD patient associations.

Question 6

- What mechanisms can be put in place to support the exchange of expertise at EU level and the adequate training for all healthcare professionals?

Answer

- To create a European network of experts on specific RD or RD groups, holding regular meetings in collaboration with EUCERD.
- Among its other duties, the network above will prepare and publish online (in Orphanet's website or elsewhere) the principles of best practices on clinical and laboratory diagnosis, as well as therapeutic treatment for each RD.
- Within the network above, a committee should be formed to deal with preparing and publishing a curriculum that will be deemed ideal for sufficiently training health professionals at European level.

Question 7

- Is your country supporting the participation of national experts in developing international guidelines to guide diagnosis and treatment of RD at national level?

Answer

- Greece appoints its national representatives. However, it is not known whether Greek experts actually take part in developing international regulations on governance of diagnosis and therapeutic treatment of RD at national level.

Suggestions

- To make a list of related national experts and to form a committee within the Ministry of Health and Social Solidarity for coordinating them. This committee must include people from patient associations and PESPA.

- To provide for regular update of interested parties by related national experts.

WORKSHOP 3: RESEARCH ON RARE DISEASES AND ORPHAN DRUGS

Facilitator (Chair): Mr. D. Yannoukakos, Director of Research, National Centre of Scientific Research “Demokritos”

Present at the workshop with active participation were:

- Mrs Kolyva, representing the General Secretariat for Research & Technology of the Ministry of Education and Life Long Learning
- Mrs Plessa, representing the General Secretariat for Research & Technology of the Ministry of Education and Life Long Learning
- Mr K. Frouzis, Vice President of Novartis Hellas and Secretary General of the Greek Association of Pharmaceutical Companies
- Mr A. Kypraios, General Director of Genzyme Hellas and Vice-President of the Greek Association of Biotechnology
- Mrs E. Gavriil, Production Manager of Pharmaceutical Institute for Research & Technology
- Ms. G. Georgiadou, Head of Pharmaceutical Prescriptions, Welfare & Insurance Organisation for Farmers
- Members of patients associations, as well patients or relatives and students.

Having taken into account the European Council recommendations, the EUROPLAN recommendations and indicators, the workshop first answered the questions posed in the slides, and then proceeded with discussing the current situation in Greece with RD and set out proposals appropriate and relevant for Greece.

With reference to the Europlan Indicators

Slides 10, 11, 12 answers

ACTIONS	INDICATORS	TYPE	ANSWERS (GREECE)
Building research programmeme for Rare Diseases	Existing of RD National/Regional research programmemes	Process	Not RD research programmeme
	RD research programmeme monitoring	Process	Not existing, not clearly stated
	Number of RD research projects approved by year (if possible yearly starting the year before plan commencement)	Outcomes	Not existing

ACTIONS	INDICATORS	TYPE	ANSWERS (GREECE)
Building a research programme for Rare Diseases	Clinical trials funded by public bodies	Outcomes	No actions have been taken
	E-RARE joining	Process	• Ongoing
	Including public health and social research, in the field of rare diseases	Process	Under discussion •
	Research platforms and other infrastructures are also funded by the research programme	Process	• Under discussion
ACTIONS	INDICATORS	TYPE	ANSWERS (GREECE)
Existence of national policy in support of the recruitment of young scientists / Researchers specifically for Rare Diseases	Number of young scientists recruited every year to work specifically on rare diseases	Process	There are some but not specifically assigned to RD
Allocate funds for the RD research programme	There are specific public funds allocated for RD research	Process	• No
	Funds specifically allocated for RD research actions /projects per year since the plan started	Outcomes	• No

RESEARCH (IN GREECE)

1. The main problem identified is the zero absorption of EU funds available for research in general (as well as in RD) through the National Strategic Reference Framework (NSRF) 2007-2013 due to failure of management of funds. While the absorption of funds should have started in 2007 no programmes have been allocated to research because a missing infrastructure of the Management Authority.

2. Pharmaceutical companies expressed the desire for funding research in Rare Diseases in Greece, but under conditions of transparency and coordinated response from all concerned ministries (Health, Education, Development, and Labour) through the creation of an inter-ministerial coordinating body.

3. Representatives of the General Secretariat for Research and Technology reported that the E-Rare2

unlike E-Rare1 encourage social research and funding of RD registries, in addition to basic research on rare diseases. The Greek government is involved with 200,000 euros in R-Rare2. There was a programme with Greek coordinator funded from E-Rare1.

4. Complete lack of research policy by the Ministry of Education and Life Long Learning (formerly the Ministry responsible for research was the Ministry of Development) for translational research, particularly for rare disease.

5. Representatives of the General Secretariat for Research and Technology reported the European scale programmemes associated with the Innovative Medicine Initiative (IMI). This is the biggest public-private initiative in Europe to accelerate the development of innovative, better and safer drugs for patients.

The IMI supports collaborative research and builds networks of industrial and academic experts to strengthen the pharmaceutical innovation in Europe. IMI is a joint venture between the European Union and the European Association of Pharmaceutical Industries (EFPIA). The fields are defined by the pharmaceutical industry. There will be such a notice soon and in Greece.

6. There was also discussion about the programme for biobanks on rare diseases, which is developing in Greece under the European Programme BBMRI of the European Strategy Forum on Research Infrastructures (ESFRI). But it was found that the programme was still at a very early stage.

Orphan Drugs

1. There is an urgent need to set up a steering committee across all ministries with the rare diseases as its main objective and include all parties: ministry officials, patients, pharmaceutical companies, doctors, researchers, etc.

2. The possibility of a non-profit organisation promoting and funding research on rare diseases was also discussed.

3. Urgent need for RD registry using different approaches such as electronic prescriptions, patients' associations, scientific societies.

4. Inclusion in the law 3816 for zero participation of patients in the cost of Orphan Drugs.

5. Measures to speed up approval of new drugs for RDs. Framework for therapeutic protocols.

6. Hospital price for Orphan Drugs sold through pharmacies.

7. Correct billing system to prevent parallel exports and ensure the availability of orphan drugs nationwide.

8. Building and expanding the role of the Institute of Pharmaceutical Research and Technology (the

major portal for introduction of many Orphan Drugs entry to Greece) for wider access to medicines, removing the causes of difficulty in functioning (high debt of hospitals to IPRT).

WORKSHOP 4: EVALUATION OF CARE PATHWAYS FOR RARE DISEASES- CENTRES OF EXPERTISE AND EUROPEAN NETWORKS FOR RD

Facilitator (Chair): Dr. J Traeger-Synodinos, DPhil, Assist. Prof of Genetics, Athens University Medical School.

Present at the Work shop with active participation were

- Dr. S. Youroukos (Pediatric neurologist, active in the field of RD)
- Dr. E Papadopoulou-Alataki (Assistant Professor, Pediatrics-Immunology, active in the field of RD)
- Ms A. Papayiannopoulou, representative of the Ministry of Health and Social Solidarity, Department of Public Health.
- Ms D. Sotiri, representative of the Hellenic Centre of Infectious Disease Control and Prevention (KEELPNO), Greek Representative for e-Rare.
- Ms A. Gliati (Rapporteur) representative of a patient organisation (Crohn’s disease).
- Members of patients associations, as well patients or relatives and students

Having taken into account the European Council recommendations, the EUROPLAN recommendations and indicators we first answered the questions asked in the slides and then went on to discuss issues with reference to our subject and come out with proposals.

With respect to the slides

Slide 10: EUROPLAN INDICATORS FOR CENTRES OF EXPERTISE

ACTIONS	INDICATORS	TYPE	ANSWERS
Improve the quality of health care by defining:	Existence of a policy for establishing centres of expertise at the national/regional level	Process	• Not existing, not clearly stated
appropriate centres with experience on RD - pathways that reduce the diagnosis delay and facilitate the best care and treatments	Number of centres of expertise adhering to the policy defined in the country	Outcomes	Number of reference centres – >30 departments/clinics but NOT CoE’s with criteria of EURORDIS
	Groups of rare diseases followed up in centres of expertise	Outcomes	Covering only some rare diseases –a few diseases !

Slide 11: **EUROPLAN INDICATORS FOR CENTRES OF EXPERTISE**

ACTIONS	INDICATORS	TYPE	ANSWERS
Improve the quality of health care by defining: appropriate centres with experience on RD - pathways that reduce the diagnosis delay and facilitate the best care and treatments	Centres of expertise adhering to standards defined by the Council Recommendations - paragraph d) of preamble	Outcomes	Percentage of centres of expertise adhered by the total of centres of expertise designed- NO GREEK CENTRES ADHERE TO THE STANDARDS DEFINED BY THE COUNCIL RECOMMENDATION
	Participation of national or regional centres of expertise into European reference networks	Outcomes	Index based on number of centres of expertise NO GREEK CENTRES PARTICIPATE IN EUROPEAN REFERENCE NETWORKS

Based on the answers to slides 10 and 11, it can be concluded that the current situation in Greece with respect to the CENTRES OF EXPERTISE (CoE's):

- There is no policy for establishing CoE's at the national/regional level
- There are DEPARTMENTS/CLINICS (not fully CoE's) for some RD's, including: Gaucher & Fabry, Histiocytosis, Hemophilia, Pulmonary Hypertension, Retinopathies, Cystic Fibrosis, Crohn's Disease, Congenital Cardiopathies, Keratoconus, Prader Willi, Hereditary Metabolic Disorders, Rheumatoid Arthritis, Primary Immunodeficiencies (Pediatric), Thalassemia, Sickle Cell Anemia.
- None of the Greek CoE's adhere to the standards defined by the Council Recommendations
- There is no participation of national or regional centres of expertise into European reference networks

The needs (numbers and location) of CoE's cannot be easily predicted, since there are **no official lists of RD nor centralised patient registries in Greece** (see reports from Working Groups 1 and 2 and 5)

SUGGESTION: the establishment of interim "Centres of Coordination" for RD patient care, as predecessors of CoE.

Proposal criteria for "Centres of Coordination" in Greece

1. Locations

To meet the care needs of both childhood and adult RD patients, ideally the "Centres of Coordination" should be in large public general hospitals with both pediatric and adult clinics.

To cover as much as possible all regions throughout Greece it is proposed to establish CoC's in Athens and Thessaloniki.

In **Athens** it is proposed to create 2 separate “Centres of Coordination”, one for children (at the existing largest children’s hospital complex in Athens, (Aghia Sophia- Aglaia Kiriakou) and one for adults (at one of the existing large general Hospitals in **Athens**, e.g.Evangelismos, Hippocratio, General-State-Hospital – to be selected).

In **Northern Greece** the creation of a single “Centre of Coordination” is proposed at a large general Hospital which offers both Pediatric and adult clinics i.e. Papageorgiou Hospital **Thessalonica**.

The advantage of housing these coordinating centres at big hospitals becomes obvious from the main actions outlined below a “Centres of Coordination” for children with RD, as follows:

2. Aims of the “Centres of Coordination” for RD patient care

To facilitate the coordination of all activities required for COMPLETE care and treatment of patients with RD. The coordination will also include the clinical diagnostic procedures and long-term patient follow-up.

3. Main Activities of the “Centres of Coordination” for RD patient care

- i. Creation of a database in order to register patients with RD, following all rules and regulation in order to comply with confidentiality/of personal data. Efforts will be made to ensure that this patient registry will be co-ordinated with all other patients registries throughout Greece to a) preclude overlap of cases recorded and b) support National epidemiological information on RDs.
- ii. In addition appropriate levels of access to the database will be given to e.g. the Ministry of Health, scientific research teams, official patients associations e.t.c.
- iii. To facilitate close cooperation with all hospital children’s units in the Attica region, as well as with any existing units and departments that offer services to patients with RD.
- iv. Development of specialised information on RD (e.g. diagnostic and care protocols, guidelines for correct medical practice), and distribution to health care professionals.
- v. Development of printed information on RD and distribution to families of patients with RD, following examples from other EU countries experienced in RDs.
- vi. Develop of Greek language webpage with information to help patients and health care professionals (eg translate Orphanet webpage – see Working Group 2).
- vii. Communication with hospital units of the region in order to exchange information and printed matter.
- viii. Close cooperation with RD patient organisations (e.g. PESPA) in order to exchange information, printed matter and electronic information.
- ix. Aim at involving help from volunteers from the patient’s societies or associations to support better services for patients with RD.
- x. Facilitate families of patients with RD by providing them with information related to health care services in their locality for the disease that their child suffers from.
- xi. Help families of patients with RD by providing them with information related to their access to medication, sickness/invalid benefits, committee’s e.t.c.
- xii. Networking with European centres of reference for RD with the aim to include Greece’s participation in CoE networks, registry networks, therapeutics protocols e.t.c.
- xiii. Facilitate and support research for RD in Greece (see Working Group 3).

Additionally, it is proposed that the Ministry of Health and Social Solidarity will support all clinics/departments ALREADY EXPERIENCED in the caring of RD patients to work towards:

- a) Fulfilling the Eurordis criteria for CoE’s
- b) Achieving recognition of CoE status by the Greek Ministry of Health and Social Solidarity.
- c) Networking with other centres specialised in RD care in Greece as well as throughout Europe

Requirements for a “Centre of Coordination” for Children with RD in ATHENS, the following INITIAL requirements are foreseen

Core Personnel

- 1. 2 general pediatricians, full-time employment.
- 2. 2 secretaries, full time employment.

In future, depending on the needs, the Medical and/or Secretarial Personal may be increased, with possible extension in to social services, psychologists, health visitors e.t.c.

Infrastructure

- 1. Establishment of an office, which will be located, in preference, in the children’s Hospital (Aghia Sophia/Aglaia Kyriakou).
- 2. Relevant technical infrastructure (computers, telephones, fax, internet connection, software, website ,e.t.c).
- 3. Access to information to help to identify clinics and clinicians, to allow rapid referral of RD patients and address all their care needs (from diagnosis, to routine and emergency treatment, long-term follow-up etc)

Requirements for a “Centre of Coordination” for Adults with RD in Athens

The same principle requirements relative to personnel and infrastructure as for CoC for Children with RD in Athens; the location is to be decided.

Requirements for a “Centre of Coordination” for Children and Adults in N. Greece, proposed at the Papageorgiou Hospital, Thesalonika.

The same principle requirements relative to personnel and infrastructure as for CoC for Children with RD in Athens.

- In addition the Clinics and Departments that are not fully adhere to the standards for CoE’s defined by the Council Recommendations to achieve these standards , including the capability to participate in national or regional centres of expertise into European reference networks

Slide 12: EUROPLAN INDICATORS FOR DIAGNOSIS

ACTIONS	INDICATORS	TYPE	ANSWERS
Develop Screening Policies	Number of diseases included in the neonatal screening programmeme	Outcomes	Number of diseases = FOUR UNDER THE AUSPICES OF THE NATIONAL HEALTH SYSTEM = PKU,

			G6PD, HYPOTHYROIDISM, GALACTOSEMIA,
	Number of diseases included in the neonatal screening programme properly assessed	Outcomes	NONE
Ensure quality of RD diagnosis laboratory	Existence of a public directory/ies of both genetic tests on Rare Diseases	Process	• NO- Under discussion FOLLOWING THE EUROPLAN MEETING
	Proportion of laboratories having at least one diagnostic test validated by an external quality control	Outcomes	NONE UNDER THE NATIONAL HEALTH SYSTEM (SOME WITHIN THE PRIVATE SECTOR)

Based on slide 12, it can be concluded that in Greece, with respect to the **EUROPLAN INDICATORS FOR DIAGNOSIS**

- There is no public directory(ies) of genetic tests on Rare Diseases
- It is not known what proportion of laboratories, if any, have at least one diagnostic test validated by an external quality control

SUGGESTION: for RD patient DIAGNOSIS, it is proposed that the Ministry of Health and Social Solidarity support the following actions:

1. Co-ordinate a survey to identify all public hospital laboratories providing genetic tests in order to define the numbers of laboratories and also for which RD genetic tests exist in Greece.
2. Support all laboratories already providing genetic tests to achieve Accreditation (ISO 15189) and participate in annual External Quality Assessment schemes run by e.g. NEQAS, EMQN, Eurogentest etc
3. Establish costs for RD genetic tests and include them within the cover offered by the public health insurance schemes.

WORKSHOP 5: Experience and expertise pooling on RD at a European Level - Patient Associations
Empowerment - Support Sustainability

Facilitators (Chairs): M. Lambrou, V. Biliou

Present at the workshop with active participation were

- Ms. I. Tassopoulos, Ministry of Health and Social Solidarity
- Ms Th. Stavrou, Ministry of Health and Social Solidarity
- Ms D. Delga, Nurse MSc, Attikon Hospital, Member of the Greek Alliance for Rare Diseases
- Ms A. Georgadi Representative of the Hellenic Organisation of | | patients with Keratokono

The workshop was attended by numerous members of patients associations, patients or relatives and students.

EUROPLAN INDICATORS ON PATIENT EMPOWERMENT

ACTIONS	INDICATORS	TYPE	ANSWERS (GREECE)
Promoting the existence of a RD patients' organisation that represents all RD patients' associations	Number of umbrella organisations specific on rare diseases	Process	Existing only one organisation
	Having a directory of RD Patients' organisations	Process	Yes. Listed with all other associations by the Ministry of Health
	Number of RD patients' associations	Outcomes	18 associations, members of PESPA
	Number of diseases covered by patients' associations	Outcomes	20-30 diseases

ACTIONS	INDICATORS	TYPE	ANSWERS (GREECE)
Patients' Organisations involvement in decisions affecting RD	Permanent and official patients' representatives in plan development, monitoring and assessment	Process	Representation has not been yet clearly defined, but the Secretary General of the Ministry of Health has committed verbally to establish institutional representation in all RD areas.
	Participation of patients' organisations in the development of RD research strategies	Process	No

ACTIONS	INDICATORS	TYPE	ANSWERS (GREECE)
Support the activities performed by including patient organisations, such as : - Awareness raising - Capacity building and training - Exchange of information and best practices - Networking - Outreach very isolated patients	Resource (funding) provided for supporting the activities performed by patient organisations	Outcomes	Not exist
	Support to sustainable activities to empower patients, as stated before	Outcomes	Not even a plan exists

ACTIONS	INDICATORS	TYPE	ANSWERS (GREECE)
Building - supporting the existence of comprehensive help line for patients	Availability of Help line for RD patients	Process	No formal decisions have been taken

ACTIONS	INDICATORS	TYPE	ANSWERS (GREECE)
Compensating disabilities caused by rare diseases	Existence of official programmes supporting patients and families with disabilities	Process	Not exclusively for RD.
	Existence of an official directory of social resources for patients with disabilities	Process	Yes, but not specialised on RD

ACTIONS	INDICATORS	TYPE	ANSWERS (GREECE)
Supporting Rehabilitation programmes for RD patients	Existence of programmes to support rehabilitation of RD patients.	Process	Yes, and they include financial support, however not specialised for RD.

ACTIONS	INDICATORS	TYPE	ANSWERS (GREECE)
Supporting social services aimed at rare disease patients and their families.	Existence of national schemes promoting access of RD patients and their families to Respite Care Services	Process	No
	Existence of public schemes supporting Therapeutic Recreational Programmes	Process	No
	Existence of programmes to support integration of RD patients in their daily lives	Process	No

RECOMMENDATIONS

PATIENTS AND THEIR REPRESENTATIVES PARTICIPATION TO DECISION-MAKING

How can patients participate and in particular get empowered?

- Representation by PESPA at institutional level in the NP elaboration.
- Inventorying of RD and RD patients in our country and PESPA-driven management of this directory in collaboration with the Ministry of Health.
- Establishment of coordination inter-sectoral committee involving all concerned Ministries: Health, Education, Labor - Employment and Development, exclusively on Rare Diseases.
- Establishment of an RD Division within the Ministry of Health.
- Operation of a single assessment committee on RD, composed by doctors of various specialisations.
- Single policy of insurance funds.

-
- Creation and operation of Centres of Expertise/European Reference Networks.
 - Update on recent clinical researches by means of representatives participating therein.
 - Co-financing of patients' associations by state and private funds, allocated equally by means of a dedicated monitoring team made up of representatives from the Ministry of Health, PESPA and SFEE (Hellenic Association of Pharmaceutical Companies).
 - On-going update of classification procedure (ICD, Orphanet etc).
 - Building a telephone help line.

Finally, given that even small-scale actions may facilitate patients:

- Development of medical background card
 - Provision of treatment at the Hospital where the patient's file belongs (not at a hospital in full-day shift)
 - Provision of assistance at home for families with RD.
 - Incentives for training courses addressed to doctors and therapists in RD.
-

IV Summary and Conclusion of the Final Report

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

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

CONCLUSIONS:

The Greek National Plan exists as a (preliminary) written document with a fairly clear structure (requiring some modification). HOWEVER it has no legal binding up to date, which precludes a monitoring and evaluating system

Theme 2 - Definition, codification and inventorying of RD

Sub-Themes

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

CONCLUSIONS:

There is no official definition of RD in Greece. Recently the ICD10 classification was officially adopted, but is not yet widely used. There are some non-state inventories and registries existing in Greece, but there is no state funding, support or co-ordination.

1.2. Information and training

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

CONCLUSIONS:

There are no official information sources, no web pages, no official Helplines existing in Greece, neither for healthcare professionals nor for the public/patients. There are some initiatives, for example from PESPA, but there is no state financial support.

With respect to adequate training for healthcare professionals, this is very limited and inconsistent. For example, most importantly there is no officially recognized specialisation on Clinical and Laboratory Genetics.

Theme 3 - Research on RD

Sub-Themes

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

CONCLUSIONS

Funding policy and funding management for research in Greece in RD (amongst all other topics) is almost non-existent.

Theme 4 - Standards of care for RDs - Centres of Expertise (CoE)/ European Reference Networks (ERN)

Sub-Themes

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

CONCLUSIONS

There is no policy for establishing CoE's at the national/regional level. There are DEPARTMENTS/CLINICS (not fully CoE's) for some RD's but none of the Greek CoE's adhere to the standards defined by the Council Recommendations. Finally there is no participation of national or regional centres of expertise into European reference networks. The needs (numbers and location) of CoE's cannot be easily predicted, since there are no official lists of RD nor centralized patient registries in Greece, in which case the EUROPLAN conference recommends the formation of Centres of Coordination to precede CoE's.

4.1. Orphan Drugs (OD)

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

CONCLUSIONS

There is no official policy for OD in Greece. Access of RD patients to orphan drugs, pricing and reimbursement is sporadic and often inconsistent.

Theme 5 - Patient Empowerment and Specialised Services

Sub-Themes

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

CONCLUSIONS

Despite some impressive initiatives and achievements by patient organisations, there is presently minimal official involvement of patients and their representatives in decision-making processes in the field of RDs, nor easily available support to the activities performed by patient organisations, nor specialised social services, nor helplines.

Horizontal Themes

Theme 6 – Sustainability

CONCLUSIONS

On the crucial subject of sustainability for the National Plan and overall policy for RD, the answer is that there is NO current budget and NO allocated source of funding in Greece, presenting a serious obstacle for the prospect for sustainability.

Theme 7 - Gathering expertise at the EU level

CONCLUSIONS

This theme was not widely relevant under the present circumstances in Greece and was not widely discussed.

SUGGESTED PRIORITIES

A Greek National Plan for RD **does exist**. However it needs a legal/policy framework and needs to become legally binding.

There have been substantial efforts made by PESPA and many of its member patient-associations, as well as other patient associations in Greece, for example initiating registries, some improvements in a few areas for patient care, patient networking and unofficial “helplines”, usually supported by private donations, volunteers and some donations by pharmaceutical companies. However, without the support of the state any effort remain fragmented and uncoordinated, leaving many gaps in the needs of RD patients and their families on many, many levels.

In the light of the current severe economic situation in Greece, the Greek Europlan meeting suggests the following initial priorities which have no or minimal financial investment, which will help the future goal towards a fuller implementation of a National Plan for RD as follows:

- Make the National Plan legally binding for all the official stakeholders, notably the Ministry of Health and Social Solidarity and the Ministry of Labour and Social Affairs, as well as the Ministry of Education and Life Long Learning and the General Secretariat for Research and Technology
- Create an external committee under the auspices of the Ministry of Health and Social Solidarity to include representatives of all major Stakeholders in RD. The role of the committee will be to monitor, evaluate and deliver opinion on the progress of the Greek NP for RD.
- Co-ordination by the Ministry of Health and Social Solidarity of Registries for RD and RD patients, such as that already initiated by PESPA and some hospital clinics, which will be officially recognised
- Promote improved care for RD patients through initially creating “Centres of Coordination” for RD patient care, as predecessors of CoE (see Workshop 4), as well as support the current Clinics and Departments that exist in Greece to become full CoE’s according to the criteria recommended by the Council of Europe and EURORDIS
- Promote timely diagnosis of RD patients by mapping the current diagnostic centres (most importantly genetics laboratories), fortifying their capabilities and supporting their accreditation (ISO 15189)
- Suggest all the Orphan Drugs to be excluded from the general legislation from the common drugs (as it is in the present situation) and to have a privilege status
- Support the development of awareness, information and education both for health professionals and the general public (Webpages, helplines, seminars etc)
- Direct the General Secretariat for Research and Technology to take a more proactive role, to support research for RD in Greece

An overall assessment of the usefulness of the EUROPLAN Recommendations for the advancement of a national strategy in the country.

In Greece, where there is no legal national strategy in existence, it is obvious that the EUROPLAN Recommendations will be extremely useful. The EUROPLAN Recommendations will help to put on track a basic national strategy and thus help the cause for RD and RD patients in Greece.

The transferability of the EUROPLAN recommendations in your country.

The main difficulties in the transferability of the EUROPLAN recommendations in Greece are anticipated to be a) the financing of any actions and b) their promotion and implementation by the political leadership and Greek government. However, of note, through the EUROPLAN meeting we achieved, for the first time, a wide participation of major stake-holders, including key Ministries and government departments, and a very positive response by them and all the other stake-holders, substantially increasing the awareness of the Greek general public about RDs. In conclusion, everyone involved has begun to understand the necessity of the existence of a National Plan in Greece.

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