

**2012 REPORT ON THE STATE OF THE ART  
OF RARE DISEASE ACTIVITIES IN EUROPE  
OF THE  
EUROPEAN UNION COMMITTEE OF EXPERTS  
ON RARE DISEASES**



**STATE OF THE ART OF RARE DISEASE ACTIVITIES IN  
GREECE**

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More information on the European Union Committee of Experts on Rare Diseases can be found at [www.eucerd.eu](http://www.eucerd.eu).

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## ACRONYMS

### General

CAT - Committee for Advanced Therapies at EMA  
CHMP - Committee for Medicinal Products for Human Use at EMA  
COMP - Committee on Orphan Medicinal Products at EMA  
DG - Directorate General  
DG Enterprise - European Commission Directorate General Enterprise and Industry  
DG Research - European Commission Directorate General Research  
DG Sanco - European Commission Directorate General Health and Consumers  
EC - European Commission  
ECRD - European Conference on Rare Diseases  
EEA - European Economic Area (Iceland, Switzerland, Norway)  
EMA - European Medicines Agency  
ERN - European reference network  
EU - European Union  
EUCERD - European Union Committee of Experts on Rare Diseases  
EUROCAT - European surveillance of congenital anomalies  
EUROPLAN - European Project for Rare Diseases National Plans Development  
EURORDIS - European Organisation for Rare Diseases  
FDA - US Food and Drug Administration  
HLG - High Level Group for Health Services and Medical Care  
HTA - Health Technology Assessment  
IRDiRC – International Rare Diseases Research Consortium  
JA - Joint Action  
MA - Market Authorisation  
MoH - Ministry of Health  
MS - Member State  
NBS - New born screening  
NCA - National Competent Authorities  
NHS - National Health System  
PDCO - Paediatric Committee at EMA  
RDTF - EC Rare Disease Task Force  
WG - Working Group  
WHO - World Health Organization

### Pilot European Reference Networks

Dyscerne - European network of centres of expertise for dysmorphology  
ECORN-CF - European centres of reference network for cystic fibrosis  
Paediatric Hodgkin Lymphoma Network - Europe-wide organisation of quality controlled treatment  
NEUROPED - European network of reference for rare paediatric neurological diseases  
EUROHISTIONET - A reference network for Langerhans cell histiocytosis and associated syndrome in EU)  
TAG - Together Against Genodermatoses – improving healthcare and social support for patients and families affected by severe genodermatoses  
PAAIR - Patients' Association and Alpha-1 International Registry Network  
EPNET - European Porphyrin Network - providing better healthcare for patients and their families  
EN-RBD -European Network of Rare Bleeding Disorders  
CARE-NMD -Dissemination and Implementation of the Standards of Care for Duchenne Muscular Dystrophy in Europe project  
ENERCA - European network for rare and congenital anaemia – Stage 3

# GENERAL INTRODUCTION TO THE REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE OF THE EUROPEAN UNION COMMITTEE OF EXPERTS ON RARE DISEASES

The 2012 Report on the State of the Art of Rare Disease Activities in Europe was produced by the Scientific Secretariat of the European Union Committee of Experts on Rare Diseases (EUCERD), through the EUCERD Joint Action: Working for Rare Diseases (N° 2011 22 01), which covers a three year period (March 2012 – February 2015).

The report aims to provide an informative and descriptive overview of rare disease activities at European Union (EU) and Member State (MS) level in the field of rare diseases and orphan medicinal products up to the end of 2011. A range of stakeholders in each Member State/country have been consulted during the elaboration of the report, which has been validated as an accurate representation of activities at national level, to the best of their knowledge, by the Member State/country representatives of the European Union Committee of Experts on Rare Diseases. The reader, however, should bear in mind that the information provided is not exhaustive and is not an official position of either the European Commission, its Agencies or national health authorities.

The report is split into five parts:

Part I: Overview of rare disease activities in Europe

Part II: Key developments in the field of rare diseases in 2011

Part III: European Commission activities in the field of rare diseases

Part IV: European Medicines Agency activities and other European activities in the field of rare diseases

Part V: Activities in EU Member States and other European countries in the field of rare diseases

Each part contains a description of the methodology, sources and validation process of the entire report, and concludes with a selected bibliography and list of persons having contributed to the report.

The present document contains the information from Parts II and V of the report concerning Greece. A list of contributors to the report and selected sources are in annex of this document. For more information about the elaboration and validation procedure for the report, please refer to the general introduction of the main report<sup>1</sup>.

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<sup>1</sup> <http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArRDActivities.pdf>

# RARE DISEASE ACTIVITIES IN GREECE

## Definition of a rare disease

Stakeholders in Greece generally accept the European Regulation on Orphan Medicinal Products definition of a prevalence of no more than 5 in 10'000 individuals; however no official definition has been proposed or accepted.

Clinician stakeholders, for the precise evaluation of the burden of a RD on public health, propose the use of supplementary indices such as the annual rate of births of affected new-borns for genetic diseases with short survival and/or population specificity and age group prevalence for diseases prevailing either in children and adolescents or in adults and old patients.

## National plan/strategy for rare diseases and related actions

A commission composed of government officials, health professionals and patient representatives was formed in 2007 following requests by the Greek Alliance for Rare Diseases (PESPA) to help draft the Greek National Plan for Rare Diseases. PESPA members presented a draft to the Committee, which was then modified by officials of the Greek Ministry of Health and Social Solidarity to the format of the current Plan. An outline for this National Plan of Action for Rare Diseases (to run over the period 2008-2012<sup>2</sup>) was presented by the Greek Minister for Health in February 2008: this document identified and outlined eight strategic priorities:

- Acknowledgement of the specificity of rare diseases (registration on the list of chronic long-term disorders),
- Increase the knowledge of the epidemiology of rare diseases and establish a National Registry of Rare Disorders,
- Develop information for patients, health professionals and the general public concerning rare diseases,
- Upgrade services for diagnosis, therapy and rehabilitation of rare diseases patients (training for health care professionals to improve diagnosis and access to quality health care),
- Organise screening and access to diagnostic tests,
- Promote research and innovation regarding rare diseases and specifically effective new therapies,
- Respond to the specific needs of people living with rare diseases,
- Generation of an integrated platform for action in the field of rare diseases at a national level and the development of European partnerships.

The provisions of the Greek National Plan for Rare Diseases (2008-2012) were discussed in detail during the Greek National Conference on Rare Diseases<sup>3</sup> co-organised by the Greek Alliance for Rare Diseases (PESPA) and EURORDIS, was held in Athens (26-27 November 2010) in the framework of the Europlan project. The priorities listed during the conference included: the need for a legal framework of the National plan and a steering committee, the need for a policy to establish centres of expertise, the need to complete the map of diagnostic laboratories, the need to establish universal access to orphan medicinal products, the need to officially recognise the specialty of Clinical and Laboratory Genetics, the need to fully reimburse diagnostic tests (including molecular diagnosis), the need for therapy and rehabilitation, the need for price adjustment of orphan medicinal products in order to continue to be available in the Greek market, the need for funding of rare disease research and the need for more information on rare diseases in Greece.

Although an initial estimate for the budget required was made, no funding has been officially allocated to the National Plan of Action for Rare Diseases, and none of the eight strategic priority actions have yet started. As of yet, there is no legal framework for the Plan so no progress has been made.

In fact, most of the objectives of the proposed nation plan of action for RD are or could be incorporated in the existing structure and function of Greek national health system (GNHS). Implementation of strategic priorities for RD is coordinated by the Ministry of Health and mainly by the Hellenic Centre of Disease Control and Prevention (KEELPNO) jointly to those of common diseases.

In 2010 the new Scientific Committee for Rare Diseases appointed by KEELPNO organised (with minimum funding) a program to evaluate the implementation and efficacy of the main strategic priorities defined in the scope of the Europlan project that are incorporated in the Greek NHS. The program started in

<sup>2</sup> [http://www.ygeianet.gov.gr/HealthMapUploads/Files/SPANIES\\_PATHISEIS\\_TELIKO\\_LOW.pdf](http://www.ygeianet.gov.gr/HealthMapUploads/Files/SPANIES_PATHISEIS_TELIKO_LOW.pdf)

<sup>3</sup> [http://download.EURORDIS.org/europlan/2\\_EUROPLAN\\_Guidance\\_Documents\\_for\\_the\\_National\\_Conference/FINAL%20Report%20Greece%20EUROPLAN%202010.pdf](http://download.EURORDIS.org/europlan/2_EUROPLAN_Guidance_Documents_for_the_National_Conference/FINAL%20Report%20Greece%20EUROPLAN%202010.pdf) (accessed 28/01/2011)

2011 with the following main objectives: i) to identify the expertise centres (CEs) involved in the care of patients with rare diseases, describe their structures and activities, evaluate the quality of health services they provide; ii) to investigate measures to upgrade the quality of the provided care; and iii) to introduce applicable strategies defined by Europlan, which are not included in the activities of expertise centres. Preliminary data from the survey are reported.

Health services already provided by Greek National Health System (NHS) can be classified in two main types. The first covers children and adolescents aged 0-19 years old and the second adults and older patients (over 20 years old). In the first type, primary health care is provided by family pediatricians and in the second by internists and physicians with basic specialties in Internal Medicine. For patients with life threatening disease and chronic debilitating diseases, such as rare diseases, hospital care is provided in two levels: for children and adolescents in paediatric clinics of rural hospitals for common and non severe diseases and University Departments of Paediatrics and Children Hospitals operating divisions of pediatric specialties, subspecialties, special reference units and basic and research laboratories. For adults and older patients hospital care is provided in general regional hospitals and in University Hospitals and Referral General Hospitals with departments, divisions and special referral units, supported by routine and specialised research laboratories.

In both branches of Greece's NHS the care of patients with rare diseases is multidisciplinary and homogeneous to that of patients with relative common diseases of similar pathogenesis.

Special units in Research Institutes and private laboratories contribute to pre and post natal diagnosis for a number of rare diseases.

Other national actions related to rare diseases include a National Programme for haemoglobinopathies (covering thalassaemia and sickle cell diseases) which includes carrier detection, prenatal diagnosis, patient diagnosis and therapy. This Programme is split into two areas, a prevention programme organised in the late 1970s and implemented in the 1980s, and a treatment programme implemented gradually in the 1970s.

### **Centres of expertise**

Within the national health system special units providing expert services for groups of a limited number of diseases including specific rare diseases/groups of rare diseases have been organised during the past few decades. The Hellenic Centre for Disease Control and Prevention (KEELPNO) started to collect data on the nature and activities of these units and aims to complete collection in 2012. Greece is working to provide better access to treatment for rare disorders, including the accreditation and creation of centres of expertise for rare disorders.

By the end of the 2011 the multidisciplinary centres of expertise (basically day care clinics) for the management of thalassaemia, cystic fibrosis, neuromuscular disorders, hereditary bleeding diseases and primary immunodeficiencies were identified and evaluated. Identified centres of expertise include:

- Thalassaemia: 15 centres (Two follow more than 400 patients, four 150-200 patients and nine 70-150 patients)
- Cystic Fibrosis: 3 centres (2 for children and adolescents and 1 for adults)
- Neuromuscular Diseases: 6 centres (2 for children and adolescents, 2 for adults and 2 for all ages)
- Hereditary Bleeding disorders: 4 centres (3 for adults and 1 for children and adolescent) and
- Primary immunodeficiency: 2 centres for children and adolescents

These centres of expertise collaborate with the following expert laboratories.

- The Laboratory of Medical Genetics of the University of Athens, "Agia Sophia" Children Hospital, for the molecular pre and post natal diagnosis of thalassaemia, cystic fibrosis, and neuromuscular diseases; it also serves as national reference laboratory for a number of genetic diseases.
- The national Thalassaemia Prenatal Diagnosis Centre covering 60-70% of prenatal diagnosis of thalassaemia.
- The laboratories of a) Genetic Neurological Diseases and b) Muscle pathology of the Department of Neurology, Athens University serving also as the National Reference Laboratory.

According to the evaluation of new Scientific Committee for Rare Diseases, certain centres of expertise involved in the management of these five diseases/groups of rare diseases, fulfil the EUCERD Recommendations on Quality Criteria for National Centres of Expertise for Rare Disease in Member States.

### **Pilot European Reference Networks**

Greece participates, or has participated, in the following European Reference Networks for rare diseases: Dyscerne, ECORN CF, ENERCA, EUROHISTIONET, EN-RBD and TAG.

### **Registries**

There is currently no national registry for rare diseases in Greece. One of the main tasks of the new Scientific Committee, mainly consisting of clinicians caring for patients with rare diseases, is to set up a national registry, according to the international standards.

In the absence of a national registry for rare diseases, scientific societies covering rare diseases, appointed working groups which, in collaboration with respective centres of expertise and patients organisations, have created registries for a number of rare diseases. Up to now the Scientific Committee on rare diseases reviewed and evaluated data collected and registered for thalassemia, cystic fibrosis, neuromuscular disorders, hereditary bleeding diseases and primary immunodeficiencies.

These registries do not receive national financing. Greek teams contribute to the European registries EUROCARE CF and EIMD.

### **Neonatal screening policy**

Neonatal screening covering around 98% of neonates in Greece and is provided by the Institute of Child Health, Athens, for congenital hypothyroidism, phenylketonuria, G6PD deficiency and galactosaemia. Recently, the neonatal screening is expanding in the private sector covering a number of inborn errors of metabolism, cystic fibrosis, adrenal hyperplasia and biotin deficiency, as well as screening for the early diagnosis and treatment of congenital deafness. Data on the extended neonatal screening program in regard to efficacy and neonatal population coverage are not yet available. The policy of neonatal screening was not further developed in 2011 in the Greek NHS.

### **Genetic testing**

Genetic testing is carried out in different laboratories specialising in the diagnosis of different rare diseases. There are neither official reference laboratories nor guidelines. Tests are reimbursed through insurance (public and private) schemes and genetic testing is possible abroad. Genetic tests provided by special laboratories of the Greek NHS covering centres of expertise are official reference laboratories and fulfil European guidelines.

Diagnostic tests are registered as available in Greece for 104 genes and an estimated 160 diseases in the Orphanet database<sup>4</sup>.

### **National alliances of patient organisations and patient representation**

PESPA (the Greek Alliance for Rare Diseases) is an umbrella non-profit organisation established in 2003, by health professionals and presidents of 20 rare disease patient associations (national or regional) with the help of EURORDIS.

In Greece, numerous national (Pan-Hellenic) patient organisations exist mainly for the more prevalent rare diseases. They have their own websites and are members of the relative International and European federations. The Hellenic Thalassemia Federation, the Association of Patients with Haemophilia, the MDA Hellas and the Society of Cystic Fibrosis are some of the indicative examples. In addition to national, there is also a considerable number of patients and parent-patient associations for rare diseases that autonomously organise their activities and conferences.

Alliances of friends of patients with rare diseases or group of rare diseases also exist. Few of them as the association of Friends of Children with Cancer "ELPIDA" and "FLOGA", MDA Hellas, Friends Association of Children With Chronic Rheumatoid Diseases are amongst the organisations which provide funding for the organisation and functioning of centres of expertise and expert units. (i.e., ELPIDA donated to "Aghia Sophia" Children's Hospital a modern and well-equipped Unit, "The Paediatric Oncology Unit. Marianna Vardinoyiannis –ELPIDA", for the multidisciplinary care of children and adolescents with cancer: the Unit with a capacity of 126 beds started operating in January 2011).

There are currently no public funding schemes to support patient organisations activities in Greece.

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<sup>4</sup> Information extracted from the Orphanet database (September 2011).

## **Sources of information on rare diseases and national help lines**

### ***Orphanet activities in Greece***

Since 2004 there is a dedicated Orphanet team in Greece, currently hosted by the Institute of Child Health's Department of Genetics (from 2009). The team was designated as the Greek national Orphanet team by the Ministry for Health and Social Solidarity in 2010. This team is in charge of collecting data on rare disease related services (specialised clinics, medical laboratories, ongoing research, registries, clinical trials and patient organisations) in their country for entry into the Orphanet database. Greece participates in the Orphanet Joint Action and allocated the amount of €50'000 for the translation of the rare disease encyclopedia for experts in the Orphanet website in Greek. The Orphanet Greece national website in Greek was launched in 2011<sup>5</sup>.

### ***Official information centre for rare diseases***

There is no official information centre for rare diseases in Greece apart from Orphanet

### ***Help line***

There is no official help line for rare disease information in Greece; some services, mainly voluntarily are offered by PESPA members who provide psychological support and general information.

### ***Other sources of information***

The PESPA website offers information on rare diseases and a list of some rare diseases in Greek. Every specialised unit produces information leaflets for the disease(s) of its expertise.

### ***Good practice guidelines***

Some scientific societies have published or renewed guidelines for specific rare diseases in local professional journals. All centres with expertise in rare diseases follow the international guidelines. There are national guidelines for thalassaemia.

### ***Training and education initiatives***

Rare diseases is a topic included in the general curriculum of undergraduate and postgraduate studies of Medical Schools in Greece and is basically addressed in the training for specialities and sub-specialities in Paediatrics and Internal Medicine. Scientific societies also organise courses and workshops in order to educate physicians, nurses and students on specific rare diseases.

### ***National rare disease events in 2011***

National rare disease events are either events for the patients and the public to disseminate basic information on clinical signs and treatment of rare diseases or scientific events addressed to expert physicians and scientists.

The National Association of Rare Diseases (PESPA) announced Rare Disease Day 2011, which was held under the auspices of President of the Republic Karolos Papoulias. A speech was given by Professor of Cancer Prevention and Professor of Epidemiology at the University of Harvard, Mr. Dimitrios Trichopoulos, Member of the Greek Academy of Sciences, and a retrospective exhibition of paintings by Katerina Lambrou, at the Cultural Center of Athens with donations going to the National Association of Rare Disorders and the Greek Society of Tuberous Sclerosis. PESPA organised a national conference on "Autoimmune Diseases: Today and Tomorrow" on 25-26 November 2011.

Events related to rare diseases are organised by most of the national patients organisations on the day devoted to the particular disease. National patient organisations organised during 2011 their annual meetings, conferences or congresses and discussed with specialists the recent advances in management and other topics related to the interest of the societies.

A considerable number of educational events on rare diseases aimed at physicians and the scientific community are organised each year by university departments, research institutes, expertise centres and scientific speciality societies in the form of meetings, workshops, lectures, training courses etc. Scientific events are usually focused on a single rare diseases or a number of homogeneous rare diseases.

For the more interesting educational events, proceedings are published.

Sponsorship for these events comes mainly by pharmaceutical companies and occasionally from patient organisations

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<sup>5</sup> <http://www.orpha.net/national/GR-EL/index/%CE%B1%CF%81%CF%87%CE%B9%CE%BA%CE%AE-%CF%83%CE%B5%CE%BB%CE%AF%CE%B4%CE%B1/>

### **Hosted rare disease events in 2011**

No rare disease related events hosted by Greece were announced in *OrphaNews Europe* in 2011.

### **Research activities and E-Rare partnership**

#### ***National research activities***

The General Secretariat for Research and Technology (Ministry of Education, Life Long Learning and Religious Affairs) has been funding research projects coping with all aspects of rare diseases (rare cancers included) in the framework of “biomedical research”. However, there are no specific programmes for rare disease research and thus, it is very difficult to determine the funding allocated to rare diseases research only.

#### ***Participation in European research projects***

Greece participates, or has participated, in European rare disease research projects including: BIOMALPAR, BNE, EPINOSTICS, EUROGLYCANET, EUROPEAN LEUKEMIA NET, EVI-GENORET, GEN2PHEN, GETHERTHAL, HDLomics, IPF-AE, ITHANET, MYASTAID, NEUROPRION and TRANSPOSMART.

#### ***E-Rare***

Greece, through the General Secretariat for Research and Technology (GSRT), participated in the 2<sup>nd</sup> Joint Call of E-Rare-1. In this context, one project coordinated by a Greek team (with a total funding of around €140,000) was approved following peer-review evaluation and is in progress. Greece currently participates in E-Rare-2, and is represented by two institutions: GSRT and the Hellenic Center for Disease Control and Prevention (KEELPNO). GSRT participated in the 3<sup>rd</sup> Joint Transnational Call launched in 2011 with the amount of €200 000. Two Greek teams were approved for funding after the evaluation of the call.

#### ***IRDiRC***

Greek funding agencies are not currently committed members of the IRDiRC. Nevertheless the possibility to join IRDiRC through E-Rare-2 is under consideration.

### **Orphan medicinal products**

The Greek National Organisation for Medicines (EOF<sup>6</sup>) ensures the public health and safety of all medicinal products, including orphan medicinal products. Orphan medicinal products that are not found on the market in Greece are imported by the Greek Institute of Pharmaceutical Research and Technology, and transferred to the patients requiring these drugs.

#### ***Orphan medicinal product committee***

No specific information reported.

#### ***Orphan medicinal product incentives***

No specific information reported.

#### ***Orphan medicinal product market availability situation***

The following orphan medicinal products are available in Greece: Afinitor, Aldurazyme, Arzerra, Atriance, Busilvex, Carbaglu, Cayston, Celpen, Cystadene, Diacomit, Elaprase, Esbriet, Evoltra, Exjade, Fabrazyme, Firazyne, Firdase, Flioland, Glivec, Ilaris, Increlex, Inovelon, Kuvan, Litak, Lysodren, Mepact, Mozobil, Myozyme, Naglazyme, Nexavar, Nplate, Onseno, Orfadin, Pedeo, Peyona, PhotoBarrl, Prialt, Replagal, Revatio, Revlimid, Revolade, Rinocacept, Regeneron, Savene, Siklos, Soliris, Somavert, Sprycel, Tepadina, Tassigna, Thalidomide, Celgene, Torisel, Tracleer, Trisenox, Ventavis, Vidaza, Volibris, Wilzin, Xagrid, Yondelis, Zavesca. Of the orphan medicinal products authorised by the EMA, 42 are readily available on the Greek market, whereas 11 more are imported by the Greek Institute of Pharmaceutical Research and Technology. The remaining 8 OD could be imported on request by the Greek Institute of Pharmaceutical Research and Technology.

#### ***Orphan medicinal product pricing policy***

No specific information reported.

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<sup>6</sup> <http://www.eof.gr>

### **Orphan medicinal product reimbursement policy**

All antineoplastic and immunomodulatory agents (29 drugs from the relative list of the *Orphanet Report Series: List of Orphan Drugs in Europe, January 2011*<sup>7</sup>), plus one drug for myoclonic epilepsy (Diacomit), one for cystic fibrosis (Cayston), one for beta-thalassemia (Exjade), one for Wilson disease (Wilzin) and two for Gaucher's disease (Vpriv and Zavesca) are 100% reimbursed (a total of 35 drugs). For the rest (26 drugs), some are 100% reimbursed, whereas some are reimbursed at around 90%.

### **Other initiatives to improve access to orphan medicinal products**

There are currently no programmes to facilitate access to Orphan medicinal products. The Greek Alliance PESPA has put in place some awareness raising campaigns concerning orphan medicinal products.

### **Orphan devices**

No specific information reported.

### **Specialised social services**

Patients have limited access to respite care services, but these are not specifically for rare disease patients. The patients sometimes have to financially contribute to these services which are run by national institutions, patient associations and non-governmental organisations. A few therapeutic recreational programmes are available, organised by the same types of organisations, and the patient must also financially contribute to this. Limited help with household chores, psychological support, help with shopping and mobility assistance can be sought by patients with special needs (suffering from rare diseases or not) and are provided by local authorities or NGOs. PESPA provides some psychological support (with the help of professionals who are voluntary) to patients with rare diseases and their families.

## **DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2011 IN GREECE**

### **National plan/strategy for rare diseases and related actions**

The Greek National Plan of Action for Rare Disorders (to run over the period 2008-2012<sup>8</sup>) was adopted in 2008. No funding has been officially allocated to the National Plan of Action for Rare Disorders, and none of the eight strategic priority actions have yet started. As of yet, there is no legal framework for the Plan so no progress has been made.

In fact, most of the objectives of the proposed nation plan of action for RD are or could be incorporated in the existing structure and function of Greek national health system (GNHS). Implementation of strategic priorities for RD is coordinated by the Ministry of Health and mainly by the Hellenic Centre of Disease Control and Prevention (KEELPNO) jointly to those of common diseases.

In 2010 the new Scientific Committee for RD appointed by KEELPNO organised (with minimum funding) a program to evaluate the implementation and efficacy of the main strategic priorities defined in the scope of the Europlan project that are incorporated in the Greek NHS. The program started in 2011 with the following main objectives: i) to identify the expertise centres (CEs) involved in the care of patients with rare diseases, describe their structures and activities, evaluate the quality of health services they provide; ii) to investigate measures to upgrade the quality of the provided care; and iii) to introduce applicable strategies defined by Europlan, which are not included in the activities of expertise centres. Preliminary data from the survey are reported.

<sup>7</sup> [http://www.orpha.net/orphacom/cahiers/docs/GB/list\\_of\\_orphan\\_drugs\\_in\\_europe.pdf](http://www.orpha.net/orphacom/cahiers/docs/GB/list_of_orphan_drugs_in_europe.pdf)

<sup>8</sup> [http://www.ygeianet.gov.gr/HealthMapUploads/Files/SPANIES\\_PATHISEIS\\_TELIKO\\_LOW.pdf](http://www.ygeianet.gov.gr/HealthMapUploads/Files/SPANIES_PATHISEIS_TELIKO_LOW.pdf)

### **Centres of expertise**

By the end of the 2011 the multidisciplinary centres of expertise (basically day care clinics) for the management of thalassemia, cystic fibrosis, neuromuscular disorders, hereditary bleeding diseases and primary immunodeficiencies were identified and evaluated. Identified centres of expertise include:

- Thalassemia: 15 centres (Two follow more than 400 patients, four 150-200 patients and nine 70-150 patients)
- Cystic Fibrosis: 3 centres (2 for children and adolescents and 1 for adults)
- Neuromuscular Diseases: 6 centres (2 for children and adolescents , 2 for adults and 2 for all ages)
- Hereditary Bleeding disorders: 4 centres (3 for adults and 1 for children and adolescent) and
- Primary immunodeficiency: 2 centres for children and adolescents

These centres of expertise collaborate with the follow expert laboratories.

- The Laboratory of Medical Genetics of the University of Athens, “Agia Sophia” Children Hospital, for the molecular pre and post natal diagnosis of thalassemia, cystic fibrosis, and neuromuscular diseases; it also serves as national reference laboratory for a number of genetic diseases.
- The national Thalassemia Prenatal Diagnosis Centre covering 60-70% of prenatal diagnosis of thalassemia.
- The laboratories of a) Genetic Neurological Diseases and b) Muscle pathology of the Department of Neurology, Athens University serving also as the National Reference Laboratory.

According to the evaluation of new Scientific Committee for Rare Diseases, certain centres of expertise involved in the management of these five diseases/groups of rare diseases, fulfil the EUCERD Recommendations on Quality Criteria for National Centres of Expertise for Rare Disease in Member States.

### **Sources of information on rare diseases and national help lines**

#### ***Orphanet activities in Greece***

The Orphanet Greece national website in Greek was launched in 2011<sup>9</sup>.

### **National rare disease events in 2011**

National rare disease events are either events for the patients and the public to disseminate basic information on clinical signs and treatment of rare diseases or scientific events addressed to expert physicians and scientists.

The National Association of Rare Diseases (PESPA) announced Rare Disease Day 2011, which was held under the auspices of President of the Republic Karolos Papoulias. A speech was given by Professor of Cancer Prevention and Professor of Epidemiology at the University of Harvard, Mr. Dimitrios Trichopoulos, Member of the Greek Academy of Sciences, and a retrospective exhibition of paintings by Katerina Lambrou, at the Cultural Center of Athens with donations going to the National Association of Rare Disorders and the Greek Society of Tuberous Sclerosis. PESPA organised a national conference on “Autoimmune Diseases: Today and Tomorrow” on 25-26 November 2011.

Events related to rare diseases are organised by most of the national patients organisations on the day devoted to the particular disease. National patient organisations organised during 2011 their annual meetings, conferences or congresses and discussed with specialists the recent advances in management and other topics related to the interest of the societies.

A considerable number of educational events on rare diseases aimed at physicians and the scientific community are organised each year by university departments, research institutes, expertise centres and scientific speciality societies in the form of meetings, workshops, lectures, training courses etc. Scientific events are usually focused on a single rare diseases or a number of homogeneous rare diseases.

Sponsorship for these events comes mainly by pharmaceutical companies and occasionally from patients organisations.

### **Research activities and E-Rare partnership**

#### ***E-Rare***

Greece currently participates in E-Rare-2, and is represented by two institutions: GSRT and the Hellenic Center for Disease Control and Prevention (KEELPNO). GSRT participated in the 3<sup>rd</sup> Joint Transnational Call launched in

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<sup>9</sup> <http://www.orpha.net/national/GR-EL/index/%CE%B1%CF%81%CF%87%CE%B9%CE%BA%CE%AE-%CF%83%CE%B5%CE%BB%CE%AF%CE%B4%CE%B1/>

2011 with the amount of €200 000. Two Greek teams were approved for funding after the evaluation of the call.

***IRDiRC***

Greek funding agencies are not currently committed members of the IRDiRC. Nevertheless the possibility to join IRDiRC through E-Rare-2 is under consideration.

## LIST OF CONTRIBUTIONS<sup>10</sup>

### Contributions in 2010

Michael Petersen and Sofia Dougzou (*Orphanet Greece, Institute of Child Health, Athens*)  
Marianna Lambrou and Jan Traeger-Synodinos (*PESPA - Greek Alliance for Rare Diseases*)

### Contributions in 2011

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The Scientific Committee for Rare Diseases of the Hellenic Centre for Disease Control and Prevention (KEELPNO)  
The Office for the Hellenic Cancer Registry and Rare Diseases  
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### Contributions in 2012

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Jan Traeger –Synodinos (*PESPA Greek Alliance for Rare Diseases*)

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## SELECTED BIBLIOGRAPHY AND SOURCES<sup>11</sup>

- Proposal for a Greek National Plan for Rare Diseases (2008-2012)  
[http://www.ygeianet.gov.gr/HealthMapUploads/Files/SPANIES\\_PATHISEIS\\_TELIKO\\_LOW.pdf](http://www.ygeianet.gov.gr/HealthMapUploads/Files/SPANIES_PATHISEIS_TELIKO_LOW.pdf)
- Hellenic Centre for Disease Control and Prevention – Office for Rare Diseases  
<http://www.keelpno.gr/el-gr/voσηματαθέματαυγείας/σπάνιανοσηματα.aspx>
- National Organisation for Medicines  
<http://www.eof.gr>
- Institute of Child Health  
<http://www.ich.gr/el/>
- Orphanet Greece national website  
<http://www.orpha.net/national/GR-EL/index/%CE%B1%CF%81%CF%87%CE%B9%CE%BA%CE%AE-%CF%83%CE%B5%CE%BB%CE%AF%CE%B4%CE%B1/>
- Greek Alliance for Rare Diseases – PESPA  
[http://www.pespa.gr/en/pespa\\_en.htm](http://www.pespa.gr/en/pespa_en.htm)
- Europlan Greek National Conference Final Report  
[http://download.EURORDIS.org/europlan/2\\_EUROPLAN\\_Guidance\\_Documents\\_for\\_the\\_National\\_Conference/FINAL%20Report%20Greece%20EUROPLAN%202010.pdf](http://download.EURORDIS.org/europlan/2_EUROPLAN_Guidance_Documents_for_the_National_Conference/FINAL%20Report%20Greece%20EUROPLAN%202010.pdf)

<sup>10</sup> The contributors and validators of the report have contributed information which is accurate to the best of their knowledge. However, readers should take note that the contents of this report are illustrative and not exhaustive.

<sup>11</sup> All websites and documents were last accessed in May 2012. A more detailed list of sources is available in the full report:  
<http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArtRDActivities.pdf>