2014 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE

STATE OF THE ART OF RARE DISEASE ACTIVITIES IN PORTUGAL

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The European Union Committee of Experts on Rare Diseases (EUCERD) was established in 2009 and its mandate ended in 2013. It is replaced from 2014 by the Commission Expert Group on Rare Diseases. The EUCERD Joint Action continues to support the activities of the new Expert Group until 2015.

More information on the activities of the former European Union Committee of Experts on Rare Diseases and the EUCERD Joint Action can be found at www.eucerd.eu.

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ACRONYMS

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
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
GENERAL INTRODUCTION

This document 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 European Union Committee of Experts on Rare Diseases (EUCERD) was established in 2009 and its mandate ended in 2013. It is replaced from 2014 by the Commission Expert Group on Rare Diseases. The EUCERD Joint Action continues to support the activities of the new Expert Group until 2015.

The present 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 2013. 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 Commission Expert Group on Rare Diseases. The reader, however, should bear in mind that the information provided is not exhaustive and is not an official position of the European Commission, its Agencies or national health authorities.

The report is split into six parts:

Part I: Overview of rare disease activities in Europe
Part II: Key developments in the field of rare diseases in 2013
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
Part VI: Activities at National level in each EU Member State and other European countries in the field of rare diseases

Parts I – V also include a description of the methodology, sources and validation process of the entire report, and 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 Portugal. 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.

Each year, there are around 15 000 downloads of the different sections of the report combined.
RARE DISEASE ACTIVITIES IN PORTUGAL

Definition of a rare disease
Portugal accepts the definition of rare disease, as stated in the European Regulation on Orphan Medicinal Products, as a disease with a prevalence of no more than 5 in 10,000 inhabitants. This definition has been adopted by the National Plan for Rare Diseases.

National plan/strategy for rare diseases and related actions
In November 2008 the Portuguese Minister of Health approved the National Plan for Rare Diseases ("Programa Nacional para as Doenças Raras") coordinated, since November 2011, by the Department for Quality in Health at the Directorate-General of Health (DGS).

Its main objectives are the establishment and improvement of national measures, in order to satisfy the needs of people with rare diseases and their families vis-à-vis medical services and care, as well as the improvement of the quality and equity of healthcare provided to those people.

Such objectives will be achieved by establishing reference centres for rare diseases, by improving the access of patients to adequate care, by strengthening knowledge and awareness on rare diseases, by promoting innovation in the treatment of rare diseases and in the accessibility to orphan medicinal products, and finally, by ensuring cooperation at national and international levels, including EU countries and those countries having Portuguese as their official language.

This Plan was foreseen to cover all rare diseases, though it was hoped that it would be articulated with other priority national plans, namely with the National Plan for Oncologic Diseases ("Programa Nacional para as Doenças Oncológicas").

An important step in terms of implementing the National Plan for Rare Diseases took place in 2012: a specific card for the identification of people with rare diseases ("Cartão para a Pessoa com Doença Rara") was developed, aiming at disclosing clinical information to medical doctors and also in emergency situations. This card issued by DGS, is currently being distributed. From December 2013 to March 2014 199 cards have been issued.

DGS, together with the former Office of the High Commissioner for Health, co-funded from 2008 to 2011, for a total amount of €1.9 million, projects on rare diseases, which are still being developed by several patient organisations.

The National Programme for Rare Diseases, published by the Directorate-General of Health in 2008, has developed its activity focusing only on the services depending on the Ministry of Health, thus meaning, today, it must be replaced by a broader national strategy with integrated actions, both at inter-sectoral and inter-institutional level. A new Integrated Strategy for Rare Diseases 2014-2020 is currently being finalised, which replaces the previous Programme and aims to ensure that people with rare diseases have better quality of care, based on the evidence that science has been producing, as well as greater celerity and variety of social responses adapted to each case. This new strategy further aims to guarantee that, in an inter-ministerial, inter-sectoral, inter-institutional and integrated way, priorities in the global approach to rare diseases be refocused, bringing together the contributions of competences and resources of all relevant sectors, in order to cause, in a progressive way, a real change in the complex conditions of the people who suffer from these diseases.

Centres of expertise
Legislation is presently being finalised, in order to identify and officially recognise reference centres that might integrate future European Reference Networks.

Registries
The following commissions and/or registries operate under supervision of INSA: National Commission for the Portuguese Registry of Paramyloidosis, National Commission for Lysosomal Storage Diseases, National Registry of Congenital Anomalies (RENAC) and National Newborn Screening Commission.

RENAC data are available since 1996. The most recent report covers the 2008-2010 period. Another report, updating existing information, is being prepared. Two informative newsletters were sent to the hospital services that collaborate with RENAC; to the remaining services, letters were sent renewing the invitation for their participation in RENAC. The information collected on RENAC was also integrated in the report of EURO-

PERISTAT system. Moreover, RENAC participated in the European Surveillance of Congenital Anomalies (EUROCAT) by sending data related to the Southern region of Portugal; it further participated in the annual meeting of that European Register.

The use of Orpha codes, to code rare diseases in health information systems in Portugal, has been proposed to DGS to be used in the NHS, though it is not yet fully explored.

Upon their own initiative, many patients are also included in international registries. A few Portuguese institutions also participate, or have participated, in European registries, such as E-IMD, TREAT-NMD, EUROCARE CF, EUROCAT, EBAR, SCNIR, CHS, SPATAX, EUROWILSON and EHDN (the European Huntington Disease Network).

**Neonatal screening policy**

The National Programme for Early Diagnosis (“Programa Nacional de Diagnóstico Precoce”) started in 1979 at the former Institute of Medical Genetics and, initially, it only included the screening of phenylketonuria. INSA is nowadays responsible, at both organizational and laboratory levels, for the national newborn screening programme, which covers ca. 100 % of all children born in Portugal.

Currently, the following 26 diseases are screened: Congenital Hypothyroidism, Phenylketonuria (PKU)/Hyperphenylalaninemia, Tyrosinemia Type I, Tyrosinemia Type II/III, Maple Syrup Urine Disease (MSUD), Citrullinemia Type I, Argininosuccinic Aciduria, Argininemia, Classic Homocystinuria, Methionine Adenosyltransferase Deficiency (MAT deficiency), Propionic Aciduria (PA), Methylmalonic Aciduria type methylmalonic acidemia (MMA, Mut-), Isovaleric Aciduria (IVA), 3-methylcrotonyl-CoA Carboxylase Deficiency (3MCCD), Glutaric Aciduria Type I (GA I), Methylmalonic Aciduria type Cbl C/D (MMA, Cbl C/D), 3-hydroxy-3-methylglutaric Aciduria (3-HMG), Malonic Aciduria, Medium Chain AcylCoA Dehydrogenase Deficiency (MCAD), Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD), Long Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)/ Trifunctional Protein Deficiency (TFP), Short Chain Acyl-CoA Dehydrogenase Deficiency (SCHAD), Primary Carnitine Deficiency (CUD), Carnitine Palmitoyl Transferase I Deficiency (CPT I), Carnitine Palmitoyl Transferase II Deficiency (CPT II/CACT), Glutaric Aciduria Type II (MADD).

**Genetic testing**

Genetic testing is available for many rare disorders, though, as in other countries, there is a significant flow of genetic testing over the borders. Diagnostic tests are registered as available in Portugal for 685 genes and an estimated 793 diseases, in the Orphanet database.

Genetic tests are carried out in genetic laboratories within the National Health System (NHS), as is the case of INSA (considered the national reference laboratory), as well as in laboratories located or associated with genetic services in public hospitals, and also in private laboratories; besides these, a certain number of labs offer genetic testing at universities and research institutions.

Whenever a specific test is not available in Portugal, there is a formal procedure to perform it abroad. Orpha codes are being implemented on the request form, to ascertain what is done and where, and laboratories’ quality.

In 2013, the number of clinical cases sent abroad for referral amounted to 159, especially for molecular study and laboratorial genetic testing.

Genetic testing in Portugal is regulated mainly by Law no 12/2005, of 26 January 2005. This law defines, among others, the concept of health information and genetic information, the circulation of information and the intervention on human genome within the health system, as well as it establishes the procedure to collect and preserve biological products, for purposes of genetic testing or of research.

A proposal of a Decree that will regulate the aspects of this Law is under preparation. The same process is underway for the licencing of medical genetics laboratories.

As a member of OECD, Portugal is subject to the OECD Best Practice Guidelines for Molecular Genetic Testing, which are to be transposed into national law, through the Decree that will regulate Law 12/2005; Portugal also signed and ratified the Oviedo Convention, but not yet its Additional Protocol concerning Genetic Testing for Health Purposes (2008), currently at its final stage.

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2 Information extracted from the Orphanet database (January 2014).

3 http://www.insa.pt/sites/INSA/Portugues/AreasCientificas/Genetica/Paginas/LaboratorioDeReferencia.aspx
National alliances of patient organisations and patient representation

Portugal has two alliances on rare diseases: FEDRA – Portuguese Federation of Rare Diseases (“Federação Portuguesa de Doenças Raras”) and APADR – Portuguese Alliance of Rare Diseases Associations (“Aliança Portuguesa de Associações de Doenças Raras”).

APADR was officially established in 2009, and since then, has developed several actions and activities with the aim of improving Portuguese health policy in the field of rare diseases, and also raising awareness to this problem, namely TV and radio spots and a conference on the rare disease day.

Fedra published a series of booklets called “Genetic Diseases A to Z”, with articles prepared by expert professionals. It has organized photo exhibitions in the National and in the European Parliament, in order to raise awareness for Rare Diseases problems and for people living with rare diseases, as well as a conference with the participation of Eurodis, FEDER, “European Phd Programme on rare diseases”, Brains for Brain Foundation, European Commission, and the European Commissioner of Health.

Sources of information on rare diseases and national help lines

Orphanet activity in Portugal

The national team of Orphanet has kept available and updated in Portuguese all menus from the international site, all the diseases names, the emergency guides and summaries of diseases (validated by experts in each area). This is an important resource also for all countries of official Portuguese language and the Portuguese communities spread throughout the world (about 240 million Portuguese speaking persons). The team also maintained and kept updated the Orphanet-Portugal website page, and a Facebook page, which have included updated news about initiatives on rare diseases in the country and in Brazil.

By the end of 2013, 1065 abstracts of rare disease and 18 emergency guidelines have been translated and validated into Portuguese and entered in the international database; about 200 new abstracts were also translated, and are now waiting to be validated.

During 2013, Orphanet-Portugal continued to collect and validate and significantly increased the information available in the country on national resources and activities related to rare disease and orphan drugs. These included 145 specialised centres, 111 laboratories (33 diagnostic laboratories and 78 research laboratories), 1041 diagnostic tests, 153 research projects, 23 clinical trials, 70 patient organisations and 19 registries and biobanks, by December 2013; the list of the orphan drugs available in the country (currently 49, in a total of 88 different presentation and/or dosages) is regularly uptake from each one.

The Orphanet-Portugal team also contributed to the development (in English) and subsequent translation and validation into Portuguese of a leaflet on “Genetic Testing Related Health”, produced by the Council of Europe and prepared by EuroGentest (a network of excellence funded by the European Commission) and the PPPC (Public and Professional Policy Committee) of the ESHG (European Society of Human Genetics).

This brochure (in Portuguese) was released during the Rare Disease Day 2013 and is available from the Orphanet national webpage.

The national scientific advisory board of Orphanet-Portugal proved to be a valuable resource, providing important information as well as the validation of some data whenever requested.

In addition, as in previous years, the national team has been committed to the diffusion of the Orphanet portal and services to professionals involved in rare diseases, as well as to patients, families and the general public. This is achieved both through the organisation of specific events, and the participation in several scientific meetings and courses with oral or poster communications and lectures.

In 2013, 6 presentations about Orphanet were made at such meetings, one organised (together with Aliança) by Orphanet Portugal (the Rare Disease Day 2013), and one interview about “Orphanet in Portugal” and published in the magazine “Pontos de Vista”, distributed by a daily Journal.

All this information is intended to help improving and shortening the time before a diagnosis is obtained, and specialised care and treatment of patients with rare diseases is reached, as well as planning and improving the care for persons with rare diseases in the country.

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4 [http://www.fedra.pt](http://www.fedra.pt)
5 [http://www.orpha.net/consor/cgi-bin/index.php?lng=PT](http://www.orpha.net/consor/cgi-bin/index.php?lng=PT)
6 [http://www.orpha.net/national/PT-PT](http://www.orpha.net/national/PT-PT)
7 [https://www.facebook.com/Orphanet-PT](https://www.facebook.com/Orphanet-PT)
**Official information centre for rare diseases**

Orphanet is specifically mentioned several times in the National Plan for Rare Diseases, one of the declared aims of which is to “propose measures to support the development of Orphanet Portugal, making it the reference portal for information on rare diseases in Portuguese”\(^8\).

**Help line**

The patient organisation RARÍSSIMAS, with funding from DGS, implemented in 2009 a dedicated call centre, called ‘Rare Line’ (“Linha Rara”).\(^9\) It has been distinguished by Eurordis, for the second year, in the Caller Profile Analysis.

Several other patient associations, as the Portuguese Haemophilia Association, also give advice and support to patients and their families, either online, by email or by phone.

**Other sources of information on rare diseases**

The Orphanet-Portugal team also deals with many requests for information coming from the country and other Portuguese speaking countries, as well as many Portuguese patients and families living abroad.

**Guidelines**

DGS, in collaboration with the Portuguese Medical Association (“Ordem dos Médicos”) has been developing a number of clinical guidelines; among these, and in the specific area of rare diseases, three guidelines were issued in 2012, which refer, specifically, to the diagnosis and follow-up of Cystic Fibrosis and Pulmonary Hypertension.

**Training and education**

There is no formal programme in this area, but some medical schools have lectures on rare diseases and information resources (such as Orphanet), and much work is done in high schools and by patient organisations and health services. All national medical schools have curricular units on basic and/or medical genetics and, some, also on clinical genetics.

The medical specialty of Clinical Genetics was recognised in 1999, at the Portuguese Medical Association, (“Ordem dos Médicos”) and implies a 5-year full-time residency program.

A Professional Master’s Course in Genetic Counselling was initiated at the University of Porto, which is one of the five in the EU already accredited by the EBMG (European Board of Medical Genetics). This two-year post-graduation course is an innovative initiative in Portugal for professionals with diverse clinical backgrounds, including clinical psychologists, nurses and others. This full-time programme encompasses bioethics, clinical and genetic epidemiology, genetic counselling principles and techniques, clinical psychology, research methodologies and clinical rotations, as well as one-year training in a recognised medical genetics service and a research project, with special emphasis on rare diseases.

A professional association of genetic counsellors has already been created. Efforts are being made to recognise this new profession both at the national and the EU level.

The Portuguese Society of Human Genetics also approved in 2013 a commission for clinical genetics laboratory, to harmonise education criteria and program for laboratory geneticists; this program has already been accepted by the EBHG.

Two Genetics in Family Medicine courses were organised in 2013, at the Institute of Molecular and Cell Biology (IMCB), in Porto, part of a series of spring and autumn courses, directed mainly to general practitioners, but also other physicians and health professionals. IMCB also has a long-standing programme to receive high school students and teachers for presentations and guided visits to genetic services and research laboratories, all throughout the year.

Some patient associations also organise one-day receptions for medical students, so that they are made aware of rare diseases and rare disease patients.

Raríssimas submitted a project to EEA Grants funding for NGO’s (Programa Cidadania Ativa/Fundação Calouste Gulbenkian) in 2013 and has had the approval for its Marcos’s Centre – Information and training, to run from 2014 until 2015. Several programmes are being defined to address different training needs, including patients, families, health and education professionals, students and volunteers.

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\(^9\) [www.linharara.pt](http://www.linharara.pt)
2014 Report on the State of the Art of Rare Disease Activities in Portugal

National rare disease events in 2013
A conference was held to mark Rare Diseases Day on the theme of “Rare disorders without borders: National and European Realities” on 23 February 2013 in Porto, organised by Aliança Portuguesa de Associações das Doenças Raras (APADR) and Orphanet Portugal. A large number of stakeholders participated and the day ended with discussions on the status of the National Plan and centres of expertise. The Day was well covered in the national press and media.

Hosted rare disease events in 2013
Amongst the rare disease events hosted in Portugal and announced by OrphaNews Europe was the First International Primary Immunodeficiencies Congress (IPIC) (7-8 November 2013, Estoril).

Research activities and E-Rare partnership

National research activities
The Foundation for Science and Technology (“Fundação para a Ciência e Tecnologia” - FCT), a partner of Orphanet Portugal, runs several programmes to fund research on rare diseases. In 2012, FCT funded scientific projects in a total amount of € 583,000. FCT is recommending, after proposal of Orphanet Portugal, that “rare disease” and the respective Orpha code(s) are included as keywords in the application forms for projects, if applicable.

Participation in European research projects
Teams in Portugal have participated/participate in 16 rare disease related FP7 projects.

E-Rare
Portugal, represented by FCT and DGS, joined the E-Rare-2 Consortium in 2009. In December 2012, DGS was replaced by INSA in this Consortium. FCT participated in the 4th Joint Transnational Call launched in 2012, funding 2 Portuguese teams participating in 2 out of the 11 selected research projects, with a total budget of €341,581. In 2013 Portugal joined the 5th Joint Transnational Call but no Portuguese teams participated in the selected projects.

IRDiRC
Portuguese funding agencies have not yet committed funding to the IRDiRC, however through the E-Rare Group of Funder, Portugal was represented at the IRDiRC as of the end of 2012.

Orphan medicinal products
In Portugal, regulation of orphan medicinal products is the responsibility of INFARMED. A partnership was established in 2010 between INFARMED and ORPHANET-Portugal for a monthly update on all orphan medicinal products already approved and available in the country, and the volume of these actually used. Following legislation establishing the access of patients to therapies involving diseases with enzymatic deficit, INSA coordinates and further authorizes these pharmacological treatments. In 2012, the delivery of enzyme replacement therapy amounted to €47.5 million. A list of enzymatic diseases, which benefit from free of charge treatment in public hospitals, is also available.

Orphan medicinal product committee
There is no such Committee in Portugal.

Orphan medicinal product incentives
Presently INFARMED is not aware of the existence of any specific incentives.

Orphan medicinal product market availability situation
A list of all orphan medicinal products available and quantities consumed in Portugal is published at the ORPHANET-Portugal entry site. This data is provided and regularly updated by INFARMED.10

Orphan medicinal product pricing policy
Orphan medicinal product pricing policy falls under the responsibility of the Ministry of Health. There are no specific provisions in place to define the price of this kind of medicines.

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10 http://www.orpha.net/national/PT-PT/index/lista-de-medicamentos-orfãos-disponíveis-em-portugal/
**Orphan medicinal product reimbursement policy**

There are no specific provisions in place for the reimbursement of orphan medicinal products. All Portuguese citizens are covered by the National Health Service. The investment of the NHS in orphan medicinal products between January and December 2013 was around €75 million, which represents 7.7% of the total consumption of medicines in hospitals. It should be noted that there was an increase of 19.4% in this group of medicinal products, as compared to the same period of 2012. However, there are special programmes in place to facilitate access to growth hormone therapy, enzymatic therapy and familial amyloid polyneuropathy.

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

A Special Use Authorisation (SUA) procedure is in place to provide access to certain orphan medicinal products. If an orphan medicinal product is not marketed in Portugal, the treating hospital can request a special authorisation from INFARMED; if the use is approved, the hospital is directly supplied by the manufacturer and there is no co-payment from the patient.

**Other therapies for rare diseases**

There are no other initiatives for improving access to other therapies for rare diseases, though several national and multicentric international clinical trials are already under way or being prepared.

**Orphan devices**

Under the Directive for Medical Devices, there is no specific regulation for this kind of devices. Instead, there is the possibility of issuing an exceptional authorisation, for the specific utilisation of certain devices, for which the conformity assessment procedure, so as to obtain CE marking, has not yet been completed.

In addition, regarding diagnostic tests for rare diseases, these are usually considered as ‘in-house tests’ because they are manufactured and used only within the same health institution and on the premises of their manufacture, or used on premises in the immediate vicinity, without having been transferred to another legal entity.

**Specialised social services**

Respite care services exist in the public, private and social sectors, and patients must pay for some services. Other respite facilities are run by patient organisations and some projects have been established with public support.

Generally, all patients with rare disease, and depending on their level of functional ability, have access to the same benefits, as any other citizen in the same situation of dependency. **RARISIMAS** has two multidisciplinary centres, which provide clinical care and therapies to patients and families with rare diseases, one in Oporto and the second in the Azores islands.

Besides these centres, Raríssimas has now opened Casa dos Marcos, the first Resource Centre for Rare Diseases in Portugal, gathering social and healthcare services and planning to respond in the educational area as well. Casa dos Marcos has both residential services and ambulatory care and is establishing several partnerships, nationally and internationally, developing innovative projects in various domains. In fact, it has a unique model of assistance with a mix offer that includes services under contract with the State (a long-term care unit, a residential unit, an occupational activity centre and an autonomous residential unit) and private services (respite centre; rehabilitation centre; medical and non-medical consultations). It also includes an information and training centre and a research centre on rare diseases. Holiday camps for patients are also run. Casa dos Marcos costs about €10 million (global investment) and Raríssimas raised about €8 million from private companies.

There are some therapeutic recreational initiatives organised by hospitals with the support of public or private organisations, which are paid through public and private funding; many programmes are organised by several patient organisations, such as the Portuguese Association for Paramyloidosis. There are some projects to help the integration of patients in daily life, and this offer will hopefully improve under the National Plan for Rare Diseases. Some other patient associations organise respite camps.

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12 EMINET – Initial investigation to access the feasibility of a coordinated system to access orphan medicines, C. Habl, F. Bachner (2011), p 57
DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2013 IN PORTUGAL

National plan/strategy for rare diseases and related actions
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13  http://www.orpha.net/consor/cgi-bin/index.php?lng=PT
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Orphan medicinal products
Orphan medicinal product reimbursement policy
There are no specific provisions in place for the reimbursement of orphan medicinal products. All Portuguese citizens are covered by the National Health Service. The investment of the NHS in orphan medicinal products between January and December 2013 was around €75 million, which represents 7.7 % of the total consumption of medicines in hospitals. It should be noted that there was an increase of 19.4 % in this group of medicinal products, as compared to the same period of 2012.14

Specialised social services
Raríssimas has now opened Casa dos Marcos, the first Resource Centre for Rare Diseases in Portugal, gathering social and healthcare services and planning to respond in the educational area as well. Casa dos Marcos has both residential services and ambulatory care and is establishing several partnerships, nationally and internationally, developing innovative projects in various domains. In fact, it has a unique model of assistance with a mix offer that includes services under contract with the State (a long-term care unit, a residential unit, an occupational activity centre and an autonomous residential unit) and private services (respite centre; rehabilitation centre; medical and non-medical consultations). It also includes an information and training centre and a research centre on rare diseases. Holiday camps for patients are also run. Casa dos Marcos costs about €10 million (global investment) and Raríssimas raised about €8 million from private companies.

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15 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.
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- FEDRA - Portuguese Rare Disease Alliance
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  [http://aliancadoencasraras.org](http://aliancadoencasraras.org)
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16 All websites and documents were last accessed in May 2014.